5.2 Genetic Disorders and Decisions about Having Children

A *genetic disorder* is a genetically based abnormality or disease.

A couple may question whether they should have their own biological children because

- they have already had a child affected by a genetic disorder;
- there is a family history of a certain disease, birth defect, or disability, for one or both spouses;
- one spouse has, or both spouses have, undergone genetic testing and tested positive for a genetic disorder;
- they are members of an ethnic or racial group in which there is a high incidence of a particular genetic disorder. (1)

Genetic disorders cannot all be lumped together. They vary significantly in terms of genetic basis, symptoms and severity, and inheritance patterns (see below). Thus a couple must get accurate and complete information about the particular genetic disorder with which they are dealing.

A couple may visit a genetic counselor. A genetic counselor may adopt a *nondirective approach* to working with clients. The counselor will give clients relevant medical information, offer testing, and describe reproductive options which are available but will not tell clients which course of action to take. (2) A genetic counselor may well mention the option of using reproductive technologies that are not in accord with Church teaching to avoid transmission of a genetic disorder. Thus it is important that *Catholic couples be well informed on Church teaching regarding reproductive options when undergoing genetic counseling*. Here the Catholic pastoral counselor plays a key role.

Basic Information about Types of Genetic Disorders and Patterns of Inheritance

A human being has twenty-three pairs of *chromosomes*. One member of each pair is received from the father, and the other member from the mother. Chromosomes carry *genes*, the basic units of heredity. (3)

Chromosomal pairs one through twenty-two are called *autosomes*. The twenty-third pair constitutes the *sex chromosomes*. There are two types of sex chromosomes, X and Y. A male is XY, while a female is XX. (4)

A person may *have the abnormal gene(s) for a particular genetic disorder and actually have the symptoms of the disease*. A person may *have the abnormal gene(s) for a particular genetic disorder but be asymptomatic* either because the onset of the disease will occur later in life (late onset genetic disorder) (5) or because the disease is under control (6). An individual may be a *carrier* of a genetic disorder, that is, have one copy of a gene associated with a recessive genetic disorder and one copy of its normal counterpart. A carrier typically never shows symptoms of the disease because the normal copy of the gene is sufficient for normal functioning, but can pass on the defective gene to children. (7)

In the case of an *autosomal dominant* genetic disorder, an abnormality in one member of a pair of genes is sufficient to cause the genetic disorder to appear. Even though one member of the gene pair is normal, the abnormal form of the gene "dominates" over the normal one. (8) Autosomal dominant diseases include Huntington's disease, neurofibromatosis, polycystic kidney disease, and Marfan syndrome. (9) When one parent is affected by an autosomal dominant disorder and the other is not, the child has a 50% chance of inheriting the disorder. (10)

In the case of an *autosomal recessive* genetic disorder, both members of the gene pair must be abnormal for the disorder to occur. Both parents typically have a single abnormal gene on one chromosome but a normal copy of the gene on the other chromosome which prevents the appearance of the disease in the carrier parent. A child must get one abnormal gene from each parent in order to be affected by the disease. (11) Examples of autosomal recessive disorders are phenylketonuria (PKU), cystic fibrosis (CF), Tay-Sachs disease, sickle cell disease, and hemochromatosis. (12) Parents who are both carriers of an autosomal recessive disorder have a 25% chance of having a child affected by the disorder, a a 50% chance of having a child who is not affected by the disorder but who is a carrier of the genetic abnormality, and a 25% chance of having a child who will not be affected by the disorder and who will not be a carrier. (13) These probabilities apply to each conception.

An *X-linked recessive* genetic disorder involves an abnormal gene on the X chromosome, one of the sex chromosomes. Since males have only one X chromosome, an abnormal gene on it is sufficient to cause the genetic disorder to occur. In females, who have two X chromosomes, both chromosomes must carry the abnormal gene for the genetic disorder to occur. Because it is unlikely that females will have two altered copies of the gene, males are affected by X-linked genetic disorders much more frequently than females. (14) Hemophilia, muscular dystrophy, fragile X syndrome, Menkes disease, and Lesch-Lyhan syndrome are examples of X-linked recessive genetic disorders. (15)

The pattern of inheritance is complex for an X-linked recessive genetic disorder. If the mother is a carrier of the X-linked genetic disorder but the father has a normal copy of the gene on his X chromosome, then, for each conception, the following is the case:

- There is a 25% chance that a son will be conceived who has inherited the Y chromosome from his father but the X chromosome from his mother carrying the genetic abnormality. In this case, the son will be affected by the genetic disorder.
- There is a 25% chance that a son will be conceived who has inherited the Y chromosome from his father and the X chromosome from his mother carrying the normal gene. In this case, the son will not be affected by the genetic disorder.
- There is a 25% chance that a daughter will be conceived who has inherited the X chromosome from her mother carrying the genetic abnormality and the X chromosome from her father carrying the normal gene. In this case, the daughter will not be affected by the genetic disorder but will be a carrier.
- There is a 25% chance that a daughter will be conceived who has inherited the X chromosome from her mother that carries the normal form of the gene and the X chromosome from her father carrying the normal gene. In the case, the daughter will not be affected by the genetic disorder and will not be a carrier. (16)

Looked at another way, if the woman is pregnant with a son, there is a 50% chance that the son will be affected by the genetic disorder and, if pregnant with a daughter, there is a 50% chance that the daughter will be a carrier of the genetic disorder. (17)

On the other hand, if the father is affected by an X-linked recessive genetic disorder but the mother has normal copies of the gene on both her X chromosomes, then, for every conception, the following is the case:

- All sons will inherit a normal copy of the X-linked gene from their mother and the Y chromosome from their father. They will not be affected by the genetic disorder.
- All daughters will inherit a normal copy of the X-linked gene from their mother and the abnormal X-linked gene from their father. They themselves will not be affected by the genetic disorder but will be carriers of it. (18)

An *X-linked dominant* genetic disorder again involves an abnormal gene on the X chromosome. In females, who have two X chromosomes, an abnormal gene on one of her X chromosomes is sufficient to cause the genetic disorder to occur. In males, who have only one X chromosome, an abnormal gene on that chromosome causes the genetic disorder to occur and, in most cases, males experience more severe symptoms of the disorder than females. (19) Rett syndrome, incontinentia pigmenti, and congenital generalized hypertrichosis are examples of X-linked dominant genetic disorders. (20)

If the mother is affected by an X-linked dominant genetic disorder while the father has an X chromosome with the normal form of the gene, then, for each conception, the following is the case:

- There is a 50% chance that a child (whether a son or daughter) will inherit the X chromosome from the mother with the abnormal gene and be affected by the genetic disorder,
- There is a 50% chance that a child (whether a son or a daughter) will inherit the normal copy of the X-linked gene from the mother and neither be affected by the genetic disorder nor be a carrier. (21)

If the father is affected by an X-linked dominant genetic disorder but the mother has normal copies of the gene on both her X chromosomes, then, for every conception, the following holds true:

- All daughters will inherit an X chromosome with the normal gene from the mother and an X chromosome with the abnormal gene from the father, and will be affected by the genetic disorder.
- All sons will inherit an X chromosome with the normal gene from the mother and the Y chromosome from the father, and will not be affected by the genetic disorder. (22)

A *Y-linked* genetic disorder involves an abnormal gene on the Y chromosome. This genetic disorder is passed on only to sons and to all of them. (23) An example of a Y-linked genetic disorder is Y chromosome infertility, which impairs fertility in males. (24)

Multifactorial disorders are due to a combination of genetic and other factors, such as lifestyle and environment. (25) Defective genes which are inherited predispose individuals to a condition, but other factors are necessary for the disease to actually occur and the condition may never in fact develop. (26) Multifactorial disorders include such conditions as cleft palate/lip, neural tube defects (e.g., anencephaly and spina bifida), diabetes, breast and ovarian cancer, and late onset Alzheimer's disease. (27)

Chromosomal disorders are of two types. Some are caused by changes in the number of chromosomes, with an individual having either an extra or a missing copy of a chromosome. (28) Examples include Down syndrome, Edward syndrome, Patau syndrome, Klinefelter syndrome, and Turner syndrome. (29) These changes are not inherited but occur as random events during the formation of eggs and sperm. (30) Other chromosomal disorders involve changes in the structure of chromosomes. For example, a segment of a particular chromosome can be deleted, as occurs in cri-du-chat syndrome. (31) Some changes in chromosome structure can be inherited while others occur as random events during the formation of eggs and sperm or in early fetal development. (32)

A couple needs to keep in mind that each conception is an independent event and that the same probability holds for each conception for having a child with a genetic disorder. In other words, already having a child with a particular genetic disorder does not decrease the probability of having another child with the disorder.

Reproductive Options

Today a couple at risk for having a child with a genetic disorder has the following reproductive options:

- Take a chance and conceive naturally, with the intent of accepting the child whatever his/her condition.
- Conceive naturally but undergo prenatal diagnosis to determine the genetic status of the fetus, with the intent of aborting a fetus affected by the genetic disorder.
- Forgo having their own biological children (and possibly adopt).
- Use an assisted reproductive technology to avoid the child being affected by the genetic disorder. This includes:

    * Using donor sperm from someone not affected by the genetic disorder to conceive a child through artificial insemination or in vitro fertilization (e.g., if the father has an autosomal dominant genetic disorder, or both mother and father are carriers of an autosomal recessive genetic disorder).

    * Using donor ova from someone not affected by the genetic disorder to conceive a child through in vitro fertilization (e.g., if the mother has an autosomal dominant genetic disorder, or both mother and father are carriers of an autosomal recessive genetic disorder).

    * Using in vitro fertilization with their own sperm and ova to produce embryos which will undergo preimplantation genetic testing, with only those embryos free of the genetic disorder transferred to the woman's body to try to achieve a pregnancy. (For

discussion of the use of this technology in the case of sex-linked genetic disorders, see section 6 Sex Selection.)

Moral Assessment of Reproductive Options

Official Church teaching neither excludes nor endorses the option of taking a chance on conceiving naturally a child who may be affected by a genetic disorder.

Christians affirm "the fully human dignity of every human person, regardless of disability or sickness." (33) At the same time, one finds contemporary Catholic theologians who urge caution with respect to the option of simply taking a chance on conceiving a child who may be affected by a genetic disorder. As ethicists Ashley and O'Rourke comment:

> In the past, some would have argued that a person or couple at risk of begetting a defective child or children, or of transmitting defective genes to future generations, should fatalistically marry and beget children and "leave it to God." This fatalism, as already pointed out, has not been as damaging to society as some eugenic enthusiasts have thought.... Nevertheless, Christian teaching does not favor fatalistic attitudes, but rather advocates parental responsibility. (34)

In the next section we will discuss factors to take into account in exercising such "parental responsibility" in reproduction.

If a couple does opt to "take a chance," undergoing prenatal diagnosis to ascertain the condition of the fetus is not precluded provided that the information obtained will be used to guide care for the child if the diagnosis reveals a genetic disorder. On the other hand, **prenatal diagnosis is not morally permissible when undertaken with the intention of aborting a child with a genetic disorder.** (35) If a couple does opt to "take a chance," they must be prepared to accept the child, whatever his or her genetic condition.

While Pope Pius XII condemned the eugenic sterilization practices which occurred prior to and during World War II (36), and condemned practices of forbidding individuals to marry on account of genetic disorders (37), he also cautioned that "it is certainly right, and in most cases an obligation, to point out to those who are bearers of extremely undesirable hereditary factors what a burden they are about to impose on themselves, their mates, and their offspring; a burden which might become intolerable." (38) In an Allocution to Midwives given on October 29, 1951 Pius XII explicitly spoke of the permissibility of forgoing having children for genetic reasons:

> Serious reasons, often put forward on medical, *eugenic*, economic and social grounds, can exempt from that obligatory service [the duty of providing for the conservation of the human race] even for a considerable period of time, *even for the entire duration of the marriage*. It follows from this that the use of the infertile periods can be lawful from the moral point of view and, in the circumstances which have been mentioned, it is indeed lawful. [italics added] (39)

Thus **the option of a couple deciding to forgo having their own biological children because of a genetic disorder may be a morally legitimate option for a couple *with certain restrictions*.**

The first restriction concerns methods of birth control. *Even in the case of a genetic disorder, neither sterilization nor artificial means of contraception is morally permissible. Couples must try to avoid the conception of a child through methods of natural family planning* (otherwise known as *fertility awareness based methods of family planning).* (40) For information about natural family planning services available in the Archdiocese of Dubuque, consult the Marriage and Family Life Office at http://www.dbqarch.org/familylife.

The second restriction comes from Canon Law governing the validity of marriage. According to Canon Law, what is required for marriage is an exchange of the *right* to children. Although this requirement is commonly expressed in terms of an *intention* to have children, the correct interpretation of Canon Law deals with *exchanging a right* rather than with the actual conception of children. Thus, if a couple *mutually agrees* not to exchange that right (i.e., mutally agree to forgo having children), their marriage is not invalidated, provided that they would accept a child should conception occur. The condition of *mutual agreement* is critical. If, in the course of the marriage, one spouse changes his/her mind and wants children, then the other spouse is not free to exclude that right and would be obligated to provide for the exercise of that right. Failing to do this would invalidate the marriage. (41)

A genetic counselor may well talk to a couple about assisted reproductive technologies which would allow them to avoid having a child with a genetic disorder. However, **none of these technologies -- use of donor sperm or ova, artificial insemination by donor, in vitro fertilization, preimplantation genetic testing – is morally permissible according to Church teaching**. See in this *Handbook* sections 1.2 Guiding Values and Principles, 2.2. Artificial Insemination by Donor, 2.3 In Vitro Fertilization, and 5.1 Preimplantation Genetic Testing.

The Morally Legitimate Reproductive Options: Guidelines for Decision Making

In accord with Church teaching, a couple at risk for having a child with a genetic disorder has two reproductive options to consider:

- **Take a chance and conceive naturally, with the intent of accepting the child whatever his/her condition.**
- **Forgo having their own biological children.**

Official Church teaching does not go so far as to guide the choice between these two options. However, guidelines for decision making can be drawn from the writings of various theologians and ethicists.

In making reproductive decisions a couple should consider not only their own desire to have a child but *the welfare of the child himself or herself***.** (42) Considering the welfare of the child means considering *the kind of life a child will have if affected by a genetic disorder*. Thus

responsible reproductive decision making will include taking the following considerations into account:

- *What is the nature and severity of the genetic disorder? What is the gravity of impairment it brings?*
- *What is the degree of risk of transmitting the genetic disorder?*
- *What therapies are available to manage the physical condition of the child?*
- *What social resources are available to ameliorate the condition of the child (e.g., modifications of the environment, special education, governmental assistance programs)?* (43)

For example, consider the genetic disorder PKU (phenylketonuria). It involves the absence of or a defect in the enzyme processing the amino acid phenylalanine. As a result, phenylalanine accumulates in brain cells, leading to serious mental deficits. PKU is a recessive genetic disorder, so that there is a 25% chance of a child being affected if the parents are both carriers. However, if diagnosed early in life, a child with PKU can be placed on a special diet extremely low in phenylalanine from birth to at least 8 years of age, which allows normal brain development. In the United States, all newborns are screened for PKU. (44) A screening program for PKU coupled with an effective therapy for it provide a very positive outcome for a child with PKU. The child's life will not be compromised by the genetic disorder of PKU.

In terms of the severity of impact on a child, a contrasting case is Lesch-Nyhan syndrome. This genetic disorder is characterized by impaired kidney function, acute gouty arthritis, and neurological and behavioral abnormalities. (45)

> Neurological symptoms associated with Lesch-Nyhan syndrome usually begin before the age of 12 months. These may include involuntary writhing movements of the arms and legs (dystonia) and purposeless repetitive movements (chorea) such as flexing of the fingers, raising and lowering of the shoulders, and/or facial grimacing. Infants who had previously been able to sit upright typically lose this ability. Initially, muscles may be soft (hypotonia) and lead to difficulty in holding the head in an upright position. Affected infants may fail to reach developmental milestones such as crawling, sitting or walking (developmental delay). Eventually, most children with Lesch-Nyhan syndrome experience abnormally increased muscle tone (hypertonia) and muscle rigidity (spasticity). Deep tendon reflexes are increased (hyperreflexia). Intellectual disability may also occur and is typically moderate…
>
> The most striking feature of Lesch-Nyhan syndrome, which has been observed in approximately 85 percent of patients, is self-mutilation. These behaviors most often begin between the ages of two and three years. …Self-injurious behavior may include repeated biting of the lips, fingers, and/or hands, and repetitive banging of the head against hard objects. Some children may scratch their face repeatedly. …Additional behavioral abnormalities include aggressiveness, vomiting, and spitting. Self-mutilating behaviors regularly lead to loss of tissue. (46)

Treatments for patients with Lesch-Nyhan syndrome include a drug regimen to control the excessive amounts of uric acid associated with this syndrome, and treatment for kidney stones. (47) At present, "no sustained treatment or drug therapy has proven uniformly effective for the treatment of the neurological problems associated with Lesch-Nyhan syndrome." (48) In terms of their behavioral symptoms, "children with Lesch-Nyhan syndrome usually require physical restraint at the hips, chest, and elbows so they do not injure themselves." (49) Their "biting of fingers and/or lips, which can lead to permanent disfigurement, may be prevented by use of a mouth guard (oral prosthetic) or the removal of the teeth." (50)

Lesch-Nyhan syndrome is an X-linked recessive genetic disorder. (51) If the mother is the carrier, then there is a 50% chance that a son will be affected by this genetic disorder and a 50% chance that a daughter will be a carrier of the genetic disorder, who can subsequently pass it on. (52)

A factor that complicates reproductive decision making is the fact that, for some genetic disorders, the severity of the symptoms varies from individual to individual. An example is Down syndrome. The risk of this chromosomal disorder occurring significantly increases for women older than their early thirties who wish to have a child. (53) Individuals affected by Down syndrome have characteristic facial features and often health problems including heart defects, intestinal malformations requiring surgery, vision problems, hearing loss, and susceptibility to infections. (54) They are typically short with round, moonlike faces, have protruding tongues, and their eyes slant upward at the corners. (55). However, "the severity of these problems varies greatly among affected individuals." (56) Individuals with Down syndrome suffer intellectual disability, but in degree this ranges from mild (IQ of 50- 69) to moderate (IQ of 35 – 50) to severe (IQ of 20 – 35). (57) The challenge posed for reproductive decision making is that one does not know in advance how severely a child will be affected by this disorder.

Another complicating factor is the fact that genetic disorders vary in terms of the time of onset of symptoms along the span of a human life. (58) In some cases, the genetic disorder is "late-onset." (59) A prime example is Huntington's disease (HD). It is an automsomal dominant genetic disorder so that, if a parent has the gene variant for HD, there is a 50% chance of a child inheriting HD. (60) The symptoms are serious and include involuntary movements of all parts of the body, deterioration of cognitive function, and often severe emotional disturbance. (61) There is currently no cure for HD (62), and the time from onset of symptoms to death averages 15 years. (63) However, the onset of symptoms usually occurs between 30 and 50 years of age. (64) Prior to this time, the individual can have a perfectly "normal" and productive life. (65)

Yet another complicating factor in reproductive decision making is that knowledge of disabilities and impairments resulting from a genetic disorder will not tell us about *abilities* a particular child may have --- artistic talents, wit, or intellect. (66) One example is Blaine Deatherage-Newsome who has spina bifida, a multifactorial disorder. Although paralyzed from the armpits down, he was an A student in high school and a national chess champion. (67) As Adrienne Asch (an advocate for the disabled) has pointed out, "Disabled people contribute to society not because of their impairments…but because in addition to their impairments they have inherently valuable qualities no different from those of the nondisabled." (68)

The risk of transmitting a genetic disorder has been mentioned as a factor to take into account in reproductive decision making. This risk is usually given in probability values, e.g., 25%, 50%. Those counseling couples making reproductive decisions need to recognize that there is a difference between *numerical probability values* and an individual's *perception* of risks. As Sidney Callahan observes:

> ...different persons can view an objective probability and a risk to be taken in very different ways. A one-in-a-hundred chance of some future happening may be perceived very differently by different persons. It also may make a difference whether the uncertain future happening is positive or negative. Subjective probability and objective probability are almost always discrepant and differentiated according to individual differences in personality and attitude. Attitudes may also change after experiencing certain events.
>
> The experience of previous outcomes makes a difference in subjective assessments of probability, but not always in a strictly logical way. Parents who have experienced some negative outcome with a previous child may view the probability of a similar event occurring again with quite different subjective assessments. Some parents would become far more conservative in ever taking such a risk again, while others might be more positive in future risk-taking. Those who were more positive might be reasoning under the faulty gambler's fallacy which hopes each event in an independent series is not independent, i.e., that after a number of boys in a family one is more likely to get a girl. Or those parents who are more positive in taking a second time risk that would be unacceptable to others, may have found by their previous experience that the supposedly negative outcome was not so dreadful after all. (69)

Various studies indicate that probability values for the risk of transmitting a genetic disorder are tempered by such factors as the desire to have children, past reproductive experiences, and the perceived burden of the disorder. (70)

In reproductive decision making, a couple may also need to consider *the potential impact on already existing children of having another child in the family affected by a genetic disorder*. Would the care of such a child prevent or substantially hinder appropriate care for already existing children? For example, suppose that a couple already has one child affected by cystic fibrosis (CF), an autosomal recessive genetic disorder (71) which affects the respiratory, pancreatic, gastrointestinal, and reproductive systems and which results in the accumulation of mucus which makes it difficult to breathe properly. (72) Daily care for a child with CF can include "lung clearance therapies like chest physiotherapy (CPT) and vest therapy, bronchodilators, steroids, anti-inflammatory medicines, or antibiotics." (73) In addition, the child may "need to take enzymes, and mineral and vitamin supplements to help with digestion and calorie absorption." (74) Taking care of a child with CF, when the child is healthy, "is a task that can take between one and two hours a day, including medicines and airway clearance," and, "when the child experiences disease exacerbations or suffers from any other condition, more time is needed for care." (75) Indeed, due to the demands of caregiving, one of the parents may stop working. (76) If a couple already has one child affected by cystic fibrosis, the demands of his or her care might not be compatible with having another CF child requiring the same level of specialized care. (77) Or even if existing children escaped inheriting the genetic disorder and are

physically normal, a couple might want to consider whether the needs of another child affected by a genetic disorder would substantially compromise what they could provide for already existing children financially, in terms of time spent with their children, and the like. Again using CF as an example, it has been noted that the parents of a child with CF "need to be particular[ly] careful since brothers and sisters of children with CF may feel left out, neglected, or jealous, due to the extra care given to their sick sibling." (78)

Finally, a couple may wish to think about *the impact on themselves of having a child affected by a genetic disorder.* For example, it has been found that "parents of children with cystic fibrosis are more likely to experience anxiety, depression, or both compared to parents in general." (79) Does the couple have the physical and emotional stamina to deal with prolonged caregiving responsibilities? Are there support groups available to assist them in coping? How will time commitments to caregiving for the child affect the couple's personal relationship with each other? Are respite services available which will give the couple time to themselves? (80)

In sum, there is no magic formula which a couple at risk for having a child with a genetic disorder can use to decide between the options of "taking a chance" and conceiving naturally (with the intent of accepting the child whatever his/her condition) and forgoing having biological children. The factors to take into account are complex, and decisions must be made on a case-by-case basis. Genetic disorders are very different in nature, and the couple must become thoroughly informed medically about the particular genetic disorder at issue for them. A couple must also consider the particularities of their own situation.

Notes

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2. American Medical Association, *Ethics Genetic Testing & Counseling* at https://www.ama-assn.org/delivering-care/ethics/genetic-testing-counseling, accessed November 2019; Encyclopedia.com, *Genetic Counseling, Ethical Issues In* at https://www.encyclopedia.com/ cience/encyclopedias-almanacs-transcripts-and-maps/genetic-counseling-ethical-issues, accessed November 2019.

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4. Laura M. Gunder and Scott A. Martin, *Essentials of Medical Genetics for Health Professionals* (Sudbury, MA: Jones & Bartlett Learning, 2011), pp. 2-3.

5. Ricki Lewis, *Human Genetics Concepts and Applications*, 5th ed. (Dubuque, IA: Mc-Graw Hill, 2003), p. 68.

6. For example, hemochromatosis is an iron storage disorder resulting in excessive iron deposits in organs but it can be effectively controlled by a regimen of phlebotomies (bloodletting). Joseph S. Alper, Lisa N. Geller, Carol I. Barash, Paul R. Billings, Vicki Laden, & Marvin R. Natowicz, "Genetic Discrimination and Screening for Hemochromatosis," *Journal of Public Health Policy* 15/3 (Spring 1994): 345-58 at 346.

7. Gunder and Martin, *Essentials of Medical Genetics for Health Professionals,* p. 212.

8. Medline Plus, *Autosomal Dominant*. https://medlineplus.gov/ency/article/002049.htm. Accessed October 2019.

9. Lewis, *Human Genetics Concepts and Applications*, p. 81.

10. *Ibid.*

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13. *Ibid.,* p. 83.

14. U.S. National Library of Medicine Genetics Home Reference, *What are the different ways in which a genetic condition can be inherited?* https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns. Accessed November 2019.

15. Centre for Genetics Education, *Fact Sheet 9 X-Linked Recessive Inheritance* at http://genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-9-x-linked-recessive-inheritance, accessed November 2019; Lewis, *Human Genetics Concepts and Applications*, p. 124.

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21. Centre for Genetics Education, *Fact Sheet 10 X-Linked Dominant Inheritance.* http://genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-10-x-linked-dominant-inheritance. Accessed November 2019.

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23. Genetic and Rare Diseases Information Center, *Y Chromosome Infertility*. https://rarediseases.info.nih.gov/diseases/185/y-chromosome-infertility. Accessed November 2019.

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26. Centre for Genetics Education, *Fact Sheet 11 Environmental and Genetic Interactions*.

27. *Ibid.*; Children's Wisconsin, *Multifactorial Inheritance* at https://www.chw.org/medical-care/ genetics-and-genomics-program/medical-genetics/multifactorial-inheritance, accessed November 2019.

28. U.S. National Library of Medicine Genetics Home Reference, *Are chromosomal disorders inherited?* https://ghr.nlm.nih.gov/primer/inheritance/chromosomalinheritance. Accessed November 2019.

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30. U.S. National Library of Medicine Genetics Home Reference, *Are chromosomal disorders inherited?*.

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33. Ronald Cole-Turner & Brent Waters, *Pastoral Genetics Theology and Care at the Beginning of Life* (Cleveland: Pilgrim Press, 1996), pp. 141-42.

34. Benedict Ashley, OP & Kevin O'Rourke, OP, *Health Care Ethics A Theological Analysis*, 4th ed. (Washington, DC: Georgetown University Press, 1997), p. 329.

35. United States Conference of Catholic Bishops, *Ethical and Religious Directives for Catholic Health Care Services,* 6th ed. (2018), no. 50: "Prenatal diagnosis is permitted when the procedure does not threaten the life or physical integrity of the unborn child or the mother and

does not subject them to disproportionate risks; when the diagnosis can provide information to guide preventative care for the mother or pre- or postnatal care for the child; and when the parents, or at least the mother, give free and informed consent. Prenatal diagnosis is not permitted when undertaken with the intention of aborting an unborn child with a serious defect." http://www.usccb.org/about/doctrine/ethical-and-religious-directives. Accessed July 2019.

36. Pius XII, Address to the Participants of the Primum Symposium Geneticae Medicae, September 8, 1953 in *The Pope Speaks The Teachings of Pope Pius XII* (Pantheon, 1957), p. 124.

37. *Ibid.*

38. *Ibid.*

39. Pius XII, "Fundamental Laws Governing Conjugal Relations," Allocution to Midwives, October 29, 1951 in The Monks of Solesmes (ed.), *The Human Body Papal Teachings* (Boston: Daughers of St. Paul, 1960), pp. 164-65.

40. United States Conference of Catholic Bishops, *Ethical and Religious Directives for Catholic Health Care Services,* 6th ed. (2018):

> Direct sterilization of either men or women, whether permanent or temporary, is not permitted in a Catholic health care institution. (no. 53)
>
> Catholic health care institutions may not promote or condone contraceptive practices but should provide, for married couples and the medical staff who counsel them, instruction both about the Church's teaching in responsible parenthood and in methods of natural family planning. (no. 52)

41. Opinion of Francine Quillin, PBVM, J.C.L., with concurring opinion of Rev. Joseph L. Hauer, J.C.L., Metropolitan Tribunal, Archdiocese of Dubuque, Iowa, August 2, 1997.

42. Sidney Callahan contends that, ethically, the parental role is one of altruistic commitment to the welfare of the child:

> ...the most ethically acceptable parental motivation involves the most conscious commitment to giving a unique child what the child needs for its fulfillment. People who want to be parents should want to be parents in order to give, not get. It is immoral and unethical to knowingly want a child to satisfy needs or desires which take no account of the child as a human person with intrinsic rights to dignity and nurturance. While many have claimed that parental giving, protecting, and advocacy of one's children within the family framework produces one of the greatest of life's satisfactions and joys, still the ethical essence of the parental role is altruism and unselfish nurturance of another. (Sidney Callahan, "An Ethical Analysis of Responsible Parenthood" in Alexander M. Capron, Marc Lape, Robert F. Marray, Jr., Tabitha M. Powledge,

> Sumner B. Twiss, & Daniel Bergsma (eds.), *Genetic Counseling: Facts, Values, and Norms* (New York: Alan R. Liss, 1979), pp. 223-24.)

Callahan traces out the implications of this view for reproductive decision making:

> Perhaps the most important ethical principle involved in reproductive risk-taking is to have a concern for the potential third party who may be involved. No potential parent or parents can judge risks simply upon their own ability and willingness to sacrifice for the consequences. Will an innocent third party, i.e., one's own child, have to bear the burden or suffer from parental risk-taking in reproduction in a disproportionate way? (*Ibid*., p. 231.)
>
> But the primary guiding ethical principle of decision-making seems as inevitable as in considerations of parental motivation, parental control and parental risk-taking. What will be best from the potential child's point of view? Any parents contemplating entry into the basically altruistic role of parenting can hardly justify putting their own motivations, desires, or even ideologies ahead of their child's welfare. (*Ibid*., p. 236.)

43. See, for example, Bonnie Steinbock & Ron McClamrock, "When Is Birth Unfair to the Child?," *Hastings Center Report* 24/6 (Nov.-Dec. 1994): 15-21 at 21; Callahan, "An Ethical Analysis of Responsible Parenthood," p. 231; Ashley & O'Rourke, *Health Care Ethics*, pp. 329-30.

44. Lewis, *Human Genetics Concepts and Applications*, pp. 81, 97; Gunder and Martin, *Essentials of Medical Genetics for Health Professionals,* p. 27; Children's Hospital of Wisconsin, *Genetic Disorders* at https://chw.org/medical-care/fetal-concerns-center/conditions/infantcomplications/genetic-disorders, accessed November 2019.

45. National Organization for Rare Disorders, *Lesch Nyhan Syndrome* at https://rarediseases.org/rare-diseases/lesch-nyhan-syndrome, accessed November 2019; Genetics Home Reference, *Lesch-Nyhan syndrome* at https://ghr.nlm.nih.gov/condition/ lesch-nyhan-sydrome, accessed November 2019.

46. National Organization for Rare Disorders, *Lesch Nyhan Syndrome.*

47. *Ibid.*

48. *Ibid.*

49. *Ibid.*

50. *Ibid.*

51. *Ibid.*

52. Centre for Genetics Education, *Fact Sheet 9 X-Linked Recessive Inheritance.*

53. Gunder and Martin, *Essentials of Medical Genetics for Health Professionals,* pp. 5-6.

54. *Ibid.,* pp. 5, 7.

55. *Ibid.,* p. 5.

56. *Ibid.,* p.5.

57. *Down Syndrome*. Wikipedia. https://en.wikipedia.org/wiki/Down_syndrome. Accessed November 2019.

58. Lewis, *Human Genetics Concepts and Applications*, p. 68.

59. "Late-onset genetic disorders Some genetic disorders are only expressed later in life, often after the reproductive years. This means that a person who may develop the disease has still been able to pass on the disorder to his descendants before he or she develops the symptoms." Centrum voor Medische Genetics, *Inheritance*. http://www.brusselsgenetics.be/late-onset-genetic-disorders?doscroll=true#L4-5603. Accessed November 2019.

60. Gunder and Martin, *Essentials of Medical Genetics for Health Professionals*, p. 47.

61. *Ibid.*

62. *Ibid.*, p.49.

63. *Ibid.,* p. 47.

64. *Ibid.,* p. 48.

65. Consider, for example, the case of Arlo Guthrie, son of the famous American folk singer Woody Guthrie:

> Arlo Guthrie seems to have taken the optimistic point of view. His father, Woody Guthrie, died of Huntington disease, which means that Arlo has a 50 percent risk of developing it himself. Despite this, Arlo decided to have children... Of course, if Arlo did not have the disease, as now seems likely, then his children would not be affected. But he could not have known this at the time he decided to procreate. Therefore, he was taking a 50 percent chance that he would be passing on the Huntington disease to his children. How could he justify this risk? Perhaps he reasoned as follows: "I haven't had a tragic life. I've had a very good life. The goodness of my life will not be destroyed if it turns out that I have Huntington. I won't regret having been born, or resent my parents for having had me. So why is it wrong for me to have kids? I can give them a decent chance

> for a good life--as good a life as I had." (Steinbock & McClamrock, "When Is Birth Unfair to the Child?," p. 20.)

66. Lecture by Adrienne Asch, College Course 25 Ethical, Legal, and Social Implications of the Human Genome Project, Dartmouth College, July 10, 1997.

67. Ricki Lewis, *Human Genetics Concepts and Applications*, 2nd ed. (Dubuque, IA: Wm. C. Brown, 1997), p. 273. Blaine Deatherage-Newsome has himself testified to the value of his life:

> I was born with spina bifida and hydrocephalus. I hear that when parents have a test and find out that their unborn child has spina bifida, in more than 95 percent of the cases they choose to have an abortion. I also went to an exhibit at the Oregon Museum of Science and Industry several years ago where the exhibit described a child born with spina bifida and hydrocephalus, and...asked people to vote on whether the child should live or die. I voted that the child should live, but when I voted, the child was losing by quite a few votes.
>
> When these things happen, I get worried. I wonder if people are saying that they think the world would be a better place without me. I wonder if people just think the lives of people with disabilities are so full of misery and suffering that they think we would be better off dead. It's true that my life has suffering (especially when I'm having one of my eleven surgeries so far), but most of the time I am very happy and I like my life very much. My mom says she can't imagine the world without me, and she is convinced that everyone who has a chance to know me thinks that the world is a far better place because I'm in it. (*Ibid.*, pp. 272-73)

68. Adrienne Asch, "Reproductive Technology and Disability" in Sherrill Cohen & Nadine Taub (eds.), *Reproductive Laws for the 1990s* (Clifton, NJ: Humana Press, 1989), p. 75.

69. Sidney Callahan, "An Ethical Analysis of Responsible Parenthood," pp. 230-31.

70. Petra G. Frets, Hugo J. Duivenvoorden, Frans Verhage, Martinus F. Niermeijer, Sophie M.M. van de Berge & Hans Galjaard, "Factors Influencing the Reproductive Decision After Genetic Counseling," *American Journal of Medical Genetics* 35/4 (April 1990): 496-502 at 496, 501; Petra G. Frets & Martinus F. Niermeijer, "Reproductive planning after genetic counselling: a perspective from the last decade," *Clinical Genetics* 38/4 (Oct. 1990): 295-306 at 301-2; Evelyn Parsons & Paul Atkinson, "Genetic Risk and Reproduction," *Sociological Review* 41/4 (Nov. 1993): 679-706 at 687, 693-94, 702, 704.

71. Gunder and Martin, *Essentials of Medical Genetics for Health Professionals*, p. 107.

72. Cystic Fibrosis News Today, *Caring for a Family Member with Cystic Fibrosis*. https://cysticfibrosisnewstoday.com/caring-for-a-family-member-with-cystic-fibrosis. Accessed November 2019.

73. *Ibid.*

74. *Ibid.*

75. *Ibid.*

76. *Ibid.*

77. Video *Genetic Screening: The Ultimate Preventive Medicine?* (KCTS/Seattle, 1980).

78. Cystic Fibrosis News Today, *Caring for a Family Member with Cystic Fibrosis*.

79. Cystic Fibrosis Foundation, *Coping While Caring for Someone With Cystic Fibrosis*. https://www.cff.org/Life-With-CF/Daily-Life/Emotional-Wellness/Coping-While-Caring-for-Someone-With-Cystic-Fibrosis. Accessed November 2019.

80. Elsa Shapiro, Charles Marques Lourenco, Neslihan Onenli Mungan, Nicole Muschol, Cara O'Neill and Suresh Vijayaraghavan, "Analysis of the caregiver burden associated with Sanfilippo syndrome type B: panel recommendations based on qualitative and quantitative data," *Ophanet Journal of Rare Diseases* 14/168 (8 July 2019): 1-9 at 7.