Deafness and Prenatal Testing: A Case Study Analysis

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Abstract

INTRODUCTION:

The Deaf culture in the United States is a unique culture that is not widely understood. To members of the Deaf community in the United States, deafness is not viewed as a disease or pathology to be treated or cured; instead it is seen as a difference in human experience.[1] Members of this community do not hide their deafness; instead they take great pride in their Deaf identity. The Deaf culture in the United States is very communitarian not individualistic. These members value their cultural group and have a very positive attitude towards being deaf.[2] Culturally, Deaf people value the use of natural sign languages that exhibit their own grammatical conventions, such as American Sign Language and British Sign Language, over signed versions of English or other oral languages.[3] The Deaf community is like every other social group. A person is a member of the Deaf community if that person "identifies him/herself as a member of the Deaf community, and other members accept that person as part of the community."[4] The one criterion is that one must be deaf.

Hearing loss is the most common birth defect and the most prevalent sensorineural condition in developed countries.[5] One of every 500 newborns has bilateral permanent sensorineural hearing loss ≥ 40 dB; by adolescence, prevalence increases to 3.5 per 1000.[6] The most common and useful distinction in hearing impairment is syndromic versus non-syndromic. Syndromic means that the hearing impairment is associated with other clinical abnormalities. Among hereditary hearing impairments, 15 to 30% are syndromic. Non-syndromic hearing impairment accounts for the vast majority of inherited hearing loss, approximately 70%. Autosomal-recessive inheritance is responsible for about 80% of cases of non-syndromic hearing impairment, while autosomal-dominant genes cause 20%. Less than 2% of cases are caused by X-linked and mitochondrial genetic malfunctions.

One gene, known as Connexin 26 (CX26) is estimated to be responsible for half of all the recessive cases of hearing loss. CX26 is a protein that is involved in the formation of gap junctions responsible for forming electrical synapses for transferring ionic currents and molecules between cells. CX26 is encoded by the GJB2 gene on chromosome 13. A mutation in the GJB2 gene that affects the coding of CX26 is common in children with sensorineural hearing loss (SNHL). CX26 mutations cause approximately half of all bilateral moderate to profound congenital hearing loss. There are over 400 known gene causes involving hearing loss. CX26 alone is responsible for about 1/3 of all the cases of genetic hearing loss. This leaves about 1/3 of all cases as non-syndromic with the remaining 1/3 as syndromic. Among the remaining 1/3 of non-syndromic cases of genetic hearing loss, 13 dominant and 6 other recessive genes have been described.[7]

Medically, when one or both genetic parents have a known genetic abnormality testing can be performed on the embryo to determine if it also carries the genetic abnormality. Preimplantation genetic diagnosis (PGD) is a genetic procedure used prior to implantation to help identify genetic defects within an embryo created through in vitro fertilization (IVF) and to prevent certain diseases or disorders from being passed on to a child. PGD was developed in the United Kingdom in the mid-1980s as an alternative to current prenatal diagnosis. PGD should be offered for 3 major groups of diseases: (1) sex-linked disorders, (2) single gene defects, and (3) chromosomal disorders.[8] PGD begins with the normal process of IVF which includes: ovary stimulation through medication and then egg retrieval. Each egg is usually fertilized by the

injection of a single sperm which is known as Intracytoplasmic Sperm Injection (ICSI). The fertilized eggs are placed in a petri dish and allowed to develop. The resulting embryos divide for three days. At the 8-cell stage, a single cell called a blastomere is removed from each embryo, leaving them with seven cells. Each blastomere is tested to see if its embryo contains the defective gene carried by one or both of the parents. The DNA is retrieved from the cell and copied through a process known as polymerase chain reaction (PCR). By molecular analysis, the DNA sequence code is evaluated to determine if the inheritance of the gene is present. Embryos determined to have the defective gene are discarded or donated for research. Embryos free from the defective gene are implanted by embryo transfer into the mother's uterus or frozen for future use. PGD is expensive. One round of IVF typically costs around $10,000 to $12,000. PGD adds another $4000 to $7500 to the cost if each IVF attempt. A standard round of IVF results in a successful pregnancy only 10% to 35% of the time depending on the age and health of the woman, and a woman may need to undergo subsequent attempts to achieve a viable pregnancy.

Recently, a married couple who is genetically deaf approached an IVF Center with a request to create 2 to 3 embryos that would be genetically deaf. They believed that a hearing child would be detrimental to their family and their Deaf community. The IVF Center contacted a number of bioethicists to ask for a consultation about whether this procedure should be allowed under the present circumstances. To address this issue, a case study has been developed that explains the facts regarding PGD to create a genetically deaf child. The names of the couple have been changed to protect confidentiality. The case study will then be analyzed medically and ethically.

CASE STUDY

Mary Beth and Dominic are a married couple in their late 20s who are genetically deaf. They are active members in the Deaf community and work as advocates for individuals who are deaf, family members of Deaf people and sign language interpreters who identify with the Deaf culture. Members of the Deaf community view deafness as a difference in human experience rather than a disability or a disease. Mary Beth and Dominic are both carriers of the gene Connexin 26 (CX26) that is estimated to be responsible for half of all recessive cases of hearing loss. Mary Beth and Dominic have decided after many conversations with counselors and physicians that they want a child who is genetically deaf. They believe that a hearing child would be problematic to their family and the Deaf community in which they are very active members. They were informed that a procedure called Preimplantation Genetic Diagnosis (PGD) is used to help identify genetic defects within the embryo created through in vitro fertilization so that the embryo free of the defective gene is placed in the uterus or frozen for future use and the embryos with the inherited genes are destroyed. However, Mary Beth and Dominic approach the fertility clinic with a request that they only want a child with the genetic gene for deafness. They would want the embryos that do not have the gene to be destroyed. This would entail creating a child who would have the gene for deafness. Medically and ethically, should the fertility clinic agree to the couple's request?

MEDICAL ANALYSIS

PGD can be performed prior to implantation to help identify genetic defects within an embryo created through in IVF and to prevent certain diseases or disorders from being passed on to a child. However, many questions remain unanswered regarding the safety and efficacy of PGD. Inherent in any IVF procedure are risks associated with the use of hormones to stimulate the ovulation and the potential for ectopic pregnancy. To improve the success rate of IVF, numerous embryos are implanted in the woman's uterus, increasing the likelihood of multiple gestations. Women who carry multiple fetuses are more likely to experience complication in their pregnancy, which could adversely affect their health, and the health of their fetuses. Additionally, little is known of the effects the removal of a cell may have on the embryo or the development of the child. Currently, approximately 20% of PGD procedures result in a pregnancy. [9]

Also, although the embryo biopsy procedure is considered safe, it may still affect the viability of the embryo and cause a decrease in the probability of pregnancy. Errors can also occur during the DNA amplification process. Allele drop-out occurs when one allele for the gene of interest fails to amplify. Partial amplification occurs when the allele for the gene of interest amplifies poorly. Both types of technical errors can result in misdiagnoses, which may lead to transferring an undesired embryo. In addition, there is the possibility of amplifying contaminating extraneous DNA. Amplification of such DNA may also lead to transferring an undesired embryo. Because of these uncertainties and undesired harms which may be caused to the mother and the future-born, PGD is typically recommended for patients over the age of 35 with known genetic anomalies. And the parents

who want to pursue PGD should have counseling done at the fertility clinic, with the procedure, its limitations, and possible adverse effects thoroughly explained. In this case, one has to wonder if the Deaf couple, Mary Beth and Dominic, has arrived at their decision based on the comprehensive understandings of PGD and IVF. If not, then it would be difficult to say they gave informed consent for the procedures.

FOUNDATIONAL ETHICAL ANALYSIS:

At the foundational level, the ethical case-analysis of clinical bioethics is grounded in our epistemic confidence that there exists "common morality," morality shared by all rational people. Nonetheless, the sense in which the status of common morality is understood among foundational ethicists is not univocal. In recent years two senses of common morality have become most prominent in academic debate: the prescriptive and descriptive senses of common morality. The former is the morality of which common feature is believed to hold normative binding force on all rational people; while the latter takes the morality as commonly practiced by all rational people here and now. However, because clinical bioethical cases are typically presented within a particular socio-cultural context, the final moral verdicts derived from the two different senses of common morality are often found homogenous. And the homogeneity is backed by the theoreticians' disclaimer that common morality of both senses does not require every single rational individual's unanimous consensus, for it is common morality, not absolute morality. On the other hand, different moral decisions are observed between the prescriptive and descriptive kinds of common morality when the moral prescription of a certain tradition in society holds a heterogeneous view from the social majority.

In our culture, the Roman Catholic tradition is the most representative kind of prescriptive morality. On the other hand, so-called the "secular liberal individualist tradition" of which the founding principle is derived from the 19th century philosopher, John Stuart Mill, provides the exclusive framework of descriptive common morality. And it should be noted that the two traditions here roughly coincide with the "conservative" and "liberal" positions about the issue in our time and place, the contemporary U.S.

To speak briefly about the reasoning of the Roman Catholic tradition, the moral prescription given to the procedure of childbirth is expressed by the inseparableness of "the unitive" and "the procreative" – that is, the child must be created through the physical union of a husband and a wife. And this norm is based on the Church's theological conviction that children are God's gifts. God decides who receives the gift, when the gift will be delivered, and what particular type of gift the couple will get. Thus, the request of the Deaf couple which involves IVF and PGD is considered morally impermissible. However, a further discussion on the Church's theological view will be proceeded in the next section, clinical ethical analysis.

On the other hand, the descriptive morality of the secular liberal individualism concerns how to translate a general social consensus into moral reasoning on a given case. In our case, the key issue is how our society (the majority members, if not the vast majority) understands the language of "defect" and "difference" related to those with physical handicap, particularly the hearing impaired. First of all, for the sake of argument, we re-construct the couple's argument as viable as it can. The Deaf couple's position is premised by the following argument. IVF and PGD have become legally and ethically accepted practices in our society. The both procedures have aided people suffering from infertility to have babies free of genetic defects or diseases. And they can also be used to enhance citizens' quality of life and ensure our happiness. Deafness is not a defect, as exhibited in the popular slogan of our society: "The handicapped are not defective but different people." Thus, what is requested is the medical assistance to help produce a normal deaf baby just different from a normal non-deaf baby. As a result, the clinic must agree to the demand.

The Deaf couple's argument is valid only when it is evidenced by the fact that the general public understands and uses the terms, "defectiveness" and "difference," in the manner they present. It is true that medical science or everyday-living itself does not offer the answer to whether a particular bodily feature is "normal" or "defective." The science can show that the gene CX26 is responsible for a given individual's loss of hearing, not that the individual is functionally defective or not. When the society sees deafness not as a functional defect but as a mere discomfort, it is not a defect. Also it is no doubt that people with hearing impaired experience many inconveniences in every living; however, not to say that the person is having a defective or abnormal lifestyle. It is the society's view that determines on the normality of it For example, left-handedness in the U.S. culture was once considered a defect but now considered a normal, yet different trait. Also more and more people in American society are seeing albinism not as a defect any

more, perhaps due to the societal effort to be "politically correct" about race and color issues. In the case of deafness too, it seems that the vast majority of American society does not consider deafness to be not defective but merely different. If so, can the couple's argument be valid? Do we really not see deafness as a functional defect?

We do see deafness as a functionally defective trait. What our society tries to change or improve is the malevolent idea that medical handicap, including deafness, is equivalent to the diminished sense of human dignity. In other words, the functional defectiveness does not lead to defectiveness in human dignity. This moral commitment has been reflected through active public campaigns in our society like "The handicapped are not defective but different people." That is, the handicapped people are not defective human beings but hold different challenges and concerns in everyday living because of their functional defects. Thus, according to the standpoint of descriptive common morality, the Deaf couple is requesting for a defective child. As a result, the couple argument is found be invalid. Let us now move to clinical ethical analysis.

CLINICAL ETHICAL ANALYSIS

IVF and PGD are two procedures that are highly controversial based on the notion of when personhood begins. The Catholic Church is morally against both procedures because the Church argues that a child must be the fruit of the conjugal union and nothing can separate the unitive and procreative dimensions of marriage. IVF would be viewed as a substitute for the marital act and thus a domination of technology. The Church argues that, "Human embryos obtained in vitro are human beings and subjects with rights: their dignity and right to life must be respected from the first moment of their existence. It is immoral to produce human embryos destined to be exploited as disposable 'biological material'. In the usual practice of in vitro fertilization, not all of the embryos are transferred to the woman's body; some are destroyed. Just as the Church condemns induced abortion, so she also forbids acts against the life of these human beings."[10] Second, PGD would not be acceptable because the embryos are obtained through IVF. This is not to say that the Church is against all prenatal testing. The Church argues that, "prenatal diagnosis makes it possible to know the condition of the embryo and of the foetus when still in the mother's womb. It permits, or makes it possible to anticipate earlier and more effectively, certain therapeutic, medical or surgical procedures. Such diagnosis is permissible, with the consent of the parents after they have been adequately informed, if the methods employed safeguard the life and integrity of the embryo and the mother, without subjecting them to disproportionate risks. But this diagnosis is gravely opposed to the moral law when it is done with the thought of possibly inducing an abortion depending upon the results: a diagnosis which shows the existence of a malformation or a hereditary illness must not be the equivalent of a death-sentence."[11] Therefore, neither IVF nor PGD would be morally acceptable or permitted in any Catholic facility.

Many secular hospitals and clinics have reproductive technology centers that perform both procedures. Examining this case from a secular perspective using the ethical principles of respect for life, beneficence/nonmaleficence and justice, one can argue that allowing this couple to implant only embryos that are genetically deaf violates the basic principles of ethics. "Respect for persons" refers to the right of a person to exercise self-determination and to be treated with dignity and respect. The principle of respect for persons divides into two separate moral requirements: the requirement to acknowledge autonomy and the requirement to protect those with diminished autonomy.[12] No one can argue that parents have the right to test their embryos under the principle of autonomy. As mentioned above, PGD is frequently recommended for patients over the age of 35 and for couples who have known genetic anomalies. The problem is that in the United States there are limited professional guidelines for the practice of PGD. In addition these are only guidelines and have no means of enforcement. Medically, many people would argue that it is morally responsible not to bring a child into the world who will suffer from some genetic anomaly when this anomaly could be eliminated. The problem for some ethicists is that the embryos that test positive for the genetic anomaly are destroyed. Depending on one's definition of personhood this could be morally problematic. Many Catholic ethicists believe that personhood begins at conception so each embryo is a person or a potential person with moral rights. To destroy embryos that have a specific genetic anomaly would be using some persons as a means to an end. This would violate the principle of respect for persons. These ethicists would argue that the embryos with a genetic defect are truly vulnerable persons with diminished autonomy and deserve added protection. Opponents, who believe personhood does not begin until viability or at birth, would dismiss this argument because these embryos lack any moral or legal rights. The only individual with legal rights is the woman undergoing the procedure. Thus the argument

focuses on when one believes personhood begins. The problem with this case is that the embryo with the genetic anomaly is not destroyed. The embryos that are without the genetic anomaly are destroyed. In this scenario both Catholic and secular ethicists would object to this procedure, because it does violate respect for persons whether one believes personhood begins at conception or after viability. Intentionally creating a child with a hearing impairment is a clear violation of a vulnerable person. To allow this procedure violates a vulnerable person's human rights and the basic dignity and respect that every person deserves.

The principle of beneficence involves the obligation to prevent, remove, or minimize harm and risk to others and to promote and enhance their good. Beneficence includes nonmaleficence, which prohibits the infliction of harm, injury, or death upon others. In medical ethics this principle has been closely associated with the maxim primum non nocere ("Above all, do no harm"). To create a child intentionally with a genetic hearing impairment because the parents believe a hearing child would be detrimental to their family and the Deaf community is not minimizing risks and harms nor promoting or enhancing the good. The parents would argue that "the good" is this situation is that the Deaf child will be fully incorporated into the family and the Deaf community. If the child does not have the defective gene CX26, the child will always be an outsider and this could cause harm to the child, the family and the Deaf community. Parents have the legal and moral rights to do what is in the best interests of their child. The direct intention of creating a child with a genetic defect that will impact the child's entire life with no consent from the child is a direct infliction of harm and injury on this child. Failure to recognize this fact is a failure not only of the test of beneficence; it may also be a failure of the test of nonmaleficence.

The principle of justice recognizes that each person should be treated fairly and equitably, and be given his or her due. This case also focuses on distributive justice: the fair, equitable, and appropriate distribution of medical resources in society. At a time when reforming healthcare in this country has become a high priority, failure to initiate preventative measures that would save medical resources and possibly social resources in the long-run violates the principle of distributive justice. The parents will argue that they have the right to create a child that would be a good addition to their family and community. They would argue that this is just and they must be given their due. Opponents would argue that intentionally creating a hearing impaired child not only violates the child's right to be treated fairly and equitably but also violates the rights of all Americans. If this child is born hearing impaired the child is going to need substantial medical, social and educational resources for the rest of his/her life. These resources will be provided by the state and federal government and the costs will be shared by members of society as a whole. Physicians, clinical researchers and the medical profession have an ethical obligation to use available medical resources fairly and to distribute them equitably. Failure to do so is ethically irresponsible and morally objectionable. To compromise the basic ethical foundations upon which medicine stands is destructive not just to this child and the parents but to society as a whole.

In addition, there is also a concern for the slippery slope. In a slippery slope argument, a course of action is rejected because, with little or no evidence, one insists that it will lead to a chain reaction resulting in an undesirable end or ends. If this couple is permitted to create a child that is intentionally deaf then what is to stop parents from creating a child that is intentionally blind or has Downs Syndrome, or some other genetic anomaly. Once a precedent is established, it logically follows that it can become applicable to other related situations.

The difficult ethical question in this situation is that the medical professionals at this reproductive clinic must decide if doing PGD in this situation is in the best interest of the child, the parents and society as a whole. Ethically, after examining the medical procedures and the potential results, it does not appear that doing PGD with the intention to create a child that is hearing impaired in in the best interest of the child or society as a whole. The viable option would be for the parents to adopt a child who is hearing impaired to meet their needs and the entire society.

CONCLUSION:

The issue of allowing parents to request the intentional creation and implantation of embryos with a particular genetic anomaly is a complex issue that involves medical and ethical aspects. In the best interest of the child, the parents and society as a whole some resolution has to be reached that will resolve this issue sooner rather than later. We have examined the issue from some varied angles. From the medical perspective, a reasonable moral suspicion is rendered whether the couple's request is grounded in their thorough apprehension of PGD and IVF and possible adverse effects. At the foundational theoretical investigation,

their request is found ethically unjustifiable at the both prescriptive and descriptive senses of common morality. From the clinical ethical standpoint, it is clear that the direct intention to create a child with a specific genetic anomaly that could be avoided is not in the best interest of the child or society as a whole. To allow for this procedure sets a very dangerous precedent that could have long term, detrimental effects for individuals and for our society. There is a viable option for these parents. They could adopt a child that is hearing impaired. This alternative would be in the best interest of the child, the parents and society as a whole.

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