

Genetic Testing and Screening: What do we choose to know?

Gathering

HYMN

(See hymn suggestions on p. 119)

PRAYER

In a world of wonders, we ponder where we have come from and where we are going. We wonder at the gifts of life and love, O God of the Universe, and we wonder at grace that draws us up and out and forward. Our genetic foundations and our species history speak of how we arrived here, but the wonder of your future for us remains mystery; we thank you. Teach us to pray, to hope, and to labor for the kingdom. Amen.



HEARING THE WORD

Psalm 139:1-18, 23-24

DISCERNING THE WORD

Silence

Discernment

What did you hear in this reading? Is there a word of God for us here?

Introduction

Session summary

This session invites you to learn more about genetic testing and screening and to reflect about the personal challenges it presents. Three examples are provided from real life situations. The session is designed for you to select one contemporary illustration: cancer genetics, prenatal testing, or teen at risk. After reading about the situation, read the “Common Territory” under “About Genetic Testing and Screening” and then the discussion paragraph for that illustration. The subsequent section is organized in the same way, but reflects on the issues in relation to theological matters. The questions at the end of the session are organized in the same pattern in order to allow discussion and deliberation on the illustration you have chosen.



Real life stories

1. Testing for disposition to cancer?

Fred has just learned that he has advanced prostate cancer at age 38. His sister died of ovarian cancer at 32, in 1995. After their parents died in an accident when they were in their 30s (both parents had been in good health), Fred and his sister were raised by neighbors because there were no living grandparents, aunts or uncles. Fred’s daughters are 14 and 11 years of age. Someone asks whether there might be a genetic predisposition to cancer in his family.

They have heard of a test that can inform people about such risks. Fred and his wife Glenda meet with a genetic counselor to learn more about their options. They learn that the test might show “nothing unusual,” which would mean either that his family has no special risk factor or that it exists but is outside the range of current testing technology. The test might show something that is “different from usual” but of uncertain significance, possibly important or possibly only a normal variation. The test might reveal a “specific abnormal gene structure” that brings with it high risks for cancer of the prostate, breast, and ovary.



The psalmist says, “I come to the end—I am still with you” (Ps. 139:18). What comfort do you find in these words amid the pokes and proddings of our medical lives?

They also learn that Fred's results might have profound meaning to his daughters. If he is tested and the results are "different from usual" or show "specific abnormal gene structure," it might open a path to reveal whether one or both of them have inherited a high risk of cancer from him. The geneticist explains that testing is discouraged for people who have not yet become adults and established their own plans as to what information they want and what they would prefer to leave unexplored.

While there are federal and state anti-discrimination laws, being labeled as a high-risk person can be uncomfortable. What does Fred want to know, and when does he want to know it? What does Glenda think? Should someone be talking to the deceased sister's son and daughter? How does Fred feel about all this, now that his treatment is completed but he is being asked to worry about the risks his daughters may have inherited from him?

2. Screening a pregnancy?

Fran is 36 years old and has two school-age children at home. She and her husband Greg had not planned it, but a new pregnancy is under way. They are inclined to accept this as a joyful challenge.

Her obstetrician proposes, along with what she considers ordinary obstetric care, genetic screening designed to learn whether this pregnancy faces higher than usual risks for chromosome abnormalities (e.g., trisomy 21, which causes Down syndrome) and/or birth defects (e.g., spina bifida, which can be crippling and cause developmental delays). She explains that the screening cannot reveal whether these problems are actually present, but only whether the *chance* of their being present is high enough to suggest that definitive testing should be offered. The screening involves one more tube of blood along with the others being taken from a vein in Fran's arm. The definitive testing would involve an amniocentesis, insertion of the needle into her abdomen to remove some of the amniotic fluid, so that specific genetic tests can be performed.

Since Fran has no known risk factors, amniocentesis will be offered only if her screen came back "positive" (elevated risks). In such settings, amniocentesis results usually come back normal, meaning that the alarm was a "false positive" rather than signaling an actual problem with the pregnancy. The doctor also encourages ultrasound examinations to monitor growth and development, but warns that ultrasound may reveal structural abnormalities known to cause severe disability, or other abnormalities of uncertain significance. What do Fran and Greg want to know about their pregnancy? When do they want to know it? Until now they have felt optimistic about their own health and the apparent normalcy of this pregnancy.

3. Testing a teenager?

At age 14, Tommy is approaching six feet tall. He is an honor student, and well coordinated with considerable talent for sports. Everything is going his way as he starts high school and looks forward to playing basketball; the team has a long history of being scouted by the best colleges.

Coach Jensen is eager to maintain her award-winning team and has heard about Tommy from his middle school teachers. But when she meets him, an inner alarm sounds. She notices his long flexible fingers and a slight curve to his spine when he removes his shirt on his way to the locker room. Having heard that Tommy's mother, who was also tall and slender, died in childbirth of a heart attack, she calls his father and asks that Tommy be evaluated for possible Marfan syndrome before participating on the basketball team. He is both alarmed and angry at the suggestion that they might have overlooked some important health risk and that Tommy's future in sports (and sports scholarships) may be in question.

An orthopedist announces that Tommy has Marfan syndrome. A cardiologist says that the echocardiogram reveals nothing abnormal in Tommy's heart and aorta, but warns that at his age the changes, which include dilating of the aorta, might not yet have developed; he offers to start prescribing medication which may protect the aorta from developing problems. A consultation with a geneticist is offered, but Tommy and his father worry that

a confirmation might prevent his continuing in sports, and they feel adequately protected by the cardiologist's opinion and treatment plan. What should they do? What should Coach Jensen do? Tommy's mother may have died of a ruptured aortic aneurysm (common in pregnancies of women with Marfan syndrome); should someone be talking to her surviving siblings, nieces, and nephews? Tommy's cousin Jane is over 6 feet tall.

About genetic testing and screening

Common territory

All of us have complexities that are evident to everyone who takes a glance at us, others that only we know quietly in our hearts, and still others that are outside the view of all but God. These mysterious factors, some favorable and others not, are tied up in a great bundle that is the gift we are from God to the world.

Our genome radically determines the limits of our possibilities. My genes will never allow me to fly like an eagle, or dive like a whale; technology allows me to imitate these activities, but not to do them myself. If I am severely damaged by mutations or by poisons or by traumas, the limitations on my possibilities will be narrowed irrevocably. Some of the ways in which I am genetically endowed make me stronger than you, while others weaken me. Some will manifest disorder and disability from even before I am born, others during my childhood, and others only much later in life.

Since we have some 3 billion subunits of information in each cell, and most of these are not directly involved in visible phenotypic features, there are millions of ways in which one person may be genetically different from another person, without either of them being impaired. Eye color is an obvious example. Green eyes are not "normal" as compared to brown eyes; one color is not "better" than the other biologically. The millions of differences between any two individuals make it possible to use genetic analysis to identify an individual at least as well as finger prints ever did—better, really.

Every child born in the United States is subjected to newborn genetic screening to learn if one of several dozen biochemical abnormalities might be present. Many of the screens flag children who in fact are "normal" and a more definitive test shows this, removing the concern that a disorder might be present. The standard definition of what should be screened in newborns is that the condition should be subject to interventions that improve the outcome for the child. Such interventions may be dietary, medical, or therapeutic.

Cancer genetics

The notion of genetic predisposition is crucially important. Everyone accustomed to climbing stairs knows that occasionally a foot is not quite lifted high enough and one trips. Usually one does not drop one's burdens or actually fall, but recovers quickly and continues climbing. Sometimes one drops or spills things, sometimes one falls, and sometimes one is seriously hurt in the process. Some people trip on the stairs more often than others, through clumsiness, blindness, carelessness, interference, etc. Some just do it more often because of being *predisposed*. Tripping on the stairs is somehow just more a part of who I am than who you are.

People who are genetically predisposed to develop cancer will have a higher risk for it than others, but this does not mean that they will certainly develop it, nor does it mean that those others will not develop it. If my risk is 20% while yours is 10%, I may yet be more fortunate than you in the long run or I may die in an accident or a war or from an evil choice and not live long enough to show my proneness to tumors.

Genetic risk factors associated with breast cancer also illustrate another subtle but important detail about hereditary predispositions: changes in most genes are inherited and transmitted to future generations regardless of gender, both males and females receiving them and passing them along. But some of these risk factors are very different in their phenotypic effects as a function of the gender of the person receiving them (due to the internal hormonal environment and the details of anatomy).

Prenatal screening

Modern technology makes it possible to observe considerable anatomic detail in a fetus through the use of ultrasound, and to determine genetic characteristics by a sampling of amniotic fluid or by conducting small biopsies from the placenta (or even the fetus) which give us cells to grow in the laboratory. Some methods involve the measurement of chemicals that, when higher or lower in concentration, are correlated to abnormalities in the developing fetus. These latter measurements are called “screening” techniques because they do not (cannot) prove or exclude an abnormality. They merely draw our attention to elevated risk rather than an actual abnormality, so that we can focus on those pregnancies for which definitive testing can be offered. Not every pregnant woman wants either tests or screens to learn of abnormal processes underway in their pregnancy; they prefer to wait until the time of birth to learn these things. They cannot forever avoid the knowledge, but they can postpone it.

Teen at risk

Marfan syndrome is caused by mutation in a gene responsible for formation of one of the components of the connective tissue—bones, ligaments, walls of blood vessels, scaffolding of the lens suspended inside the eye, etc. There are dozens of physical characteristics that are commonly found in Marfan syndrome, most of them things that are also seen in many people who do not have the syndrome. Features shared with other people include tall stature, long arms, loose joints, spinal curvature. Several features that are seldom seen in non-Marfan people carry more weight in making the diagnosis: enlargement of the root of the aorta, dislocation of the lens of the eye, and an abnormally generous amount of space for the spinal column inside the lower back. Coaches are concerned about Marfan syndrome because healthy, tall, slender teens can have catastrophic events with dissection of the aorta and drop dead on a basketball court. But clinical delineation of the syndrome is complex because of the considerable phenotypic overlap of people who have Marfan syndrome with people who just have some physical features that bring Marfan syndrome to mind.

Reflections for consideration

Common territory

Notions of determinism enter the discussion at all major turning points in personal and corporate life. Do I act selfishly because of the ways in which I am constructed genetically, or the ways in which I have been shaped behaviorally, or with a radical freedom that leaves me wholly responsible for my bad choices? Questions of this kind await us around every corner in a discussion of genetics and faith. And if the answer is that my genes make me do what I do, then I must decide whether I think that is the result of blind accidents through evolutionary time or of direct planning and enforcement by God. Lutherans typically opt for a middle way, in which the force of events within and around me is acknowledged, but I am still expected to accept all the implications of freedom, which opens me to opportunities for sinfulness while foreclosing the excuses of victimhood.

Cancer genetics

Watching closely and helplessly as loved ones are felled by cancer in their 40s can condition a person to feel fated to suffer the same events. Family dynamics can further complicate the picture; if I have been out of communication with my sister for 20 years because of an old dispute, will I contact her to warn her about the cancer risk I have just learned is moving through our family? In such situations, where are faith and grace in my life?

If my overall attitudes toward life are optimistic and I am prone to take charge of problems to seek out their solutions, I may be more inclined to opt for surgical efforts to reduce those areas of the anatomy at risk (e.g., breasts, ovaries, colon), rather than to watch with imperfect monitoring methods. If I am more passive in my life style, I may not be so assertive about risk management. And these different personality types themselves

probably stem from complex multi-factorial traits: genes, upbringing, environment, the history of accidental and decisional events in one's past.

Prenatal screening

We may enjoy poetic reflection on Psalm 139, which marvels at how we have been knit together in our mother's wombs, but we should not allow that to overshadow the combination of joy and hope along with dread that is experienced when a pregnancy is underway. Parents are charged with the responsibility of providing prenatal, neonatal and pediatric care that includes nutrition, protection, education, nurturing, etc. How individual parents meet the challenges of these many arenas varies depending on their own internal capacity, and their available resources (financial, psychological, etc.).

Most of us most of the time would lay down almost anything in order to save our children from suffering and death. But some of us some of the time are faced with information that may lead us to think that death is preferable to birth, and so to elect pregnancy termination after prenatal diagnosis. Some of us do not want that information because we are certain that we could never be induced to elect termination or because (perhaps subliminally) we do not want to face such a temptation. If we do not know about the problem until it is too late to make termination decisions, then we narrow the field of options. This either enhances our heroism in making selfless choices, or it betrays cowardice in avoiding situations in which we might not choose heroism.

It is important to remember that prenatal testing is not designed to set up abortions, though the outcome after an abnormal result may be the choice to terminate. In a much larger context, these tests allow some families to better prepare for a child with special needs by reading and by speaking with other similarly affected families. In a few diagnoses, there are even prenatal interventions to improve prognosis.

Teen at risk

The competing and overlapping roles of fear, hope, guilt, ambition, denial, and confusion can make decision making complex and painful. No sane parent looks for ways to place his or her children at risk, but who can say that there is not a strong temptation to avoid confronting facts that frighten by raising painful memories or uncovering our worst worries after years of clinging to fondest hopes? Where is my hope located—in my own best efforts? In my genes? In a risen Christ whose presence may seem elusive while the genome looms as pervasive? Who will fault the widower Dad who begins to suspect that the killer of his beloved wife might be emerging in his beloved son? Where does he turn now? Do we glibly invite him to prayer, or do we try with little hope of success to enter the maelstrom with him?

Invitation to conversation, prayer, and action



A QUESTION COMMON TO EACH SITUATION

- When scientific advances allow me to know more about what health problems are more or less likely to develop in my life, to what extent do my decisions about how to employ them depend on curiosity, fear, or the sense that I must take responsibility to be well informed in order to then pursue upright choices?

QUESTIONS FOCUSED ON EACH STORY

- Should I seek out knowledge of my cancer risks at the genetic level in order to make better-informed career plans? To decide about whether to have children? To advise close relatives of the risk that they also might face?
- Is delving into the genetics of a pregnancy underway a means of expressing a sense of partnership with God in the creative enterprise? Or is it an interference with God's manifest will in the existing conditions to which I should not raise my hand in objection?

- How much testing and seeking is enough? What level of risk should a parent take in decisions about pursuing a possible genetic problem in a teenage child when physicians called for consultation differ in their perspectives? What is appropriate when major life options may open or close on the basis of the answers obtained?

Closing prayers

INVITATION TO INTERCESSORY PRAYER

Pray for genetic counselors, health care providers, and parents who struggle to understand what is best to do for their children, born and unborn.

PRAYING WITH THE TRADITION

Gracious and holy God, give us diligence to seek you, wisdom to perceive you, and patience to wait for you. Grant us, O God, a mind to meditate on you, eyes to behold you, ears to listen for your word, a heart to love you, and a life to proclaim you through the power of the Spirit of Jesus Christ, our Savior and Lord. Amen.

(from Evangelical Lutheran Worship, page 76)