

## Chapter Three—Genetic Testing at the Beginning of Life

Some blood had been drawn during Sarah’s regular visit to her doctor about a week ago when she was 16 weeks pregnant. Sarah was anxious about the results, but her obstetrician had told her not to worry. If the screen showed an abnormality, she could learn more about it at that time. When her doctor called her that evening, she could tell by the tone of his voice that something was wrong. “Sarah,” he said, “the results of your triple screen have come back ‘screen positive.’ You might want to consider having amniocentesis performed.” He then gave her instruction about how to proceed. Sarah did not hear much else that he said other than the fact that her baby might have something called Trisomy 18, also known as Edwards Syndrome.

### Edwards Syndrome (Trisomy 18)

A syndrome associated with the presence of an extra (third) number 18 chromosome. It occurs in approximately one of 8,000 live births and involves multiple abnormalities. Common characteristics include low birth weight, mental retardation, low-set and malformed ears, small jaw, hand abnormalities, congenital heart disease, hernias, and others. Few infants survive beyond the first year.

The next morning Sarah called for an appointment and was told that she needed to see a genetic counselor. She had no idea what that was, but she managed to get an appointment for that afternoon. Her husband, Allen, took off work so he could join her. At 3:00 p.m., they were introduced to Jill, the genetic counselor. Jill explained to them that the

triple screen results indicated a slight risk that their baby might have Edwards Syndrome. Only an amniocentesis could determine it definitively.

As they talked with the genetic counselor, it dawned on them that having had the triple screening, they were now confronted with whether or not they should pursue having a test to determine, before the birth of their baby, if it had this condition. Sarah was not sure she wanted to know. Allen, on the other hand, was wondering if the anxiety of not knowing would be too much to think about for another five months. Together they discussed with the genetic counselor whether or not they might think about termination if they found out the baby had Trisomy 18. Sarah and Allen had never even thought about it. They were not sure what they would do. They did not even really know what children with Edwards Syndrome were like.

### Genetic Counseling

Genetic counselors (a relatively new profession) hold a master’s degree that prepares them for counseling patients undergoing medical procedures involving genetics. A genetic counselor follows a few basic tenets. These include respect for autonomy and privacy of the individual, the need for informed consent and confidentiality, and the goal of non-directiveness.

Sarah and Allen were overwhelmed at the thought of having to make the decision whether or not to terminate a much-wanted pregnancy. Jill suggested they think about the amniocentesis more before coming to any conclusion. That evening the couple talked more about the triple screen results, now armed with enough information to make an educated decision. In the end, they decided they would go through with the amniocentesis. When they had the chromosome results, then, and only then, would they be able to think about what they should do.

The amniocentesis was performed two days later. Allen and Sarah waited impatiently for the results of the chromosome studies. As far as they were concerned, life could not go on until they had the answer. The genetic counselor called them

early the next week. “I’m sorry I have to give you this news,” said Jill. “I know you were hoping to be reassured by the results. The baby has an extra chromosome 18. This confirms Edwards Syndrome. . . .”

Sarah was devastated, even more so than when she had received the results of the triple screen. Jill suggested that she and her husband come for another appointment. Sarah agreed. She knew the information about the condition would be beneficial, but she also wondered how much she really wanted to hear it at this point.



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The couple tried to think of others with whom they might be able to talk. They had little previous preparation for making this decision. They knew that this information could help them prepare for the birth of their baby with a severe abnormality. But the news also had introduced abortion as an option. Or, could they look into adoption? What should they do? What did their faith tell them to do? Since Sarah was 18 weeks along in her pregnancy, they had only a few weeks to make a decision. . . .<sup>1</sup>

## Introduction

Medical genetic testing has been the most prevalent way in which “genetics” has affected the immediate lives of people today. The use of the triple screen mentioned above represents a prime example and the expanding knowledge of genetic science will permit the development of dozens of tests and screens for other syndromes or predispositions to disease. It is important to divide these tests between those associated with the beginning of life and adult testing. This chapter considers the first while a later chapter will explore the second. The tests related to the beginning of life can be broken down further into prenatal, neonatal (newborns) and carrier testing (see sidebar for further explanation). Attention here will be directed largely to the first and third category because they pose the most troubling questions. The case of Allen and Sarah will help illustrate what could become a common part of the parenting experience. The purpose is to help the reader better understand and reflect upon the complexities posed by these uses of genetic testing. Attention will center on several of the following questions.

Genetic testing is offered or even encouraged because it permits, like other medical tests, greater information and choice. This information can be exceedingly useful. Yet, some critics contend that often genetic knowledge creates the “illusion of choice” and makes all pregnancies tentative. Pregnancy becomes tentative because it is increasingly difficult for prospective parents to commit themselves to an unborn child before a diagnosis is offered. These critics argue that such important decisions are truly free only after a commitment has been made to the unborn. In this sense, medical technology has actually set up a bias, even though it is seemingly neutral. If this is true, should medical practice be altered in some way?

Yet the dilemmas can predate pregnancy. If family history warrants it, prospective parents can have carrier testing done in order to determine whether or not they carry certain inherited conditions that could affect their offspring. (See sidebar for examples.) Since most of these conditions are recessive, however, carrier tests often can indicate only the statistical chance for an offspring to suffer from that condition. It is quite possible, increasingly so, for prospective parents to face the following dilemma: Should we avoid conceiving a child because as carriers we have a 25 percent chance of having one with a problematic medical condition? Or should we conceive, but anticipate an abortion if the odds go against us?

The related and obvious question is that of abortion. Should it ever be performed in these cases? Could aborting the pre-born, known to have a severe condition, ever be the loving thing to do? What are the criteria?

### About Tests and Screens

The procedures of testing and screening are often blurred under the term “testing.” A test, technically speaking, is a procedure to identify specific information about an individual’s DNA or the specific proteins it produces. Screens, by contrast, are a population-wide procedure that look for telltale products of genetic material known to cause disorder. The triple screen mentioned here looks for signs of three genetic disorders by identifying proteins in a blood sample of the expectant mother. The findings of a screen often include false positives, and only an additional test of the fetus’ chromosomes can be determinative.

### Categories of Genetic Tests

Medical protocol offers *prenatal* screens or tests early in a pregnancy when certain conditions such as advanced maternal age or ethnic disposition are present. Such screens or tests look for telltale indicators of such conditions as Edwards or Down Syndrome. *Carrier* tests are done before pregnancy. The testing of high-risk ethnic groups are examples, including African Americans (sickle cell anemia), Scandinavians (cystic fibrosis), and Eastern European Jews (Tay Sachs). *Neonatal* testing is exemplified by the screen used to look for possible signs of Phenylketonuria (PKU) in newborns. (See glossary for descriptions of these in medical terms.)



Genetic testing adds increasing pressures to have a “perfectly healthy” baby. If testing is easy, abortion available, and social pressure strong, will that result in social discrimination against even mildly “imperfect” children? To take a specific case, could governmental and charitable dollars disappear for families raising children with Down’s Syndrome? Will, or should, insurance companies refuse to provide coverage for those with pre-existing “expensive genes?” Such cases of refusal have been reported.<sup>2</sup>

Finally, what are the pastoral and congregational challenges here? Could Allen and Sarah go to a pastor and receive the kind of sensitive and informed attention they need to help sort out the issues? Would this couple find support in most Christian congregations? Would they find acceptance even if they decided to have an abortion?

## Personal Experience and Values

Each one of us confronts challenges like those faced by Sarah and Allen with a backpack full of personal experiences, emotional responses, values, and judgments. These are related to upbringing, economic class, race, and other factors of our lives, and are sometimes called “*pre-understandings*.” The point is that every person brings such pre-understandings to any issue and it is important to become aware of them within oneself. This awareness will permit a more open reflection or discussion. The following are a couple of important questions to reflect upon when engaging this topic.

What experience do you bring to the question of whether or not the use of medical technology can set up a bias? Have you had any experiences similar to Sarah and Allen’s in which you have felt entangled by the choices shaped for you by medical knowledge or technology? Consider how medical technology has changed your life. In your judgment: *Is medical technology truly neutral, or can it involve unintentional bias?*

Because few cures exist for many prenatally diagnosable diseases, it is impossible to separate genetic testing from the issue of abortion. Clearly, abortion is one of the most divisive issues in contemporary society, and Christians will have very diverse perspectives and strong emotions about it. Constructive discussion about such a challenging issue begins with awareness of one’s own perspective and the search to understand others’ perspectives, even when drastically different from one’s own. *What convictions do you bring to this topic?*

## Gathering Input


### Science and Medicine

Much of the technical and scientific information needed for understanding these issues was covered in the chapter entitled *A Primer in Genetics*. A return to that chapter may refresh your grasp of genes, alleles, and other concepts. However, several additional points about genetics will aid in understanding the complexities of these issues. The first concerns why genetic tests often cannot give definitive answers, and the second how genetic tests can be used.

#### Gene Functions are Complex

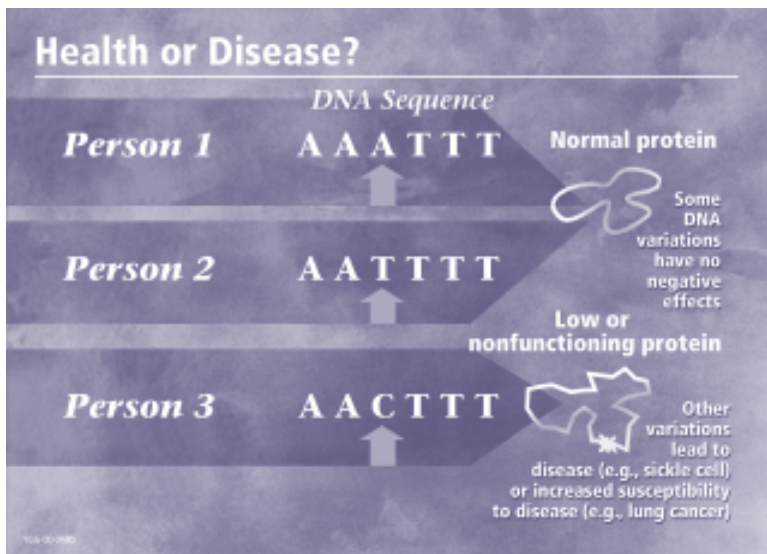
While a gene or combination of genes that create a specific condition may be tested, in many cases, the test cannot predict the exact severity. One dominant reason for this is that mutations occur in a variety of configurations, and the actual *expression* (the process by which a gene’s coded information is translated into a body’s actual functioning) of any given gene can vary greatly. The graphic on the next page demonstrates how variations at different locations on the same gene can lead to a wide range of effects. (Some genes are known to have more than 300 different variations.)

This fact adds significant ambiguity to the factors parents have to weigh when considering the results of a genetic test. This should remind us that genes are extremely dynamic. Mental, behavioral, and even physical characteristics of a child depend for their development and expression on environmental and social factors. Geneticists indicate this in two ways. They talk about the importance of *penetrance*—a term meaning the likelihood that a given gene will actually result in disease. They also make a distinction between *genotype* and *phenotype*. Genotype is the actual genetic material carried by an individual, whereas phenotype is the observable expression of a genetic characteristic that results from the interaction of a gene with its environment.



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In carrier testing, the reason that tests cannot give clear-cut answers requires a review of recessive and dominant alleles. The graphic below indicates why even an affirmative test response can result only in a statistical knowledge. Both parents are known to be carriers of a recessive gene disease (cystic fibrosis, for instance) and thus half-shadowed. Neither shows any sign of the disease because a normal allele (“A”) functions perfectly well, but each also carries one altered allele (“a”—the recessive gene). Therefore, each child they conceive will receive one out of four possible combinations of their genes (AA, Aa, aA, aa). One combination could be a double set of recessive alleles (aa), and this child will be affected. There are two chances in four that a child’s genes will make him or her a carrier, but he or she will

be disease-free. There is one chance in four a child will have two copies of normal allele and could never even pass on the problematic allele to his or her offspring. Recall that this statistical chance is repeated with the conception of each child.

### About Medical Technology

On the question about how genetic tests are used and whether they are neutral, it is important to consider how specialists in this field regard them. Dr. Robert Lebel, a medical geneticist writing in an Evangelical Lutheran Church in America (ELCA) publication, notes the following: “We should combat the naive assumption that these testing methods are useful only for identifying pregnancies to be targeted for termination [abortion]. Such an assumption is a grave error because it overlooks the opportunity some families appreciate—to prepare themselves psychologically, financially, spiritually, and socially for the birth of a child with a handicap. In some few instances, it also allows for specific prenatal therapeutic efforts to be undertaken. In severe conditions, with survival impossible, it may provide the basis for “do not resuscitate” plans when delivery occurs.”<sup>3</sup>

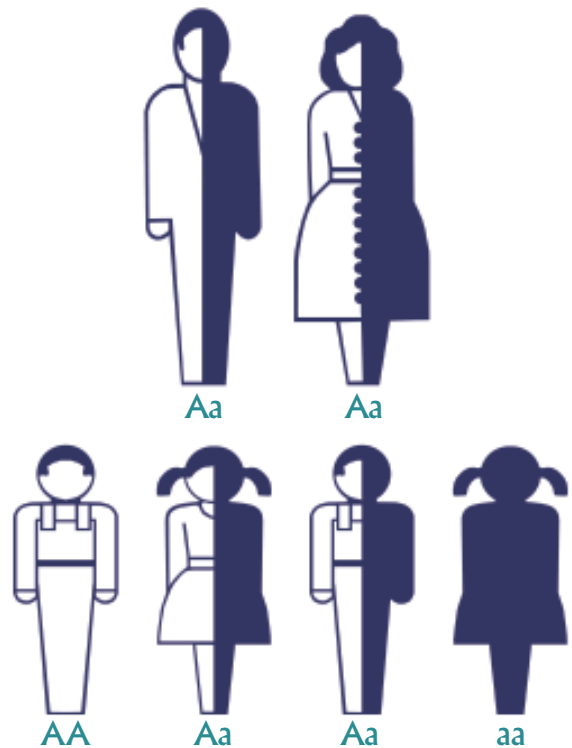
### Moral and Theological Considerations

In considering where they stand on these issues, Christians will include the facts and perspectives of science and medicine, but will also look to resources in their faith traditions. Several relevant aspects will be sketched here: what factors to consider, guidance on the question of abortion, and the challenge to practice Christian friendship.

### Factors for Decision Making

Dr. Lebel, now wearing the hat of an ethicist, reminds patients and caregivers of the many complex factors to consider in discerning wise decisions in these matters. His list for families to consider includes:

a) the strengths and weaknesses of the individual and the couple to meet foreseeable challenges, b) the family’s financial resources, c) the impact on existing children, d) the benefits and burdens predictable in future children, e) the impact on society, f) the impact on the future gene pool, and g) the meaning of reproduction in the broad context of God’s activity.<sup>4</sup> These considerations will need to be given varying



weights, but all are important in attempting to discern God’s loving intention for the situation. To ignore any of these is to miss an aspect of the big picture.

Lebel recognizes that such a list poses a rigorous challenge. He believes, however, that Christians are called to strenuously seek to be guided by the Christ-like principles of sacrifice, creativity, and generosity. He notes that in each specific case, the most creative option will not necessarily be obvious or easily discerned. But he believes such a rigorous effort can help move Christians toward faithful decisions and that grace can empower such full and careful moral consideration.

### **Contrasting Perspectives on Aborting the Severely Ill**

The ELCA has provided guidance on the thorny question of abortion in “The Social Statement on Abortion” adopted by the Churchwide Assembly in 1991. Among other things the statement designates several pertinent convictions that include the following:<sup>5</sup>

“The act of intentionally terminating a developing life in the womb is an issue about which members of the Evangelical Lutheran Church in America have serious differences.

“All human beings, created in God’s image, have intrinsic value and dignity and are to be treated with respect.

“As such, there is, therefore, a strong Christian presumption to preserve and protect life.

“Yet, there may be compelling circumstances wherein abortion may be an option of last resort, yet seen always as a tragic option. What is determined to be a morally responsible decision to abort in one situation may not be in another.

“In circumstances of extreme fetal abnormality, which will result in severe suffering and very early death of an infant . . . the parents(s) may responsibly choose to terminate the pregnancy.”

The statement does not detail what constitutes “compelling circumstances” or “severe suffering.” It does not engage in discussion of criteria about when personhood begins *in utero*, etc. It clearly has a preference for protecting life, but allows that exceptions do exist. While it gives guidance, it does not prescribe exactly what a couple like Allen and Sarah should do in their situation.

One Lutheran thinker, Hans Tiefel, is sympathetic to the reasons for the ELCA position, but respectfully disagrees. He asks whether it could ever be loving to abort a seriously ill pre-born, and concludes that it could not. As he writes: “I conclude that our faith may require more of us in the context of genetic testing than we are inclined to believe or accept. Our traditional responsibilities to the dying, our including the afflicted unborn in the community of faith . . . our commitment to God’s will over our own (a God who does not explain our misery, but shares it) . . . all seem to point us toward keeping company with the dying child until their condition brings death.”<sup>6</sup>

Tiefel recognizes that this view is painful, and possibly cruel for parents of genetically affected children. The medical costs and emotional burdens can seem like more than flesh and blood can bear. Nevertheless, he argues that love may well require this of both parents and the church community. He notes that invoking the help of God and the body of Christ is an indispensable necessity in such a situation. He would argue that Allen and Sarah should not have to bear their burden alone.

### **The Challenge of Christian Friendship**

Many dilemmas that result from genetic knowledge will not involve such extreme difficulties as Edwards Syndrome, but in all cases, they will raise the challenge of Christian friendship. Theologian Philip Hefner, has written about this idea in relation to genetic matters, and defines it in this way: “By this ‘friendship,’ I refer to the challenge to every Christian to offer respect, care, and support to Christian brothers and sisters who face the difficulties and traumas that come with genetic medicine—even when one disagrees sharply with the brother or sister, or even when the brother or sister has chosen a mistaken path.”<sup>7</sup>

Hefner believes the Lutheran insistence that none are justified except by God’s grace through faith is the basis for such a concept and practice. This view recognizes several things. It recognizes that hard decisions must be made, that life is ambiguous, that sin is prevalent in all human actions, and that God has given his people a community to live in—the church. He acknowledges that this form of Christian friendship is a tremendous challenge. This is partially the case because persons with genetically related problems, like Allen and Sarah, are often reluctant to share their burdens with their local religious communities. The

reasons given often stem from fear. Couples like Allen and Sarah fear that their burdens are too great, too ambiguous, or too disagreeable for their friends to bear. They fear that disclosure will weaken their ties with the community of the congregation, and so they hide their situation from the friends who are most important to them.<sup>8</sup>

Clearly, when issues are as emotionally charged as in some cases involving genetics, it may be especially difficult to turn this concept of friendship into acts of care. Yet Hefner suggests that it is perhaps here—in communities of care for those whose lives have been marked by such ambiguities and disappointments—that we find the major challenge of genetic testing. As he concludes: “There is no greater treasure in the church today, however, than the possibility for Christian friendship . . . God has given no greater gift of grace than the love that can sustain such friendship.”<sup>9</sup>

### A Common Theme

While the four sets of thought offered in this section may respectfully differ at certain points, a common theme does emerge. All agree that Christians are called to make decisions by criteria richer than the prevailing cultural idea of rights and self-fulfillment. All challenge the idea that each couple should simply choose in isolation to do what is “fulfilling for us.” A society may consider “the right to do as I choose” as an adequate moral criterion in medical situations, but such reasoning is not adequate for Christian thinking. Couples are the ones to make their own decisions, but they are called to factor into their thinking both the community as the broader context for making their decisions and the criteria of Christ-like love. In so doing, they will seek to discern God’s loving intention. Thinking beyond rights and individual choice may even be counter-cultural, but could this be what it means to be Christian in a society practicing genetic testing and screening?

### Deliberation

As an exercise in moral imagination, answer the question: *What would I do if I were in Allen and Sarah’s situation? Why?*

It is absolutely true that no one will ever know exactly what he or she would do in a situation until actually in it. However, imagining oneself in a situation is one excellent way to extend moral understanding. Such an exercise offers a means to move beyond just thinking abstractly about ideas. Therefore, take up the challenge to determine what you would do. Try to determine the factors to consider in making such a decision. What moral criteria would be important? Recall that although different means may be used to obtain an answer, the central moral question for Christians is: What is the godly decision in this case? One term for approaching a godly decision is called “discernment.” One other way, then, to ask the question is: “What is your discernment in this situation?” Bring the input from above into this discernment. Bring additional insights available from personal experience into the reflection. Bring both humility and openness into this exercise.

### Taking the Conversation With You

In order to take the conversation beyond the thinking stage, consider what actions might be possible for you or your study group. One way to consider this is to ask the following questions and then act upon them: What would Christian friendship mean in my congregation? What actions might it entail: Toward families whose children have a congenital condition? Toward the unborn with a congenital condition? Toward a couple who has chosen to have an abortion in such a situation? With some answers to these in mind, you might consider speaking with you pastor and reflecting together about how your congregation could further Christian friendship.

### For Further Investigation

Committee on Medical Ethics, Episcopal Diocese of Washington. *Wrestling With the Future Our Genes and Our Choices*. Harrisburg, Pennsylvania Morehouse, Publishing, 1998. This book is designed to provide guidance for those within the Anglican tradition who are faced with decisions about genetic testing. The contents often overlap with the ELCA volume, *Genetic Testing and Screening: Critical Engagement at the Intersection of Faith and Science*, but give less attention to the broad social issues and theological questions that genetic testing poses. Its narrower focus, however, as a guide to making biomedical decisions offers richer reflection on those specific dilemmas.



Evangelical Lutheran Church in America, *A Social Statement on Abortion*. (1991)

Willer, Roger A., *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science* (Kirk House Publishers, Minneapolis, 1998). Further investigation of the ideas above may be pursued especially in the chapters written by Philip Hefner, Robert R. Lebel, and Hans Tiefel.

## Citations

1. Kirstin Finn Schwandt, "Personal Stories: Cases From Genetic Counseling." *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science*, ed. Roger A. Willer. (Minneapolis: Kirk House Publishers, 1998), 41f.
2. Ted Peters, "Love and Dignity: Against Children Becoming Commodities." *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science*, ed. Roger A. Willer. (Minneapolis: Kirk House Publishers, 1998), 119.
3. Robert R. Lebel, "A Geneticist's Synthesis: Evolution, Faith, and Decision Making." *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science*, ed. Roger A. Willer. (Minneapolis: Kirk House Publishers, 1998), 161.
4. *Ibid.*, 160.
5. Evangelical Lutheran Church in America, *A Social Statement on Abortion* (1991), 2f.
6. Hans O. Tiefel, "Individualism Vs. Faith: Genetic Ethics in Contrasting Perspectives." *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science*, ed. Roger A. Willer. (Minneapolis: Kirk House Publishers, 1998), 147.
7. Philip Hefner, "The Genetic "Fix": Challenge to Christian Faith and Community." *Genetic Testing & Screening: Critical Engagement at the Intersection of Faith and Science*, ed. Roger A. Willer. (Minneapolis: Kirk House Publishers, 1998), 92.
8. *Ibid.*
9. *Ibid.*