When Christian Faith and Genetics Meet

A Practical Group Resource

The Canadian Council of Churches
Le Conseil Canadien des Églises

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The Canadian Council of Churches

Founded in 1944, The Canadian Council of Churches is the largest ecumenical body in Canada, now representing 24 churches of Anglican, Evangelical, Free Church, Eastern Orthodox and Oriental Orthodox, Protestant, and Catholic traditions. We are one of the few ecumenical bodies in the world that includes such a range of Christian churches. The officers and staff of the Council are drawn from the whole diversity of traditions represented by the member churches.

Member churches believe in the Lord Jesus Christ as God and Saviour, according to the Scriptures. Members seek to fulfill together their common calling to the glory of one God: Father, Son, and Holy Spirit.

Biotechnology Reference Group

The Biotechnology Reference Group (BRG) was established by the Governing Board of The Canadian Council of Churches in 1999 as a reference group, a clearinghouse for the gathering and exchange of information on biotechnology. Both the Commission on Faith and Witness and the Commission of Justice and Peace are represented on the Biotechnology Reference Group and see the work of the Biotechnology Reference Group as their own.

Purposes of the Biotechnology Reference Group

- To gather information from churches, sister ecumenical organizations, and prominent research institutes, engaging existing expertise.
- To share the information broadly.
- To minimize duplication of work by churches, sister ecumenical organizations, and other related organizations.
- To encourage a forum for theological and ethical reflections.
- To assist in the churches' learning on these issues, for example, by providing advice on helpful research and other written sources.
- To bring together representatives from the churches who are working on these issues.

Acknowledgements

This curriculum first came to the attention of the Biotechnology Reference Group (BRG) of the Canadian Council of Churches (CCC) in December 2007 at the *Global Consultation on Genetics and New Biotechnologies and the Ministry of the Church* in Johannesburg, South Africa. This conference brought together representatives from churches and church councils around the world to discuss pressing ethical, social, and theological issues brought about by new genetic technologies. BRG member Jim Rusthoven struck up a discussion with a representative of the National Council of Churches in the United States, David Lesley. As director of Oregon Ecumenical Ministries (OEM), David was showcasing a curriculum on genetic technologies that had recently been piloted in multiple denominations represented in OEM. The curriculum began as the idea of Marc Marenco and Lisa Sardinia, professors of philosophy and biology, respectively, at Pacific University, Forest Grove, Oregon. They felt that there was a need to better educate those in the body of Christ in the new genetic technologies that are beginning to affect an increasing number of lives. Moral, theological, and social issues have arisen that trouble many Christians and non-Christians alike. The need for such a curriculum came originally from local pastors who were being asked by their parishioners to give advice regarding the ethical and theological issues raised by new biotechnologies. Over time, Marc and Lisa focused the design of the curriculum for use by parishioners themselves.

Marc and Lisa were able to secure resources from Pacific University and from the National Institutes of Health to craft a first version of this curriculum. This was accomplished with the help of various external consultants including Allen Verhey, Audrey Chapman, Ted Peters, and Ronald Green. They recruited David and OEM to approach their churches about participating in pilot testing the curriculum. Formal feedback was obtained by the time the curriculum was being displayed at the Johannesburg meeting. David kindly allowed Jim to return to Canada with a hard copy of the curriculum and the BRG discussed its potential merits in the Canadian ecumenical setting. After discussions with Marc Marenco, including a personal visit to a BRG meeting, it became clear that, so long as no commercial profit was foreseen, the BRG could use the initial version of the curriculum to move forward with its own revisions and pilot phase without engaging in any copyright infringements. The BRG agreed to fully acknowledge the prior work of the American group and its sponsors in creating the initial American version from which the final Canadian version could be developed. The Canadian version includes various content updates in the science section and changes in all sections that make the curriculum current and suitable for the Canadian community of Christian believers.

In recognition of their pioneering efforts, we would like to sincerely thank Marc, Lisa, the consultants for the American version, Pacific University, and the National Institutes of Health for their contributions to the curriculum in its initial form. The Canadian version has been crafted by members of the BRG who have dedicated many hours of volunteer time and talent in providing an educational resource for the churches of Canada. It is a resource readily available on the CCC website and includes a helpful guide for facilitators to organize and implement it in any congregation. We would also like to thank the Reid Trust and the Canadian Conference of Catholic Bishops for their generous financial contributions to the project, without which the resources that ensured the success of this project could not have been secured.

The current Canadian version was assembled through many hours of cooperative work of BRG members, external consultants, and volunteer facilitators. Members of the BRG, CCC staff, and interested additional contributors who constituted the writing group and shepherded the Canadian version through several critical stages include Moira McQueen, Mary Marrocco, George Tattrie, Isaac Kawuki Mukasa, Mark Boulos, Jaya Kirstmeyer, Anne Mitchell, and Jim Rusthoven. Expert and timely administrative support was provided by Bambi Rutledge of the Canadian Catholic Bioethics Institute and Mary Delph, administrative assistant for the Canadian Council of Churches. During the pilot phase of the project, volunteer facilitators led small groups within church groups of specific denominations and gave invaluable feedback of their experience in that phase of development. These facilitators and their participating church groups included BRG member Moira McQueen (Our Lady of Lourdes Roman Catholic Church, Toronto), Gus Pappas (St Silouan the Athonite Orthodox Church, Toronto), Jason Taekema (Ancaster Christian Reformed Church, Ancaster) and Jacob Plantinga (Ancaster Fellowship Christian Reformed Church, Ancaster), Jaya Kirstmeyer and Anne Mitchell (8 – 12 members of the Toronto Monthly Meeting Friends (Quakers)), and John Wilton (St. Augustine of Canterbury Anglican Church, Toronto). Thanks also to Katarina Prosenjak, German pastor and intern with the CCC who provided assistance during the pilot phase and valuable commentary on post-pilot phase version of the curriculum.

Following the pilot phase, the curriculum was revised according to the valuable input of facilitators and their respective church groups. Others were brought in to aid in providing objective critique and helpful suggestions for improvement. External consultants Gus Pappas, Minya Milanovic, and Jason Taekema provided the necessary arms-length advice during various phases of the project. Gus, Minya, and Jason provided invaluable critique and suggestions for improving the science section. Our thanks especially to editor Patrick Gallagher, who took the curriculum from final draft to finished manuscript by overseeing the final editing, adding sections such as the Letter

to Facilitators, ensuring the educational usefulness of the curriculum, and offering valuable and timely insights into the whole. Special mention goes to BRG member Stephen Allen whose well-crafted introduction sensitively sets the Canadian context and tone of the curriculum. We would also like to thank CCC staff member Peter Noteboom for his steady support of the organizational processes by which this project could incrementally proceed over time and in helping to update the Governing Board of the CCC with respect to the progress of the curriculum. Finally, we would like to give thanks to translator Louis Remillard for his French translation of the curriculum and to Thomas Hentrich of CCC for his proofreading of the French translation. None of these many contributions would have been possible without the unwavering support of the CCC Governing Board, its President during most of this project, Rev. Bruce Adema, and its General Secretary, The Rev. Dr. Karen Hamilton.

The BRG and the CCC see this curriculum as God's work, done through the gifts given to his servants within the body of Christ, his Church. As an ecumenical effort, the challenges of historical, ecclesiastical, and theological diversity have been met with an effort to incorporate the strengths of each denomination into the content of this curriculum. We pray that the small gatherings that use this curriculum will be blessed by the power of the Holy Spirit to find collective wisdom in responding to the ongoing challenges confronting church members individually, congregations, denominations, and the Church at large.

James Rusthoven, MD, PhD

Genetics Curriculum Writing Group Member
Former Chair, Biotechnology Reference Group, The Canadian Council of Churches

Contents

Preface

Preface

This curriculum has been prepared by committed and thoughtful colleagues in the Canadian Council of Churches' Biotechnology Reference Group (BRG). Individuals in the BRG represent their respective denominations and bring a variety of skills and gifts as scientists, health care professionals, theologians, and ethicists.

The various applications of biotechnology, including crops and food, genetics, molecular biology, nanotechnology, synthetic biology to name a few, have global dimensions. Canada is a significant player in biotechnology and much of the research and development in Canada is publicly funded. In addition, the federal government and a number of provincial governments offer generous tax incentives to encourage the development of biotechnology companies.

Canada – A Major Player in Biotechnology

Here are some examples of recent developments in Canada.

- An international team of scientists from Canada, China, Japan, the U.K. and the U.S. has been collaborating since 2002 on what is known as the HapMap Project. Research published in 2007 allows scientists to detect minute fractions of genetic materials that vary among individuals – these variations could explain the differences in disease susceptibility and drug response. As the lead Canadian scientist, based at McGill University explained: This (HapMap) is really a map to study the genetics of common diseases. (www.genomecanada.ca). As a result of this research, scientists have identified the genes involved in Type 2 diabetes and colon cancer.

- AquaBounty Technology, a company established as a result of research at Memorial University in St. John's, has genetically modified a salmon that grows twice as fast as its wild counterpart. The corporation is seeking approval by the U.S. Federal Drug Administration to market its AquaAdvantage salmon. The company is in the early stages of seeking approval by the Canadian Food Inspection Agency. ("A drug with gills? U.S. agency reshapes debate on biotech fish" by Jessica Leeder, *The Globe and Mail*, Sept. 4, 2010).

- The so-called "Enviropig™" could soon be the first genetically modified (GM) (also called genetically engineered or GE) food animal on the market. Enviropig™ is the trademarked industry name for a pig that has been genetically engineered to excrete less phosphorous in its feces. Enviropig™ was developed by researchers at the University of Guelph in Ontario.

Sixteen Canadian universities are part of a network of more than 100 teaching hospitals and research institutions involved in biotechnology and its application to human health. Departments in a number of universities carry out research in biotechnology and agriculture. Over 530 Canadian biotechnology companies are involved in research and development in a number of areas including: human health, agriculture and food processing, and the environment. This represents the second highest number of such companies in the world; of these, 58% are involved in human health and 24% in agriculture and food processing. (Industry Canada Life Sciences Gateway – Canada's Biotechnology Industry see www.ic.gc.ca. See also BioCanada www.biotech.gc.ca). Most of us learn about major breakthroughs through the media. The use of a vocabulary and of concepts that are not familiar to us can be frustrating. More importantly, the lack of understanding of genetics may make it difficult for many of us to make appropriate choices for our own health or that of a loved one when these technologies become available.

Federal Government Involvement in Biotechnology

In 2007, the federal government launched Science and Technology Strategy – Mobilizing Science and Technology for Canada's Advantage. This set out the government's priorities and is intended to "improve the quality of life of Canadians and strengthen the economy." (See Industry Canada, "Mobilizing Science and Technology to Canada's Advantage: Progress Report 2009," www.ic.gc.ca) The federal government views biotechnology as an important platform in the economy of the 21st century. This is consistent with previous governments. The lead government ministry responsible for biotechnology is Industry Canada. It's the hub - other government departments are the spokes.

The Federal Government's Role in Public Oversight

Ultimate oversight rests with Parliament, which has the responsibility to provide the mandate and the regulatory framework to ensure that the laws and regulations governing biotechnologies are followed. This includes approving new drugs and other products. A number of government departments, such Agriculture Canada, Environment Canada, Health Canada and the Department of Justice, have specific oversight responsibilities. There are a number of Standing Committees in the House of Commons and the Senate that might conceivably deal with biotechnology. For example, the House of Commons Standing Committee on Health tabled a report on November 23, 2010 that included a number of recommendations, one of which directed Health Canada to develop a program to ensure that Canadians have the appropriate information to make informed decisions about the safety and efficacy of stem cell treatments, especially

those not available in Canada or in countries where there is strong regulatory oversight. (Standing Committee on Health – Ninth Report – November 23, 2010. www2.parl.gc.ca/HousePublications/Publication).

Standing Committees provide an important access point for organizations and citizens to participate in discussions about biotechnology and public policy. The Canadian Biotechnology Advisory Committee (CBAC) was created by the Liberal Government in September, 1999, to advise the government of the day and to engage Canadians regarding the development and regulation of biotechnologies for the benefit and protection of the public. CBAC had its sceptics who felt that CBAC accepted biotechnology too uncritically, but at least it sought to be consultative. The Conservative Government closed CBAC in 2007 and replaced it with an advisory body of scientists that reports to Industry Canada.

What do Canadians Think?

Several years ago, the Department of Justice commissioned a survey to learn about citizens' expectations of genetic privacy. One important finding was that Canadians expect government to have laws and policies in place to protect the privacy of individuals' genetic information. ("Genetic Information and Privacy", Valerie Howe, Senior Research Officer, JustResearch No. 10, Department of Justice, www.justice.gc.ca)

There is a paucity of recent surveys of Canadians' attitudes on biotechnology. This doesn't necessarily mean that Canadians don't care or don't think about biotechnology and what impact it might have on their lives. We tend to be more attuned to biotechnology when there is an issue that grabs our attention – like Dolly the sheep or an announcement about a cure for a disease thought to be incurable.

Unfortunately, there are currently few opportunities for citizens to participate in public conversations about biotechnologies that are changing the way we live. Some research institutes provide opportunities for the public to attend lectures and to learn about the issues through their web sites. This is commendable but inadequate to meet the need to learn more so that all citizens have a better understanding of how the commercialization of biotechnologies will affect their lives. (See Ontario Genomics Institute www. ontariogenomics.ca and Genome Canada www.genome.canada)

The Church: A Place for Moral Discernment

The Church is one community that has a mandate (as taught in Scripture) to think deeply about issues that touch our lives. For those of us who are Christian (as with society in general), keeping up with the many developments in biotechnology is impossible. This risks uncritically welcoming new developments in genetic technologies. On the other hand, we should not view every new development with suspicion or reject a new development without thoughtful reflection. We need tools to critically assess the opportunities and the risks of biotechnology. This curriculum is intended to be such a tool to assist us in learning about and grappling with genetic technologies that are changing our lives. You will find words like DNA, gene, genome, nanotechnology and synthetic biology in this curriculum, words that you won't find in the Holy Bible.

But in the Holy Bible, you will find words like compassion, creation, God, hope, humility, Jesus, Holy Spirit, love, mystery, prudence, sacrifice, suffering and wisdom. What better foundation as we discuss and discern the theological and ethical implications of biotechnology and its many applications in the 21st century!

Stephen Allen

The Presbyterian Church in Canada

Shared Statement

Shared Statement

As Christian churches, we need to address questions raised by medicine and science, including the relatively new, and constantly developing, field of genetic science. The Church has a responsibility, through God's call, to support life and oppose all that harms life, protecting all of creation. Through Christ, we know God incarnate in the world, and we know that being human involves both body and soul; all this is under our care. This responsibility means we need to listen to science and come to understand it, including genetic technologies as they emerge. We also need to speak to science, for these technologies affect life itself, the life of all. We live in a broken world, where genetic sciences can be used to do either harm or good. In this conversation between faith and science, raised by genetic technologies, we acknowledge our limitations and approach our quest to understand the mysteries of creation with humility. Both our knowledge and our limitation bring us closer to God, reminding us of God's loving power.

We have approached this conversation not independently, as separate denominations, but working together out of our various Christian traditions. What unites us is greater than what divides us. We can do much work together, and can say much together, in our quest to understand the relationship between faith and science. When we speak together authentically, the Church's voice is strengthened. This common voice, this fruit of our shared work and wisdom, reminds the wider society that churches cooperate and work together for the common good. At the same time, we come to understand each other better, and this we need to do.

Genetic technologies affect us all. We need careful communal reflection to meet the challenges of judging the good or harm made possible by such technologies. It is valuable to hear the interpretive wisdom of each Christian tradition. Our diverse voices and our shared voice can enrich the scientific conversation. It is beneficial to bring together the Body of Christ, of which all churches are part. By doing so, we help give

some answer to our longing for full Christian unity, echoing Jesus' prayer that all be one (John 17).

We are created in the image and likeness of God. Yet we fall short of God's original intention for us, and harm ourselves, and the world that God has given us to look after. We understand that the pursuit of knowledge in genetic science and other biotechnologies may yield many benefits for us to the glory of God, if we allow the power of the Holy Spirit to guide us in discerning the right paths to follow. But we also know that such knowledge may be used in evil ways, to abuse the weak and vulnerable and to destroy God's creation. As the Body of Christ, we must heed God's calling to protect God's creation and uphold the equal value of all human life. We believe that in the death, resurrection and ascension of Jesus Christ, good ultimately triumphs over evil.

Together in this faith and understanding, we present the following curriculum on Faith and Genetics as a guide to addressing theological and ethical concerns raised by genetic technologies.

Letter to Facilitators

Letter to Facilitators

Dear Facilitator:

On behalf of the Canadian Council of Churches, we would like to thank you for taking on the role of facilitator for this "Faith and Genetics" curriculum. You are doing important work and we are grateful for your generosity.

We have prepared this resource to help you, but you should feel free to make it your own. Each group that uses it will have different interests and concerns and, as the group's facilitator, you should respond to the needs of the people in your group and adapt this resource to suit them.

Here is some information we think you will find helpful.

Organization of the Resource

The program consists of five different sessions:

1. Genetics, Faith, and Human Dignity
2. Genetic Technologies, Information, and Personal Identity
3. Genetic Technologies and Research on Human and non-Human Subjects
4. Genetic Technologies and the Engineering of Future Generations
5. Genetic Technologies and Social Justice

Each session follows the same simple structure.

Introduction

The introduction to each session is a brief orientation intended to set the stage for what follows. This sometimes includes background information and questions for consideration about the scenarios to be presented.

Question to Think About

After the Introduction, a question is presented that is meant to provide focus and inspire thought and reflection at the beginning of the session as well as highlight a foundational principle that is meant to help ground the discussion.

Narrative

Each scenario includes a narrative that provides a human context for approaching and considering the ethical and scientific issues that the session focuses on. Each narrative allows the group to explore a different facet of the session's topic. Usually the reader is given a role to play in these narratives.

Discussion Questions

After the narrative, a number of discussion questions are suggested to help guide and direct the responses and ideas of the group to the issue they are considering.

Neither you nor anyone else is expected to have a scientific background or expertise. For that reason, the Resource also offers additional material as a help to you and the other participants: several relevant background articles on genetics and a glossary of terms.

Genetics Background Information

Seven brief genetics background information sections have been prepared to help the participants understand some of the science involved in the session issues.

Genetics 101: The Molecular Basis and Implications of Genetic Variation

Behavioural Genetics

Inherited Disease and Genetic Testing

Transgenics

Embryonic Development and Genetic Engineering

Population Genetics

Genetics for Guiding Therapy

References to this science information are included at the point in particular sessions where they are most relevant, but participants will benefit from reading these sections ahead of time on their own.

Glossary

The resource includes a glossary of science and genetic terms for quick reference.

Time Lines

For most people who have used the resource, a session lasts for about two hours, including time for a break. This might seem like a lot, but the time will pass quickly. The sessions raise important, difficult, and often complicated issues. Everyone who participates is interested in them, but some people will have a strong personal interest in a particular topic or have strong opinions about what is raised. Here are some suggestions to help everyone make the best use of the time.

- Encourage people to read the material for each session and especially the science material before the session begins. If people are familiar with the material and prepared with questions and discussion ideas ahead of time, everyone will find that the time they spend together will be very fruitful.

- Each session is full of ideas and issues, far more than you can cover in one meeting. Before the first session, try to find out what your group is especially interested in and choose the narratives and the discussion questions that explore those issues. Feel free to adapt and modify them to match the needs of your group or to take into account something current in the news. (For example, you may wish to discuss at the beginning of each session whether the duration of the session should be fixed, given a flexible end time with extra time available if further discussion is needed, or even whether to schedule an additional session so the discussion can continue later.)

Participants

You will find among the people who come for the sessions those who have a deeply personal involvement in an issue and those who have an intellectual curiosity; those who have at least some scientific expertise and those who know little science except for the realization that something important is happening; those with a strong theological grounding and those whose religious connection is less secure. In short, each group will be unique because each person in it will be unique.

It's important that you make everyone feel that they are personally welcome and that their contribution is welcome, too. Here are some suggestions.

- At the first session, make sure participants are invited to introduce themselves and say what they are looking for from the sessions. This allows each person to feel part of the process from the beginning and also allows you to make sure that particular interests or concerns are included. If nametags are available, they may be helpful for the first couple of sessions.

- Make sure there is a break. Not only does this give everyone a chance to relax but it also creates an opportunity for the kinds of unscheduled and unexpected conversations that often are the most important part of a meeting. If possible some kind of food or refreshment would be a wonderful addition.

- The discussions will be greatly enriched if you encourage those who have prior or specialized knowledge about a topic or issue to share that knowledge with the group. Be careful, though, that those with specialized knowledge of genetic science, ethics, or theology do not dominate the discussion.

- Be sure that everyone is invited to speak. Some people thrive in group situations; others find speaking in groups difficult. But all participants should have an opportunity to ask a question or say what they think.

Role of Faith Communities

These scientific developments are so dramatic and their social, political, personal, and religious implications so profound that the Canadian Council of Churches believed it was important that its members work together on this curriculum. We hope that by sharing our expertise, our experience, and our prayers, we will be helpful to all Christian people and all people of good will.

We want anyone who takes part in these sessions to feel that not only have they personally gained from the experience but that they have also grown as members of their faith community. If the question, "How do we, as a Christian community, understand and respond to these issues?" is one they feel they have at least begun to answer, then one of the goals in offering this curriculum will have been met.

- An important component of this Resource is the appendices that the individual Christian communities have prepared for their members. These reflect the particular responses of individual churches to some of these

difficult and challenging issues and can offer helpful guidance to participants, who are strongly encouraged to read and make them part of the sessions. Participants are also invited to connect with the national office of their church community or with a Biotech Reference Group member of their denomination who helped develop this Resource.

- Make sure that your pastoral team is a partner when offering this curriculum. Keep its members informed and draw on their experience and strengths to help you in your role as facilitator. For example, you can expect that some of these issues will have immediately and directly touched some participants. Some may have used IVF; some may have had to make genetics-based life decisions. Be prepared for this yourself and if something comes up in a session, be ready and willing to speak privately to the person. You can also invite a pastoral team member to get involved.

Success

This is not a school program with a test at the end and marks that measure levels of accomplishment. The curriculum will be a success if all the participants, including you, feel that they know more than when they started; that they have shared time with others in a respectful and supportive way; that they can afterwards participate more fully, both as individual citizens and as members of a Christian community, in some of the major ethical and religious issues of the day.

Thank you again for your generosity. We hope you find the experience of leading these sessions to be enriching, educational, and rewarding.

Yours sincerely,

Biotechnology Reference Group

Session One: Genetics, Faith, and Human Dignity

Introduction

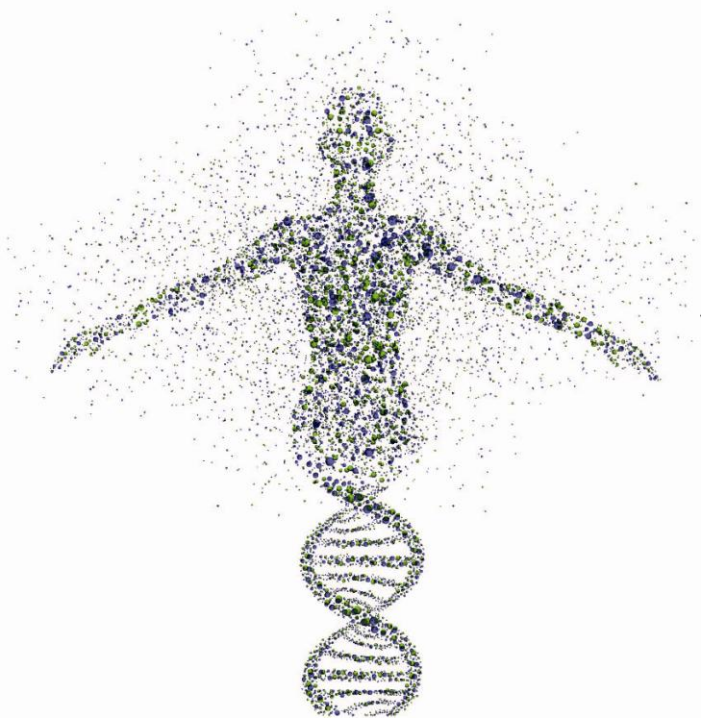
The focus of our first session is on faith: faith and science, faith and genetic technologies, faith and human dignity. This may not be an easy discussion, but it is certainly an important one.

Let's begin by considering this comment by sociologist Alex Mauron:

"It is claimed that our genome is important in a way that everything else isn't. The genome is construed as the ontological hard core of our being, the main determinant of our individual and species characteristics, the necessary and sufficient cause that makes us. The genome has practically become the secular equivalent of the soul."

This claim states that our genetic material is the primary determinant of who and what we are.

- How do we respond to such a claim?
- How does such a claim affect our self-understanding?
- Are we predetermined to be who we are because of our genetic makeup?
- Does genetic science or science in general say all there is to say about being human?



Scenario 1

Question to Think About

How do the values of our faith affect our ethical thinking and decision making about genetic procedures?

What, for you, is faith? Does it involve a radical acceptance of everything in your life, trusting that God will sustain you and be present to you in it? Or does your faith involve a radical trust that God will sustain you and be present to you if you act to *alter* the circumstances of your life in the light of what you believe to be true for you and to be in conformity to God's will for you? This first question is meant to help you articulate your understanding of faith and how your faith might make a difference in your thinking about genetic science and technology.

Narrative: An Invitation to Make Church Policy

Karen has been an active member of her church for most of her forty-five years. As someone knowledgeable about the science involved in genetic research, she has been nominated to be on a church policy review and development committee dealing with genetics, theology, and the Church. Her assignment is to write an essay showing how developments in genetic research affirm the faith of the Church and are in conformity with God's will.

Discussion Questions

If this assignment were given to you, what would you say? Does faith make a difference in approaching the legal, ethical, theological, and policy questions raised by genetic science? Could scientific discovery pose a threat to Christian faith? Could it pose a threat to your faith?

Scenario 2

Question to Think About

Does the science of genetics contribute to or contradict our belief that we are created in the image of God?

This question comes out of a very old dialogue between faith and science. There is a sense in which the questions raised by the concept of "genetic inheritance" are the same as those raised by Copernicus when he claimed that the Earth revolves around the sun or by Darwin when he claimed that we have evolved as a species from other species. What does it mean when we say that as created human beings we bear the marks of God? As we learn more about biology, neuroscience and genetics, is there room for thinking of humans as beings having the kind of freedom, dignity, and stature supported by religious communities and expressed in declarations of human values and human rights?

Narrative: The Book of Genesis and the Origins of Humankind

For years you have been an active member of your church and have participated fully in the communal and prayer life of your congregation. Recently, your growing knowledge of genetic science is leading you to use the lens of genetics to think about the teachings of your church regarding the origins of humankind. Within the context of your faith community you want to engage in this kind of reflection honestly and productively. Two questions stand out for you.

- How does the confession that we are created in the image and likeness of God connect with discoveries in genetic science that associate certain genetic markers with certain behavioural traits?
- How can we reconcile our belief in human freedom and free will with the constraints and limits indicated by our genetic profiles?

Discussion Questions

1) Does genetic science, as you understand it, pose any real threat to the belief that we are created in the image of God? Do faith and science describe two very different and incompatible ways of understanding our origins? Does genetics add something to the older debates about faith and reason, revealed and natural theology, religion and science? (You may find it helpful to read Behavioural Genetics: Genes and their Environment, to get a better understanding of the link

between genetics and behaviour characteristics that we may acquire through the expression of our genes.)

2) It may be that some genetic alleles invariably lead to particular outcomes. For example, if you have the allele for Huntington's disease, it is widely believed that you will get Huntington's disease. Other alleles are more complex. They do not create certainties but probabilities. Women with the BRCA1 or BRCA2 allele have an increased risk for breast cancer. Most of the alleles associated with behavioural traits are of this type. Does this mean that no matter what the behaviourally relevant allele is, if we resist the behavioural tendency we can preserve our free will and dignity as a human being? (You may want to review the remainder of the science section entitled Behavioural Genetics, particularly the subsection Genetic Variation and the Environment: Complex Interactions, where genetically linked disorders are discussed, including Huntington's disease, cystic fibrosis, and sickle cell anemia.)

3) Does the fact that most conceptions do not lead to live births influence your thinking about human life as a creation of God?

4) Does a basic understanding of genetic science make it difficult to maintain a traditional faith?

Scenario 3

Question to Think About

What ethical questions arise from the use of genetics in reproductive technologies and what are some of the theological responses?

In discussions about the use of genetics in reproductive technologies the phrase "playing God" frequently occurs. This term reflects the opinion that we are not meant to do certain things, even though we can. For example, scientists have recently reconstructed the genetic makeup of a primitive single-cell organism, using the minimum required number of genes to allow the cell to replicate and function. Is this "playing God" or is it an acceptable way of discovering new types of living organisms that might help people or the environment? This question makes us realize that we must think about the kind of world we want to live in and use our knowledge in a modest and resourceful way toward building that world. With every increase in our knowledge combined with our increasing ability to use that knowledge comes an increase in moral responsibility.

Review Genetics 101. This section contains a brief history of the discovery of genes and their fundamental -molecular backbone, deoxyribonucleic acid or DNA. It also gives some general structural information about the location and structure of genes in each cell of the body and familiarizes you with some basic terms used in genetics to distinguish the genes themselves and the characteristics associated with their functioning in the body. This section also will familiarize you with the impact of genetic variation on body functioning, using the example of genetic determination of various blood types. This is particularly important when someone needs a blood transfusion. In the glossary you will also find important terms.

Narrative: Disability and Genetic Counseling

Ten years in the future, Jim and Joan are seated at a table with a genetic counselor. Their daughter, Sarah, is in a stroller beside them. They conceived Sarah using a method once considered normal – through sexual intercourse. Sarah was born deaf. Her parents are moderately well off and so they could have had testing to find out their carrier status and could have had prenatal testing to find out if Sarah had a genetic disorder. They belong to a community of faith and sincerely believe that tinkering with reproduction in this way is a moral affront to God. They love Sarah and cannot imagine life without her, but feel terribly guilty about her suffering, suffering that they could have avoided if they had not played a kind of

genetic Russian roulette by conceiving via sexual intercourse. They want another child but do not want to risk having a child with a preventable genetic disorder. They are not sure they've made the right choice in asking for this meeting and avoid looking at each other as they wait to begin their interview.

The genetic counselor is knowledgeable and sympathetic. He tells Jim and Joan that they have available three of twelve embryos that are free of all known major genetic diseases. They have done genetic profiles on the three and Jim and Joan can choose from among them. Embryo #1 is a male. While normal in most ways, the embryo does have one of the alleles now associated with addictive behaviour. Embryo #2 is female. She has an allele that will produce insufficient human growth hormone (HGH) and is likely to be normal in all other ways, but will never grow beyond five feet, perhaps no taller than 4' 9." Embryo #3 is also female. Her profile also indicates a predisposition to addictive behaviour.

You may find it helpful to review Embryonic Development and Genetic Engineering. This section will give you a basic understanding of what an embryo is, how embryos are being used in stem cell research, and how they could be used in cloning animals and, potentially, human beings.

The counselor tells Jim and Joan that they can genetically modify the embryo with the HGH defect. Jim and Joan did not anticipate these kinds of choices and become increasingly uncomfortable as the counselor speaks. Finally, as the counselor talks about genetic modifications to create a child with a so-called normal height, Joan breaks down in tears and runs from the room. Jim follows.

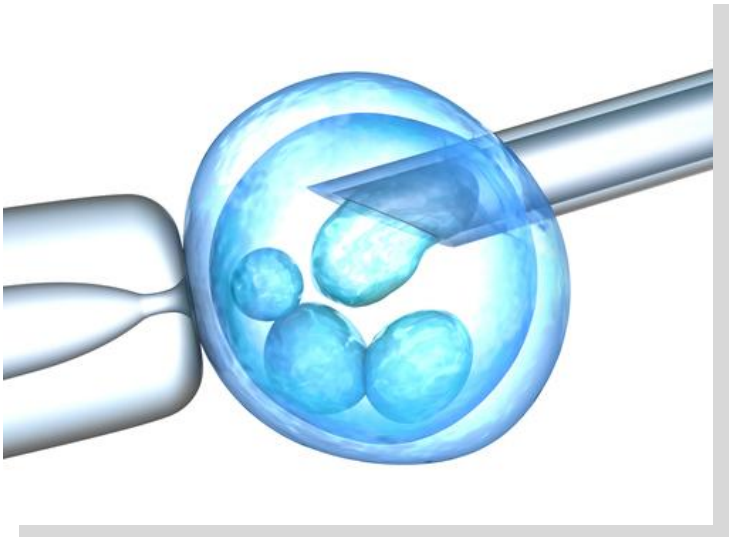
Discussion Questions

1) Jim and Joan feel that trying to manipulate the reproductive process in the way the genetic counselor suggests is like playing God. They believe that selecting more desirable embryos or modifying existing embryos is interfering with the act of creation itself and is both dangerous and morally wrong. What do you think?

2) Jim and Joan love each other. They want their love to produce a child. Intercourse for the purpose of creating a child with all the longings, hopes, fears, and mystery that go with the act runs very deep in them. What does being a person of faith mean in the context of human reproduction?

3) Some would say that this scenario is an affront to Sarah and all people with disabilities and genetic disorders. Is the message to Sarah: "If we could have done so, we would have not had you?" Is the message to the disabled: "If we could, we would live in a world without you as you are?" What do you think? Is prenatal and pre-implantation genetic manipulation different from other kinds of nature-altering activities (e.g., seeking medical treatment for cancer or heart disease)

4) What is embryo adoption? What is it meant to do? Is it an appropriate response to the ethical issues arising from in vitro fertilization (IVF)?

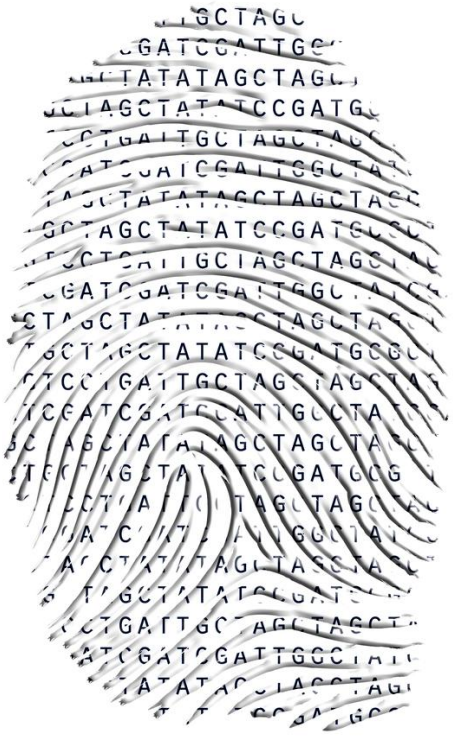


Session Two: Genetic Technologies, Information, and Personal Identity

Introduction

The film "Gattaca" follows a familiar storyline. Boy meets girl. Girl likes boy. Boy and girl perform the dating ritual. They disclose information about each other. They talk about their dreams.

The story takes an unexpected turn when girl tells boy she's had boy genetically sequenced and profiled. Girl apologizes. Girl then confesses to boy that she has a heart problem. Boy seems unmoved by girl's confession. Girl pulls a hair from her head and gives to boy and says, "If you don't believe me, take this. If you're still interested, let me know." Boy holds the hair, looks at it for a moment, looks at girl, lets the hair go and says, "Sorry, the wind caught it."



We are now able to test for well over a thousand genetic diseases, disorders, and traits. New tests are developed almost every day. Techniques for doing many tests at once and doing them cheaply are in the works. In this session, we will discuss a few of the many puzzles that arise because of our rapidly expanding ability to create comprehensive genetic profiles of individuals. The infamous O. J. Simpson case catapulted the concept of DNA evidence into the public eye. It is now a routine part of forensic science. But this is just the tip of the proverbial iceberg.

According to technology critic Neil Postman, every technology has within it at least one big idea. What is the big idea embedded in genetic technology? Is it that everything of value to us can be discovered in the structure and function of our genes? We might not like our genetic profile, but according to that big idea it is immutable. How is our capacity to sequence and analyze our DNA likely to affect our self-understanding and the way we interact with each other? Will it change our social institutions, our way of judging the suitability of people to be husbands, wives, fathers, mothers, sons, daughters, employees?

Scenario 1

Question to Think About

How will increased knowledge of genes and their significance affect our social lives?

Although the first narrative focuses on dating, it is easy to extrapolate from this to other social rituals and institutions. How will daily life change as genetic profiling becomes increasingly possible?

Narrative: The New Dating Ritual

Ginny and Kevin have been dating for some time and now they are both ready to take their relationship to another level. Kevin proposes and Ginny accepts. They realize that they are now committed to a common destiny and will need to plan their future together. Before they announce their intentions to their families and friends, they agree that they need to undergo medical examinations to ensure their compatibility and expectations of a healthy life together. They agree that along with tests for HIV and other sexually transmitted diseases they should also find out everything they can about their genetic profiles. Kevin and Ginny consider this to be simply a matter of taking responsibility.

Review Genetics 101 to refresh your memory on the relationship between genes and genetic traits. The section on Genetic Testing will also be helpful to understand the basics of testing for various genes associated with certain traits.

Discussion Questions

1) One purpose of dating is for couples to get to know each other to see if they are compatible. Couples learn about each other's history and each other's families. Is learning about each other's genetic traits simply another part of that?

2) Embedded in every technology is an idea – usually a very powerful idea – that has the capacity for changing the way we think about life. What is the idea embedded in our ability to learn about someone's genetic profile?

3) As genetic testing becomes routine and affordable this scenario will become increasingly plausible. Should there be regulations about such testing? What kind, and how would they be enforced?

Scenario 2

Question to Think About

Is genetic testing for those planning to have children a responsible thing to do?

You may want to review Genetics 101 and review terms in the glossary such as *allele*, *phenotype*, *genotype*, and other terms mentioned in Genetics 101.

Scenario Two looks at pre-conception genetic decision-making. When a couple thinks about having children and the future of their children, the issues become complex. There are some instances where both potential parents may be carriers of a specific disease, for example, and their offspring may well develop it. With diseases such as Tay-Sachs, a couple may decide to remain childless or even not marry.

Parents want to provide the best possible future for their children. This scenario asks how far that hope should be carried.

Narrative: Pre-conception Screening

You are a pastor in a clinic. Julie and Frank have made an appointment with you to talk about their plans to have a child. They are not yet pregnant. Julie is a university student and is taking a course on genetics. She has learned a great deal about genetic science and understands that their children will inherit alleles from both parents, and that these combinations could be detrimental to their offspring. Julie wants them both to undergo an extensive battery of genetic screenings before conceiving, so they can assess the genetic risks associated with conceiving a child together. Neither Frank nor Julie wants to have an abortion, so they hope that by screening themselves they can decide whether to conceive or adopt.

Julie says that a couple they know have three children and each child has the same genetic disorder, which has led to stunted growth and developmental problems. One of the children developed an autism-related disorder and is institutionalized. Neither parent had any family history to suggest that one or both of them were carriers for this disorder. Julie hands you a brochure from a genetic testing advocacy group that makes the argument that, given the state of genetic testing technologies, it is irresponsible for any prospective parent not to take these tests in order to avoid genetic tragedies like that which befell Julie's friends.

Frank is not in favour of the genetic screenings but admits that he does not know much about genetics and is willing to do what Julie wants provided you, their pastor, support this important decision.

Discussion Questions

1) How should you respond to the genetic information that Julie brings to the conversation? To what extent is this science relevant for your thinking? Is it necessary for Frank to get up to speed with the science? Can you effectively proceed with your counseling role in this case without having some understanding of genetics?

2) What might be the psychological and social consequences if Julie and Frank decide to proceed or not to proceed with these tests?

3) What counsel would you give Julie and Frank?

4) If Julie and Frank were to instead come to you for pre-marital counseling, would your mind change on any of these issues?

Scenario 3

Question to Think About

Will genetic knowledge change the way we think about reproduction?

This question explores the larger issue of reproduction itself. In this scenario, a couple has reproductive choices put before them by their insurance company, the imaginary GenLife Insurance Inc. The ideas found in this scenario are implied in the logic of the insurance company and the natural desire on the part of potential parents to do what they can to bring a healthy child into the world.

Narrative: Pre-implantation Screening

Before you consider this story you may wish to review Embryo Development and Genetic Engineering. In addition, you may wish to review in the glossary some of the terms used in this scenario, for example, *in vitro fertilization*, *pre-implantation genetic screening*.

George and Melinda live in Ontario ten years in the future. They have decided they want to have a child. Melinda has health benefits through her employer, a major university. When they examine her policy this is what they find.

GenLife is committed to quality health care at an affordable price. You want a healthy child and we will help you achieve this goal. We will cover 100% of your prenatal and birthing costs provided you meet the following conditions:

- *You must agree to have your embryo or fetus screened for all known genetic disorders, defects, and disabilities. We will do this either in utero or through in vitro fertilization (our much preferred method). All costs associated with this screening will be assumed by GenLife.*
- *If the screening reveals genetic markers raising the probability that your child (if you bring the embryo to term) would develop a genetic disorder or disability you will have two options:*

1) *Discard the embryo/fetus and repeat the process. GenLife will cover the cost of discarding the embryo/fetus and work with you as many times as necessary to produce an embryo free of potentially costly genetic anomalies that could pose a threat to your future child 's health and greatly increase health care costs.*

2) *Keep the embryo and assume full financial responsibility for the treatment of the discovered condition(s) should it (they) develop.*

- *GenLife WILL NOT cover prenatal or birthing costs should you elect to carry an unscreened embryo to term.*
- *GenLife WILL offer health insurance to your unscreened child but at a cost commensurate with increased risks associated with unscreened embryos.*

Be aware that your unscreened child will always be a member of a high-risk pool and will be required to pay higher premiums. Also be aware that if you choose to carry an embryo to term with a known risk for a disorder or disability they will never be covered for that disorder or disability by GenLife.

THIS PROCEDURE IS NOT MANDATORY. If you elect not to have your embryo or fetus tested we WILL cover your child once born, but premiums will be commensurate with his or her unscreened risk pool, which is significantly higher than for screened pools. We also offer two enhanced prenatal programs described below.

GenLife Fetal Selection Program (GFSP)

Under the GFSP program we will help you produce 20 embryos through IVF. As with the standard screening program these embryos will be carefully screened for genetic disorders and disabilities. In addition we will test for all known genetically influenced traits, such as sex, height, skin, hair and eye colour, etc. We will then assist you in selecting not only the healthiest but also the most desirable based on your own needs. We understand that you want to give your child every possible advantage. We're here to help. COST: $45,000

GenLife Fetal Enhancement Program (GFEP)

Under the GFEP program you not only receive the benefits of the standard screening and Fetal Selection programs, you are also able to take advantage of the latest developments in germline intervention technology. This technology uses artificial chromosome technology to actually tailor the genetic profile of your child. There are currently 6 alterations known to be safe and effective. Many others are being developed. With current technology you have the ability to virtually guarantee a healthy baby with traits known to be advantageous in today's competitive world. Our extensive research shows that your child's genetic profile is the single most important factor for health, longevity and quality of life. We are so confident that you will be pleased with the results that we will reduce the standard screened embryo premium by half and guarantee these rates for the life of your child. Expensive? Yes. But how much is your child's health worth? COST: $180,000.

Discussion Questions

1) What do you think about an insurance company that tells you that if you use in vitro fertilization you must agree to have your fetus or embryo screened for genetic defects?

2) Are the options GenLife offers morally acceptable to you? Why? Why not?

3) Assuming such programs would be legal, what do you think about GenLife's Fetal Selection and Fetal Enhancement Programs?

Session Three: Genetic Technologies and Research on Human and Non-Human Subjects

Introduction

As we come to understand the genetic basis of specific diseases we will learn how to address these diseases at the genetic level. There is no doubt that even now genetic research is generating promising new treatments for diseases. Research with stem cells, for example, could lead to growing organs for transplantation that would not be susceptible to rejection. For many people, the research being done on both human adult and embryonic stem cells raises ethical questions. Some make no moral distinction between the two types of cells. Others do not condone the use of embryonic stem cells, which requires the destruction of human embryos. Like abortion, this issue can be understood to centre on the question of who is a person. In Canada, a fetus is not legally a person (and therefore the subject of rights) until it has been completely delivered from the birth canal of its mother. Society is divided on this legal position, with those opposing it arguing that a fetus should be considered to be a person from the time of conception. Both views have implications for the use of embryonic stem cells in research and experimentation. For those who oppose embryonic stem cell research, this experimentation is morally wrong since the embryo is a person who is killed as a result of such experimentation.

The use of adult stem cells does not involve this moral dilemma. These cells can be harvested from specific human organs and used to develop treatments for cells that have been destroyed by disease or that are genetically abnormal. Moreover, recent scientific advances in reprogramming adult somatic or mature stem cells to a state similar to that of embryonic stem cells is changing the moral landscape, especially since this method also seems to solve the problems of rejection and tumour formation, both of which have impeded progress of the use of embryonic stem cells. In this session we will discuss how genetic technologies are being used – and are likely to be used in the future – in research on human and non-human subjects. The scenarios that follow are both fascinating and troubling. Each asks a difficult question raised by our developing technologies.

Scenario 1

Question to Think About

Should we create transgenic beings?

Our first scenario is based on a remarkable true event that occurred in 2001. The Oregon Primate Center announced the first successful germ-line engineering in a non-human primate. The Center had created a "transgenic" animal: a monkey with the inserted DNA of another species – a jellyfish – in every cell in his body. The experiment showed it is possible to genetically modify non-human primate embryos.

Narrative: ANDi & the Jellyfish

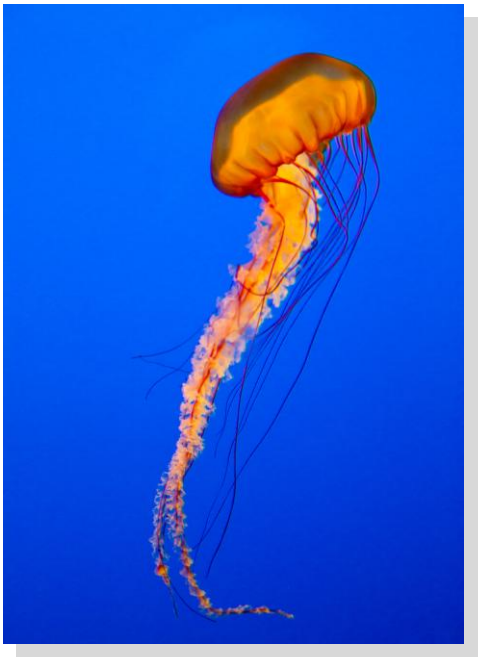
Review Transgenics, noting especially the distinction between mixing genes from two or more organisms versus cloning the entire organism. The section on Human Engineering and Cloning in Embryonic Development and Genetic Engineering will also be helpful.

You are on an ethics panel at a prominent medical research centre. The research proposal being reviewed by the panel is from the lab of Dr. Chan, whose research team has made remarkable strides in genetic technology, including the successful birth of the first cloned nonhuman primate using the technique of in vitro blastomere separation. The experiment they want to perform involves the insertion of genetic material from a jellyfish (GFP) into the eggs of a Rhesus monkey. After the genetic material is inserted into the eggs, they will be fertilized and then implanted in the monkeys. If the experiment is successful the offspring will have the genetic sequence from the jellyfish in every cell of their body. GFP has been successfully inserted into mice without harmful side effects. The justification for the research is that if it is possible to insert DNA sequences into the germline of nonhuman primates, then it will eventually be possible to design nonhuman primates that are susceptible to human diseases. Such an animal will be very effective for research purposes and the research will also require far fewer animals.

Discussion Questions

1) What questions would you ask if you were a member of an ethics board faced with this proposal?

2) Are there ethical concerns that you might have because the experiment was being performed on a Rhesus monkey rather than, say, a mouse or a nematode? What are the principles you would use to distinguish between species if you were developing research regulations for different classes of animals?



3) Should society allow transgenic experiments? We have already inserted human genes in some species (cows for instance) to produce proteins used to treat human diseases. The UK allows enucleated cow eggs to be combined with human material for experimental purposes. What are the possible risks of inserting human DNA sequences into a Rhesus monkey? Is it possible that we might create a half human-half monkey hybrid? What are the ethical implications of such a possibility?

4) Genetic manipulation of germline cells is perhaps the ultimate trajectory of genetic research. What does this kind of potential for control over our own evolution and the evolution of other species mean?

Scenario 2

Question to Think About

Who is a person? What is the status of a human embryo?

Some of the recommendations of the 1994 Royal Commission on Reproductive Technologies were incorporated in the Assisted Reproduction Act, 2004. This Act allowed experimentation on "spare" embryos in Canada, for therapeutic but not reproductive purposes. Earlier, in 1978, Pierre Soupart had submitted a proposal to do research on human embryos. His proposal was instrumental in the development of the first policy statement on embryonic research in the United States. In 1983, in defense of research on human embryos, he wrote, "Because of its human origin the embryo undoubtedly deserves to be paid a high degree of respect when treated as a research object. What higher form of respect could be paid to human embryos than to ask them to provide vital information leading to the alleviation of some types of human infertility, the prevention of birth defects, contraceptive and cancer research, and the actual causes of natural embryonic losses in man?"

Embryonic stem cell research remains a controversial ethical topic and we continue to ask questions about what we now know about the human embryo. Is there anything distinctive about its status or its use in research? What do we mean by human "personhood"? The question of when a fetus becomes a person is complicated and troubling, and is answered in different ways by different groups. In Canada a fetus is not legally a person until birth, and therefore embryonic stem cell research is allowed, since the embryo has no legal rights, including a right to life. Here are two scenarios that raise the question of what it means to be a person.

Narrative: Use of Embryonic Stem Cells and Fetal Tissue in Medical Treatment

1) You are a genetic counselor and Tom and Jackie have come to you with a dilemma. Their six-year-old daughter, Molly, has Fanconi anemia, a rare genetic disorder that prevents the production of bone marrow and can kill at a very young age. A bone marrow transplant from a matching sibling has an eighty-five per cent chance of curing Molly. Tom and Jackie have conceived a child with the intention of using stem cells from the umbilical cord and placenta after the birth to try and save Molly. They had not wanted a second child but had no doubt that this was the only and best option for Molly. They want to use pre-natal testing to find out if the fetus, a) has the same disorder as Molly and b) is a good match for transfusion.

2) Mark and Anna have come to you with the intention of testing a fetus they have conceived with the intention of aborting if it is a good match for Anna's father who is dying of Parkinson's disease. They want to use the fetal tissue in what they have been told is a very effective treatment for Parkinson's.

Discussion Questions

1. German philosopher Immanuel Kant talks about the ethical principle of never using a human being as a means to an end, however great the end might be. Can this principle be applied in these two scenarios? Is there a moral difference between Scenario I and Scenario II?

2. Abortion is permitted under the Criminal Code in Canada. The motives of women asking for abortion services are regarded as their business. Is there anything different about the two cases under discussion here?

3. Should processes like these be regulated? If so, what kind of regulations should be developed?

Scenario 3

Question to Think About

Should human embryos be cloned for medical research?

Ian Wilmut made international headlines in 1996 when he announced that he and his team had successfully cloned a sheep using somatic cell nuclear transfer technologies. The Roslin Institute was granted a license, the second in the UK, to clone human embryos for research purposes. Wilmut's work raises fundamental questions, not just about the disposability of human embryos in research, but also about the application of cloning technologies to human beings.

There are two broad categories that frame the debate on human embryo cloning:

a) cloning for the purpose of research, in which case the embryo is ultimately destroyed, and

b) cloning for the purpose of reproduction. To date, cloning for reproduction is illegal in most countries.

The focus of this session is on using embryos for research. Should we allow scientists to clone human embryos if such research might lead to breakthroughs in treatment for human diseases (for example, motor neuron disease)?

Another important distinction is that between cloning using blastomere separation and cloning using nuclear transfer. Blastomere separation occurs in nature and leads to identical twins, or even quadruplets. In 1993 human embryos were cloned using technology based on blastomere separation. Nuclear transfer involves the direct transfer of genetic material into an enucleated egg, which is then coaxed into a totipotent state' that is, it becomes an embryo. The purpose of this type of cloning is for experimentation, and the same moral questions arise here as in the use of "spare" embryos.

Narrative: Cloning Human Embryos & Motor Neuron Disease

You are a member of the federal board established to regulate the use of genetic technologies. Two researchers have come to the board to request permission to clone human embryos for research on motor neuron disease.

They want to clone embryos using tissue from sufferers of motor neuron disease so they can learn something about the developmental mechanisms of this terrible disease. They have located women willing to donate eggs for this project and are ready to proceed if the licensing board will agree. The board has already granted one laboratory a licence to clone human embryos, but you are having doubts about the morality of this kind of research. You have just finished reading some research on human reproduction and have had great difficulty working out in your own mind how to think about human embryos. One of the researchers makes the following argument: the best way to learn about this disease is to clone human embryos. If we can understand this disease we can cure it. Thousands of people and their families will benefit.

You are aware that recent stem cell research may make the need for cloning redundant. Further, use of these stem cells does not incur the same moral problems as the use of embryos.

Discussion Questions

1) Bioethicist Andrea Bonnicksen writes about human embryos that, "we cannot talk about germline therapy without considering the policies on embryo research. And much of the concern relates to the sanctity or the non-sanctity of the embryo. What the embryo is will determine what people believe about what should be done with it." Discuss.

2) Mary Mahowald suggests in the *American Journal of Bioethics* that it might make a moral difference if researchers allowed embryos to die before extracting stem cells, since this action would preserve the letting die/killing distinction. She also suggested that some form of ritual at the disposal of embryos used in this way might allay the moral concerns of some groups. Here is the quote: "Allowing embryos to die before retrieving their stem cells thus provides a means by which some individuals can preserve their moral integrity. Extra embryos may also be dealt with respectfully or disrespectfully. Presumably, the notion of 'respect' or 'disrespect' for early embryos makes no sense for those who regard them as having no moral value or status. For those who do, however, respectful disposal is surely possible." Discuss.

3) Here are two suggestions on how to derive stem cells from embryos without destroying them. Would either suggestion solve the moral problem of embryonic stem cell research?

    a. Remove one cell from the embryo and derive stem cells from that instead of destroying the whole embryo.

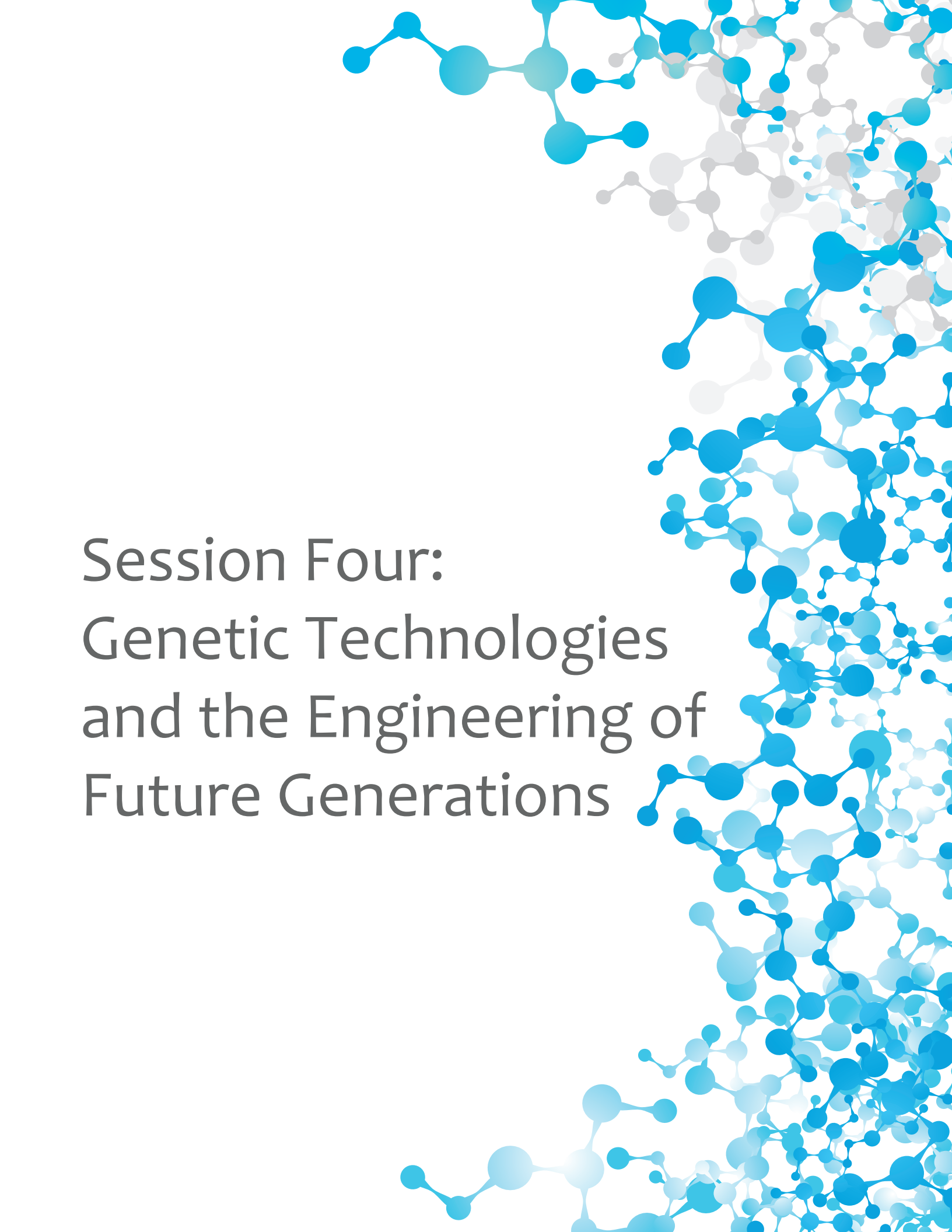
    b. Create human embryos that cannot be brought to term, even if we wanted to.

4) In some ways, the debate about human embryos is similar to the older debate about abortion. Do you see ways in which the discussion about human embryos can move forward in ways that the abortion discussion cannot?

5) The debate about embryo-derived stem cells is part of a larger conversation about embryos generally. When we do research on embryos, they die. When we use embryos obtained through IVF, many are destroyed or stored and later destroyed. When we derive stem cells from embryos, the embryo dies. How should we develop policies that affect human embryos?

6) What would you say to the researchers?

Session Four: Genetic Technologies and the Engineering of Future Generations



Introduction

Mary Shelley's famous novel, *Frankenstein*, is among the first literary responses to genetic engineering – humans tinkering with the mechanisms of human creation. The theme carries straight through to the Ridley Scott classic science-fiction film, "Blade Runner." In both stories human arrogance and hubris create powerful and dangerous creatures that become their creator's worst nightmare. The film "Gattaca" gives an indication of what a society of genetically engineered citizens could be like. Movies and stories like these can make us think that genetic science takes us into areas of scientific experimentation that we are not meant to explore and that cannot be controlled.

In this session we will consider the extent, given our genetic knowledge and power, to which we should regulate decisions that could affect future generations and perhaps even human nature itself. James Watson who, along with Francis Crick, is credited with discovering the overall structure of DNA, believes that genetic engineering is inevitable and a good thing, since nature, left to its own devices, can make some pretty horrendous mistakes from time to time. He calls our current method of reproduction "roulette," which implies that anyone who does not take advantage of the new technologies is simply foolish. What do you think? The case studies that follow take us quickly to the heart of the controversy.

An illustration of the double helix structure of DNA. It looks like a twisted ladder.

Scenario 1

Question to Think About

What are the ethical boundaries for genetic testing?

Eugenics is the science of improving the quality of human beings through interventions in the processes of reproduction. There are two types of eugenics: positive and negative. Negative eugenics involves the selective destruction of unwanted embryos or fetuses based on one or more of their genetic traits. Positive eugenics involves actual modifications to the embryo, which are intended to change the resulting human being for the better, either by eliminating an unwanted disorder or disease or by enhancing some capacity in the future person, such as greater height, intelligence, athletic ability, and so on. The first narrative considers negative eugenics, but with an unusual twist.

Narrative: Genetic Testing and Disability

You have been the pastor for a deaf couple, Roger and Sally, for three years. You are fortunate to have someone who knows sign language on your staff. Sally has just found out she is pregnant. They are happy but are concerned about the likelihood that their child will be deaf also. You have just attended a conference on genetic testing and know that there are tests that screen for many genetic disorders that cause deafness. You tell Roger and Sally about this option and give them a business card of a clinic that can do the screening. As they tell you their story you suddenly realize that they were not worried about having a deaf child, but a hearing child. They want to screen for and abort any fetus that will be able to hear.

Review Inherited Disease and Genetic Testing to refresh your memory on testing for predispositions for different diseases and the implications of knowing whether an embryo, fetus, infant, or adult might have inherited a gene that may increase the risk of the associated disease.

Discussion Questions

1) Would your response have been different if Roger and Sally were blind? Why or why not?

2) What implied message does permitting selective abortions give to disabled people? A blind bioethicist, Adrienne Asch, argues that all prenatal screenings should be banned based largely on this implied message. (Compare this question with Session 2, Scenario 2, which raises similar ethical questions.)

3) We often use words like *disease*, *disorder*, *condition*, *disability*, and *trait* when referring to different sorts of bodily features. Is deafness a disease? A disorder? A condition? A disability? A trait? What are the different messages suggested by these terms?

4) If people are allowed to selectively abort embryos or fetuses to ensure a "healthy" child, does this imply that people should also be allowed to selectively abort to ensure having a "disabled" child? Is the principle the same? What is the principle used in the defence of selective abortions of any type?

5) The more control people have over reproductive outcomes the more significant the questions become concerning the rights of the person most affected by these decisions. Should there be rules established to regulate selective abortion based on a single trait or a set of traits?

Scenario 2

Question to Think About

What does your faith tell you about the procedure Bob and Carol are thinking about using?

This scenario proceeds on several assumptions:

- that IVF is morally acceptable
- that selection of apparently healthy embryos from a raft of embryos is morally acceptable
- that sex selection is up to the parents.

Carol is concerned about genetic enhancement, which raises further ethical questions. How would you respond to these different layers of morality?

Narrative: Genetic Testing and Disability

You are a genetic counselor who works with members of faith communities. Bob and Carol are married athletes who have decided to have a child and are using IVF procedures to screen embryos for various genetic disorders. After selecting a few embryos that seem relatively free from potentially harmful alleles they finally decide on a female. They name her Sarah.

The clinic representative then gives Bob and Carol a brochure on the "Gold Medal" program, a method for genetically modifying the selected embryo to give them the "Gold Medal" advantage. For example, genetic science can now explain why some people, like champion Canadian cyclist Ryder Hesjedal, are "built" for endurance and maximum energy output. The average

lung capacity of a healthy male is about 6 litres, Ryder Hesjedal's capacity is 8.3 litres. That amount of extra air translates into more oxygen in the blood, which leads to enhanced performance.

Bob and Carol look at each other and confer. Bob wants to do it. Carol does not. They decide to postpone the decision for 24 hours and have made an urgent call to you. Bob's argument is simple. The kinds of traits he wants to modify are already found in nature. Some people have some of them. Some people have others. Some people seem to have more of the right alleles for athletics than others. Bob feels that, since they have a choice, they should give Sarah every advantage that they can afford.

Should winning athletes give back gold medals because nature gave them certain genetic traits? They still have to work hard. So would Sarah. If Bob and Carol can afford to give her the "Gold Medal" advantage, why shouldn't they? Carol says she is uncomfortable with "genetic fiddling," as she calls it. She was already uncomfortable with the screenings and choices they had to make that day about which embryo would survive and which would not. While she hopes her daughter will be interested in sports and will do well, Carol just does not think they should try to engineer all that. She just wants a child to love and care for, whatever her problems and her athletic abilities might be. It just feels "creepy" to her to do all this foisting of their desires on this child, even before it is implanted in her womb. Bob responds with, "being a good parent means giving your child every advantage and opportunity you are in a position to give." They stop talking, and wait for you to speak.

Discussion Questions

1) Bob and Carol are decent people trying to do the right thing. They each have a reasonable principle underlying their opposing points of view as they try to agree on what they should do. What do you think the principle is in their positions? What do you think of their arguments?

2) Bob makes the point that the genetic changes they will make already occur through natural methods of reproduction. What do you think of this argument? If something is found in nature are we morally justified in reproducing it scientifically? What principle is Bob using? Do you agree with it? Would it make a difference to you if one or more of the enhancements suggested involved a novel genetic innovation, say a method for rapidly removing waste products in the blood not found in nature?

3) The collection of genetic traits offered by the "Gold Medal" program has nothing to do with potential disorders or diseases. Does this make a difference to you? If "health" means psychological and social health as well as physical health, do you think that genetic engineering should take these factors into account and not just focus on physical diseases?

4) Some scientists believe that we can insert genes that do not "turn on" unless they are in the presence of a certain hormone. If this technology were used to delay the development of the desired traits in Sarah, delaying the decision until later, perhaps even involving Sarah's wishes as well, would this make a difference to you? Why or why not?

5) When the clinic representative handed the brochure to Bob and Carol, there were certainly commercial motivations. Should we always be presented with every possible option in the case of genetic modification? Should there be regulations guiding what can or cannot be offered by IVF clinics to the consumer?

6) What are the deeper implications of this type of genetic engineering for society? What are the implications for what we think it means to be a good parent?

7) Finally, what does this case imply about human nature, about what it means to be a human being? What do we, as people of faith, have to say to Bob and Carol about the goal and purpose of life?

Scenario 3

Question to Think About

If cloning were both possible and legal, do you see any ethical problem in using a dead child's DNA to clone another child, who would be the dead child's genetic "twin?"

Cloning is proving to be more difficult than at first anticipated, but, assuming that the technique could be perfected, we would then have to consider the outcome of using other people's DNA for cloning. Embryos and children developing from them would be genetic replicas. In cases such as in this scenario, couples might feel they owe it to the child who died to somehow use his or her DNA to reproduce the original child. What would happen to our experience of being unique, or of being wanted for ourselves, as opposed to being the product of someone else's DNA?

Narrative: Reproductive Cloning

You are the pastor of a large urban church in the near future. One of your parishioners, Alice, has come to your office for counseling on a specific issue that has come up with her business. Alice is an embryologist and the owner of an up-and-coming embryo research lab. They have successfully cloned non-human primates. The lab has a partner in Asia where laws regulating research are not as strict. The company is struggling financially, but Alice feels confident that they are working on important research that will ease human suffering. Her work on cloning human embryos for therapeutic purposes has produced very good results. Alice believes that she now has reliable techniques for performing nuclear transfer cloning. She tells you the following story.

Larry and Linda came to the lab a few weeks ago. Last year their 11-month-old son, Ted, died in the hospital. Because of an accident Larry is no longer able to produce sperm so they will not have another chance for biological offspring. The hospital was determined to be largely at fault. Larry and Linda were already very well off and the suit they filed produced an award of more than 30 million dollars. They had preserved tissue from Ted's body and presented it to Alice at the interview. They want Alice to attempt to use the tissue to produce a genetic clone of Ted. They are willing to put up 50 million dollars to carry out this genetic experiment. Alice's company can't do this work, but cloning for reproductive purposes can be done by her Asian partner. Larry and Linda seem to understand that this clone would not be "Ted" but his genetically identical twin.

Alice told Larry and Linda she would consider their offer and call them. At first Alice was inclined not to accept, but as she began to examine the idea she found herself having

less and less resistance to it. They would simply insert Ted's DNA into a donor egg, which would then be implanted in Linda's uterus. For all practical purposes the child would be their own son, raised with all the love and care that their child deserves.

They certainly have the means for raising a child and the means for taking advantage of cloning technologies. As far as safety goes Alice is confident that the risks are not greater than other legal reproductive technologies She has come to you because she remains unsure of the ethics of reproductive cloning. She wants your counsel as she approaches this important decision.

Discussion Questions

1) This case is not as far-fetched as it may sound at first. The desire to reproduce can be a very strong human drive. For people with money, for whom other techniques either do not work or do not provide enough genetic continuity, somatic cell transfer will be a considered option. What is your initial reaction to this? What are the reasons for your reaction?

2) Larry and Linda are wealthy. Is this important?

3) Assume that somatic cell transfer could be shown to be no more risky than other IVF techniques. Is this morally relevant?

4) Are there theological concerns that bear on this case? What does your faith tradition offer that might address the question of reproductive cloning? Is the method itself the problem? Safety concerns? What it implies about our self-understanding as human beings?

5) This scenario implies that privately funded research on reproductive cloning is not regulated. Do your concerns about cloning as part of IVF treatment lead you to believe it should be regulated? Banned?

6) Alter the scenario. Imagine that Larry wants to clone himself, instead of his dead son. The baby would be Larry's twin brother and son. How would this change your thinking? Why do "brother" and "son" produce a kind of uneasiness in many? Is this uneasiness connected to a moral concern or is it more a kind of "man, that's just creepy" reaction?

Session Five: Genetic Technologies and Social Justice

Introduction

This session asks us to think about genetics and faith from a social justice perspective. We have already considered some ethical questions from the point of view of individuals and from the point of view of couples thinking of marriage. Parents are also faced with difficult situations when prenatal or pre-implantation genetic diagnosis indicates that their child, either at the embryonic or fetal stage, suffers from a serious illness or disability.

But what are the implications for policies that involve society as a whole? Health care is a good example.

Given the amount of money spent on health care in Canada, and the pressure to spend more, it is possible that emerging genetic technologies will not be covered by our health care system. Even now, new and expensive therapies are being carefully assessed for effectiveness and toxicity risks compared to the therapies they are meant to replace or supplement. Decisions for approving funding should be based on important outcomes such as improved survival or curability. Difficult decisions will be made that will likely exclude funding for some patients.

What type of genetic testing should be admissible and who should be considered eligible for such testing? Should it depend on whether a test is deemed life-saving (e.g., needed to determine urgent treatment)? What if it is requested for non-therapeutic reasons (e.g., the demands of insurance companies for a battery of tests)? Should these types of tests be publicly funded?

If expensive genetic technologies are publicly funded, making them more accessible, what health services would they displace? If genetic technologies are privately funded, how would we regulate the worst effects of the disparity in access? Will the ability to screen for more and more diseases and disabilities more and more cheaply lead to changes in our attitudes toward disability? Will there be increasing pressure not to produce any child with a disability? Will health

insurance, especially employer-based health insurance, pressure employees to take genetic tests to find pre-existing conditions? Will genetic knowledge change our ideal of equality as we discover great differences in our "natural endowments"?

We know from history what can happen when eugenics becomes acceptable policy. In the mid-twentieth century, Nazi Germany targeted people they considered not worthy of life and eliminated the mentally and physically disabled through sterilization and euthanasia.

Some people still believe that society should be cleansed of its mentally and physically "unproductive" members. This can come in the form of pressure to abort fetuses with physical or genetic evidence of mental or physical disabilities or threatening not to subsidize their care with public funds if they are allowed to live. Yet most of us become disabled in some way if we live into old age, no matter how strong and vigorous we have been in our youth. Will we, in turn, be perceived as "liabilities" and users of scarce resources? This is an important question for a society in which the proportion of elderly citizens is steadily increasing.

The question of scarce resources concerns us all. With shrinking budgets and expanding medical and surgical possibilities, how do we decide what gets funding? Most of us believe our health care system operates on a fairly equitable basis, but some circumstances pose problems. For example, how do we decide who should receive donated organs when there are not enough to go around? Should younger people have priority? Is this discrimination against older people justified and if so, should other eligibility criteria be added?

Another social justice question is raised by pressure to introduce in Canada a fee-for-service system, or privatized health care, where a treatment may be prioritized on the basis of the ability to pay rather than need. The inability of poorer citizens with no extra insurance coverage to pay for drugs or dental care is also an ongoing social justice issue. No health care system is perfect and these issues are real and challenging.

Scenario 1

Question to Think About

Will genetic technologies affect ethnically related social problems? If so, how?

There is a widespread belief that ethnicity is a genetic rather than a social characteristic, and, while it is true that certain ethnic groups are carriers of some specific diseases, it is not true that such diseases are confined only to those ethnicities. There is the danger that certain groups could be stereotyped or that, for example, intermarriages would be avoided out of fear of passing on genetic problems.

Francis Collins writes that "…race is an imperfect surrogate for ancestral geographic origin, which in turn is a surrogate for genetic variation across an individual's genome."

He notes that many think that race and ethnicity are too flawed as concepts to be helpful in working out race-health concerns. Although some diseases seem to be specific to some races or ethnicities, we have to be careful not to presume too much, nor to perpetuate stereotypes or prejudices linked to some diseases.

Narrative: The Use of BiDil

For the past two years you have been a member of a Health Canada committee that approves new drugs. You are currently charged with making a recommendation regarding the drug known as BiDil (isosorbide dinitrate and hydralazine hydrochloride). You are convinced that the drug is sufficiently safe and effective. You are concerned, however, about how the drug is to be marketed. Its maker, NitroMed, intends to market this drug to a particular ethnic population, based on studies that show it to be particularly effective in that group. Little was done to isolate the particular genetic profile that might make this drug more or less effective. You have done enough reading to understand the debate that approval of this drug will raise since it is being targeted for use in a particular group as though it were established that ethnicity is a genetic rather than a social category. Further, studies show there is more genetic variation within that group than in other ethnic communities. Overall, further studies show that BiDil has been effective in treating African-Americans, but it is feared that since only African-Americans participated in clinical trials, some think it is only effective within that group, whereas it may be more universally beneficial.

Discussion Questions

1) Even though you understand that there is a preponderance of evidence in favour of BiDil's safety and effectiveness, you also understand that the science behind the marketing decision is shaky. You know that the marketing will inevitably reinforce the dubious idea that ethnicity is a genetic category. You also understand that there will no doubt be a public controversy about the decision should the drug be approved for use in that particular community. How will you weigh these considerations?

2) Do you think your role on the committee should focus only on the safety and effectiveness of a drug or should you also consider social issues?

Scenario 2

Question to Think About

What effect will genetic technologies have on the social and economic spheres?

According to Neil Postman, every technology has winners and losers, those who benefit and those who suffer. The film, "Gattaca," portrays a future society that is divided into two groups: the genetically engineered who are designed for the highest skilled jobs, and the "invalids" who are given menial tasks to support the work of the engineered class. Is the world of Gattaca a logical extension of the social and economic system we have today? Who will benefit from the genetic revolution? Who will suffer? What factors will shape how we use genetic technology?

Narrative: Genetic Testing and Employment Benefits

Janet is applying for a job at Maplekey, a large computer manufacturer. She is excited about this opportunity and looks over the job application very carefully. The company offers extraordinary benefits, particularly in the area of health. It pays 100% of all health related costs for employees and all immediate family members. There is just one caveat. The "health history" portion of the application includes genetic testing. All her immediate family members must be sequenced and profiled. The job is not dependent on the outcome of this test unless it reveals a life threatening or seriously debilitating disease which would affect Janet's job performance in the near future and cost the company a good deal of money. If the tests reveal the presence of a genetic marker linked to the high probability of less serious illnesses or disabilities Janet will still be offered a job, but her health premium and those of her family members will be based on the actuarial tables for the set of probabilities—the "risk pool" revealed in the genetic profiles. Janet seems to be in good health and she is not aware of any major health issues on either side of her family or that of her husband. Janet decides to discuss the situation with her husband, the kids, and you, her close friend.

Discussion Questions

1) This narrative is similar to the GenLife narrative in Session Two. Here, Janet and her family are being asked to disclose genetic information in exchange for a potentially good situation, a good job with great health coverage. As a close friend, what advice would you give to Janet?

2) It is a common, and some would argue reasonable, request that insurance applicants disclose pre-existing conditions. Physical exams and family health histories are sometimes part of the process of assessment and the assignment of a risk pool. How is genetic testing different, if at all, from this kind of risk assessment process?

3) What are the risks for Janet and her family when they take these tests? Apart from the possibility that Janet may be turned down by Maplekey, what other problems could arise?

4) Some argue that genetic technologies will either accelerate the demise of private insurance as a way of delivering health care or further isolate those with a less than ideal genetic inheritance. What do you think?

5) Does genetic testing have any place in assessing suitability for employment? Are there safeguards that could be put in place?

Scenario 3

Question to Think About

How will genetic technologies affect the allocation of health care resources?

Demands for testing for genetic information may affect health care budgets. For example, the daughters of women with breast cancer traceable to the BRCA1 and BRCA2 genes can be tested. Such testing has prompted many women to request pre-emptive mastectomy surgery based on probabilities of their developing breast cancer in the future. Even though not all the women would develop cancerous tumours, it's easy to understand the internal pressures that would lead to such requests. As more research uncovers genetic sources of disease, more of us will find ourselves anticipating outcomes and undergoing pre-emptive treatments where available. Since this will be statistically based, our health care resources will be further stretched, in some cases unnecessarily.

Narrative: Statistical Assessments of Successful Treatment

A 50-year-old man had surgery recently to have a malignant tumour removed from his colon. Certain features of the tumour suggest there is a high risk of the cancer recurring in the next five years. A treatment with some potentially troublesome side effects is available that can reduce that risk by 20%. However, patients whose resected tumour possesses a certain mutation are much more likely to benefit from the treatment; that is, the tumour recurrence rate is reduced by 60% in patients whose tumour has the mutation while the rate is less than 5% in patients whose tumour does not have the mutation. A reliable test for this mutation is available but is very expensive due to patent protection. Colon cancer is a relatively common cancer, so clearly, more lives can be saved by preventing cancer recurrence in patients whose tumours have the mutation. In addition, considerable health care funds are saved or deferred among treated patients whose tumours had the mutation. Therefore, the ministry of health decided to fund the genetic test that can discriminate which patients are more likely to benefit from the treatment.

Discussion Questions

1) What are the financial pros and cons of paying for tests for all patients who have a particular cancer that will indicate that the recurrence rate in some patients will be reduced by a certain treatment?

2) What would you do if a member of your family were told that she or he was ineligible for treatment?

3) Patent protection makes some treatments very expensive. Under our health care system, is non-funding of such treatments on the basis of expense (as opposed to efficacy) morally justified?

Genetics 101

Early History

Why do certain characteristics run in families?

Humans have always been curious about inheritance. Until the 1800s, the mechanism of *how* traits were passed from parents to children was debated by philosophers and theologians, but almost no scientific analysis was performed.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

In the mid-1800s, there was as yet no knowledge of genes or their molecular basis. However, a central European monk named Gregor Mendel became intensely curious about the mechanism of genetic inheritance. Born in what is now the Czech Republic and educated in a local monastery, Gregor excelled in the natural sciences and remained at the monastery into adulthood. One area of particular interest for him was an understanding of the mechanism by which certain physical traits were passed on from parents to their offspring. In pursuit of this, he began performing experiments with field peas in an attempt to determine how certain traits such as flower colour, plant height, and pea shape were passed from parent plants to offspring plants.

Although he had no knowledge of genes or the DNA of which they are mainly composed, Mendel was able to develop certain principles of inheritance that are now known to apply also to other living creatures, including humans. For these discoveries and others, he has become known as the "Father of Modern Genetics."

Nearly 100 years after Mendel's experiments, Francis Crick and James Watson discovered the structure of **DNA (deoxyribonucleic acid)**. Understanding the molecular composition and function of genes has led to a much better understanding of the relationship between genes and characteristics that they express as well as the interaction between genes, the environment, and human health.

The Composition of a Human Cell

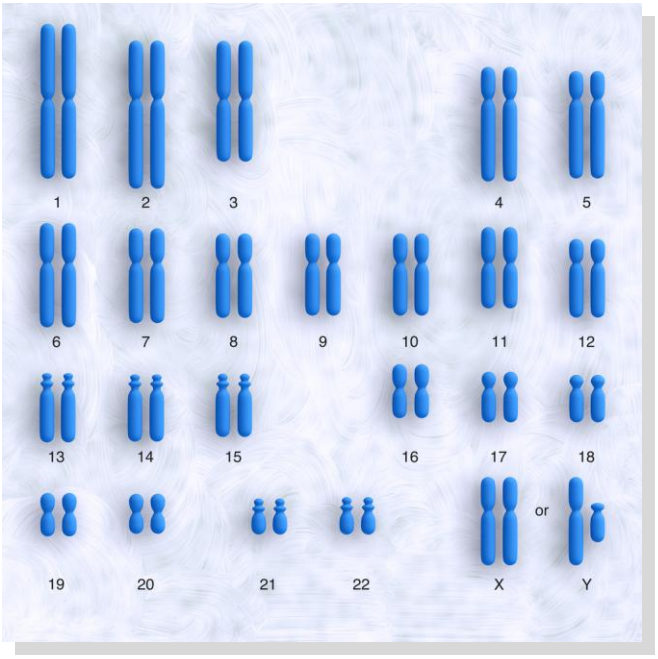
To understand the importance of genetics, one needs to understand some anatomy at the organ and cellular levels.

In the human body, each organ (e.g., the lungs, kidneys, brain, heart, etc.) is made up of tissues. These are types of cells that function differently but work together to keep each organ functioning normally.

Although the cells that make up different parts of the body – skin, bones, brain, heart, lungs, and everything else! – have different structures and compositions, all cells of the

human body share some basic physical and biological attributes.

All cells are composed of a liquid, known as the cytoplasm, within which are located the nucleus and other smaller structures. This cytoplasm is surrounded by a membrane that keeps the cell intact. The **nucleus** acts as a control centre for the cell. Each nucleus contains a full set of **chromosomes**, known collectively as a **genome**. The nucleus of all body cells contains 23 pairs of chromosomes (46 total chromosomes), including a pair that determines the gender of the person. (The exceptions are **germ cells** (sperm and eggs), which have only 23 chromosomes each.)



An illustration of a normal complement of human chromosomes

Each chromosome pair contains genes, the backbone structures of which are DNA. This DNA guides the formation of proteins, which are building blocks for the structure and functioning of each cell. Each gene has two copies or **alleles**, one on each of the paired chromosome. These alleles may be structurally identical or may vary to different degrees. Thus, each allele of each gene has an identical or structurally different "twin" on the other chromosome in that pair. Structural variations of normal alleles of a given gene are called **mutations**. Mutations that cause an allele to differ even slightly from the normal allele may have a profound impact on the expression of that gene.

Although each gene has only two alleles in each cell (except germ cells which have one allele per gene), there may be many different varieties of alleles in a human population. This is the case with traits such as height as well as hair and eye colour and explains the wide variety of these traits in some populations.

The genome of each person is identical in the nucleus of every cell in the body. So your brain cells have exactly the same genes as the cells in your lungs, your inner ear, and the tendons of your big toe. However, your brain appears different, and functions differently, from your big toe because not all of the same genes are at work in your brain cells as are at work in the cells of your toe. That is, during your development as an embryo and fetus, inherited genetic instructions in certain genes turned off the functioning of other genes (called silent genes). Different genes were turned off in different types of cells, allowing the functioning of the different genes in each type of cell. This variation in the genes turned on and turned off in each type of cells led to the diversity of appearance and function necessary for your development as a unique newborn human being.

A Sidebar: DNA Outside of the Nucleus

Besides the nucleus, cells also have small, distinct structures in the cytoplasm called **organelles**, each type of which has its own function. One type, known as the mitochondrion, carries important energy-producing components of the cell. Each mitochondrion contains a very small amount of additional genetic material containing DNA. This genetic material is passed on through the mother's genetic line because the egg from the mother carries nearly all of the cytoplasm that is contained in the **zygote** (that is, the cell produced by the union of a sperm and an egg at conception).

Mutations of this mitochondrial genetic material have been linked to an increasing number of distinct health disorders, although there are also mitochondrial disorders due to DNA in the nucleus. One or more organs may be affected and symptoms vary among disorders. Symptoms that may occur include muscle weakness and lack of coordination, visual or hearing problems, nerve problems, mental disorders, and diabetes. We will not discuss this type of DNA further but for those interested in more information on this interesting new area of mitochondrial gene-related diseases, see: http://www.ncbi.nlm.nih.gov/books/NBK1224/.

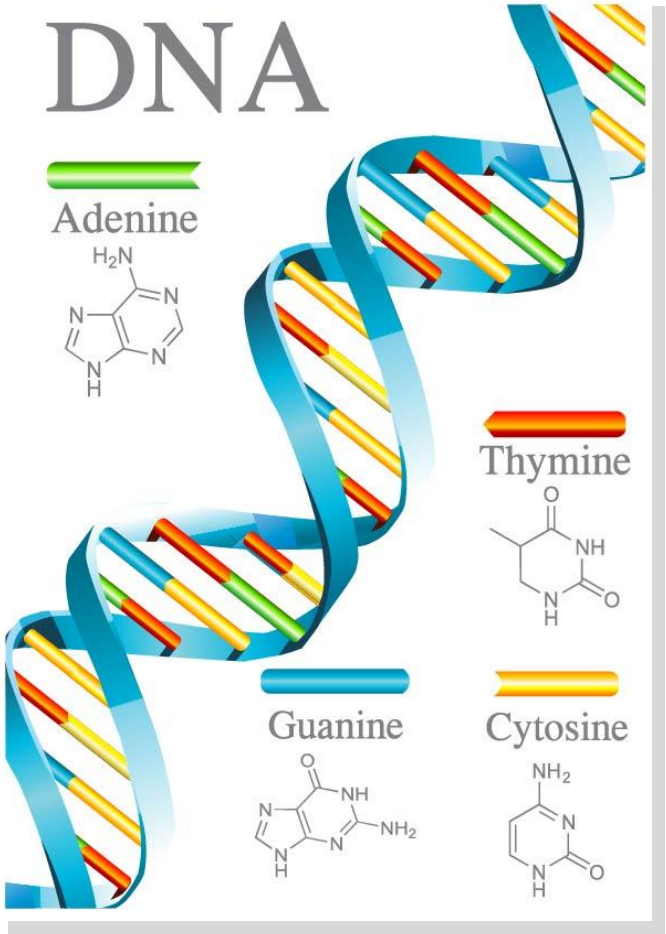
Composition, Characteristics, and Effects of Chromosomes and Their Genes

Chromosomes are composed of **chromatin**, a complex of molecules made up of DNA that is wrapped around special proteins known as **histones**. DNA is the backbone of this complex that contains the information of genetic inheritance. When a cell divides into two cells, each pair of chromosomes can double and separate so that each new (also called daughter) cell has a normal number of the same paired chromosomes that were present in the original (also called parent) cell. While many different kinds of cells are

capable of dividing and replicating in this way during life, sperm and egg cells are unique in that they each have only one of each type of chromosome rather than a pair. When a sperm fertilizes an egg, one set of chromosomes in the sperm (from the biological father) is paired with one set of chromosomes in the egg (from the biological mother). This combination of a sperm and an egg results in a unique set of paired chromosomes in the fertilized egg, now known as a zygote.

How the structure of DNA is translated into proteins that become the molecular basis for functions such as movement, speech, sight, and other workings of our bodies

DNA is a polymer, a type of molecule made up of repeating subunits, like beads on a string. DNA has four subunits: **adenine, guanine, cytosine,** and **thymine** (or A, G, C, and T). These subunits are like codes that determine the production of certain molecules called **amino acids**. These amino acids are like blocks for building proteins. Different combinations of subunits produce different amino acids and different combinations of amino acids determine the formation of different proteins.



The DNA subunits are arranged on the DNA like steps on a ladder. The A and T subunits are always paired to form a ladder rung, as are the G and C subunits. Thus, DNA is like a ladder composed of pairs of subunits that make up the rungs of a spiral ladder. It takes three of these pairs or rungs to make a single amino acid and different sequences of pairs produce different amino acids, which in turn produce different proteins. Multiple layers of combinations can produce a huge number of different proteins that, when combined with other molecules like sugars and fats, constitute the backbone of many bodily functions.

In the cell, the DNA of each gene is translated into specific proteins through the

mechanism just described. These proteins then build vital structures in the cell that serve many essential functions in our bodies. Each gene produces a protein or set of proteins distinct from those of other genes. As mentioned earlier, each gene has two alleles. If each allele is identical, they produce identical proteins. However, if one allele is different (usually due to mutation), the proteins produced by each allele may differ. For example, differences in traits such as eye colour are determined in large part by the structural or functional differences or similarities between the proteins produced by the pair of alleles on a particular gene. In the next section we will explore in more detail the concept of mutations and their important genetic and functional diversity.

Mutations: Alterations in the Order of DNA Subunits and Their Consequences

An alteration in the order of subunits in the DNA of a gene is called a **mutation**. Such mutations can produce an altered subunit of the protein normally produced by that gene, which in turn can sometimes lead to altered structure and function. In a cell, that change or mutation will be inherited by any new cells created when the mutated cell divides. Whether that mutation results in a change in the structure or functioning of these new cells will depend on a number of factors, including the type of gene that was mutated, whether the gene was a functioning or silent gene, what function it had before it was mutated, and so on. Mutations occur all the time. Right now, you are accumulating mutations in your DNA. But fortunately, most mutations are repaired before they can be translated into abnormal proteins. If they are repaired, the mutations have no adverse consequences for the cell, for its progeny that result from cell division, or for the person to whom the cell belongs. This is the case when a mutation occurs in parts of the DNA that don't contain genes or when a structurally altered protein still functions normally.

Sometimes, however, a mutation produces a protein that functions abnormally. For some mutations, these proteins may have no noticeable effect on the person with the mutation. Other mutations may cause the death of the cell itself. Such cell death may have little or no consequence for fully developed persons but could result in the death of a developing embryo or fetus. Still other mutations can cause a change in a desirable or undesirable characteristic such as hair colour or height. Occasionally mutations result in new qualities or functions not typical for that type of person or species.

Spontaneous versus Acquired Mutations

Mutations can occur spontaneously in any cell and at any time. If such a mutation

occurs in a cell other than a sperm or egg cell, that mutation is known as a **somatic mutation** and cannot be transmitted to that person's children. The mutation may have no noticeable effect, may affect only a specific type of tissue or organ, or may noticeably affect the whole person. However, if the mutation occurs in a sperm or egg cell, it is a called **germ-line mutation**. As noted earlier regarding the chromosomes, each egg or sperm has just one allele of each gene rather than the two alleles found in somatic cells. So if a sperm with a mutation of an allele merges with an egg without the mutation on its allele, the gene of the resulting embryo will grow to adulthood having one mutated allele and one normal allele in each cell of its body for the rest of its life.

In a person with a genetically linked disease, the disease may occur through spontaneous mutation of only one allele of a particular gene during embryonic development. In other cases, the disease may occur through the inheritance of an allele that mutated in an earlier ancestor. In still other cases, the disease may occur when a person who inherits a mutation on one allele of a particular gene experiences a spontaneous mutation on the other allele of that gene As we will learn later (in the section, Inherited Disease and Genetic Testing, Predicting Breast Cancer by Genetic Testing) the strength of expression of the mutation for causing the disease will determine whether one or both alleles must carry the mutation for the disease to develop.)

Differences in Genes Can Become Differences in Our Characteristics

Genetic information is passed from parent to offspring mainly by DNA. In cells, this information is translated into proteins that build structures and fulfill important functions for the cell, for the organ or tissues where the cell resides, and for the body as a whole. These structures and functions are evident in characteristics such as eye colour, facial appearance, height, and other features. Because we get our DNA from both of our biological parents at conception, our features and traits often are similar to those of one or both of our parents. A difference (or mutational change) in the molecular structure of a gene is a **genotypic** difference between the original gene and the changed gene. If that genotypic difference results in a difference in the characteristic or function associated with that gene, such as eye colour, that difference is a **phenotypic** difference. In this case, the eye colour would be different between the original gene and the changed or mutated gene.

Epigenetic Changes

Until very recently, geneticists thought that only changes in the DNA of genes (that is, mutations) could lead to changes in phenotypic expression of those changes. They have recently discovered that changes in the non-DNA, or histone portions of the

genome, can alter gene expression. Such changes in histones are known as **epigenetic** changes because they can cause changes in gene (or phenotypic) expression without altering the DNA sequence of the genes

Epigenetic changes can also be brought on by environmental factors. For example, epigenetic changes have been linked to altered stress responses in the brains of individuals who experience childhood abuse. In turn, these changes have been linked to an increased risk of suicide later in life. Very importantly, these changes can sometimes also be inherited by subsequent offspring. Since epigenetic changes can be acquired changes and then transmitted to offspring, this newly understood dimension of genetics has changed fundamentally our concept of genetic change and inheritance.

Along with certain sections of DNA, epigenetic changes appear to be involved in the mechanisms by which different types of cells know which genes to turn on and off as these cells multiply and differentiate into different functions during embryonic and fetal development. Such epigenetic regulation can be involved in the control of normal cell division and tissue growth. However, they can also be involved in disturbances of cell growth in which epigenetic changes can lead to uncontrolled cell growth as cancers.

It is now known that many of the changes of the genome that can lead to uncontrolled growth of a cell and its progeny involve subtle molecular changes of histone proteins associated with DNA. This knowledge is now being exploited to develop new cancer drugs that can alter or negate such epigenetic changes in cancer cells and thus suppress or stop cancer growth. There are already therapies available that target such changes in the genome of cancer cells and more are being developed and approved for use every year.

Different Proteins for Different Functions

Different genetic information in DNA is translated into different proteins. Structural differences in these proteins are associated with different physiological functions. Proteins made in your muscle cells interact with one another to allow your muscles to contract. Proteins in neurons, the main cells that make up the nervous system, produce

the neurotransmitters that allow you to know what to say next. Proteins in the retina of your eye produce molecules that change shape when light hits them, allowing you to see. Fingernails and hair are composed of the protein keratin. Hemoglobin is a protein that transports oxygen from your lungs to the rest of your body. Amylase is a protein made by saliva-secreting cells in your mouth that allows you to digest starch in various foods such as potatoes or crackers.

Practical Implications of Genetic Variation: The Story of Blood Types

Blood can be distinguished into types according to different molecules associated with red blood cells. One system of typing human blood can be instructive in understanding the concept of genetic inheritance. On one of our chromosomes, there is a blood-type gene that contains instructions for producing a protein whose function is to add sugars to the surface of red blood cells. Like other genes, this gene has two alleles, one inherited from the biological mother and the other from the biological father. Each allele can be one of three minor structural variants. The A variant has the recipe for an enzyme that adds A-type sugars to the red blood cells. A mutation that occurred in the far distant past may be the reason for the existence of a second variant, called the B variant, that adds B-type sugars instead of A-type sugars to the surfaces of red blood cells. A third variant (or O variant), possibly also the result of a past mutation, produces a non-functional enzyme that cannot add sugar to the red blood cell surface.

If each allele of your blood type gene has only the A variant (the gene is then designated AA), you and your gene are **homozygous** for Type A blood. If one allele has the A variant and the other the O variant (the gene is designated as AO), you and your gene are **heterozygous** for Type A blood. In both cases, only A-type sugars are present on your red blood cells because variant B is not present and variant O is cannot add sugars. The same pattern follows for the B variant, in which case only B-type sugars are present on the red blood cells and in either homozygous (BB) or heterozygous (BO) situations, the person has Type B blood. If you have neither A-type nor B-type sugars on your red blood cells (or OO), you'll have Type O blood. Finally, if one allele has the A variant and the other the B variant, both sugars will be present and the blood type is AB.

These genetic variants are analogous to different recipes for oatmeal cookies. The allele for blood type A is like a recipe for cookies with raisins (red blood cells with type-A sugar) while the type B blood is analogous to a slight change in the cookie recipe, resulting in cookies with chocolate chips instead of raisins. The O variant is the simplest recipe, with no raisins or chocolate chips included. In the absence of raisins (one A allele) or chocolate chips (one B allele), the resultant oatmeal cookies are just plain (and less flavourful!)

The A and B Variants are Dominant and the O Variant Recessive

Some alleles of particular genes are expressed (that is, result in a characteristic such as eye colour or blood type) more dominantly than other alleles of the same gene. When a gene is heterozygous and one allele is more **dominant** than the other, the dominant allele may be expressed while the other, **recessive** allele is not. A recessive allele can be expressed only when the gene is homozygous for that allele (both alleles have the same recessive mutation), in which case a more dominant allele is not present. In blood typing, the A and B alleles are both dominant and the O allele is recessive.

Summary of the genetics of the ABO blood typing system

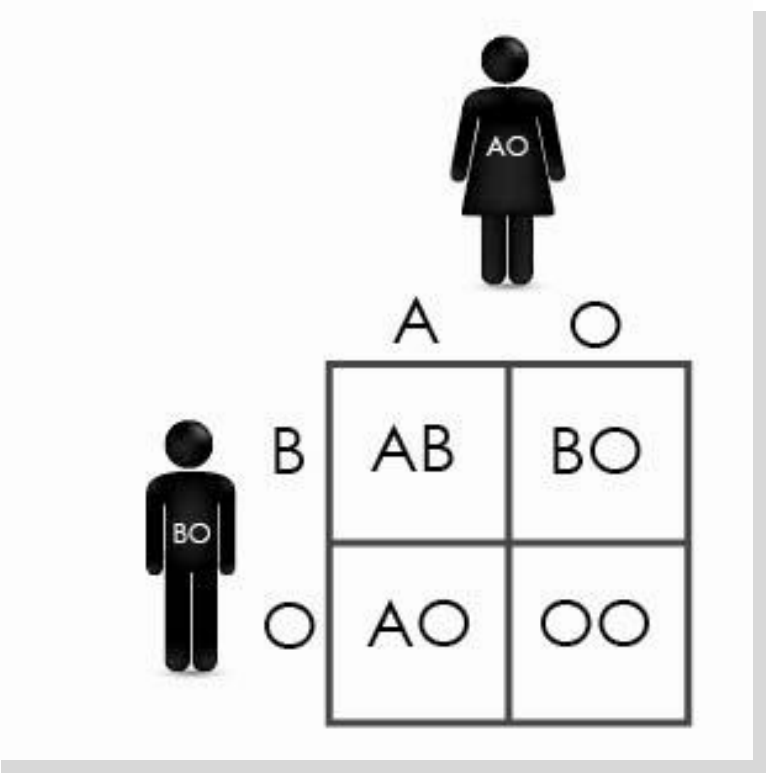
1. In the ABO blood typing system, if you are homozygous or heterozygous for only the A variant (that is, AA or AO), you will express only the A sugar on your red blood cells and will be considered to have Type A blood.

2. If you are homozygous or heterozygous for only the B variant (that is, BB or BO), you will express only that sugar on your red blood cells and will be considered to have Type B blood.

3. In the absence of both A and B alleles (i.e., OO), you have Type O blood. If you have one A allele and one B allele, you have both sugars and thus Type AB blood (or oatmeal cookies with both raisins and chocolate chips)

Predicting the Proportion of Offspring with Traits of a Particular Gene

The Punnett Square is a helpful way to explain the relationship between dominant and recessive genes (see figure). Named after biology professor Reginald Punnett, it is used to predict the genetic contribution of parents to their offspring and the resultant genotypes of those offspring. If two people with the blood types A and B have a child, the results can be predicted using a Punnett Square (see figure). The mother's known alleles (A and O in this case) are placed along the top of the square and the father's known alleles (B and O) are placed along the side of the square. The four squares represent the possible genotypes of their children. Here we see that this couple could have children with any of the four possible blood types and that each time they have a child, there is a 1 in 4 or 25% chance that the child will have blood type AB, O (represented by OO), B (BO) or A (AO).



Similarly, if the parents each have the A and O alleles and therefore each have type A blood (that is, each have AO), the letters can be plugged into the Punnett Square. You will see that there is:

- a 75% chance of their child having blood type A (one out of four squares or 25% has two A alleles while two out of four or 50% have the A and O alleles together) and
- a 25% chance (one square out of four) of having both alleles or blood type O.

This type of inheritance, where one gene controls one measurable characteristic, is called simple Mendelian inheritance, after the monk, Gregor Mendel, who first deduced the mechanism of single-gene inheritance.

Behavioural Genetics

Genes and Their Environment

"The primary goal of Behavioral Genetics is to establish correlational relationships between genes and behavior" [1]

Is there a gene for bungee-jumping? Is alcoholism a genetic trait just like blood type? Are there genes for schizophrenia? Is a person's personality a series of chemical reactions in the brain that are determined by their genes?

Scientists who study these kinds of questions are called **behavioral geneticists**. Most of these geneticists would say the data suggest that personality traits are influenced by, but not determined by, genes.

Most human characteristics are not determined by a single gene. Many human characteristics are influenced by several genes working together. In such cases, the simple Punnet Square that can be used to determine blood type won't work easily for determining the frequency of inheritance for such characteristics. And, to make things a bit more complicated, a human characteristic is frequently the result of the interaction of one or more genes and the environment. When we say environment here, we are not talking about just the outside world—where you went to school, what you had for lunch, whether or not you exercise. The environment of a human gene includes 1) the other genes in that cell, 2) hormones and other chemicals to which the cell is exposed, 3) interactions with other cells and tissues, and 4) the environment outside the body. Studies of identical twins can be helpful to study the influence of environment. Since identical twins have identical genes/DNA, scientists can learn a lot by studying how twins that are separated after birth are affected by being raised in two different communities/environments.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

Polygenic Inheritance: Inheritance Involving More than One Gene

Hypothetical Example of Multiple Genes Affecting Height

Let's look first at specific traits that are determined by several genes, a phenomenon known as **polygenic inheritance**. A good example is height. Height is a continuously varying characteristic. That is, not all humans are either 5 feet tall or 6 feet tall. Rather,

[1] (Bazzett, Terence J. *An Introduction to Behavior Genetics*. Sunderland, MA: Sinauer Associates, 2008. ISBN 0-87893-049-3)

human heights are distributed through a range. Multiple genes contribute to height. Indeed, geneticists have learned that many genes, scattered widely over multiple chromosomes, appear to contribute additively to the genetic determination of height.[2]

To understand multiple gene involvement in the inheritance of polygenic traits, let's consider a simplified and hypothetical case. Let's assume that only three genes interact with each other to control height and that each gene has two different alleles. For each gene, one allele adds height (additive) while the other allele does not (nonadditive). In addition, the allele that adds height is dominant over the one that does not. Because every child inherits one allele of each gene from each parent, you can look at all three genes and even prepare a Punnett Square for each gene. A couple who each have three dominant alleles and three recessive alleles among the three genes could end up with children who were the same height as they are, but could also be significantly shorter or taller. While more than three genes are normally involved in height determination, this hypothetical example gives the general idea. In fact, with more genes involved, one can see greater extremes in height in offspring than if only a few genes were involved, even when the parents are of fairly average height.

You may be thinking, doesn't environment affect height? Yes, but negligibly. The vast majority of the characteristic of height is genetically determined. If your parents had fed you protein shakes as a child and sent you to a Montessori school, you still would not likely be 7 feet tall. Environment will result in relatively minor adjustments to the genetic underpinnings that determine a person's height.

Other Polygenic Traits and Risks of Illness Influenced by Environmental Interactions

Many characteristics are determined polygenically, including skin colour, weight, blood pressure, and blood cholesterol levels. Of course, these characteristics also have an environmental component. Determining whether a trait is determined by genes or an interaction between genes and the environment can be difficult, but not impossible. In some cases, it is actually quite simple, as when only one or two genes interact with the environment.

For example, the risk of stroke by a blood clot can be related to certain alleles for two genes that control the production of proteins involved in the clotting process (one is called prothrombin while the other is called factor V). Each gene has some alleles that are associated with changes in the nature or production of these proteins. Some of these protein changes may considerably increase the risk of blood clots, particularly if

[2] (Visscher, PM, et al. Am J Hum Genet. 2007 Nov; 81(5):1104-10).

the person with those proteins is exposed to certain environmental factors. Some kinds of oral contraceptives are a good example. They may act as environmental factors by interacting with such proteins after being swallowed and absorbed into the body.

Taking contraceptive pills can have a much greater effect on clotting in women carrying genes that produce the more risky proteins. For example, among women who have *not* inherited the risky proteins, there is a *three times greater risk of stroke* from a blood clot when taking oral contraceptives compared to women who don't take oral contraceptives. However, among women who inherit the risky proteins, taking oral contraceptives increases the risk of a stroke *150 times higher* than similar women with the risky protein who do not take oral contraceptives. Clearly, oral contraceptives can have a substantial environmental effect when interacting with certain genetically inherited proteins

Interactions between Genetic Variation and the Environment: The Case of Behavioural Genetics

Most of the time, however, the interaction between genes and the environment is more complicated, often because of unknown factors. Such unknowns can include the number of genes involved, the percentage of genetic variation in a trait, and the percentage of variation in a trait due to environmental influence. Nowhere is this truer than with behavioural genetics.

Behavioural traits include abilities, feelings, moods, personality, intelligence, and how a person communicates, copes with anger, and handles stress. Disorders with behavioural symptoms are wide-ranging and include phobias, anxiety, dementia, psychosis, addiction and mood alteration. While most illnesses associated with abnormal behavioural traits involve multiple genes, a few such conditions can be traced to a single gene. Huntington disease is a rare example of such a condition.

Huntington Disease: Behavioural Disorder Due to a Single Genetic Mutation

Huntington disease (HD) is a fatal, progressive, neurodegenerative disease caused by a dominant mutated allele. Individuals who are heterozygous for HD usually develop symptoms in their late 30's or 40's. Some early symptoms of HD are mood swings, depression, and irritability or trouble driving, learning new things, remembering a fact, or making a decision. As the disease progresses, concentration on thinking and speaking becomes increasingly difficult and the affected person may have difficulty feeding himself or herself and swallowing. Angry outbursts are the hallmark characteristic of this disease.

Since a mutation in a gene can result in behavioural traits, it is clear that genes can be linked to human behaviours. Unlike HD, however, most behavioural disorders are not the result of a single mutated gene.

Challenges of Identifying Genetic Associations with Variations in Behaviour

Investigating the genetics of behavior is more difficult than understanding a disorder such as sickle cell disease or HD in which an abnormal protein clearly disrupts physiology in a particular way. One of the reasons that such investigations are difficult is that many behavioral disorders share symptoms, which can complicate diagnosis. For example, poor concentration may be a symptom of attention deficit disorder (ADD), major depressive disorders, or post-traumatic stress disorder, to name a few. Further to this, many symptoms, including poor concentration, can be considered variations of normal behaviour – surely everyone from time to time has a hard time concentrating or experiences mood swings when under some degree for stress for a time.

Another challenge to understanding the relationship between genes and behaviours is the highly subjective nature of studies that rely on self-reporting of symptoms by study subjects. A person can also, unintentionally, copy someone's unusual behavior, because he or she does not realize it's unusual. Such sources of confusion do not occur with diseases such as cystic fibrosis, where strictly physical symptoms such as shortness of breath and cough are characteristic manifestations of the disorder.

Although it is necessary to be cautious when assigning a genetic cause to a behaviour, it is still possible to examine genes that contribute to a particular behaviour. Typically, scientists attempt to identify behaviours that appear to be inherited, then focus on identifying and describing candidate genes. (More information on these behavioural disorders can be found in the section Inherited Disease and Genetic Testing.)

Example of Research Exploring the Association between Genetic Control of Nerve Transmission and Behaviour

How are the experiments performed to determine whether a candidate gene is actually involved in a behavioural trait? Let's look at the gene for the serotonin transporter.

Serotonin is a molecule that transmits signals from one nerve cell to another. One cell (the sending cell) produces and releases the serotonin. A nearby nerve cell (the receiving cell) then binds the serotonin and this cell responds to the serotonin signal in a certain way. The longer it takes the serotonin to move from the sending to the receiving cell, the more signaling occurs to the receiving cell.

A particular gene controls the production of a certain protein (called a serotonin

transporter) that can bind to serotonin when it is between the two cells, acting like a ferry that can return it back to the releasing cell. This process of returning the serotonin back to the releasing cell shuts off the initial signal and thus prevents the signal from becoming continuous. Normally, just enough serotonin reaches the receiving nerve cell to cause the appropriate amount of signaling for normal nerve functioning. However, if too much signaling occurs, there can be problems in nerve conduction that result in behavioural disorders such as depression, anxiety, and other mood disorders.

Now certain drugs can slow down the rate at which the serotonin is returned to the releasing cell. These are called selective serotonin reuptake inhibitors (SSRIs) and the antidepressive medications Prozac and Paxil are examples of such drugs. Such drugs can give relief to patients with depression by slowing the transport of serotonin back to the sending cells in certain parts of the brain. This slows the signaling frequency by allowing for a slightly longer time for signaling between nerve cells. Therefore, studying the serotonin transporter gene may give us a better understanding of the mechanism behind these behavioural disorders and lead to better therapies for such disorders.

Clinical Studies Can Find Associations between Different Alleles and Types of Behaviour

It turns out that there are two alleles for the gene associated with the serotonin transporter, called the long and short alleles. The long form is more active and more quickly mops up serotonin from the space between neurons. That is, people with the long form of the gene have a shorter signaling time than people with the short form of the gene. The question then is: can scientists detect a behavioural difference between people with these two forms of the gene?

Researchers at the National Institutes of Health conducted a study in which people's transporter genes were examined. The participants also took a standardized test that measures neuroticism, a term for emotional instability that includes obsessive-compulsive disorder, anxiety neurosis, and a variety of phobias. Each individual was then given a neuroticism score. Not surprisingly, when the scores of all the people were plotted, the scores formed a generally bell-shaped curve. Some people were extremely neurotic, some were extremely tranquil, but the majority of people were somewhere in between. When the neuroticism scores of people with the short allelic form of the serotonin transporter were plotted separately from those with the long form, we see that both sets of people formed a somewhat normal distribution. A careful examination of the graph, however, showed that the average neuroticism score of individuals with the long form of the allele was slightly higher than the average neuroticism score of individuals with the short form of the allele.

Statistical analysis of the results suggested that approximately 1% of the variation in neuroticism scores among humans was due to a variation in the gene for the serotonin transporter. This difference is small, but appears to be real. This result suggests that other genes are also involved as well as the environment. Such careful and methodical study is necessary in order to learn how many genes may be involved and how they interact to result in abnormal variations in human behaviour.

Inherited Disease and Genetic Testing

In this section we will learn about different types of genetic abnormalities using a specific disorder as an example for each type. Some types involve one gene and one mutant allele that is recessive (sickle cell anemia) or dominant (Huntington disease), others involve one gene but many different mutant alleles (cystic fibrosis), while still others involve multiple genes and multiple mutations (cancer).

Diseases Caused by Single Mutations with Recessive Expression

Sickle Cell Disease

Red Blood Cells Carry Oxygen

As we said earlier, DNA mutations can lead to altered proteins that may or may not work correctly for the benefit of the cell and organism. As modern molecular techniques now make detecting these mutations relatively easy, more genes are being detected that are associated with particular disorders. For example, you may recall that hemoglobin is a protein found in red blood cells that transports oxygen from your lungs to the rest of your body. As red blood cells pass through capillaries in the lungs, oxygen passes from the airways into the capillaries, then into the red blood cells. Bound to the hemoglobin, this oxygen is delivered by the blood supply to various tissues. The alleles of the normal gene for hemoglobin are designated HbA and thus a normal gene is designated Hb AA (or HbA/HbA).

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

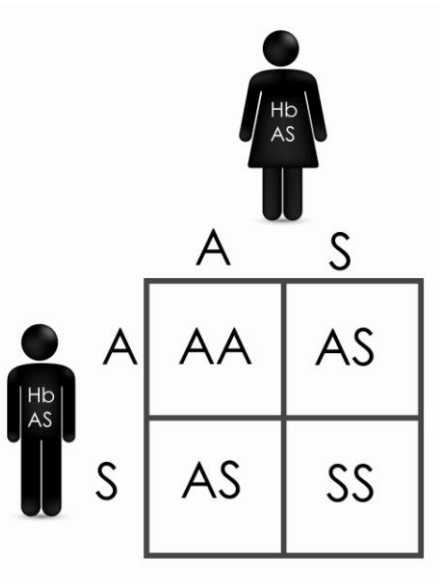
Sickle Cell Mutation Can Disturb Oxygen-Carrying Capacity

With a certain single subunit change or mutation, the normal allele is changed to a different allele called HbS. This allele is recessive. Thus, for carriers with only one allele, the gene is designated HbAS. As shown in Session 1 (see Genetics 101, "Practical Implications of Genetic Variation: The Story of Blood Types"), this means that the full characteristic or disease associated with the mutation occurs only when both alleles are affected, that is, when the gene is said to be homozygous for the mutation (designated as HbSS). (Among those with the HbAS gene, life expectancy is normal and they are not anemic, but some symptoms of the disease may occasionally occur if the person is unable to breath in enough oxygen, as happens with some lung diseases.)

Hemoglobin molecules produced by the HbS allele tend to stick to one another, producing long, rod-shaped structures that cause the red blood cells to assume a stiff,

elongated shape that looks like a harvesting sickle. These "sickled" cells tend to get trapped in the tiniest blood vessels or capillaries. As a result, the tissues supplied by those capillaries become starved of oxygen and the tissue dies. In carriers, the presence of one HbA allele and its normal hemoglobin molecule offsets the effect of the more recessive HbS allele and sickle cell disease does not occur. Clinical manifestations of those with sickle cell disease (that is, those with HbSS) include shortened life span, severe anemia, impaired growth and development, and unpredictable episodes of severe pain involving joints, abdomen, or chest during sudden occlusion of capillaries in those areas.

Sickle Cell Carrier versus Sickle Cell Disease



Persons homozygous for the HbS allele will have signs and symptoms of sickle cell disease early in life so testing for the mutation will not be necessary. While those homozygous for the normal HbA allele (HbAA) will not have the disease, people who are heterozygous also will not have symptoms of the disease, but are carriers of the HbS allele and could pass it on to one or more offspring. As seen in the Punnett Square constructed in the figure to the right, if two heterozygous individuals have a child, there is a 1 in 4 (25%) chance that the child will receive HbS alleles from both parents and as a result will develop sickle cell disease. If one parent is homozygous for the normal allele while the other is heterozygous, having a child runs a 25% chance that the child will carry sickle cell disease. However, as mentioned above, such a child would not have sickle cell disease.

Other Consequences of Sickle Cell Mutation

While genetic testing can be particularly helpful to determine the likelihood of parents passing on the sickle cell trait or disease to their children, genetic testing is also helpful to determine when genetic screening should be performed in a given population. For example, some ethnic and geographical populations have a very low prevalence of the disease while others, particularly those in Africa and those living around the Mediterranean Sea, have a much higher prevalence. Interestingly, malaria is also often prevalent in the latter populations and there is some evidence that heterozygous carriers of the sickle cell allele (HbAS) are partially protected against malaria. Since the abnormal hemoglobin in sickled cells carries oxygen less efficiently, parasites that infect red blood cells that carry the sickle cell trait seem to survive and replicate less well in

such oxygen-depleted conditions. Homozygotes with the sickle cell disease, however, do not benefit because of the severity of sickle cell disease itself outweighs any protection against malaria. As a consequence, testing citizens of these areas may be helpful in reducing the prevalence of those with the disease. In some countries, such as Cyprus, the prevalence of disease has become such a burden on the cost of health care that young people who carry the trait are discouraged from marrying someone with the trait in order to reduce the prevalence of disease.

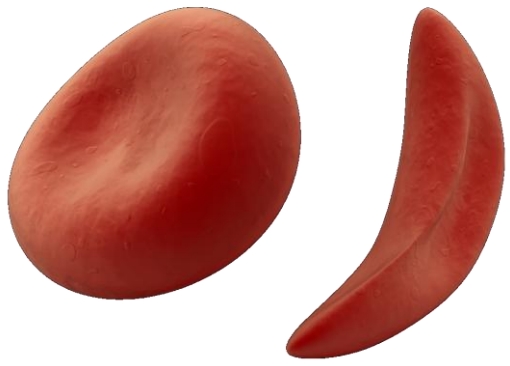
Cystic Fibrosis

Normal Function of the Cystic Fibrosis Gene

Cystic fibrosis (CF) is also a genetic disorder that is the result of a mutation in a single gene. In this case, the protein produced by the gene transports salt into and out of cells. If a mutation in the gene results in an abnormally functioning protein, this transport may be disrupted, in which case thick mucous can accumulate outside the cells. In some organs, this abnormality has no impact on function. In the lungs, however, the thick mucous causes difficulty in breathing and provides an environment in which bacteria can grow, leading to pneumonia. In the digestive system, the mucous prevents the secretion of digestive enzymes, leading to digestive problems.

Comparisons with the Sickle Cell Mutation

Unlike the case in sickle cell disease, where the same mutation is present in nearly everyone with the disorder, different mutations of the salt transporter gene can lead to cystic fibrosis. Currently, just over 1400 different mutations have been identified in the salt transporter gene and the severity of the disease varies depending on the mutation involved. However, over 70% of individuals diagnosed with cystic fibrosis have one particular mutation. Because the frequency of the other mutations is so rare, they are not routinely tested. Like the sickle cell mutation, all of these mutations of the salt transporter gene result in recessive alleles. So for an individual to develop CF, both alleles of the salt transporter gene must be mutated. Also like the sickle cell mutation, the prevalence of the mutation varies among different ethnic groups and nationalities. For example, in the United States, one in 25 (4%) Caucasians of northern European descent (EuroAmericans) carry the mutation. By



A typical human blood cell (left) contrasted with a sickle shaped cell (right).

contrast, the prevalence among African Americans is less than half that figure (1.5%) and less than one third of the rate found in Asian Americans.

Uncertainties with CF Testing

Unlike the test for sickle cell mutation, a negative test does not mean that an individual is definitely not a carrier because only the most prevalent mutation of CF is routinely tested for. Those who test negative may in fact be heterozygous for another mutated allele not tested for. For example, following testing, the carrier rates of cystic fibrosis for African Americans drops by one third, from 1.5% to 0.5%, but still does not reach zero. As in the case of the sickle cell mutation, such testing might be helpful in identifying higher risk groups. However, it also may miss carriers of rarer CF mutations and thus not give the full picture of carrier rates among different ethnic groups.

Disease Caused by a Single Autosomal Dominant Gene: Huntington Disease

Huntington Disease

The mutated allele that results in Huntington disease (HD) is dominant over the normal allele. This means that a person with only one mutated allele is not just a carrier, but will also develop the disease associated with the mutation. However, unlike persons with sickle cell disease or CF, those with Huntington disease typically develop signs and symptoms around the age of 40 or later rather than in infancy or early childhood. Consequently, genetic testing has a very different meaning for those with the HD mutation.

As with sickle cell disease and CF, individuals who appear healthy but who test positive for the HD allele know that they may pass on the mutation to their offspring. However, as carriers of a dominant allele like HD, they also know that they will eventually develop a deadly, progressive disease sometime later in life and that some offspring will receive the allele and develop the disease – doubly devastating news. This means that someone carrying only one allele has a 50% chance of passing on that allele (and therefore the disease) to any child, regardless of the genotype of the other parent.

Thus, for children with a parent with HD, testing for the HD mutation can be a difficult decision. Some children may choose not to be tested and thus not risk having to live with the knowledge that someday HD will develop. Such testing also has implications for marriage since having the HD mutation carries distinct risks for children to develop HD while not having the mutation carries no risk of future offspring.

Genetically Inherited Increased Risk of Cancer

Genetic Testing for Disorders Involving Single versus Multiple Genes

Although the majority of genetic tests currently available are for single gene disorders like sickle cell disease, cystic fibrosis, and Huntington disease, the majority of disorders associated with genetic changes are not the result of a mutation in a single gene. Multiple genes may be involved and/or genes may interact with the environment to produce a particular type of disorder, such as cancer, diabetes, heart disease, Alzheimer's disease, and others. While genetic testing for these disorders is much more complex and may not provide the precise answers obtained from testing for a single gene disorder, such testing may become more helpful in the future as our understanding of such complex interactions between genes and the environment increases.

BRCA1 Gene and Breast Cancer

Let's use cancer as an example. The development of cancer is a complex process, but the first step is usually a mutation in one of only a few important genes that control normal cell replication. An example of such a gene is the BRCA1 gene. This gene produces a protein that is involved in the regulation of cell division, particularly in certain cells in the breast, by repairing damaged DNA and helping cell division to occur in an orderly fashion. Cancer develops when cells begin dividing uncontrollably. Certain mutations of the BRCA1 gene result in a non-functional protein that fails to repair damaged DNA, resulting in a greater tendency for cells to replicate uncontrollably and a much higher risk of breast cancer. In other words, if a mutation of the gene results in the loss of the ability to stop cell replication, uncontrolled growth may result in cancer development.

Predicting Breast Cancer by Genetic Testing

These genes can develop mutations that markedly increase the risk of breast and ovarian cancer, even if only one allele is affected. The increased cancer risk associated with the inherited, mutated gene may be due to the mutation of the other BRCA1 gene but could also be due to the interaction of the inherited mutated protein with other proteins involved in cell replication. In either case, a woman who inherits a mutation of one of her BRCA1 gene alleles has a very high lifetime risk of developing breast cancer or ovarian cancer. Estimates of risk for breast cancer before the age of 70 vary from about 40% to 70% or greater (the risk of ovarian cancer may be as high as 40%). In addition, such cancers are also more likely to occur at an earlier age than those not linked to a mutation of BRCA 1.

Thus, women with breast cancer that develop at a very early age, or those with a first degree relative who develop breast cancer at a very early age, are eligible for genetic testing. If they test positive for a BRCA1 mutation, they may wish to consider preventative therapy to prevent cancer from recurring. In addition, daughters of such patients may wish to be tested to know whether they have inherited increased susceptibility to develop breast or ovarian cancer.

Diseases Involving Mutations of DNA Located Outside of the Nucleus

As mentioned earlier (in Genetics 101, *A Sidebar*….) there is a small amount of additional genetic material containing DNA outside of the nucleus of cells but within the energy-producing organelle called the mitochondrion. This genetic material is passed on only through the mother's genetic line because the egg carries nearly all of the mitochondrial DNA in its cytoplasm. Genetic testing on this type of DNA has been used to confirm the identity of deceased persons by comparing their DNA with that of a suspected female descendent. An increasing number of distinct health disorders have been associated with mutations of this genetic material and genetic testing for determining the susceptibility of the descendants of affected individuals may be possible. (For more information, see http://www.ncbi.nlm.nih.gov/books/NBK1224/.)

Testing Human Embryos for Genetic Mutations

Sometimes, a couple asks to have genetic testing conducted on embryos produced for them by **in vitro fertilization**. This is called **pre-implantation genetic diagnosis** and is performed by carefully removing one of the zygote's cells at the 8-cell stage. The cell is then examined for genetic abnormalities. The remaining 7-celled zygote is allowed to continue to divide and can mature into what appears to be a normal infant at birth. While such zygotes appear to develop normally thereafter, this area has not been well studied. This procedure also has raised ethical issues since the cell that was removed for testing was possibly still totipotent and thus capable of forming an identical infant to the one produced by the remaining 7-cell zygote (see also the section Embryonic Development and Genetic Engineering, Conception and Development of the Embryo).

Transgenics

Genetically-Modified Organisms (GMOs): Modification within a Species

In the past, before modern genetic technologies became available, genetic modification of organisms came about through inbreeding of a species over many generations. For example, improvements in crop resistance to insect damage were developed in this way. Genetic material such as DNA can also be exchanged naturally between organisms of the same or similar species, such as bacteria, resulting in genetic modification. For example, viruses, which are smaller than bacteria and usually must live in a cell to survive, can infect bacteria and incorporate some of their DNA into their bacterial host (some viruses have a molecule similar to DNA known as RNA, which can function in a similar way to DNA). This can result in changes in the bacterium's resistance to antibiotics. In more complex organisms, such infected cells usually die from the infection without passing the viral DNA to the next generation. **Genetically-modified organisms** or **GMOs** may be organisms that have been modified in the laboratory through the insertion of a mutation of a gene of the same species associated with a desired trait that would take generations to breed into the species by conventional methods.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

This has become a common and sometimes controversial technology in agriculture and animal husbandry where the results of introducing such GMOs can result in environmental imbalance and instability, and socioeconomic disruption. Use of GMOs in agriculture can result in the replacement of natural strains of plants by genetically modified strains. While the GM strains may carry desired characteristics, such as resistance to certain fungi or bacteria, the GM strain may also be less hardy in other respects over time. In addition, companies that produce GM strains have been known to gain a monopoly on the production of such plants in specific localities, resulting in economic and cultural damage and the loss of livelihood especially among some indigenous peoples.

Transgenic Organisms

Today, genetic technology allows us to take DNA from one species into the genome of another, resulting in a **transgenic organism**. Attaching DNA from one individual or species to a virus and injecting the virus into the recipient cell of another individual or species is one of the most efficient ways to introduce a new foreign gene into a cell. Before injection, most of the virus genome is removed and replaced with the desired gene. The virus is then injected into the cell of a new individual. Because the type of

virus used normally inserts its genetic material into the genome of cells it infects, the new gene gets inserted into the genome along with any remaining viral genetic material. If this is done to the germ cells, such as the sperm or the egg, this DNA from another species will likely be transferred to offspring and will be present in all cells of those offspring. To date, although viruses insert DNA more efficiently into cells, they cannot insert the foreign DNA very precisely into the recipient genome yet. In fact, the foreign DNA may end up anywhere in the genome, on any of the chromosomes. As a result, it may not function correctly in its location or it may function in an abnormal way because of its proximity to other genes. The development of some cancers, including leukemia, has been associated with abnormal gene placements on chromosomes.

Difference between Hybrids and Chimeras

When large amounts of DNA are combined between species, additional ethical concerns arise. Recently, there has been controversy in the United Kingdom over whether it is morally acceptable to combine human and non-human cells to produce a unique organism. In such an organism, called a **chimera**, cells of each species would exist side-by-side but function together for the health of the whole hybrid organism.

By contrast, when large amounts of DNA from one species such as a human are injected into the nucleus of an early embryo of a non-human species, all of the subsequent cells of that developing organism will have DNA from both species and the organism is considered to be a **hybrid** of both species.

Such combining of human and non-human DNA into one organism brings up basic ethical issues about what is means to be a human being.

Example of a Transgenic Organism

In 2001 scientists at the Oregon Regional Primate Center produced the first transgenic primate, named ANDi, a rhesus monkey that had a jellyfish gene in each cell of his body. They injected 224 rhesus monkey egg cells with the jellyfish gene. The egg cells were then fertilized with rhesus monkey sperm by **in vitro fertilization**. Forty fertilized eggs reached the stage at which they could be implanted. These 40 embryos were transplanted into the uteri of 20 female rhesus monkeys. Five of the monkeys became pregnant. From the five pregnancies, three monkeys were born alive. One of those monkeys, ANDi, was found to have the jellyfish gene in each of his cells, but the gene was not functioning. In jellyfish, the gene contains the information for the green fluorescent protein that allows the jellyfish to glow. But ANDi did not glow.

Can/Will Transgenic Humans Be Developed?

If this process eventually works, and transgenic non-human primates are produced, there is no theoretical barrier to producing transgenic humans. A transgenic human would not only contain a new gene from another species in every cell of his or her body, he or she would transmit that gene to any offspring. This modification is called **germline modification** because it affects not only the individual being modified, but also that individual's offspring.

Why Does Almost the Same DNA In Primates and Humans Result In Very Different Creatures?

The DNA in the human genome shows perfect identity with 99% of the chimpanzee genome. But if our genes are so similar chemically, why do we seem so remarkably different? We humans can build cities and develop political systems, write great literature and music. Perhaps most importantly, we have self-reflective consciousness. What is it, then, that this small 1% genetic difference can tell us about the difference between us and chimpanzees? Perhaps other factors affect the functioning of these genes in the two species. However, scientists also have recently discovered 49 regions in human DNA (called Human Accelerated Regions or HARs) that show faster nucleotide substitution than the rates of normal genetic evolution would predict. Within these regions scientists have found genes involved in advanced functioning such as the development of speech, overall brain volume, manual dexterity required for tool use and making, digestion of starch and lactose required in agricultural societies, and so on. As this exciting new area of research reaches deeper into our genome, we see that the differences between chimpanzees and humans are significant, even if based on only a small amount of our overall genetic material. That 1% may not be so insignificant after all.

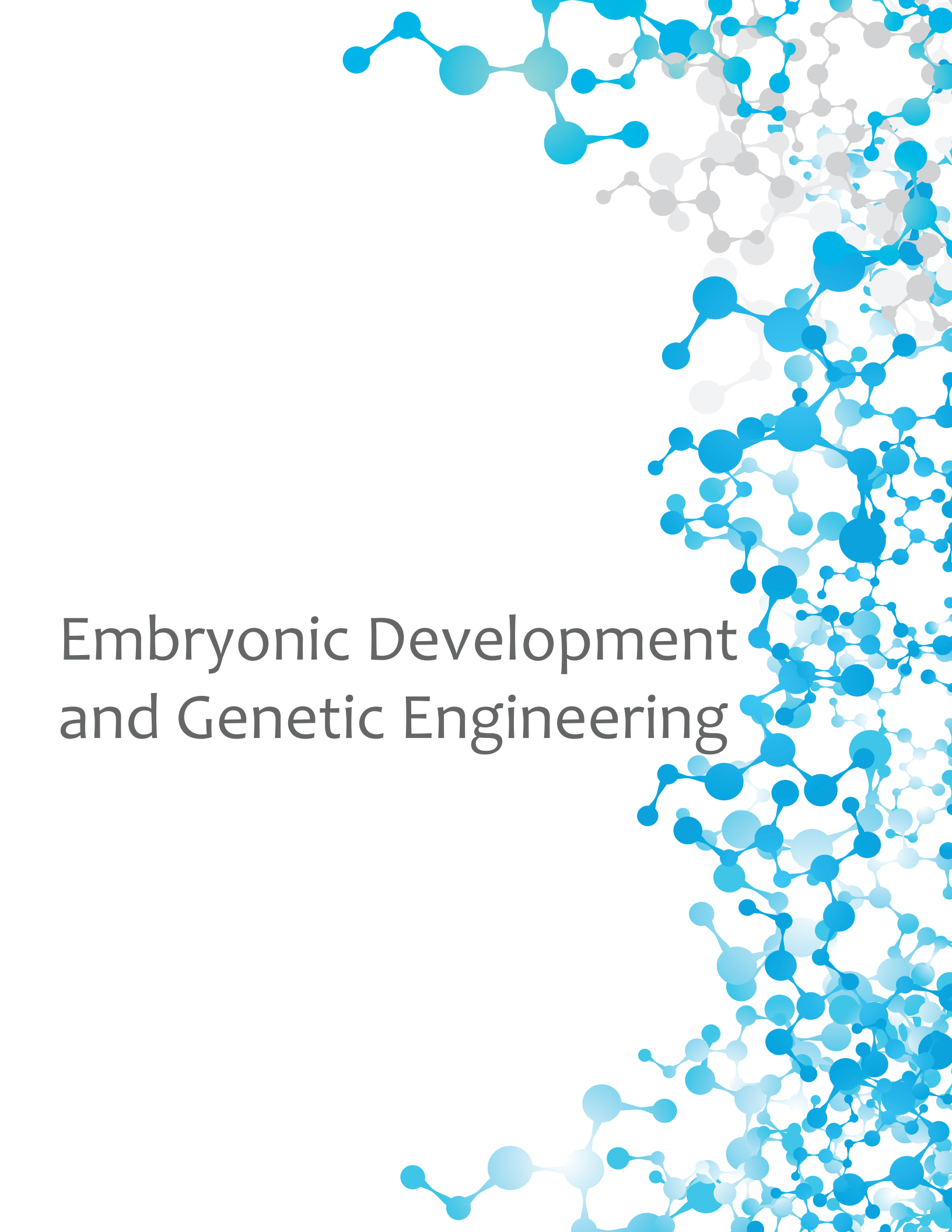
Animal Cloning

Finally, let's talk about cloning. A number of mammals have been cloned, including mice, sheep, cats, and mules. The process by which they were cloned is called **somatic cell nuclear transfer** or **SCNT**. Everything except egg cells and sperm are considered

somatic cells. Describing the technique is fairly simple, getting it to work is trickier. In somatic cell nuclear transfer, the nucleus containing its genome is removed from an egg cell. Then a somatic cell (that is, not a sperm or egg cell) is removed from a donor. The nucleus containing its genome is removed from the somatic donor cell and inserted into the egg cell whose own nucleus has been removed. An electrical current is applied to activate the newly nucleated egg, which may then begin to divide and produce an embryo. The embryo is implanted into the uterus of a surrogate mother (that is, a female of the same species but not the biological donor of the egg) and, if it fully develops, it becomes an individual genetically identical to the individual who donated the somatic nucleus. No primates have yet been cloned using this method due to technical difficulties. But theoretically, this method could be used to clone primates, including humans.

The Need for Greater Reflection on the Implications of Transgenic Research

Transgenics is a particularly difficult area of science and research for many people, including Christians. Much of the research is carried out with the goal of developing better plants, animals, or medical therapies. However, the research often moves ahead without sufficient reflection on the short- and long-term consequences of transgenic organisms and their products on ourselves, and on the environment in which we live and for which we are responsible. Since we believe that caring for Creation is our God-given mandate as human beings, it is our responsibility to advocate for careful reflection on the purpose of all transgenic research and the benefits and risks of transgenic organisms and their products on all aspects of the created order.



Embryonic Development and Genetic Engineering

Totipotency

A human egg can be fertilized by a human sperm either naturally by sexual intercourse or artificially in a laboratory (known as **in vitro fertilization** or **IVF**). A fertilized egg is called a zygote. This cell is **totipotent**. That is, it has the ability to divide and specialize into all the cell types found in a human being and into all of the cell types that form the extra-embryonic tissues, such as the placenta, umbilical cord and amniotic sac. In other words, a zygote has the potential to develop into, and support the development of, a newborn infant if allowed to implant into a functioning uterus. The process by which a zygote develops into an embryo, a fetus and then an infant is, essentially, a process of cell division and increasing specialization of different groups of dividing cells. An infant is composed of trillions of cells, most of which are highly specialized cells in muscles, nerves, the liver, the brain, and so on. The single cell that constitutes the initial zygote is completely unspecialized but has the potential to specialize.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

The fertilized egg divides into two cells. These two cells are still unspecialized and therefore are still totipotent. We know this because occasionally identical twins develop in the uterus when these two cells separate completely, divide to produce their own separate supporting tissues (placenta, umbilical cord, and so on), and eventually develop into two infants. Usually, however, the two-celled zygote remains intact and divides into 4 cells. We know that those 4 cells are still totipotent because, by the same mechanism just described for twins, the 4 cells can very rarely separate and develop into 4 individual infants.

Pre-implantation Genetic Diagnosis (PGD)

If the 4-celled zygote divides again into 8 cells, these cells are usually, if not always, still totipotent because identical octuplets have very rarely occurred. These 8-celled zygotes have also been used for a process known as **pre-implantation genetic diagnosis** (**PGD**). In fertility clinics, infertile couples can use IVF to create a zygote. Eggs from a woman are combined with sperm from a man to create zygotes. These zygotes then begin to divide to become embryos in a Petri dish in a lab. Sometimes, the couple asks to have genetic testing conducted on the embryos. PGD can be performed by carefully removing one of the cells at the 8-cell stage. The cell is then examined for genetic abnormalities. The remaining 7-celled zygote is allowed to continue to divide, usually resulting in what appears to be a perfectly normal infant. However, it also means that the single removed cell could also have the potential to form an identical twin of the

remaining 7-cells zygote, presenting an ethical problem for some who consider such a totipotent cell to be a potential human being.

Harvesting Embryonic Stem Cells for Research Purposes

When the embryo consists of about 150 cells, known at this stage as the **blastocyst**, the outer cells have differentiated (specialized) into cells that will only develop into supporting cells such as the umbilical cord or placenta. The remaining inner cells are now considered **pluripotent** rather than totipotent. They are still capable of differentiating into a wide variety of cells under the proper environmental and genetic influences. However, if separated from the outer cells, they cannot form a human infant if placed in the uterus and can no longer differentiate into the umbilical cord or placenta.

It is these inner cells that are so sought after by scientists who do research with human embryonic stem cells. The main source of such cells is extra embryos that are produced during IVF and are no longer wanted for producing human infants. Scientists separate out this inner cell mass from the outer cell layer, destroying the embryo in the process. These inner cells are then cultured in laboratory dishes as embryonic stem cells, which are used in experiments. Such destruction of human embryos raises important ethical problems for many people. Some feel that embryos are not fully human beings, using that claim to justify killing them for research to develop therapies to help others later. Others feel that the embryo has the status of a human being and that killing the embryo is immoral.

Alternative Sources of Human Embryonic Stem Cells

Somatic Cell Nuclear Transfer (SCNT)

In recent years scientists have tried to develop other sources of cells that have similar physical, physiological, and genetic characteristics to those of embryonic stem cells One of the first such sources involved making an embryo in the laboratory by a method called **somatic cell nuclear transfer** *(*or **SCNT***),* the same method by which animals such as Dolly the sheep have been cloned (some call the cell cluster produced by this method an **embryoid** rather than **embryo** to distinguish it from an embryo produced from the merger of a sperm and an egg. Some consider the use of embryoids to be less morally objectionable than the destruction of embryos as a source of stem cells for research). In this technique, a nucleus from an ordinary cell, like a skin cell, from a particular animal is joined with an egg cell from that same species whose nucleus had previously been removed. The new cell is stimulated, usually by electrical current, causing it to divide and develop into cells that look like, and function like, embryonic

stem cells produced by joining a sperm cell and an egg cell. Although this process has been successful in mammals like Dolly the sheep, it has not yet succeeded in humans. If it did succeed in humans, the result would be an embryo from which stem cells could be obtained. In this case, the stem cells would be genetically identical to the person who donated the nucleus for the somatic cell nuclear transfer (There is one exception: you'll recall from the previous discussion in Genetics 101, A Sidebar: DNA Outside of the Nucleus, that a very small amount of DNA is located not in the nucleus but rather is in the mitochondria. This DNA would be derived from the donated egg).

Induced-Pluripotent Stem Cells

More recently, scientists have discovered newer ways to construct cells in the laboratory that also have many characteristics similar to those of embryos. Instead of being created by the union of a sperm and an egg or by somatic cell nuclear transfer as described above, these cells are produced from an ordinary body cell like a skin cell, and not just its nucleus. These ordinary somatic cells are manipulated in the laboratory to lose their normal functions while taking on characteristics similar to those found in pluripotent stem cells. These now undifferentiated (unspecialized) cells can then be "induced" (that is, directed under special laboratory conditions) to re-differentiate (re-specialize) into a cell with a new, desired function that may be completely different from its original function. For example, a skin cell might be de-specialized, then re-specialized to function like a cell that now produces insulin, like certain cells normally found in the pancreas. This cell could then be replicated (multiplied) in the laboratory and could be transplanted into a person with diabetes whose own insulin-producing cells no longer work.

These **induced-pluripotent stem (iPS) cells** are considered by some to be more ethically acceptable for developing new therapies. For at least two important reasons, some consider this method more ethically acceptable than using leftover embryos after IVF or using SCNT: 1) these cells are not produced involving the use of natural embryos, produced from sperm or eggs that must be destroyed to harvest the stem cells and 2) this method does not require a supply of human eggs that would involve ethical problems such as risky drug therapy to produce the eggs from female donors and tempting women in need of money to take those risks and accepting payment for donating their eggs.

Human Engineering and Cloning

It has been proposed that combining stem cells and embryo cloning could allow **germline genetic engineering**. In this process, pluripotent stem cells would be removed from an embryo or embryoid (that is, stem cells produced by SCNT from iPS cells

described above). These cells would then be infected with a virus that contains whatever genes someone desired to transfer into the cells. The cells could be tested for successful incorporation of the desired gene or genes into the nucleus of those cells. Cells that have been successfully engineered could have their nuclei removed and transferred to denucleated eggs (recall that this is the somatic cell nuclear transfer technique described in Transgenics, Animal Cloning). The resulting cell could be allowed to develop into an embryo, then implanted into the uterus of a surrogate mother. If a fetus developed fully and was born, that newborn would carry the engineered genes in every one of his or her cells. If that newborn grew to adulthood and reproduced, his or her offspring would also possess these introduced genes. It is important to note that this process has not at this point been achieved in primates, including humans, but scientists are currently working to develop this process.

Population Genetics

How Can Understanding Genetic Racial Distinctions Be Helpful?

What is the genetic basis of race? Are Africans genetically different from Asians? Are there race-specific genes?

The more closely researchers examine the human genome the more most of them are convinced that the standard labels used to distinguish people by race have little biological meaning. Although it may seem easy to tell at a glance whether a person is Caucasian, African or Asian, when geneticists probe beneath surface characteristics and scan the genome for DNA hallmarks of race, that seemingly obvious conclusion disappears. Humans have spread out over the world in a relatively short time. Therefore there has simply not been enough time for the human species to divide itself into separate biological groups in any genetically significant way.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

Ninety-nine per cent of the human genome is similar. Within this similar portion, about 75% of all the genes come in only one allelic (monoallelic) form and are identical in everybody. Using the blood typing example explained earlier (Genetics 101, Practical Implications of Genetic Variation: The Story of Blood Types), such single allelic genes would be like having a blood type consisting of only the A allele, which would mean that everyone would have type AA blood. Because of this 99 % genetic similarity, individual variations among human beings are accounted for by the remaining 1%. Within that 1% of genomic variability, 85% exists within any local population, be they Italians, Kurds, Koreans or Inuit. Of the remaining 15%, about 7% variability can be found within any given continent while the remaining 8% in variability occurs between large groups living on different continents. That means two random Koreans may be as genetically different as a Korean and an Italian and there is not much additional variation within a continent compared to that between continents.

The way we measure human variation genetically is to look and find all the different allelic variations of a gene and then see what percentage of each variant of that gene occurs *within* populations and *between* populations. To illustrate this, let's look at blood types again. To examine genetic differences in blood types between two populations, start by examining the percentages of the allelic variations in those two populations. For example, the distribution of blood types among people from the Philippines is nearly identical to the distribution of blood types among the population of China. There is genetic variation within each population but not much variation between the two populations. Alternatively, 100% of Peruvian Indians have blood type

O, while among Blackfoot Indians, 82% of individuals have blood type A and 18% have blood type O. Within each of these two populations, there is little or no genetic variation, but there is a great deal of variation between the two populations.

Generally, most genetic variability can be found within populations but different populations can have very different degrees of variability. In fact, about 93% of all of the genetic variability that exits on this planet occurs within Sub-Saharan Africans. So, if there were a catastrophe that destroyed the rest of the world's population, 93% of the genetic variability in the world would still be present in Sub-Saharan Africans.

Medical Implications of Racial and Geographically-distinct Human Groupings

Sickle Cell Anemia Again

Are there genetic differences between human groups that are medically important? If race has any bearing on health at all, it may simply be a marker for the geographic origins of certain populations. In the Eastern Hemisphere, where it is thought humans have lived for at least 2 million years, differences that developed in skin colour were closely correlated with latitude and exposure to sunlight. The same pattern is not apparent in the Western Hemisphere, to where anthropologists suggest humans migrated only about 35,000 years ago.

This kind of knowledge can be helpful in understanding diseases such as sickle cell anemia. This disease is often found in African and Mediterranean peoples but also among immigrants or ancestors of immigrants from these regions to North America. The higher prevalence in these peoples is thought to be as least partly due to a health advantage for persons with the sickle cell anemia in battling malaria, which is endemic to those areas. Malaria parasites do not survive as well in sickled cells and therefore those infected by mosquitoes with the parasite that causes malaria may not develop malaria or may have a milder form of the disease. Sickle cell anemia is rarely seen in descendants of people from northern Europe, where malaria is rare or absent.

Genetic Ethnic Distinctions and Other Common Diseases

So the genetic link between sickle cell anemia and protection again illness caused by malarial parasites is clear-cut. But for the major diseases that cause or contribute to most deaths and disabilities, the genetic contribution is harder to pinpoint. For these diseases, such as heart disease, high blood pressure and cancer, multiple genetic mutations are thought to increase the susceptibility of some individuals, but environmental factors, such as diet and lifestyle, also play an essential role in

development of these diseases.

It's much harder to make the case that high blood pressure is a bigger burden in some ethnic groups because of their genetic makeup. For example, high blood pressure more severely affects people of African descent in Canada compared to those of European descent.[3] African-Canadians are also much more likely to die of stroke than Canadians of European descent. Some have speculated that African slaves who were better able to retain salt were more likely to have survived the deprivations of diet and sanitation on the slave ships transporting them to North America. If this were true, then the same genetic makeup that helped them survive, when passed on to their offspring, may have put later generations of black Canadians at risk for developing high blood pressure (also called hypertension). But there are also a number of societal and cultural factors that might predispose African-Canadians to hypertension including the stress of living in a prejudiced society, lack of access to health care, poor diet, etc. Complicating matters is that no one really knows which combination of genes is responsible for susceptibility to hypertension. It's likely that a large number of mutated genes may contribute to high blood pressure, but that not all patients may have all those mutations.

[3] Brophy, Kathleen Marion, Scarlett-Ferguson, Heather, and Webber, Karen S. Clinical Drug Therapy for Canadian Practice, Lippincott Willimas & Wilkins, 2010, p. 779.

Genetics for Guiding Therapy

Genetic Testing of Cancers for Improving Therapy

Improved Understanding of the Link between Genetic Mutations and Cancer Formation

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

Genetic testing can be very helpful in cancer diagnosis and treatment. Testing the genetic composition of tumours can help to determine what tumours are more effectively destroyed by certain therapies than other tumours, and thus which patients may benefit more from such therapies. It is now known that cancers develop because of genetic and epigenetic changes in normal cells. In virtually any organ in the body, normal cells can develop mutations, either spontaneously or under environmental influences, such as cigarette smoking. Some of these mutations involve genes that control cell division; some stimulate cell division and thus tissue growth; while others suppress or turn off the mechanism causing cell division.

Knowing What Mutations Disturb Normal Tissue Growth Can Lead to Targeted Cancer Therapies

Knowledge of the genes that most commonly mutate during cancer development is gained by testing the genetic composition of different types of cancers and studying which normal communication pathways for controlling cell division and tissue growth are disturbed. Once these genetic mutations are identified and their effect on cell division is understood, therapies targeted at these mutations and their effects can be developed in an effort to slow or stop tumour growth.

Such targeted therapy can be particularly helpful in two ways.

1) Conventional therapy treats cancer cells that are dividing, but many normal cells also divide each day to replace old, worn-out cells. Thus conventional therapy cannot discriminate between "bad" dividing cells (cancer cells) and "good" dividing cells (normal cells). If a particular tumour has a mutation against which an existing targeted treatment was developed, the person with that tumour is much more likely to benefit from that targeted treatment compared to someone whose tumour does not have the mutation.

2) Similarly, conventional chemotherapy can be quite toxic, resulting in hair loss, diarrhea, and infection, because it often kills normal cells of hair growth, the lining of the gastrointestinal tract, and immune cells along with cancer cells. By contrast, toxicity related to such targeted therapies tends to be much less

because such therapies do not kill normal cells but only tumour cells with the mutation against which the therapy was developed.

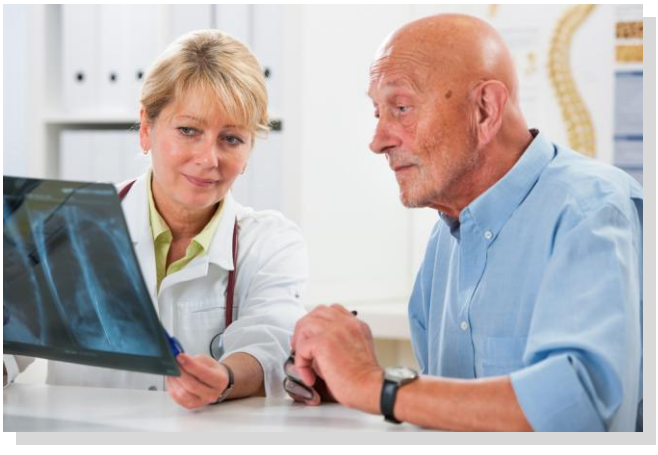
By testing tumours for specific mutations after they have been surgically removed, it is now possible to choose patients most likely to benefit from treatment while sparing those unlikely to benefit. Internationally recognized practice guidelines are beginning to recommend pretreatment genetic testing of tumour samples to identify patients most likely to benefit.

Testing for an Important Mutation in Breast Cancers: HER2/neu and Herceptin

An example of genetic testing for therapeutic guidance involves a mutation known as the **HER2/neu** mutation, found in 20-25% of breast cancer patients. This mutation is associated with more aggressively growing and more chemotherapy-resistant tumours. Patients whose tumours have this mutation will very likely benefit from a targeted treatment known as trastuzumab *(*the more easily pronounceable trade name is Herceptin*)* whereas those without the mutation will not. Herceptin is a manufactured antibody that blocks the protein of the mutated gene that contributes to uncontrolled growth of the tumour. In addition, Herceptin is generally very well tolerated, in large part because it affects only cancer cells and not normal cells of the body. Thus, this very expensive therapy is very cost-effective; it can be targeted only to those patients most likely to benefit and has little risk of toxicity when administered.

Genetic Testing of Cancer Patients (Rather than their Tumours) to Reduce Treatment-related Toxicity

Identifying Patients with Increased Risk of Therapy-related Toxicity



Genetic testing of the normal cells of cancer patients (rather than the tumour cells) can also be helpful in determining which patients are at risk for therapy-related toxicity based on the presence of different alleles on the patient's chromosomes. For example, patients with colorectal cancer are often treated with a combination of chemotherapy agents that include a drug known as irinotecan. Initially, safe doses of the drug were determined on the basis of an acceptable level of toxic effects regardless of knowledge

of the genetic makeup of the patients. However, it is now known that patients who are homozygous for a certain recessive genetic allele (i.e., known as UGT 1A1 *28) are predisposed to an increased risk of infection due to extremely low white blood cell counts experienced one to two weeks after the drug is administered. This appears to be a consequence of reduced metabolic processing and reduced disposal of irinotecan by the body, associated with the presence of the allele on both chromosomes.

Because of this genetic information, patients who are at less risk for infection using conventional doses of irinotecan (i.e., those who are only **heterozygous** for the allele or who do not carry the allele at all) can now be identified. Clinical studies are underway to test the safety and efficacy of cancer therapies in such genetically-selected patients so that the dose and frequency of therapy is tailored to the presence or absence of the toxicity-predicting allele.

Glossary

The Genetic Science Glossary

Adenine (A) One of the four bases, or building blocks, found in DNA. A d e n i n e Analogy: Adenine is one of the letters in the four-letter DNA alphabet. Adenine (A), Guanine (G), Thymidine (T), and Cytidine (C) are the four different molecules base molecules that compose DNA. The smallest unit of DNA consists of one base molecule, one sugar molecule, and one phosphate molecule. Since the DNA molecule is a double helix, each base must be paired with one other base to form the "rungs of the ladder" that makes up the helix. Adenine is always paired with Thymidine and Guanine with Cytidine. When a cell prepares to divide, the DNA unwinds its helix and each strand of DNA is seen as a linear series of codons (ex: ACC; GTC; AAT, etc). Each codon consists of three base molecules that form the code linked to a particular amino acid (see definition of codon below). The sequence of different combinations of codons on the chromosomes determines what amino acids will be incorporated into specific proteins according to a specific order. Thus, the DNA determines the way proteins are built up for that person's cells.

Allele Alternate forms of a gene. We receive two copies of each gene, one copy from each parent. Some genes have more than one form of a trait (e.g., brown eyes or blue eyes). These alternative forms of a gene are called alleles. Allele analogy: Genes are like recipes for proteins. An allele is like a variation of that recipe. There could be a recipe for oatmeal cookies that has two variations: one with raisins and one without. A gene for skin colour could have two variations: one with freckles and one without.

Amino Acid There are 20 amino acids. Each amino acid is one of 20 kinds of building blocks that form proteins. Protein shape and function are determined by the combination of the amino acids. The order of bases in DNA, the genetic code, determines which amino acids make up each protein and in what order. If the nucleotide bases are the letters of the DNA alphabet, then each codon (three nucleotide combination) is a word. The amino acid is the meaning of the word. All the words together make a sentence, like all the amino acids together make a protein.

Anticipation	A genetic disorder tending to increase in severity and with earlier onset as passed on through generations.
Apoptosis	Programmed cell death
Artificial Selection	Evolution caused by humans through choosing and breeding specific organisms based on the expression of a desired trait. Example: If a farmer wants to grow bigger tomatoes, selecting and then planting only the seeds of the biggest tomatoes the farmer grows might select the tomatoes with the genes that cause them to be larger. This could result in a crop of tomatoes that are bigger.
Autosomal Dominant	A gene on the autosomes (non-sex chromosomes) that is always expressed even if only one copy is present. Example: The brown eye allele is autosomal dominant. If you have a gene for brown eyes and a gene for blue eyes, brown eyes will be expressed.
Autosome	Humans have 23 pairs of chromosomes: 46 chromosomes in total. An autosome is one of the 44 chromosomes that contain genetic information that does not determine sex. There are a total of 46 chromosomes, 44 autosomal plus two sex chromosomes (X and Y).
Base	One of the molecules, or building blocks, that form DNA and RNA. The bases consist of adenine, guanine, cytosine, and thymine. Base Analogy: If a genome is the encyclopedia containing all the information necessary to produce an organism, DNA is the alphabet in which the encyclopedia is written. DNA is a four-letter alphabet, and each base is one of those letters.
Base Pair	Two of the four bases that are held together along the double helix DNA molecule. The four bases are: adenine (A), thymine (T), cytosine (C), and guanine (G). A always binds to, or pairs with, T and C always binds to C.
Behavioural Genetics	The study of how genes may influence behavior.
Bioinformatics	Use of advanced computing techniques to analyze genomic data.
Blastocyst	A blastocyst is an embryo that has not yet implanted, containing about 150 cells. The blastocyst is a hollow ball consisting of an outer layer of cells that will specialize to become extra-embryonic tissues

such as the placenta, umbilical cord and amniotic sac. It also consists of an inner cell mass that will specialize to become all the cells of the developing fetus. It is from the inner cell mass of a blastocyst that embryonic stem cells are derived.

Blastomere A blastomere is any of the cells that form an early embryo, after the first division of a fertilized egg. For example, at the 8-cell stage, a single cell, a blastomere, can be removed for pre-implantation diagnosis. Additionally, blastomeres can he separated at an early stage and each blastomere may develop into a separate embryo.

Cancer Diseases involving the unregulated division of abnormal cells within the body. All cancer is genetic in the sense that mutations in the genes that regulate cell division are the cause of cancer.

Carrier Someone who has an unexpressed recessive genetic trait. Example: A person with brown eyes may be a carrier of the trait for blue eyes. Two brown-eyed parents who are carriers of blue eyes may have a child with blue eyes.

Catalyst A catalyst is something that speeds up a chemical reaction. Often enzymes catalyze chemical reactions.

Chimera An organism that has cells with different genetic makeup. These organisms may be transgenic, that is carrying gene sequences from more than one species.

Chromosome The cellular structure containing the DNA molecule carrying genes. There are 46 chromosomes in the human genome. Each person receives 23 chromosomes from each parent: 22 autosomal chromosomes plus an X chromosome from the mother and either an X or a Y sex-determining chromosome from the father. Each chromosome has two arms. The shorter arm is referred to as "p," the longer arm as "q." Chromosome Analogy: The genetic material is organized into structures called chromosomes. If the genetic material of an individual is a set of encyclopedias, each chromosome is a volume of that set. Each chromosome contains information that is different from the information contained by the other chromosomes.

Clone An exact (or nearly exact) copy made of a DNA segment, a whole cell, or a complete organism. Cloning of mammals has been accomplished through the process of somatic cell nuclear transfer. In nature, cloning occurs through blastomere separation, when, at a very early stage of development the blastomere separates and forms two distinct and developing pre-embryos. We refer to these clones as identical twins.

Codominance Where two different alleles for a genetic trait are both expressed. This is the case for the alleles for type A and type B blood. The person is said to be AB because both the A and B allele are expressed.

Codon Sequences of three nucleotides (bases) code for amino acids. These triplets are called *codons*. Example: the nucleotide triplet ACC codes for the amino acid serine. The amino acids in turn make up the proteins, which in turn do the work in developing and maintaining a particular organism according to its genetic code. Codon Analogy: If nucleotides are the letters in the DNA alphabet, then codons are the words. Each word has a specific meaning, being an amino acid. Some words are synonyms, coding for the same amino acid.

Complex Trait Many traits and their development involve the expression of more than one gene. In addition many genes interact with the environment to create a trait. These traits are considered "complex."

Conserved Sequence A base sequence in a DNA molecule that has remained essentially unchanged throughout evolution. A conserved sequence could he found in one organism, such as a snail, and another organism, such as a human, and would be the same or very similar.

Cytosine (C) One of the four bases, or building blocks, in DNA.

Deletion A loss of a part of the DNA from a chromosome. This can lead to a disease, or to an abnormality leading to disability.

DNA Deoxyribonucleic acid. The molecule that encodes genetic information. DNA is a double stranded molecule held together by hoods between base pairs of nucleotides. There are four bases in DNA: adenine (A), guanine (G), cytosine (C) and thymine (T). Generally, A only bonds to T, and C to G. DNA Analogy: DNA is the

genetic material. It contains a recipe for the characteristics of a human being. If all genetic material can he considered a set of encyclopedias, the DNA is the words on each page. There are only four "letters" in the DNA alphabet but, just like the 26 letters of the English alphabet, the DNA letters can be put together to form words. Each volume of the set could he considered a chromosome.

DNA repair genes Genes that code for proteins that function to correct errors in DNA base sequences.

Dominant allele An allele that is always expressed, even if only one copy is present. For example, the Huntington allele is dominant. That is, you will get the disease even if only one of the two alleles has the defective gene.

Embryonic Stem Cells A cell found in embryos that can replicate indefinitely and transform itself into other types of cells.

Enzyme An enzyme is a protein that catalyzes reactions. Many of the functions in the human body are chemical reactions (digestion, growth, transmitting signals along nerves). These chemical reactions happen slowly (some reactions would take years) unless an enzyme is present to speed up, or catalyze, the reaction. For example, the enzyme called acetylcholinesterase catalyzes (speeds up) the breakdown of the neurotransmitter acetylcholine. Acetylcholine is released by nerve cells and received by muscle cells, causing the muscle cells to contract. If acetylcholinesterase did not speed up the breakdown of acetylcholine, all your muscles would contract continuously.

Epigenetics The study of how environmental factors change gene function without changing gene sequence.

Epistasis Genes from one location interacting with genes at another location, affecting their expression. For example, if a dog has the gene for brown hair, but does not have the gene for expressing hair colour, the brown hair will not be expressed.

Eugenics The manipulation of the gene pool through artificial selection or genetic engineering with the purpose of improving a species.

Fingerprinting	Every genome is unique because of the accumulation of mutations over time. Fingerprinting in genetics refers to flapping a set of variations due to mutation in someone to uniquely identify them. This process is useful in establishing the presence of a suspect at a crime, establishing paternity, and identifying accident victims.
Gamete	These are mature male or female reproductive cells with a full complement of chromosomes (23).
Gene	The fundamental unit of heredity. An ordered sequence of bases (nucleotides) that encode for specific proteins and the functions those proteins will carry out.
Gene Expression	The process whereby the cellular machinery converts a gene's encoded instructions into the structures and operations of a cell.
Genetic Discrimination	Prejudice against those who have or are likely to develop a genetic disorder.
Genetic Predisposition	A genetic trait that leads to susceptibility to certain diseases. These diseases may or may not actually occur.
Genome	All the genetic material in an individual cell of an organism.
Genomics	The study of genes, their origins, and their functions
Genotype	The genetic constitution of an organism. This includes the traits an organism carries and the traits an organism expresses. This term contrasts with phenotype, which is an organism's measurable traits, or only the traits an organism expresses.
Germ Cell	Sperm and egg cells and their precursors. These are the only cells that contain 23 rather than 46 chromosomes.
Germ Line	The continuation of genetic information from one generation to the next.
Green fluorescent protein	Green fluorescent protein (GFP) is a protein produced by a jellyfish. CFI fluoresces (glows) bluish-green. The gene for GFP has been isolated and inserted into the cells of other organisms, including mice, pigs and monkeys. The expression of certain genes in these transgenic animals can he monitored by examining the pattern of GFP fluorescence.

Huntington Disease	Huntington Disease (HD) is a fatal, progressive, neurodegenerative disease that usually appears in adults between 35 and 50 years of age. HD is inherited as an autosomal dominant trait. An individual who has the HD allele has a 50% chance of transmitting that allele to a child. The HD allele arises as the result of an expansion of a 3-base repeat (CAC) in the gene. Normal individuals have up to 26 repeats. Individuals with over 40 repeats will likely develop HD). A higher number of repeats correlates with earlier onset of symptoms.
Imprinting	A non-permanent alteration of a gene that varies depending upon whether the alteration takes place in a male or a female. In some cases, the particular disease one inherits depends on whether the allele is inherited from the mother or father. For instance an offspring will get either Prader-Willi or Angelman syndrome depending on whether the missing portion of chromosome 15 is inherited from the mother or father.
Junk DNA	A better term for "non-coding" DNA. These are vast stretches of DNA that do not code for the expression of amino acids. They may have regulatory functions, structural functions, or functions we have yet to discover.
Locus	The position of a specific gene on a chromosome.
Messenger RNA (mRNA)	RNA that functions as a blueprint for manufacturing specific sequences of amino acids to produce proteins.
Mitochondrial DNA	Genetic material found in mitochondria. Mitochondria are involved in the production of energy in a cell. Mitochondria (and, therefore, mitochondrial DNA) are inherited only from one's mother.
Monogenic Disorder	A disorder caused by a mutation of a single gene.
Mutation	Any heritable change in DNA sequence.

Nematod Nematodes are microscopic worms generally found in the soil. These structurally simple organisms (adult nematodes are comprised of fewer than 1000 cells) are useful model organisms for scientists studying the genetics of development.

Nuclear Transfer A procedure in which the nucleus of a cell is removed and placed within an oocyte, which then uses the new genetic information in the development of a new organism. This is how cloning in mammals was accomplished.

Nucleotide A subunit of DNA or RNA consisting of a base (A, C, T or C), a phosphate molecule and sugar molecule. These units link to form the DNA or RNA molecule.

Oncogene A gene that, when mutated, is associated with the onset of cancer. Many oncogenes are involved in the control of the rate of cell growth.

Oocyte A female gamete before it matures.

Pedigree A genetic family tree that shows how a particular genetic trait or disease has been transmitted.

Penetrance The probability of a gene or genetic trait being expressed. Complete penetrance means that a particular genotype always results in a particular phenotype. Incomplete penetrance means that a particular genotype (such as polydactyly) is expressed in only a portion of those individuals with that genotype.

Peptide Two or more amino acids joined together.

Pharmacogenomics The study of the interaction between a person's genetic profile and their interaction with specific drugs.

Phenotype A measurable characteristic (blood type, height) that is determined by, or influenced by, expression of a particular gene(s). An organism may have the genotype for blue and brown eyes, but only brown eyes are expressed (brown eyes are autosomal dominant). In this case, the phenotype is for brown eyes.

Pluripotent The potential of a stem cell to develop into more than one type of mature cell depending on environment.

Polygenic	A phenotypic trait created through the interaction of two or more genes.
Polygenic Disorder	Genetic disorders that rely on the combined action of alleles of more than one gene. Although these diseases arc inherited their actual expression is more complex than in monogenic disorders.
Promoter	A site on the DNA strand to which RNA polymerase will bind and begin the process of transcription, the first step of gene expression.
Protein	Large molecules made up of amino acids in specific sequences as determined by the corresponding gene. Proteins provide the structure, function, and regulation of cells, tissues and organs.
Recessive	A gene that will only be expressed if there are two copies of the same allele. (Note: only one copy is required for males on the sex chromosomes.) A recessive allele will only be expressed if the organism does not also have a dominant allele.
Recombinant DNA technology	A procedure for splicing genes from different organisms outside the structure of the cell and then inserting the altered sequence into a cell where it can replicate.
Regulatory Sequence	A DNA sequence that controls gene expression.
Ribonucleic Acid (RNA)	There are several types of RNA. RNA is the blueprint taken from the template DNA, which is used in the construction of amino acids, which in turn determine the shape and function of proteins.
Sex Chromosome	The X or Y chromosome in human beings that determines the sex of an individual. Females have two X chromosomes while males have both an X and a Y chromosome. These sex chromosomes comprise the 23rd chromosomal pair.
Somatic Cell	Any cell in the body except gametes and their precursors.
Stem Cell	Undifferentiated cell. They are found in embryos, placental tissue and bone marrow. Since the most useful lines of stem cells are derived from embryos, research on them is controversial.
Suppressor Gene	A gene that can suppress the action of another gene.

Telomere The end of a chromosome. Telomeres are involved in the replication and stability of DNA molecules. They are therefore thought to the involved in the process of aging.

Thymine (T) One of the four bases, or building blocks, in the DNA sequence.

Totipotent In mammals, totipotent cells have the potential to differentiate into all the cells of an adult organism as well as all the cells of the extra-embryonic membranes.

Transcription The creation of an RNA copy of DNA that may then he used to direct the binding of amino acids to one another, thus creating a protein.

Transfer RNA (tRNA) RNA that uses the information from mRNA (messenger RNA) to position amino acids in a particular order, allowing them to be bound together to create proteins.

Transgenic An experimentally produced organism in which foreign genetic sequences have been added to the germline. ANDi is a good example of a transgenic organism.

Zygote A zygote is the result of fertilization of an egg cell with a sperm cell

Appendices

Appendix A

The Anglican Church on Bio-ethical Issues

The Anglican Communion and the Anglican Church of Canada rarely make formal statements that may be described as "the official position of the Anglican Church" on ethical and doctrinal issues. Nevertheless, various conversations, statements, and resources may help us to understand the mind of Anglicans, both local and global, on a wide range of bio-ethical issues that they have engaged in order to think about them with clarity and integrity. The statements below are a sampling of what Anglicans have been saying about bioethics, genetic technology and faith.

I. What have we been saying?

The Anglican Communion has dealt with issues related to human life and bioethics since at least the 1930s. The Lambeth Conference of 1938 for example declared its "abhorrence of the sinful practice of abortion." The statement marked where the mind of the church was at that point in time. It did not end further reflection and debate on the subject. Technological developments since then have made questions about the beginning and end of human life more complex.

The 1978 Lambeth Conference acknowledged its awareness of these changes and called for studies that "emphasize the sacredness of all human life, the moral issues inherent in clinical abortion, and the possible implications of genetic engineering."

More recently, the Archbishop of Canterbury, Rowan Williams, has made some comments on the issue of the treatment of human embryos in scientific research. Archbishop Williams says:

[Christians] have many profound questions about the status of the human embryo and the proper ethical framework within which scientific research takes place...science in itself is never going to be able to tell us what the right thing is for us to do--it can tell us only what's possible.

And, despite the way some people talk in this debate, there really is a difference between what is possible, and what is right.

The Anglican Church of Canada has also made statements opposing the misuse of "excess embryos" created as a result of IVF procedures. Creating embryos solely for the purposes of experimentation, the Church observed, is "morally repugnant" because it treats the unborn as an "object for adult consumption."

Genetically modified organisms and foods, too, have been the subject of discussion in the ACC since the late 1980s, citing concerns about inadequate testing and the economic injustice suffered by local and international farmers.

II. What are our theological resources?

When we discuss issues of faith and genetics, a key question emerges: "What does it mean to be made in the image of God?" Not only are there decisions to be made about the nature and content of human life, but also about what role image-bearing creatures take in and toward the rest of creation. Are we "wreaking havoc with the order of creation" by manipulating genetic and developmental processes? Are we concerned about bringing "the year of Jubilee" to the rest of the created order?

Many Anglicans are convinced that the "image" we bear has its source in the Triune God and that, at its roots, human vocation has to do with reminding the created order of its fullest joy, namely worshiping God in spirit and in truth. This faithful God became incarnate in Jesus Christ to liberate Creation from sin. The incarnation of the Word encourages us to be self-reflective about appropriate use of genetic technologies and other scientific developments.

(Thanks to Rob Walker for researching and drafting the appendix)

Appendix B

Positions of the Christian Reformed Church (CRC) on Genetic Technologies and other Relevant Topics of Biotechnology and Bioethics

Compiled by James J. Rusthoven, representative for the CRC, Biotechnology Reference Group, Canadian Council of Churches

Obtained from the website of the CRC regarding its beliefs and positions on life issues.

Stated Positions of the Christian Reformed Church Regarding Ethical and Theological Issues in Bioscience and Genetic Engineering

Introduction

Over time, the Christian Reformed Church has stated its position on a variety of contemporary topics. The following is a summary of the denomination's doctrinal and ethical positions as stated over the years by synod regarding bioscience and genetic engineering.

This précis offers accurate and concise descriptions of the positions of the CRC. For full reports and exact statements of the denomination's position on a particular issue, the reader should look to the references provided. The material has been updated through the decisions of Synod 2011.

General Statement on Relating Synodical Decisions to the Church Confessions

Synod 1973 appointed the Committee on Synodical Decisions and the Confessions. Its mandate involved two tasks: (1) to compile materials for a publication containing pertinent synodical decisions on doctrinal and ethical matters and (2) to present a clear statement as to how such synodical decisions are related to the confessions. Synod 1975 subsequently approved the original version of the material in this section and adopted the following recommendations of the study committee regarding the relationship of synodical decisions to the confessions:

1) The Reformed Confessions are subordinate to Scripture, are accepted as a true interpretation of this Word, and are binding on all office bearers and confessing members of the church.

2) Synodical pronouncements on doctrinal and ethical matters are subordinate to the confessions and are "considered settled and binding, unless it is proved that

they conflict with the Word of God or the Church Order" (Art. 29). All office bearers and members are expected to abide by these decisions.

3) The confessions and synodical pronouncements differ in their extent of jurisdiction, in their nature of authority, in their distinction of purposes, in the measure of agreement expected, and in their use and function.

4) The use and function of the synodical decisions (i.e., interpretation of the confessions, pronouncements beyond the confessions, adjudication of a particular issue, testimony, guidelines for further study or action, or pastoral advice) are explicitly or implicitly indicated by the wording of the particular decision itself.

For the full report of the 1975 committee and synod's response to it, see Acts of Synod 1975, pages 44-45 and 595-604.

Study of Ethical and Theological Issues involving Bioscience and Genetic Engineering

In response to overtures about abortion and pregnancy-related issues as well as ethical and theological issues in bioscience and genetic engineering, Synod 1999 appointed a study committee "to examine the biblical/theological/ethical issues raised by the increasing capabilities and recent discoveries in bioscience and genetic engineering" (Acts of Synod 1999, p. 578). This study committee reported to Synod 2003 with guidelines for dispensing pastoral advice concerning life issues arising from new biotechnologies including genetic engineering. Synod recommended the committee's report to the churches for study and reflection and encouraged members "to engage governmental agencies regarding the pursuit of policies that are consistent with the guiding precepts adopted by synod and outlined in the report" (Acts of Synod 2003, p. 644).

A summary of the guidelines for pastoral advice concerning life issues were published as follows (from Acts of Synod 2003, pp. 632-35, 639, 643-44, found at www.crcna.org/pages/synodical.cfm):

- We must not recommend rules that bind the conscience in disputable matters. To do so would violate personal Christian liberty. Instead, we should prescribe only where God's will is clear. Scripture is clear that every human being is created in the image of God and is precious to God.

- Procreation should be kept within the context of the male-female, two parent, covenantal relationship of marriage.

- Although it is fitting for married couples to want to have children, and it is a blessing to have children, there are limits to the lengths to which couples may go in order to have children. Infertility is a result of the fall, and we may attempt to reverse this but only through morally acceptable means.

- While Scripture does not explicitly teach what moral protection the unimplanted human embryo deserves, it is clear implicitly that as a unique human life it warrants significant human protection.

- Recognizing the horrific nature of rape and the complex circumstances facing a rape victim, she is not necessarily morally culpable if she takes a morning-after pill. The focus of ministry in such circumstances should be on the compassionate care for the woman.

A full discussion of evidence and positions regarding the background of a broad range of procreative and genetic issues deliberated by the study committee are found in the Agenda for Synod 2003, pp. 275-313. As there was a majority report and a minority report, the main points of both and the final approval or rejection of their points are discussed in the Acts of Synod 2003, pp. 632-35, 639, 643-44. The final guidelines were distilled primarily from an earlier set of recommendations from the majority report of the study committee. However, some earlier recommendations were not approved, such as 1) a more explicit statement regarding a moral imperative to create human embryos in vitro only when every embryo so created will have an opportunity for implantation and 2) a statement condemning as morally wrong the intentional destruction of a human embryo except as a necessity to save the life of the mother after implantation.

These omissions from the guidelines as well as the more general nature of the final guidelines reflect significant differences of views on many of these issues among committee members. From this it follows that they also likely reflect the heterogeneity within the denominational membership on many life issues. The denomination continues to reflect on these issues through various forums including solicited and unsolicited denominational publications with which denominational members can work out the continued commitment to keep themselves informed and keep such discussions alive and relevant over time.

Appendix C

An Orthodox Appendix for the Faith and Genetics Curriculum

1. All Orthodox discussions of the weighty matters introduced by this curriculum must be informed by Orthodoxy's fundamentally 'theocentric' anthropology.

2. We must start by asking basic questions about what it means to be a human person. In the Orthodox Church's understanding, human persons are 'defined' by their having been created by God and by their bearing his image[1] which is indelible. However, despite this lofty point of origin, human beings in a very real sense experience life in this world as a 'fallen' reality, given that they encounter sin, sickness, suffering and death on a daily basis.[2]

3. God, out of his infinite love for the whole human race,[3] affords human beings a 'way out' of the dilemma of their fallenness. Through the life, death, passion, resurrection and ascension of his Beloved Son Jesus ("like us in all things except sin"[4]), God announces the imminence of his Kingdom,[5] addresses us with words of life,[6] visits and redeems us[7] in the dark places of our present lives,[8] and summons us to begin leading new and eternal lives[9] as his sons and daughters[10] within a veritable new creation.[11] To accomplish this goal, God empowers us to be refashioned in Christ's likeness[12] by pouring out God's Holy Spirit upon us and upon the whole of creation.[13]

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[1] Genesis 1:27. Humanity's creation is understood to be the work of God the Holy Trinity. The Image according to which humans have been formed in creation is that of the pre-incarnate Logos-Son of God who is the perfect Image of his Father (Colossians 1:15). God's "agent" in effecting humanity's creation "in the Image" is the Spirit or 'breath' of God which God "breathed into [Adam's] nostrils" so that he "became a living being" (Genesis 2:7).
[2] See Isaiah 35:10b LXX, quoted in the Byzantine-rite funeral prayer "O God of spirits and of all flesh . . ."
[3] Described in the original Greek of numerous Orthodox liturgical texts as the *philanthropía* of God who is thus the *philánthropos* –" lover of the human race"
[4] Hebrews 4:15
[5] Mark 1:15
[6] 1 John 1:1 and John 1:4
[7] Luke 1:68b
[8] Luke 1:79a
[9] John 17:3
[10] John 1:12 and Galatians 3:26
[11] 2 Corinthians 5:17 and Apocalypse (Revelation) 21:5
[12] 1 Corinthians 15:49
[13] Joel 2:28-29, quoted in Acts 2:17

4. Along our pilgrim way towards becoming "new creatures in Christ"[14] God summons us: a) to lead personal lives of self-denial and ascetic struggle, taking up our cross daily[15]; b) to learn how to love and serve all members of God's human family[16]; and c) to grow into ever-deepening loving fellowship[17] with our sisters and brothers in the one Body of Christ.[18]

5. Bearing the foregoing considerations in mind (points #2 – 4), there are a number of issues arising out of this curriculum which need to be addressed more specifically from an Orthodox Christian perspective. Orthodox communities using this curriculum need to be aware of these issues and to strive for their discussions on these matters to be informed wherever possible by an Orthodox *phronema*[19] (see points #6 – 10 below).

6. With specific reference to point #2 above, the eastern patristic tradition discerns a definite correlation between humanity's creation "in the Image" and the capacity for human persons to exercise freedom of choice (even in their apparently 'fallen' state). St Gregory of Nyssa in his treatise *On Virginity* observes that "being the image and the likeness . . . of the Power which rules all things, [humanity] kept also in the matter of a free-will this likeness to Him whose will is over all."[20] The Prodigal Son[21] (a beloved subject of Orthodox reflection every year in the immediate pre-Lenten period), despite his living in a literal pigsty of degradation and despair, nonetheless was able to "come to himself" and decide freely to "arise and go to [his] father.[22]

   From this perspective, Orthodox anthropology remains critical of any type of thorough-going determinism and therefore reacts forcefully against contemporary opinions such as those reported by sociologist Alex Mauron, to the effect that "the genome is construed as the ontological hard core of our being . . . the secular equivalent of the soul."[23]

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[14] Galatians 6:15
[15] Matthew 16:24
[16] Matthew 5:43-44 and 25:40
[17] *Koinônía* ("communion")
[18] 1 Corinthians 10:16-17
[19] "mindset"
[20] (St.) Gregory of Nyssa: *On Virginity*, chapter 12; accessed on-line at www.newadvent.org/fathers/2907.htm .
[21] Luke 15:11-32
[22] Luke 15:17-18
[23] As quoted for discussion purposes in the Introduction to the curriculum's theological chapter on "Genetics, Faith, and Human Dignity"

7. Creation according to God's image and likeness (point #2) also moves Orthodox theology to understand and describe human life as a "sacred gift"[24] freely bestowed on each one of us, on our families and on the wider human community by the God of love who is *philánthropos*[25] (point #3 above). Such a theocentric (and communitarian) point of view will condition Orthodox attitudes to a whole host of contemporary moral issues which the wider society tends to treat as falling more or less within the purview of the autonomous human subject (for example: prenatal genetic diagnosis, recourse to new reproductive technologies, abortion, assisted suicide, euthanasia and others).

   Attitudes towards the lives of actual or potential "special needs children" in particular (whether before their conception, during their gestation or after their birth) need to be informed by an outlook which views every child, no matter how 'imperfect' he or she may appear to be (genetically or otherwise), as being a gift from God and therefore as having the potential both for giving and receiving love.

8. There can be no doubt that being afflicted by (and living with) a chronic handicapping and/or life-limiting illness imposes tremendous burdens and much real hardship on the person so afflicted as well as on their parents, families and other caregivers. To seek to avoid such burdens at any cost might seem, at first sight, to be nothing more than a normal, totally understandable and very human reaction.

   However, Orthodox Christians who may be contemplating having recourse to one or other morally questionable 'new technologies' (e.g. prenatal diagnosis for abortion of fetuses with genetic disorders) do well to bear in mind and reflect upon the whole ascetical dimension of traditional Orthodox Christianity. As alluded to above under point # 4(a), we believe that we are enjoined by Christ to "deny ourselves" and "take up our cross"[26] in order to follow Christ and become his true disciples. Viewed in this way, disability and its attendant suffering, embraced willingly for Christ's sake and in witness to the Gospel, can become a way into the Kingdom for both disabled persons and their caregivers.

9. Community support (financial, material, instrumental and moral) for people and families living with disabilities (whether genetic or acquired) can go a long way towards lightening the burdens borne by these persons and their caregivers. Hopefully, Christian communities in particular would feel a special sense of commitment to those in their midst who must deal with chronic illness in themselves

[24] Cf. Fr. John Breck: *The Sacred Gift of Life: Orthodox Christianity and Bioethics* (Crestwood, NY: SVS Press, 1998)
[25] See note 3 above
[26] Matthew 16:24 and parallels

or family members.[27] Although most traditional Orthodox cultures have emphasized the virtue of providing community assistance to those in need within the extended family or village setting, these village-level communal strengths often fail to be carried over into the congregational life of the average cosmopolitan North American Orthodox parish.

10. Many Orthodox Christians will resonate with the observation that scientists making certain "futuristic" advances in genetic and reproductive technology can appear to be "playing God." In this respect (and as a concluding observation to this 'Orthodox appendix'), we should attend to these words from the curriculum's opening chapter on "Genetics, Faith, and Human Dignity." "Perhaps the use of this term ['playing God'] has to do with the belief that we are not meant to do certain things, even though we can. . . . Perhaps we are supposed to think about the kind of world we want to live in and use the knowledge we have at our disposal in a *modest and resourceful way* toward achieving that end [service to humankind]. . . . *With every increase in our knowledge combined with increasing ability to use that knowledge however we wish comes a commensurate increase in moral responsibility*."

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August, 2012

[27] See points #4(b) and 4(c) above

Appendix D

The Presbyterian Church in Canada

The Presbyterian Church in Canada affirms that the rule of its faith and life is the Scriptures of the Old and New Testaments which are the standard by which all church doctrine, policy and pronouncements are to be evaluated and tested. It also affirms that in the pages of the written word the nature of the God who was in Christ stands revealed. In the light of this revelation the church formulates its Doctrine, some parts of which bear directly on the concerns of the Genetics Curriculum. Among these are the following.

The Sovereignty of God

Scripture witnesses to a sovereign God who is the Creator and sustainer of that which is. Created to live in conformity to God's sovereign will, in all our activity we are called to reflect God's creating, loving and sustaining activity.

Stewardship

We have been mandated to live before our Creator as responsible stewards of that which has been entrusted to our care. Thus we intervene in and give shape to the natural order so as to protect, sustain and promote life. Life is a gift from God, a gift we are called to safeguard. In this process the creating, sustaining activity of a loving God is revealed, a God who wills to overcome all that mars or destroys that fullness of life that is his intent for his creation. (1)

The Image of God

As stewards of God's creation and servants of his purposes we are created in his image and likeness. (2) This means we have been created with an intelligence that can be used to discover and to understand the mechanisms of the natural order. This enables us, through progressively creative activity, to exercise a responsible stewardship of the created order and also to take responsibility for the life of the neighbour, which life has been entrusted to our care. In our intelligence, sense of responsibility and our freedom, we reflect God's image in us.

Human Dignity

The Image of God in which we are created reflects a relationship with our Creator that we cannot escape and a relationship from which we derive our dignity. Human dignity is thus an alien dignity. It reflects God's valuation of the humanity of God's creation and is

therefore a dignity that is to be affirmed and honoured. Hence, service to God requires that God's care and concern for the well being of all people be reflected in our relationships with the neighbour, near or far. The dishonouring of human dignity is a dishonouring of God.

Additionally, God has conferred on humanity the capacity to participate in the divine nature by virtue of a capacity to know and to communicate with the Creator and to reflect the very nature or qualities of the Creator in the world. This, too, is the Creator's affirmation of the human and is that in which the blessing of humanity consists. (3) In this, too, human dignity is conferred and affirmed.

To be human, then, is to be invested with God's image, to reflect this image and to live in relationship with him. It is also to live in community. As we are created for relationship with God so also are we created for relationship with others. It is in the realization of our encounter with the other that we work out our response to the question of what it means to be human. In relationship, our humanity is affirmed and realized or denied and perverted.

Sin

Scripture affirms that our relationship both with our Creator and with our neighbour is marred by sin, a condition arising from our alienation from God.(4) This means that our relationship with God and with our neighbour will always be less than it could be, should be or is intended to be. Individually and collectively, we confront the power of sin and its destructive consequences even as we struggle to live creatively, peacefully and justly.

The Grace of God

Scripture affirms that the destructive power of sin is countered by the grace of God working effectively in the life of faith through the agency of the Holy Spirit. Grace represents God's favour and the presence of God's life in our life to effect that reconciliation, healing, wholeness and peace that is our human need.

Jesus the Christ

New life is the promise of God proclaimed in the person, death, and resurrection of Jesus the Christ. Through him we receive the forgiveness of sin. The new life in Christ, then, points to the renewed creation and fullness of life in the fully realized Kingdom of God. Such "realized eschatology" is the basis of Christian hope. (5)

Justice

Scripture witnesses to a God who requires justice. (6) That is, God seeks for his people a world that, in all its parts, reflects the qualities that constitute his nature such as fairness or equity, concern, compassion and mercy. (7) Justice, then, is God's norm for human relationships and thereby establishes the framework within which these relationships are to proceed. It is in the practice and exercise of justice that the command to love the neighbour is worked out and fulfilled. (8)

Justice has to do with the affirmation and protection of human dignity (9). This means that justice opposes all that diminishes or assaults the value that God has bestowed on his creation. Justice defends the right of God's people to be human and their right to that life which is the gift of the Creator. It also witnesses to the claim of a sovereign Creator to the life of his creation (10). God's justice requires that the life he intends for his creation to be safeguarded so that his people might live to his glory and praise (11).

Truth

Scripture summons us to seek truth and to live in truth. Therefore we are to be open to the truths and insights of human skill and science. We are called to use such knowledge and skill for the common good and as an expression of our concern for the life that has been entrusted to our care. (12) Similarly, we are called to refrain from the use of knowledge and scientific and technological capability when such use can occasion great harm or when it reflects the pursuit of particular interests at the expense of the interests of the many. (13) Such activity is destructive of the community in which we are called to live for the sake of our humanity and thus constitutes an assault on the right to the life intended for us by our Creator.

It is in the light of the witness of scripture and the expression of its faith reflected in the forgoing that the Presbyterian Church in Canada has declared its position with respect to certain aspects of genetic science and has formulated a faith response to developments in biotechnology.

In 1974, the church raised concerns about the uses to which biological engineering might be put and called for the formulation of policies and principles by which new developments in this field might be evaluated. In 1979 the church adopted an introductory study with respect to Genetic Engineering and the meaning of human life. (14) The study focused on three areas: The procedures and goals of genetic science and technology, the dangers inherent in the use of technology made possible through advances in genetic science and the vision that should guide genetic research and its

technological applications.

With respect to the goals and purposes of genetic science and technology, the study identifies what it suggests are underlying assumptions on which this enterprise proceeds. One is that through the application of genetic technology humanity quality of life can be improved. A second is that the nature of humanity can be ascertained through an understanding of its biological constitution. A third is that human wholeness can be achieved through a biomedical intervention "which could stabilize and make dominant the moral and ethical propensities of man and subordinate, if not eliminate, his negative and primitive behavioral tendencies." (15) The study concludes, then, that the fundamental goal of genetic science and technology is intervention, change and controlled reproduction with a view to the creation of a new humanity.

The study also maintains that there are certain dangers associated with advances in genetic science and technology. It suggests that if the enterprise of genetic science proceeds on the basis of a deficient understanding of what constitutes humanity, it runs the risk of de-personalizing the human subject. The nature of humanity cannot be ascertained solely from a biological perspective. An adequate understanding of the human also involves an appreciation both of the mystery of the spiritual dimension of its creation and of its predicament in the world, neither of which science can fully address.

Failure to acknowledge any limitations to their understanding and to their capacity to recreate the new human and to perfect human life can lead practitioners of genetic science or biological engineering to a pride that denies responsibility to anything other than self-interest or self-will. This leads to the possibility that genetic science will become the servant of the pragmatic interests of a technological mentality "which is inclined to assess human value in terms of social usefulness and fitness." (16)

The statement proposes that the appropriate stance for the church to adopt toward a developing biological revolution in general and genetic engineering in particular is that of a "Christian realism" which cautions against the attempt to seek a transformed humanity solely through the manipulation of genetic endowments. Science and technology in themselves cannot yield a human condition free from the destructive power of sin "which impinges upon even our best efforts," (17) a sin which results from the separation of the creation from its creator and which is ultimately overcome through the agency of Jesus the Christ. In the words of the study: "God's design for us in Jesus Christ is moral and spiritual. It takes root in us through the creation of a 'new person,' not through the improvement of our genetic endowments." (18) While cautioning against uncritical acceptance of genetic research, however, the statement

also affirms its legitimacy. For such research gives rise to an increasing ability to understand the nature of defective genes. This understanding in turn helps scientists devise responses to these genes, responses that are helpful in alleviating human suffering.

The study also raised a number of questions upon which the church needs to reflect as it seeks to frame a response to developments in genetic science and technology. What does it mean to be human? Which model of humanity would inform the enterprise of creating the new humanity? What would be the cost of separating human sexuality from procreative love as occurs in cloning? What are the costs and benefits of pursuing genetic research given the reality of pressing social needs and limited resources?

The perspectives, concerns and questions raised in this study were further elaborated in a statement on Genetic Engineering, accepted by the church in 1989. (19) The statement asserts the need for the formulation of criteria in order to adequately assess and respond to the ethical dilemmas posed by ongoing advances in genetic engineering and the application of genetic technology. The statement then sets out a number of principles that should inform such an assessment and response.

Stewardship

The study calls for acceptance of new knowledge and of scientific insight and discovery as tools for an enhanced understanding of the natural world and "the particularities of our time." Such knowledge and understanding is to be used in order to fulfill the biblical mandate for humanity to exercise a faithful stewardship of the created order.

Equality

The statement affirms that no one gender, race or group is of greater value than another. This means that gene selection should not be used in an attempt to give one life greater value in the eyes of society than another. The procedure should not be used for the purpose of selecting certain genetic traits deemed desirable to be passed on to children and the elimination of traits thought to be less desirable. Gene therapy should not be used to benefit one segment of society over another. Similarly, gene splicing should not be used to create a life form for the advantage of only a few.

Dignity

Genetic engineering and technology should be used as a means of protecting and honouring the dignity of God's creation.

Reproductive technology

The statement opposes the use of gender selection as a means of reproductive control.

Human rights

The rights and freedoms of all people are to be protected. Thus, genetic screening must be voluntary and mass genetic screening of any particular social or racial group is to be avoided.

Pastoral counseling

The church should be aware of developments in genetic engineering in order to be able to offer effective pastoral counseling and guidance to those who are dealing with issues relating to genetic disorders either in themselves or in their offspring.

Embryo research

The church recommends "that embryonic research into correction of human genetic disorders using tissue encultured by in-vitro fertilization should proceed only under strict government guidelines that do not allow the indiscriminate use of fertilized embryos, but encourages development of cell culture lines from fetal material that will accomplish the same purpose." (20)

In 2000 the church adopted a study on human cloning and biotechnology that reflected on a number of questions and issues posed by ongoing developments in genetic science and technology, issues such as stem-cell research, somatic gene therapy and research in genetic screening. It also considered the appropriate relationship that should exist between human and non-human species and the extent to which we are justified in subjecting non-human species to the utilitarian needs of humanity. (21)

The study also raised the question as to whether or not there is a limit to the knowledge to which humans have a right and suggests criteria by which to determine which knowledge it is legitimate to acquire and which is not. (22)

In these and other areas of genetic research and its technological application, the church has continued to urge caution with respect to what genetic science seeks to accomplish and what use is made of its discoveries. The church has also committed itself to ongoing reflection and study of the possibilities provided by the biological revolution of our age as it attempts to be faithful to its calling to glorify the Creator and serve God's creation.

REFERENCES

1. Living Faith: A Statement of Christian Belief. The Presbyterian Church in Canada, 1984 (LF) 2.1.3
2. LF 2.2.1
3. Gen.1:28
4. Gen.3:8, Psalm 51:5, Rom. 3:23, LF 2.5.3
5. LF 10
6. Micah 6:8
7. Jer.9:24
8. Luke 10: 26ff.
9. LF 8.4.3
10. Psalm 24.1
11. LF 1.6
12. LF 2.4.1
13. LF2.4.2
14. Genetic Engineering and the Meaning of Human Life: An Introductory Study. Acts and Proceedings of the One Hundred and Fifth General Assembly, (1979) The Presbyterian Church in Canada. pp 225-232
15. ibid. p 225
16. ibid p 228
17. ibid. p 231
18. ibid.
19. A Statement on Genetic Engineering. Acts and Proceedings of the One Hundred and Fifteenth General Assembly (1989), The Presbyterian Church in Canada. pp 341-348
20. ibid. p68
21. Human Cloning and Biotechnology. Acts and Proceedings of the One Hundred and Twenty Sixth General Assembly (2000), The Presbyterian Church in Canada. pp 333-342.
22. ibid. p 334-33

Appendix E

Roman Catholic Perspectives

The Faith and Genetics curriculum of the Biotechnology Reference Group, a committee of the Canadian Council of Churches, is an interesting exercise in tackling ethical questions that arise from developments in genetic research. In general, the Roman Catholic Church welcomes progress in any area of scientific and medical research that is aimed at helping people overcome diseases and serious defects. Many cures have been found for these problems, and in the field of genetics the sequencing of the genome is seen as an encouraging contribution to developing more relief for suffering people. In principle, advances in genetic research are to be sought and encouraged, both for the cures that are developed and also for the new insights they give into the human condition, for example in showing how some patterns of behaviour have a genetic origin.

Pope Benedict XVI specifically referred to this in an address to the Pontifical Academy for Life in February, 2009: "This knowledge, the result of intelligence and the efforts of countless experts, has made possible not only a more effective and early diagnosis of genetic diseases but also treatment destined to relieve the sufferings of the sick and, in some cases even to restore the hope of recovering their health."

As in so many areas, further ethical questions tend to arise once more specific practices are developed, and once the implications of those practices become clear. The Roman Catholic magisterium has made specific pronouncements about genetic practices, and, broadly speaking, it approves procedures that are truly therapeutic and beneficial for the person receiving them. Archbishop Fisichella, the current President of the Pontifical Academy for Life (the committee responsible for the application of medical and genetic research and so on), noted that genetic research for therapeutic success is a necessity for human development. He emphasized that "… scientific progress must be accompanied by greater ethical awareness that respects the full dignity of every human person." The Vatican has frequently voiced its concerns about ethical concerns such as, for example, the possibility of the practice of eugenics based on genetic information, where those with serious genetic defects might not be considered worth treating.

In a message for the World Day of the Sick issued in December, 2003, Pope John Paul II urged the protection of every individual, thanking medical and scientific researchers who have made advances in the field of genetics, and reminding us that "No one, in fact, can arrogate to himself the power to destroy or manipulate in an indiscriminate manner the life of the human being."

In 2008, in the most recent official Magisterial document that refers to genetic matters, *Dignitas Personae*, the Congregation for the Doctrine of the Faith stated in Note 19 that: "Gene therapy is allowed if used to eliminate defects in somatic cells, but not in germ-line or reproductive cells. Risks must be carefully assessed as in any procedure. Germ-line procedures may affect future children and the possibility of future harm precludes its use." The Congregation expressed concern in Note 27 about the use of genetic engineering in humans for non-medical purposes, especially if "… it involves an attitude of being dissatisfied with certain aspects of being human." Our response should, rather, embody the attitude that the Congregation promotes, i.e., that of "…accepting human life in its concrete, historical, finite nature."

Roman Catholics are instructed to ensure that every individual be protected from any changes proposed to be made through genetic engineering that are not sought for that individual's therapeutic treatment, i.e., treatment for disease, but rather are changes aimed at altering the person "for the better," which more accurately means in accordance with the engineer's subjective view of what is "better."

Pope Benedict XVI warned in his 2009 speech to the Pontifical Academy for Life that, "… If the human being is reduced to an object of experimental manipulation from the very earliest stages of his development, this means that biotechnological medicine has surrendered to the will of the stronger." Our trust in scientific developments is always to be subject to an ethic that first and foremost protects human life at every stage of its existence.

On a more global note, Pope John Paul II called in 2003 for the protection and development of third world countries, in order to "… prevent a further source of inequality between nations, also given the fact that enormous financial resources are invested in research of this sort, resources which, according to some, could be allocated first and foremost for the relief of curable illnesses and of the chronic poverty of so many human beings. " Catholic teaching on genetics, therefore, not only encourages genetic research in the hope that cures for serious illnesses will be found, but also hopes that it will lead to an escape from poverty in less developed nations. These hopes not only raise the bar for our expectations of genetics, but also acknowledge the tremendous potential genetics has to benefit humankind individually, socially and globally.

Appendix F

The Society of Friends

Queries on Faith and Genetics

Quakers believe that "there is that of God in everyone." Many believe that this also includes the natural world. In the 21st century, as we contemplate the rapid development of biotechnologies and genetics, how should Quakers respond?

During the 1700s Quakers adopted a set of queries as a form of guidance intended to help them direct their thoughts when seeking their way in the world. These queries have been augmented and reworded as time passed and have proved their worth through to the present day. Using the same approach the following Queries on Faith and Genetics are offered for worship, prayer, discernment, and discussion.

Queries of a General Nature

1) How does God's presence in each one of us act as teacher and lead us to act in ways that lead to the betterment of people?

2) The potential to do good in the world and leave it better is present in all of us. As we live out that potential, how can we take into account self interest?

3) What must people of faith do to protect and to maintain hope for the potential good that can come from genetics and technological development?

Queries Bearing on Genetics and Technology

4) What criteria should we use to judge the positive and negative aspects of genetically related technological change?

5) As your congregation (Meeting) studies and prayerfully considers technological change, how do you include its impact on reproduction, on men's and women's bodies, their role in families and society, and on those with special needs?

6) How can we evaluate the positive and negative effects of reproductive technologies on the lives of individuals, on families, and on society?

7) What are the advantages and disadvantages of particular technologies for individuals, families, local and global human society, and for all other life?

Your financial contribution to offset the cost of this project will be gratefully received. A suggested donation of $10.00 can be made online at www.councilofchurches.ca or by cheque payable to The Canadian Council of Churches.

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