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GENETIC ANOMALY OR GENETIC DIVERSITY: THINKING IN THE KEY OF DISABILITY ON THE HUMAN GENOME

MARY JO IOZZIO

[Thinking in the key of disability reconfigures scientific presumptions to accept identified genetic anomalies as instances of the great diversity possible in the human genome. While genetic testing and diagnoses advance, the secrets of 30,000 genes in human DNA yield slowly, providing remedy only rarely. Promises aside, genomic medicine can relieve suffering or further oppress people with disabilities but Christians must reclaim those who have been exposed, sterilized, euthanized, aborted, or institutionalized by their nonconformity to an artificially constructed genetic norm.]

ARGUABLY, GENETIC DIVERSITY is as perplexing a reality as the cultural, ethnic, linguistic, racial, and sexual diversity of the pluralist and liberalized democracies of our Western world. With the advent of genomic medicine the questions of genetic diversity come to the fore, and the need to respond appropriately to popular and scientific ambivalence is as pressing as ever. On the one hand, genomic medicine—in the forms of adult genetic testing, preimplantation embryo and prenatal diagnosis, and neonatal testing—is heralded as the answer to all manner of human disease, disability, or discomfort. On the other hand, this medicine is feared as the next stage in the dehumanization and manipulation of the weak and strong. Like the questions challenging miscegenation prohibitions, forced sterilization programs, and exposure of persons malformed or of the wrong/undesired sex, genomic medicine identifies these "others" in our midst before they are even born.[1] Genomic medicine promises relief of a number

MARY JO IOZZIO received her Ph.D. from Fordham University. She is currently associate professor of moral theology at Barry University, Miami Shores, Florida. In addition to her text *Self-Determination and the Moral Act: A Study of the Contributions of Odon Lottin, O.S.B.* (Peeters, 1995), she has contributed to and edited *Considering Religious Traditions in Bioethics* (University of Scranton, 2001). She has also written on the intersection of disabilities and theology for the *American Academy of Religion Spotlight on Teaching*, *Journal of Religion, Disability and Health*, *National Catholic Bioethics Quarterly*, and *Irish Theological Quarterly*.

[1] "From prenatal screening and the selective termination of 'undesirable' pregnancies to euthanasia of disabled adults, one of the biggest threats to the rights of disabled people this Millennium lies within the field of bioethics—the ethics of advances in biological medicine and science.... Human genetics poses a threat to

of conditions but this medicine also threatens a disvaluation if not the elimination of persons already exhibiting this or that undesirable trait. The ambivalence that modern societies experience, in light of communities including increasingly visible and active disabled persons, suggests rather a moment to embrace genetic diversity as the birthright of all people.

An approach to disabilities from a predominantly medical model of disease, anomaly, or abnormality to be remedied by genetic manipulations is misinformed. Many people in the disability community hold that the medical model presumes a normate position that confounds the lives and real experiences of people with disabilities and labels them deviant. Further, the medical model denies and minimizes, through a variety of remedial accommodations, the manifold disabilities that are "acceptable" in polite society, such as aging, hearing losses, and loss of visual acuity. The medical model takes as its starting proposition the presumption that the world—the nondisabled world—would be a better world if there were no (or at least fewer) people with disabilities. The medical model also suggests that embodied differences from idealized norms ought to be corrected or eliminated.[2] Although persons with disabilities surely would welcome the genetic interventions that promise relief of chronic pain, a greater ability to negotiate physical spaces, and wider participation in those activities from which they are at present excluded, the temporarily nondisabled would also benefit from these same accommodations and yet such thinking of themselves, i.e., from the key of disability and thus subject to genetic intervention, is anathema to the nondisabled.

Nevertheless, like growing old, most people will join the ranks of disabled people. Likewise, those people who will then be newly disabled will find that accommodations like "universal design," subtitles and closed-captioning, and softer or brighter light serve some of the most basic and standard needs of living. Many will accept their disabling condition(s) and many will wax philosophical about growing old or about the nondiscrimi-

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us because while cures and palliatives are promised, what is actually being offered are genetic tests for characteristics perceived as undesirable. This is not about treating illness or impairment but about eliminating or manipulating fetuses which may not be acceptable for a variety of reasons.... Disabled people have faced enforced sterilization, prenatal termination, infanticide, euthanasia and wholesale elimination. We were left out on the hills of Sparta to die, sterilized by 'caring' doctors in the US, Scandinavia and Germany, and were the first to be driven into the Nazi gas-chambers" ("Disabled People Speak Out on the New Genetics" http://www.dpieurope.org/htm/bioethics/dpsngfullreport.htm [accessed April 23, 2004 and June 27, 2005]).

[2] "Ironically the medical model's technology actually facilitates the survival of many disabled people at the same time that it pathologizes them" (Rosemarie Garland Thomson, *Extraordinary Bodies* [New York: Columbia University, 1997] 79).

nation of either traumatic accidents or crippling disease. Perhaps just as many will curse the day that their life betrayed their former conception of themselves and of their health.

As recent thinking in theology and religious studies has begun to show, reflection in the key of disability studies or from a perspective of disability yields profound insights on God's revelation to humankind in creation, in history, and in the person of Jesus of Nazareth, the Broken Bodied Risen Christ of faith.[3] Just as liberation, feminist, and other context-based theologies have shown, doing theology from a particular key privileges a perspective that has been otherwise unknown or denied, and thereby provides another understanding of God's revelation as well as another opportunity to grow in love of the other whose perspective has been ignored—or worse still—whose very person has been oppressed. To begin, I ask questions arising from proposals of interventions and modifications to the human genome or of a particular anomaly in a somatic cell line in light of the insights gained from a theologically informed perspective of disability.

These reflections point to the impermanent and hubristic attempts of genetic science to modify so-called genetic anomalies in light of the almost always temporary state of nondisabled. These reflections also suggest some of the difficulties that genetic sciences face with regard to the human genome. I suggest that, following the data of the present state of genetic therapies, most attempts at genetic modifications are "good" for the one person on whom the modification is applied and are, as such, impermanent—lasting only for the duration of that person's life.[4] I also suggest that most attempts at genetic modifications betray a very certain insolence, an impertinent disregard against persons born with a genetic anomaly. My article considers the current state of testing and prenatal diagnosis, the options available to parents when they learn of genetic transgression, and the current state of genetic therapies available to persons bearing the more common disabling conditions like Parkinson's disease, diabetes, spina bifida, achondroplasia, and Down syndrome, among others.[5] The conclusion will return to some of the doctrinal themes of creation, history, and the

[3] See, for example, the work of Jennie Weiss Block, Flavian Dougherty, Nancy Eiesland, as well as Don E. Saliers, Edward Foley, Elisabeth Moltmann-Wendell, Hilde Lindemann Nelson, J. David Smith, Ginny Thornburgh, Jean Vanier, Brent Webb-Mitchell, among others.

[4] Some scientists will argue that even somatic cell gene therapy, while ostensibly targeting a deviation in one individual's genome, migrates into the individual's germ line DNA, and as such this therapy would be reflected in subsequent progeny.

[5] The list of even common disabling conditions is large. To add just a few, for example, think of Huntington's Chorea, ADA Deficiency, SCID (severe combined immunodeficiency disorder, aka Bubble Boy Disease), Multiple Sclerosis, Cystic Fibrosis, Phenlketonuria (PKU), Polio, Sickle Cell Disease, Muscular Dystrophy, and Fragile X Syndrome.

Jesus event of crucifixion-death-resurrection with a disabled and glorified body—he rises disabled by the crucifixion—with a renewed interest in the challenges for theological reflection that attempts at genetic modification pose to those who are or will become disabled. This reflection considers both a theology of access for people with disabilities[6] and a theology of accountability for genetic scientists and all nondisabled persons in regard to those who are disabled.

CURRENT STATE OF PREIMPLANTATION, PRENATAL, AND ADULT GENETIC DIAGNOSIS

Not since the 19th-century Augustinian monk Gregor Mendel proposed the statistical probability of inheriting specific characteristics following genetically determinable lines of parent genome expression, albeit among legumes populations, has contemporary science been as close to unlocking another degree of understanding of human generation, health, and disease. Certainly, some will argue convincingly that both cloning and stem-cell isolation provide the greater part of understanding and, as with the early 1950s insight of the double helix structure of DNA, all three findings and their technologies have been compared to the first splitting of the atom. Nevertheless, with the June 2000 announcement of the near completion of the sequence of the human genome by the Human Genome Project of the U.S. Department of Energy—five years ahead of projected schedule—a new era of understanding the characteristics of genetic normalcy and abnormality has dawned.

The Human Genome Project, a 15-year effort begun in 1990, sought to identify, among other objectives, approximately 30,000 genes in human DNA and to determine the sequences of some 3 billion chemical base pairs that comprise these genes. The expectations of the information gained from this sequencing include the ability to recognize abnormal chemical sequences of genes associated with particular diseases and disabilities that will lead to ever more efficacious treatment and/or prevention of recurrence. Lasting treatment modalities are far from being realized, however, and prevention remains closely associated with selective abortion. What is more, assisted reproduction technologies have facilitated the ability to identify and prepare for or dispose of embryos that present a particular desired or undesired characteristic. Nevertheless, understanding the diversity of the human genome holds both the promises of succor and the tools of mad science reminiscent of "The Island of Dr. Moreau" (1896), "The Leech Woman" (1960), "The Fly" (1958), "The Swamp Thing" (1982), or a *Brave New World* (1932).

[6] See Jennie Weiss Block, *Copious Hosting: A Theology of Access for People with Disabilities* (New York: Continuum, 2002).

First, a Genetics Primer

The human genome consists of 46 chromosomes: 22 pairs of somatic cell chromosomes and 1 pair of germ line or sex cell chromosomes, this last chromosomal pair determines the individual as female (XX) or male (XY). These chromosome pairs resulted from the fertilization of the female gamete ovum by the male gamete sperm. Each of these gametes carries half of the chromosomal makeup of a human genome (and each fertilization produces a new human genomic phenotype). Standard nomenclature of these chromosomes follows a pattern that includes the chromosome number (between 1–22), sex, and, where variation occurs, a code indicating addition/deletion/duplication/etc on the specific affected chromosome(s), a code indicating the short (p) or long (q) arm of the chromosome, and a band number. These 46 chromosomes make the genome in fact human. Approximately 30,000 genes, determining each individual's inherited and, in some cases, autogenerated characteristics are located on these chromosomal pairs. Each of these genes is responsible alone or in tandem for a wide range of characteristics the least of which are the most obvious like sex, skin color, height, etc. Genes are identified by the letters AGTC, representing the base chemical nucleotide pairings of Adinine, Guanine, Thymine, and Cytosine. These chemicals combine along the skeletal structure of the double helix DNA to form the characteristics peculiar to an individual; as many as 3 billion chemical nucleotide pairs comprise the human genome. Given the large number of genes per human individual, it is not surprising both to discover and to uncover great genome phenotype diversity within the human population.

When it comes to the identification of a disability by the determination of genetic anomaly, however, science takes an unapologetically marginalizing, discriminatory, and impertinent turn: the diversity of the human genome is permissible only within a narrowly proscribed range.[7] Any deviation from the range signals warnings—a monster, a floppy rag doll, an idiot.[8] The range of identifiable genetic deviations fall under four types, and can number into the millions as a result of the combined permutations

[7] Peter Singer is perhaps the most vocal of contemporary philosopher voices on the subject of personhood or the lack thereof with individuals expressing anything but a chromosomal norm. He is of a mind to actively kill defective infants. He writes, "killing a defective infant is not morally equivalent to killing a person ... and very often it is not wrong at all" (*Practical Ethics* [New York: Cambridge University, 1979] 138). He is further more inclined to advocate for the rights of nonhuman animals than for persons with disabilities (embryos and fetuses are quite disposable for him).

[8] Although my thoughts on these kinds of attitudes toward persons with disabilities are somewhat anecdotal, see Mary Jo Iozzio, "Justice is a Virtue Both In and Out of Healthcare," *Irish Theological Quarterly* 63 (1998) 162–66, as well as Simi

possible of the 30,000 genes of 46 chromosomes. Genetic deviations are characterized thus as single gene, also called Mendelian disorders, chromosomal disorders, multifactorial disorders, and mitochondrial disorders. Single gene disorders are often acknowledged and documented by experiential patterns within family histories. Chromosomal disorders, also inheritable, ordinarily occur during the nuclear division of fertilization and meiosis. Multifactorial disorders result from the interaction of a number of genes; and mitochondrial disorders result from mutations in mitochondrial DNA.[9]

Since Mendel, geneticists have been able to predict the likelihood of offspring inheriting the characteristics of their parents. Among Mendelian disorders, four types are recognizable: autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive. Thus, the offspring of a woman with a single gene/autosomal dominant disorder who becomes pregnant by a man with the same single gene/autosomal dominant disorder will inherit the gene and express the disease. If both the woman and the man are carriers of a single gene/autosomal recessive disorder the offspring has a 75 percent chance of inheriting recessive disorder carrier status and 25 percent chance of expressing the disease. Some of the most known and publicized inheritable conditions are Tay-Sachs disease, muscular dystrophy, sickle cell disease, and cystic fibrosis. Especially with the recognition of a family history of these kinds of genetic disorders, many couples request genetic testing in order to determine their carrier status for the sake of the next generation. Further, in some communities (Ashkenazi Jews and African Americans), knowledge of a family history of disease will influence decisions of marriage and/or pregnancy between carriers or those already expressing disease. Lastly, with the advent of widespread genetic testing, scientists are able to remove the uncertainty of predictions; with either preimplantation or prenatal diagnosis, parents can learn the genetic status of their developing offspring.

Chromosomal abnormalities present a significantly different constellation of genetic fact. They are accidents insofar as and although they are inheritable they cannot be predicted necessarily from parental genetic histories of anomaly. Chromosomal disorders display a transgression of the

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Linton, *Claiming Disability: Knowledge and Identity* (New York: New York University, 1998).

[9] "There are literally millions of types of abnormalities." For example, 46,XX/46,XY—Normal Female/Male Karyotype; however, 46,XX,del(14)(q23) = female with 46 chromosomes with a deletion of chromosome 14 on the long arm (q) at band 23; 47,XY,+21 = male with 47 instead of 46 chromosomes with the gain of an extra copy of chromosome 21 (Down Syndrome). See "The GAPS INDEX" at http://aspin.asu.edu/geneinfo/index.html (accessed April 23, 2004 and June 27, 2005).

norm from the way that the chemical bases of genes ordinarily align themselves on the DNA skeleton. The more common transgressions of chromosomal disorders express either a chromosomal excess, such as with Trisomy 21/Down Syndrome, or they express a chromosomal defect/deficiency, such as with monosomy aneuploidy or the partial trisomy of Wolf-Hirschhorn Syndrome (a condition expressing severe growth retardation, microcephaly, and congenital heart defects).[10] Nevertheless, most of the chromosomal disorders presently identifiable (by name or significant frequency) spontaneously abort.[11] In spite of the ability to identify these conditions, chromosomal abnormalities that present a defect in the arrangement of chemical pairs or the lack of pairing differ widely in the expression of characteristic features and severity of symptoms in live births. Further, not all chromosomal abnormalities present disease in individuals. However, deletions, often more severe than additions or sex-associated transgressions like ambiguous genitalia and hermaphroditism, may present significant psychological developmental concerns.

Multifactorial and mitochondrial disorders present the least developed arena of scientific investigation in genetics. As the name suggests, multifactorial disorders involve not only one's genetic inheritance, they also involve environmental factors such as parental diet, health, and behavior and overall climate of the fertilization event. These disorders are not as readily predictable as single gene disorders or a family history of chromosomal disease. Rather these factors are combined with population studies to determine inheritable risk. Multifactorial disorders often express as diabetes, cleft lip and palate, schizophrenia, spina bifida, and congenital hip dislocation. Multifactorial disorders may be prevented by a careful diet, supplementary nutrition, and exercise programs for women during their pregnancies. Mitochondrial disorders, as the name suggests, involve the mitochondria organelles that form the cytoplasm of every cell and that are responsible for producing energy from protein interactions between these organelles to the organs of the body. Mitochondrial disorders limit cell energy production and lead to slow or stunted growth. These disorders are inherited only through the cytoplasm of the fertilized egg; transmission follows from an affected female to either her female or male offspring. Thus, the gametes of her female offspring may transmit the disorder to her subsequent offspring but the gametes of her male offspring, who may in fact be affected, will not. Mitochondrial disorders often express as neuro-

[10] Chromosomal abnormalities reach no more than .125–.2% of all live births, for a total of .6% of the total human population.

[11] "Approximately 20% of all conceptions have a chromosomal disorder. 30% of fetuses with trisomy 21, 75% of fetuses with 45X, 68% of fetuses with trisomy 18, and 43% of fetuses with trisomy 13 have been found to be spontaneously aborted during the second trimester." The GAPS INDEX.

logical differences such as Alzheimer disease, Parkinson's disease, and autism. Both multifactorial and mitochondrial disorders vary widely in severity since expression is influenced by inheritance risk, by a particular defective combination of chemical pairing and gene mutation, and by the environment—all factors which, individually, would exert only slight effects on an individual phenotype.

Second, the Current State of Genetic Testing

Genetic testing is a relatively new field. Certainly laboratory scientists have been hard at work in trying to decipher the human genome but the application of their findings for public consumption remains mired in controversy. Who will pay for the tests? Who will have access to the results? How may bringing a pregnancy to term balance the secular pressures to abort a fetus with genetic anomaly?[12] Since such a variety of genetic variation exists within human genome phenotypes (remember 30,000 genes in 46 chromosomes = some 3 billion nucleotide pairs) only 940 conditions and a general understanding of basic symptoms of those conditions are at present identifiable. Of course, with the basics of the 46 chromosomes of the human genome it is easy enough to recognize an addition or deletion of one or another gene or chromosome abnormality, as with trisomy or monosomy conditions. What is not yet known or fully understood is the resulting severity of the expression of this or that anomaly. Nevertheless, genetic testing is the best available recourse for adults wishing to know, because of a familial predisposition, their status as carriers of predictable genetic anomalies or parents wishing to know the status of their preimplantation embryo or to undergo prenatal fetal diagnosis.

Genetic testing or DNA-based tests examine DNA samples to compare one sequence with a typical human genotype sequence. The currently available tests for 940 genetic conditions cost anywhere from hundreds to thousands of dollars.[13] Most insurance providers do not cover adult testing. However, if insurance providers do cover the cost of testing they will have access to the results. When third-party providers have this access they may use the results to increase premiums or, if they are enrolling a new client, they may deny new or additional coverage based on the insured's "preexisting genetic condition." In spite of these insurance concerns, individuals

[12] Disabled People International (DPI) expresses this concern poignantly. "We are threatened when Bob Edwards, a world famous embryologist, says: 'Soon it will be a sin for parents to have a child which carries the heavy burden of genetic disease' " (DPI Europe, "Disabled People Speak on the New Genetics").

[13] See Genetic Testing at the Human Genome Project Information website, http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetest.shtml (accessed May 3, 2004 and June 27, 2005).

may request genetic testing for a number of reasons including the determination of carrier status, preimplantation genetic diagnosis, prenatal diagnosis, newborn screening, presymptomatic testing for adult-onset disorders, diagnostic confirmation, and forensic identification. Theoretically, once a determination of genetic or chromosomal anomaly is confirmed informed health and medical decisions will follow.

According to Cynthia Powell: "The most recent edition of the *Catalog of Prenatally Diagnosed Conditions* lists 940 conditions . . . including chromosome abnormalities, congenital malformations, dermatologic disorders, fetal infections, hematologic disorders, inborn errors of metabolism, tumors and cysts, and multiple congenital anomalies of unknown etiology."[14] Some of the more disabling conditions for which gene testing is available include, from the spectrum of carrier to expression: ALS (amyotrophic lateral sclerosis), cystic fibrosis (chronic infections), Duchenne muscular dystrophy (muscle wasting), dystonia (muscle rigidity), Factor V Leiden (blood-clotting disorder), Fragile X Syndrome (leading cause of mental retardation), Huntington's disease (mid-life onset, progressive and lethal), phenylketonuria (PKU—progressive mental retardation), Prader Willi/Angelman syndrome (early death), sickle cell disease (chronic pain), spinocerebellar ataxia (reflex disorders, explosive speech), spinal muscular dystrophy (lethal progressive muscle wasting), thalassemia (reduced red blood cells), Tay-Sachs disease (fatal neurological disease). Given the primer information above, for example, testing for cystic fibrosis is confirmed with an anomaly identified on chromosome 7 q31.2 (long arm 31 at band 2), testing for Fragile X-linked mental retardation syndrome is confirmed with an anomaly on the X chromosome q27–28 (long arm 27–28), testing for sickle cell disease is confirmed with an anomaly on chromosome 11 p15.5 (short arm 15 at band 5).

In terms of prenatal genetic testing, if a woman or her physician suspects the risk of fetal development anomaly early in the pregnancy she may undergo chorionic villus sampling (CVS) between 9 and 12 weeks into the pregnancy. CVS before 9 weeks has a high incidence of the fetal harm of focomelia, a condition similar to the birth defects resulting from thalidomide use in the 1950s to the 1960s to relieve morning sickness. Aware of the possible harm CVS may cause to the fetus, another screening battery was developed: the maternal triple screen (a quadruple screen is newly available though not in widespread use) which examines three substances from a pregnant woman's blood sample. The best results of the triple screen occur with testing between 16th and 17th weeks of the pregnancy (at

[14] Cynthia Powell, "The Current State of Prenatal Genetic Testing in the United States," in *Prenatal Testing and Disability Rights*, ed. Erik Parens and Adrienne Asch (Washington: Georgetown University, 2000) 47.

the start of the second trimester).[15] Low levels of maternal alpha-fetoprotein (AFP), human chorionic gonadotropin (HCG), and estriol (uE3) (the quad screen adds a test for folic acid) point to certain defects in the developing fetus including Down Syndrome, neural tube defects, and other physical defects. Following a positive/abnormal triple screen result, a woman would ordinarily undergo a fetal ultrasound to confirm gestational timing and, perhaps another CVS or amniocentesis to confirm a suspected chromosomal abnormality. Amniocentesis may be performed safely after the 15th week of a pregnancy. The maternal triple/quad screen has become almost routine care, with many health professionals recommending the screen for all pregnancies. As the practice of screening increases, insurance providers have begun to cover routinely the expense as part of appropriate prenatal care.[16] What follows with a positive screening and subsequent definitive testing result?

PARENTAL OPTIONS CONCOMITANT TO PRENATAL DIAGNOSIS WITH POSITIVE RESULTS AND THE CURRENT STATE OF GENE THERAPY

According to Ferguson and Asch: "The most important thing that happens when a child with disabilities is born is that a child is born. The most important thing that happens when a couple becomes parents of a child with disabilities is that the couple becomes parents."[17] Many well-intentioned people seem to forget both the child and her or his parents when difference is revealed as a result of prenatal genetic testing and diagnosis. The difference itself diverts attention away from the joys and hopes that ordinarily accompany news of a pregnancy or a birth announce-

[15] See WebMD Health at http://my.webmd.com/hw/being_pregnant/aa21828.asp (accessed May 4, 2004 and June 27, 2005).

[16] "As the ease of testing increases, so does the perception within both the medical and broader communities that prenatal testing is a logical extension of good prenatal care: the idea is that prenatal testing helps prospective parents have healthy babies. On the one hand, this perception is quite reasonable.... On the other hand, as long as in-utero interventions remain relatively rare, and as long as the number of people seeking prenatal genetic information to prepare for the birth of a child with a disability remains small, prospective parents will use positive prenatal test results primarily as the basis of a decision to abort fetuses that carry mutations associated with disease or disability. Thus, there is a sense in which prenatal testing is not simply a logical extension of good prenatal care" (Parens and Asch, "The Disability Rights Critique of Prenatal Genetic Testing," in *Prenatal Testing*, ed. Parens and Asch 4); see n. 14 above.

[17] Philip M. Ferguson and Adrienne Asch, "Lessons from Life: Personal and Parental Perspectives on School, Childhood, and Disability," as quoted in Philip M. Ferguson, Alan Gartner, and Dorothy K. Lipsky, "The Experience of Disability in Families: A Synthesis of Research and Parent Narratives," in ibid. 74.

ment to responses of pity, fear, and apologies. Unfortunately, both the general public and many genetic counselors are misinformed about life with a disability. Although the *Code of Ethics for Genetic Counselors* holds a position of neutrality and nondirective information regarding parental choice, in fact, most genetic counselors are biased against decisions to carry a positive test result pregnancy to term. "Most genetics services providers . . . said they would be nondirective in counseling about most conditions; however, they were personally much more willing to abort for these conditions than were primary care physicians or patients."[18]

The willingness to abort on the grounds of a positive test result betrays a commitment to the medical model of disability, a model that is rejected by the disability community as dangerous not only to the fetus but dangerous also to all people with disabilities. This rejection is not based solely on slippery slope arguments. Rather, this rejection is based on historically real oppression, discrimination, and outright exclusion of people with disabilities.[19] As noted previously, this model suggests that even the hint of embodied differences from idealized norms that arise with a disabling condition ought to be corrected or eliminated and such thinking implies the unworthiness of continued prenatal care or parental efforts to embrace that difference. Further, this model establishes a covertly discriminatory stance toward disability and disabled people that reduces the embryo, fetus, or child to her or his condition.[20]

Nevertheless, genetic counselors, by the very nature of their scientific discipline, focus their attention on positive results for pathology. These

[18] Dorothy Wertz, "Drawing Lines: Notes for Policymakers," in ibid. 278.

[19] Consider, for example, the exclusion of people with disabilities from worship and/or priestly functions in the Hebrew Bible and the Mishnah: "Once the affinity of Leviticus 21 is questioned, however, it is clear that the better translation of *vehamah ba'im Otaharat hamiqdash* is 'but these are approaching the purity of the temple' as suggested by Florentino Garcia Martinez. . . . The passage is no longer a prohibition on those who are blind and deaf serving as priests, *but it is a polemic against allowing those who are blind and deaf from even entering the temple precincts*" (Kerry H. Wynn, "The Invisibility of Disability at Qumran," paper presented to the Consultation on Religion and Disability Studies at the AAR Annual Meeting, Nashville, Tenn., November 11–20, 2000); emphasis added.

[20] That persons have been isolated into a group by nondisabled people makes them easy prey for all manner of discrimination. " '[I]f persons with disabilities are regarded as a definable group who have faced great oppression and stigmatization, then prenatal screening may be regarded as yet another form of social abuse.' This is the essence of the disability community's challenge to prenatal genetic testing. We believe that the current promotion and application of prenatal screening has a potent message that negatively affects people with disabilities, and reinforces the general public's stereotyped attitudes about people with disabilities" (Marsha Saxton, "Why Members of the Disability Community Oppose Prenatal Diagnosis and Selective Abortion," in *Prenatal Testing*, ed. Parens and Asch 148).

counselors make pathology the starting place of their counseling sessions with prospective parents regardless of whether the session is preparatory of or follows a positive screen result or a definitive diagnosis of genetic anomaly. However, with the great diversity of phenotype expression and the indeterminate severity of any number of conditions noted above, these biases reveal assumptions that any degree of positive test result signals calamity and raises cautions that persons with disabilities and their families suffer greatly on physical, emotional, intellectual, and spiritual levels.

This misconception must be challenged. Since "an increasingly dominant body of research. . . . finds aggregate patterns of overall adjustment and well-being to be similar across groups of families with and without children with disabilities . . . a significant number of parents actually report numerous benefits and positive outcomes for their families associated with raising a child with disabilities."[21] Further, as the Disabilities Movement becomes more widely acknowledged and persons with disabilities are present and welcomed in work and social settings, many persons with disabilities themselves report very full, productive, and satisfying lives. In fact today many persons with disabilities embellish nondisabled perceptions of their disability: as Cheryl Marie Wade writes of herself in the poem "The Woman with Juice," "I'm the Gimp/I'm the Cripple/I'm the Crazy Lady . . . I'm a French kiss with a cleft tongue."[22] Finally, the medical model of disability perpetuates universalized and generic myths about people with disabilities. "Even though the prototypical disabled person posited in cultural representations never leaves a wheelchair, is totally blind, or profoundly deaf, most of the approximately 40 million Americans with disabilities have a much more ambiguous relationship to the label. The physical impairments that render someone disabled are almost never absolute or static; they are dynamic, contingent conditions affected by many external factors and usually fluctuating over time."[23] In the same way, the physical endowments that render a person "abled" or "nondisabled" are also never absolute or static; endowments too are dynamic, contingent conditions that are affected by environmental, social, educational, familial, and economic factors that fluctuate over time.[24] To reduce a person to her or his disability makes as little

[21] Ferguson, Gartner, and Lipsky, in ibid. 85.

[22] Cheryl Marie Wade, "The Woman with Juice," as quoted in Thomson, *Extraordinary Bodies* 25.

[23] Thomson, *Extraordinary Bodies* 13.

[24] Consider, for example, the experience of the Martone family, when their only daughter was hit by a car [February 22, 1998], remained unconscious for months, and has since undergone years of attentive familial and specialized care. "One moment my daughter was an extremely independent, resident assistant, Phi Beta Kappa fourth-year student at the University of Chicago who left her dorm room to go to dinner. Five minutes later she was a totally dependent severely brain-injured

sense as to reduce a person to her skin color or his sexual orientation; these are important features but they are not definitive of persons in their totality.

In spite of this pathologizing, abortion remains a problematic response to a positive prenatal genetic diagnosis. The option to abort for any reason, as U.S. laws currently allow, confounds a respect for the inherent dignity of the fetus, refuses hospitality to an as yet unknown sister or brother who sojourns intimately within a mother's womb and through wonderful developmental stages, and insults as well as blasphemes against God in whose image this new one is created. Moreover, even if one were to concede abortion for the most severe kinds of disabling conditions, like Tay Sachs or anencephaly which express both a failure to thrive and very early demise, the option to abort is tragic. Further, as the disabilities literature shows, selective abortion on fetal indications of a positive test result, first, "expresses negative or discriminatory attitudes not merely about a disabling trait, but about those who carry it. Second, it signals an intolerance of diversity not merely in the society but in the family, and ultimately it could harm parental attitudes toward children."[25] In fact, selective abortion based on a prenatal diagnosis of genetic anomaly reduces the fetus and ultimately the person with a disability to their genetic characteristics. Just as it is foolhardy to reduce moral argumentation to a static interpretation of the natural law or to a biological determinism, it is foolhardy to reduce anyone to their genetic phenotype and on that basis to project the totality of their existence or, in the case of a positive test result, to deny continued existence, birth, childhood, and life.[26]

Curiously, on the other hand, the decision to carry a pregnancy to term in which a positive test result has been made is often met with disbelief.[27] However, the decision to carry the pregnancy to term should be met with

young woman" (Marilyn Martone, "Making Health Care Decisions without a Prognosis: Life in a Brain Trauma Unit," *Annual of the Society of Christian Ethics* 20 [2000] 325).

[25] Parens and Asch, "The Disability Rights Critique of Prenatal Genetic Testing," in *Prenatal Testing* 13.

[26] "Reduced to a Gene. A person with a disability could presumably ascribe any number of different meanings to the existence of PND [Prenatal Diagnosis]. Yet there is remarkable congruence in the writing about this issue coming from people with disabilities around the world. All these writers identify this theme: 'These technologies make us feel devalued as human beings.' For people with disabilities, 'the message' implicit in the practice of abortion based on genetic characteristics is, as Deborah Kaplan outs it: 'It is better not to exist than to have a disability.' Your birth was a mistake. Your family and the world would be better off without you alive" (Saxton, in *Prenatal Testing* 160).

[27] See, for example, Martha Beck, *Expecting Adam* (New York: Penguin/Berkley, 2000); and Mary Jo Iozzio, "Justice is a Virtue" 151–66, esp. 161–66.

congratulations (remember, there is a child to be born and parents to become). On the part of the medical professionals and genetic counselors, information regarding the disability as well as meetings with children who are similarly diagnosed, meetings with parents of children with the identified disability, literature about the disability, and support group lists that are readily available on numerous websites should be made available to these parents.[28]

Genetic therapies and genomic medicine continues at a steady pace. Research protocols for somatic cell gene therapy presently dominate the field; germ line interventions require an as yet developing technical expertise in microsurgery that attempts to delete and replace a defective gene in the embryo before it is transferred to the mother's womb or to incise the ova or sperm cells of afflicted adults before in vitro fertilization. At present, the principal targets of clinical trials are those disorders which are caused by a single gene—disorders of the Mendelian type. "The first somatic cell gene therapy procedure inserted a normal gene into the DNA of cells in order to compensate for the nonfunctioning defective gene. The normal gene is delivered using a domesticated retrovirus that infects the cell, introducing the properly functioning gene. . . . The two main methods of performing germ-line therapy would be: 1) to treat a preembryo that carries a serious genetic defect before implantation in the mother; or 2) to treat germ cells (sperm or eggs) of afflicted adults so that their genetic defects would not be passed on to their offspring."[29] Nevertheless, with the Human Genome Project, April 14, 2003 announcement that the sequencing of the human genome was completed, the genomic era has dawned: anomaly expressions from the innocuous to the severe will never be approached again without recourse to the mathematical science of the prototypical human genome.[30]

[28] One of the most extensive of website resources on genetic disorders and living with a genetically disabling condition is hosted by the State of New York On line Access to Health at http://www.noah-health.org/english/illness/genetic_diseases/geneticdis.html (accessed May 4, 2004) now available by following the links at http://mentalhealth.about.com/gi/dynamic/offsite.htm?site=http://www.noah%2Dhealth.org/ (accessed June 27, 2005).

[29] Scope Note 24, National Reference Center for Bioethical Literature, The Joseph and Rose Kennedy Institute of Ethics, Georgetown University; http://www.georgetown.edu/research/nrcbl/scopenotes/sn24.html (accessed May 3, 2004) now available at http://www.georgetown.edu/research/nrcbl/publications/scopenotes/sn24.htm (accessed June 27, 2005).

[30] "The extent and pace of progress in genomics are suggested by the fact that this achievement occurred 11 days shy of the 50th anniversary of the publication of Watson and Crick's seminal description of the DNA double helix. If science, technology, and medicine have consistently demonstrated anything, it is that they proceed at an ever-quickening pace. That we have gone in the past 50 years from the

In terms of direct therapeutic genetic interventions results are thus far dismal. At least six (as many as eleven) people have died as a result of genetic therapy intervention: Jesse Gelsinger, an 18-year old with ornithine transcarbamylase (OTC) deficiency, died in September 1999 as a direct result of experimental gene therapy; six other deaths have been reported as a result of somatic line interventions; researchers in France have reported four deaths.[31] And although gene therapy protocols have been submitted to the U.S. Food and Drug Administration, no gene therapy for full-scale product development and sale has yet been approved. Following prudent recommendations, "since it remains difficult to alter genes in humans (for both technical and ethical reasons), for the next couple of decades we will generally use personalized modifications of the environment, and not of genes, to translate genomics-based knowledge into improvements in health for most of our patients."[32]

In spite of this science, what many people fail to recognize is that being born with a disability or becoming disabled as a result of an accident, as a side effect of illness, or as a result of aging is not equivalent to having a disease. And yet, much of genomic science approaches the patient-subject of a disabling condition as if he or she has a disease which needs to be cured. This approach fails to recognize that persons with disabilities are ordinarily quite healthy despite their disability. The distinction between disease and disability further points to a paradigm of diversity within the human community instead of an ableist homogeneity.[33] Certainly, I wish to be cured of cancer, to have had my brother cured of AIDS, and to have others cured of their Chronic Fatigue Syndrome or heart/lung disease—all medical conditions that have the potential to cause a disability of varying severity to occur; but do I similarly wish to be cured of my genetic phenotype as female, or as olive skinned, or as hirsute for that matter—all

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first description of the structure of our DNA to its complete sequencing gives some indication of how much the impact of genomic medicine on the health care of today's neonates will increase by the time they turn 50 years of age" (Alan E. Guttmacher, M.D., and Francis S. Collins, M.D., "Welcome to the Genomic Era," *New England Journal of Medicine* 349 (2003) 996.

[31] See Gene Therapy at the Human Genome Project Information website, http://www.ornl.gov/sci/technresources/Human_Genome/medicine/genetherapy.shtml (accessed May 3, 2004 and June 27, 2005).

[32] Guttmacher and Collins, "Welcome to the Genomic Era" 998.

[33] "A great many people with disabilities like their lives a lot. I happen to be one of them. We tend to repudiate the medical model, which views us as sick and in need of a cure, and the mechanical model, in which we are broken and require repair" (Nancy Mairs, "Learning from Suffering," *Christian Century* 6 [1998] 481, as quoted in Block, *Copious Housing* 17). I encourage the reader to visit Nancy Mairs's website entitled "Celebrating Mortality" at http://maskink.com/mairs/ (accessed May 2, 2004 and June 27, 2005).

conditions which historically could be identified as disabling? Even with these concerns, gene therapies hold promise for cures, by means of retrovirus vector offensives, of diseases that are disabling, such as severe combined immune deficiency (SCID), polio, and AIDS.[34]

Stem-cell research opens another line of interventions for persons with disabilities. Of course, embryo stem-cell research receives the lion's share of celebrity support and, at least, with so many celebrities, like Michael J. Fox (Parkinson's), Mary Tyler Moore (juvenile diabetes), and Nancy Reagan (Alzheimer's), "coming out" of their respective disabled closets, more people with disabilities are likely to benefit from celebrity-endorsed funding appeals. Fortunately for those of us who oppose experimental research on human embryos, a good deal of the success of stem-cell research has been forthcoming from adult sources of these prized cell builders; in fact, the embryonic/pluripotent stem cells have repeatedly underperformed to research expectations.[35] Adult stem-cell gene therapy holds promise as an alternative to retrovirus vectors; stem cells that have been engineered with a replacement gene would then generate "corrected" cells in the developing embryo and in the life of an individual adult.[36] In addition to prenatal applications, stem-cell research holds promise for the repair of spinal cord injuries, disorders of the central nervous system such as Huntington and Parkinson's disorders, and epidermatologic disorders such as fragile skin (epidermolysis bullosa).[37]

In and of themselves, these therapeutic interventions signal advances in reaching the goals of medicine: to relieve human suffering by the cure of disease and to chart a system of health and preventive care that supports

[34] "The biology of human gene therapy is very complex, and there are many techniques that still need to be developed and diseases that need to be understood more fully before gene therapy can be used appropriately" (Scope Note 24, National Reference Center for Bioethical Literature).

[35] "Scientific evidence has shown that stem cells coming from adults or extracted from the placenta and umbilical cord after the birth of a child are much more abundant, more easy to cultivate in laboratories, more versatile and flexible, more capable of generating different types of tissue, and more effective for distinct types of therapy than was previously believed. On the other hand, stem cells extracted by the destruction of human embryos are behaving very "unsociably"; although, it is easier to cultivate them in vitro, their handling in vivo (which is what matters for their therapeutic application) becomes very problematic. The tendency of embryonic stem cells to convert into tumoral formations is no laughing matter" (Gonzalo Miranda, "The Dilemma of Stem Cells—Seeing, Listening, Understanding," in *The Stem Cell Dilemma—For the Good of All Human Beings?*, ed. Gonzalo Miranda [Boncourt, Switzerland: Fondation Guilé, 2002] 8).

[36] Salvatore Mancuso, "Adult Stem Cells in Obstetrical and Prenatal Settings," in ibid. 34–35.

[37] Angelo Vescovi, "Neural Stem Cells", and Michele De Luca, "Adult Stem Cells in Dermatology" in ibid. 37–42, and 43–48 respectively.

the individual and social good. All the same, people with disabilities experience tension between these research goals and the care that is always present in the medical community. I remain suspicious, moreover, of the continued large budgeting expense of scarce human and funding resources to the high-tech genetic and stem-cell research protocols that looms over a gross neglect of providing services to people who are disabled here and now. As the disability community rightly declares: "the stereotyped notions of the 'tragedy' and 'suffering' of 'the disabled' result largely from the isolation and exclusion of disabled people from mainstream society. While the limitations of a disability can be difficult, it is the oppression [encountered on the institutional level in housing, employment, education, religion, health services, legal services, transportation, recreation, and within the media and on the cultural level in language, standards of behavior, logic systems, within the arts and societal expressions, and in the expression of values and norms][38] that is most disabling about disability."[39] As much as one may want to separate the genetic issues of intervention, manipulation, and cure from the lives of people with disabilities, their experience of real oppressions does not allow the luxury of this kind of abstraction. As long as the thinking of a Peter Singer (philosopher), a Bob Edwards (embryologist), or a Justice Oliver Wendell Holmes[40] predominates over what to do about "defects" in the scientific and public sphere, the nondisabled must

[38] See Block, *Copious Housing* 53; see also Parens and Asch, "The Disability Rights Critique" in *Prenatal Testing* 5–8; Thomson, *Extraordinary Bodies* 55–80; and *The New Disability History*, ed. Paul K. Longmore and Lauri Umansky (New York: New York University, 2001).

[39] I continue the quote. "In an era that offers access to improved health, longevity, social mobility, and a political voice for disabled citizens, it is ironic that the growth of the new reproductive and genetic technologies . . . provides the possibility of eliminating categories of people with certain kinds of disabilities, such as Down syndrome, spina bifida, muscular dystrophy, sickle cell anemia, and hundreds of other conditions. Laura Hershey suggests: The idea that disability might someday be permanently eradicated—whether through prenatal screening and abortion . . . has strong appeal for a society wary of spending resources on human needs. Maybe there lurks, in the back of society's mind, the belief (the hope?) that one day there will be no people with disabilities. That attitude works against the goals of civil rights and independent living. We struggle for integration, access, and support services, yet our existence remains an unresolved question" (Saxton in *Prenatal Testing* 155–56).

[40] "The worst thing that could happen is that someone enrolled in a [genetic research] study would get pregnant and not get an abortion. . . . Moreover, while eugenic practices have occurred throughout history, even in our own day, in a country where basic human rights are described as "inalienable," there have been efforts to dissuade or prevent women who suffer from anomalies such as mental retardation or epilepsy from having children. Recall, for example, Justice Oliver Wendell Holmes's infamous statement in *Buck v. Bell*, supporting compulsory sterilization of the retarded on grounds that "three generations of imbeciles are

stand with and for people with disabilities against continued marginalization through objectivization, infantilization, euthanasia, and abortion, and worse.

THEOLOGICAL INSIGHTS FROM A DISABILITY PERSPECTIVE

A theology of access and accountability requires an appreciation of disability as one of the many and diverse ways that God is revealed in humankind and in which God reveals human frailty — reminding us that our posture before God must be characterized by humility. And although diversity may be the buzz word of the early 21st century, it may be the key to appreciating, valuing, and affirming all people, regardless of their race, gender, sexual orientation, religion, health, and disability. A theology of access and accountability begins with each of us as made in the image and likeness of our triune God[41] and from there reflects upon the real concrete needs of persons with disabilities demanding nothing less than full communion and love of, for, and with all our disabled and nondisabled sisters and brothers alike.

Why a theology of access? Because people with disabilities have been denied communion with the faithful; because people with disabilities have been redeemed; because people with disabilities have been oppressed; and as such, "because the gospel of Jesus Christ is a gospel of access, [and] creating access for those on the margins is a Christian mandate."[42] A theology of access can begin to repair the systemic violence perpetrated against those whose bodies, culture, gender, race, or sexual orientation differ from the main.

Why a theology of accountability? Because power and privilege rest historically upon people without disabilities and those with power have responsibility for those without; because people with disabilities have needs of food, clothing, shelter, healthcare, education, friendship, love, and a place with the fellowship of disciples; because God makes the sun to shine and the rain to rain on the rich and poor, the abled and disabled alike; and

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enough." More recently, Laura Purdy has argued for the obligation of persons who are carriers for serious genetic disorders such as Huntington chorea not to have children" (Mary Briody Mahowald, *Genes, Women, Equality* [New York: Oxford University, 2000] 40 and 55).

[41] As Jack Mahoney has recently reminded us: "God's creation of humankind, male and female, 'in our image, according to our likeness,' as Genesis 1:26 describes it, is the theological basis for asserting the fundamental dignity of all human beings without exception, as well as recognizing their inalienable value and destiny as individuals" (Jack Mahoney, "Christian Doctrines, Ethical Issues, and Human Genetics," *Theological Studies* 64 [2003] 722).

[42] Block, *Copious Housing* 120.

because all people will be judged according to the manner in which the least among them have been loved, cherished, and cared for.[43] A theology of accountability can begin to remove the ideologies that separate and divide us from one another and from the Mystical Body of Christ, the Church, as well as it can move us to embrace the Broken Bodied Risen Christ of faith.

Further, the United States Conference of Catholic Bishops reminds us that "it is essential that all forms of the liturgy be completely accessible to persons with disabilities since these forms are the essence of the spiritual tie that binds the Christian community together. To exclude members of the parish from these celebrations of the life of the Church, even by passive omission, is to deny the reality of that community.[44] Accessibility involves far more than physical alterations to parish buildings" ("Pastoral Statement of US Catholic Bishops on Persons with Disabilities," 1978).

Theological reflection from a perspective of disability, even while temporarily nondisabled, refocuses the images, ideas, and impulses of Christian discipleship. With this perspective comes a call to hospitality toward the diversity of strangers, toward those who do not look like us, toward those with "extraordinary bodies." With this perspective comes a call for repentance and forgiveness for the sins of Spartan exposure, leper colonies, silencing, sterilization, denial of sacraments, institutionalization, objectification, avoidance and disparaging glances, stairs—all manner of violence and oppression inflicted upon people guilty of nothing more than being "different." With this perspective comes a call to find ways to include people with disabilities in schools, work, leisure, and worship and to liberate them from their oppressors (access) as their oppressors are liberated of their wayward thinking (accountability). The new images, ideas, and impulses of Christian discipleship from a perspective of disability celebrate the diversity of people redeemed by a God who gave the world a Word, enfleshed by the Spirit, abiding with us now as disabled and raised in glory.[45]

Once the People of God accepts in faith that each of us is intimately,

[43] "The moral responsibilities incumbent on humankind resulting from the doctrine of creation, and our failures to respect them, must sooner or later bring to our attention the mysterious capacity that we humans have to misuse the gifts of God, a capacity that we call sin" (Mahoney, "Christian Doctrines" 725; see n. 41 above).

[44] "When a church building is physically inaccessible, the disabled parishioner or visitor feels just as emphatically unwelcome as if the cultic laws of Leviticus [Lev 21:16–20] were still in force" (Donald Senior, "Suffering as Inaccessibility: Lessons from the New Testament Healing Stories," *New Theology Review* 1 [1988] 10).

[45] "Certainly the crucifixion was disabling. Gaping holes in hands and feet from nail wounds cause disfigurement and limit mobility, and yet the marks of Jesus' own mortality—the scars of the crucifixion—were not erased from the body of the Risen Christ" (Block, *Copious Housing* 86–87). See also Jürgen Moltmann, "Liberating

wonderfully made in the image of the triune God, then we, as that People, cannot fail to welcome, embrace, aid, comfort, and feed all. And we must do more. Given the God-created diversity of human communities, given the history of oppression that people with disabilities have endured, given the disability of the Word made flesh, the Crucified God, now is the time to go out to the highways and the outskirts of our towns to bring in those whose diversity has just dawned on normates, those whom the nondisabled have sinned against, those whose presence is needed if we are to be the community of disciples God wants us to be.[46]

How Can We Be the People of a Crucified God?

As Donald Senior has noted: "Reading the Gospels from the vantage point of the disabled can be instructive. The plight of the sick and disabled is due not simply to physical pain but to the isolation and exclusion that often comes in the wake of serious illness or disability. To exclude is to dehumanize."[47] First, we must recognize how we, the nondisabled, have dehumanized, oppressed, and otherwise marginalized people with disabilities. This step requires a healthy examination of where we are (to follow the virtue ethics method of moral inquiry and change) in terms of the integration of people with disabilities into our lives. Second, we must ask the questions of the kingdom, the banquet, and the discipleship of equals to which Jesus call us. This step requires a visionary embrace of the world as God embraces the world, an embrace that acknowledges, celebrates, and welcomes the diversity of people (what virtue ethics would identify as our goal). Third, we must explore the ways in which we can become this people of a Disabled and Glorified Christ. This step requires the practical decisions that move the nondisabled oppressor to repentance and appreciation (what virtue identifies as the means: social justice, healthcare, education, friendship, work, hospitality, among many other virtues). Finally, the disciples of the Crucified God can challenge the presumptions of the medical model of disability, the exaggerated promises of genetic interventions, and the misconceptions held by the nondisabled about life with a disability . . . and risk a full life as other too.

Yourself by Accepting One Another," in *Human Disability and the Service of God: Reassessing Religious Practice*, ed. Nancy Eiesland and Don Saliers (Nashville: Abingdon, 1998).

[46] "For I tell you, none of those who were invited will taste my dinner" (Luke 14:24).

[47] Senior, "Suffering as Inaccessibility" 13.