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The Ethics of Prenatal Diagnosis
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[1] According to a recent assessment, it is now possible through medical procedures such as amniocentesis, fetoscopy and ultrasound, to diagnose in utero and with considerable accuracy over 280 abnormalities of pregnancy.[1] These abnormalities include a range of fetal genetic disorders (e.g. Down's syndrome, Tay-Sachs disease, Lesch-Nyhan syndrome, spina bifida) which, if left unattended, will result in births of children suffering from serious physical and/or mental disabilities of varying degree. Given the current state of medical technology, intrauterine therapy for such disorders is possible only in rare instances. Under certain circumstances - for example, some cases of spina bifida - postnatal therapy can restore the hope for a reasonably normal life. But much depends in such instances on the affliction's degree of severity, a matter which typically cannot be evaluated at the time of prenatal diagnosis. For a number of afflictions, moreover, postnatal therapy cannot remove the primary symptoms of the condition. Thus, medically indicated treatment for an infant with Down's syndrome often includes the administration of antibiotics for complications associated with the condition. Yet Down's syndrome children inevitably suffer from some degree of mental retardation, though with special education they often can achieve a substantial measure of independence and in the largest number of instances do lead relatively long and happy lives. Finally, for certain afflictions such as Tay-Sachs disease or Lesch-Nyhan syndrome the symptoms are harsh, and there exist at present only minimally palliative postnatal therapies. In the most extreme cases the destinies of children suffering from these afflictions are comparatively short lives marked by considerable physical pain and profound mental retardation.

[2] This general state of affairs has given rise to a number of perplexing ethical questions. For one thing, a pregnant woman whose fetus has been diagnosed positively for any of the genetic disorders untreatable in utero faces with her family a decision of enormous moral consequence, namely, the decision whether to abort the fetus or bring it to term, in the latter case for the sake of rearing the child or giving it up for adoption. To what extent, if any, can abortion for genetic defect be countenanced from the moral point of view? What quality-of-life judgments inform any answer to this question, and how do such judgments bear on our attitudes toward persons actually born with genetic disease? Arguably the question of genetic abortion is the most significant of the ethical problems spawned by the new genetic technology. Nonetheless, the advent of modem prenatal diagnostic methods does raise other moral questions which require serious consideration. For example, under current conditions amniocentesis, the most fruitful of the diagnostic techniques,

involves certain serious risks for the fetus. Is the taking of such risks morally warranted by the concern to prevent possible genetic defect or to prepare a family for the possible birth of a child with genetic disease? Under what conditions? Finally, there are a number of ethical questions generated by proposals to make the prenatal detection of genetic disease a matter of public policy. What sorts of moral constraints ought to be placed on a screening program? Who, for instance, should have access to such a program and what will be the moral responsibilities of individual physician and client participants? In what follows I examine these and related issues with particular reference to the question of how Christian theological beliefs might direct inquiry in such matters.

Risks of Prenatal Diagnosis

[3] Discussion of the health-risk factors in prenatal diagnosis has focused for the most part on the hazards of amniocentesis, a procedure designed to detect fetal chromosomal abnormalities and other fetal disorders.[2] As is well-known, this technique involves the withdrawal of fluid from the amniotic sac by insertion of a needle through the mother's abdomen into her uterus. At the earliest, the tap may be performed during the fourteenth week of pregnancy, by which time there is sufficient amniotic fluid for extraction. From the fluid, fetal cell cultures can be developed, and these cells allow testing for chromosomal aberrations such as Down's syndrome, as well as inborn errors of metabolism such as Tay-Sachs disease and Lesch-Nyhan syndrome. In addition, the fluid itself can be analyzed for excess quantities of the substance alpha-fetoprotein (AFP), a symptom of anencephaly or spina bifida. If all goes well, results are typically available around the twentieth week of pregnancy.[3]

[4] As noted by a recent working paper, the health hazards of amniocentesis include risks to the mother, to the fetus, and to the infant during the newborn period.[4] Generally speaking, maternal complications are minor when they occur. Of over 20,000 amniocenteses surveyed in an international study, one maternal death was reported to have resulted from the procedure. A more probable health risk to the mother is the possibility of infection (amnionitis), which still occurs in less than one out of every thousand cases. All in all, the risks to the fetus are more serious. First, there is the slight possibility (roughly I percent) of injury from the needle, but for the most part the signs of such injury have been nothing more than the presence of skin dimples indicating healed punctures, though there have been isolated reports of serious injury such as the loss of an eye. More important, although there is some disagreement about the significance of the findings, evidence exists suggesting that amniocentesis markedly increases the risk of spontaneous abortion. The working paper cited above sets the risk of abortion from amniocentesis at something under 1 percent and goes on to note that the procedure may be particularly hazardous in this regard for a category of women who typically submit to amniocentesis, namely, pregnant women with elevated alpha- fetoprotein levels, the presence of which may be indicated preliminarily by maternal serum tests. Finally, there is some evidence that the procedure poses special risks for the infant during the newborn period. For example, the finding of one study indicated a correlation between amniocentesis and respiratory distress syndrome. At the same time, there is no evidence associating

the technique with an increase in stillbirths or infant deaths during the first week after birth. Neither is there any evidence suggesting that amniocentesis adversely affects infant development during the first year of life. Researchers have speculated about the possibility of impediment to long-range mental development due to sudden loss of pressure caused by fluid withdrawal. Yet up to now such speculation has been neither confirmed nor falsified. More generally, the question of the procedure's long-term developmental effects, whether harmful or beneficial, remains unresolved.

[5] Given the hazards noted here, any prudent and morally responsible deliberation about whether amniocentesis should be undertaken will involve a careful assessment of the technique's possible benefits in relation to these potential costs. For the purposes of ethical analysis it is essential to note that judgments based upon such cost-benefit assessments are inevitably value-laden, i.e., they presuppose value commitments, which can and ought to be submitted to moral scrutiny.

[6] To take one example, it is commonly proposed that pregnant women who have reached the age of thirty-five should submit to amniocentesis, the reason being that the chances of conceiving a child with Down's syndrome increase markedly at this age. Yet frequently this judgment is issued without any explicit comparative evaluation of the stated risk over and against the hazards of the procedure itself. Setting aside. for a moment the question whether selective abortion of a Down's syndrome fetus is acceptable morally, we must still ask if the risk of giving birth to a Down's syndrome child is sufficiently grave reason for taking on the hazards of amniocentesis, particularly the danger of spontaneously aborting a normal child. And when the issue is posed in this way, matters are cast in a somewhat different light. As mentioned earlier, recent assessments set the chances of spontaneous abortion resulting from amniocentesis at something less than one out of every hundred cases. At the same time, estimates put the risk of a thirty-five-year-old woman's conceiving a child with Down's syndrome at around one in four hundred.[5] Given these figures, the conclusion seems inescapable that a thoughtful judgment supporting amniocentesis and selective abortion for a pregnant woman of this age assumes - all other things being equal - that risking the loss of a probably normal child would be morally preferable to risking the less likely outcome of giving birth to a child with Down's syndrome.

[7] The point is that such an assumption raises a number of important moral questions. Is the life of a Down's syndrome child so impoverished in quality or so burdensome to family and society that fear of the relatively remote possibility of giving birth to such a child overrides any presumptive obligation to care for the probably normal child? Is an affirmative answer to this last question consistent with the realization that Down's syndrome children, though significantly limited in potential, and exceptionally dependent on familial and social resources, can with proper care lead flourishing lives?[6] Is a decision to risk the life of a probably normal child reconcilable with a moral outlook which characterizes human life as a divine gift, and which regards the bringing forth and nurturing of such life as a human analogy to the divine love issuing forth in the creation and preservation of the world? These and related questions

would need to be resolved before a responsible decision could be made.

[8] Of course, different probabilities may lead to different judgments. For instance, the chance that a woman at age forty-eight will conceive a child with Down's syndrome is one in twelve. Under these conditions, the less than 1 percent chance of spontaneous abortion may begin to look like a minor matter.

[9] Of course, whether a woman ought to submit to the procedure in this case will depend upon the ultimate purpose in so doing. If the purpose of amniocentesis is to prepare psychologically or otherwise for the possible birth of an affected child, the issue becomes one of determining whether the good which would be served by such preparation is sufficiently weighty to justify the risk of an accidental abortion. On the other hand, if upon positive diagnosis the goal is selective abortion, we are left with the question of whether such abortion is morally defensible. Indeed, in some cases (e.g. Lesch-Nyhan syndrome) the gravity of the disorder combined with a high probability of affliction (50 percent for a woman who has previously given birth to an affected child) will serve to shift the weight of moral concern decisively from the question of the procedure's hazards to the question of selective abortion on positive diagnosis.

Genetic Abortion

[10] It is likely that there will come a day in the not too distant future when advances in genetic technology will have made intrauterine therapy the standard medical response to prenatally diagnosed genetic disease. Unfortunately, while there has been some progress in this area, that day has not yet arrived. And until it does we shall be forced to come to grips with the problem of abortion for genetic defect. Of course, to characterize genetic abortion as a problem is not to raise questions about its legality; even post-amniocentesis abortion, late as it comes in the pregnancy's term, still falls within the legal limits established by the Supreme Court. At the same time, I shall be assuming here that genetic abortion does constitute a moral problem, particularly from the viewpoint of Christian ethics. Such abortion is morally problematic given the recognition of a presumptive obligation of parents to protect their fetuses from harm - an obligation which has its source not in some putative fact that the fetus is an actual or a potential person, as some contemporary philosophers utilize that term (self-conscious, autonomous being), but rather in the fact that the fetus is an unborn child whose being lays moral claim to parental care. For Christian ethics this moral claim is part and parcel of an objectively real parent-child covenant which mirrors analogically the providential covenant God has forged with the world. Since care for children typically involves protection against their destruction, this parental covenant generates a moral presumption against abortion. To note the presumption, however, is not to suggest its indefeasibility. Indeed, as far as Christian ethics is concerned, it is fruitful to regard the problem posed by genetic abortion as the problem of determining whether there are conditions under which the moral obligation to care allows a parent to set aside, albeit tragically, the presumption against fetal destruction.[7]

[11] Needless to say, actual determinations in real-life situations are fraught with empirical uncertainty, moral ambiguity, and acute human anguish. Still, despite the strong presumption against fetal destruction mentioned above, I would propose that some genetic abortions can be regarded intelligibly as expressions of parental care, though I would emphasize also that one must discriminate carefully among cases, as well as acknowledge a measure of tentativeness about particular judgments. Consider the example of Tay-Sachs disease. A child suffering from this condition appears to be normal shortly after birth but within months begins to show signs of lethargy, weakness, and arrested psycho-motor development. Eventually the stricken child is over- come by even graver symptoms, e.g., blindness, deafness, proclivity to seizures, paralysis, and profound mental retardation. Death typically occurs between the ages of three and five. Granted these circumstances, it is certainly reasonable to suggest that a decision to abort a Tay-Sachs fetus is warranted precisely by the concern to promote its best interests because it is reasonable to argue that death is preferable to continued existence from the point of view of the afflicted life. Other examples (e.g., Lesch-Nyhan syndrome, epider-molysis bulloso lethalis) could be cited. The general point is that for certain diseases detectable in utero the prospective symptoms are so harsh that it is meaningful to speak of selective abortion not as the abandonment of parental care, but quite the contrary, as the very manifestation of such care.

[12] At the same time, there are other genetic afflictions in relation to which an argument for genetic abortion is not so easily made - if it can be made at all. Take, once again, the case of Down's syndrome. True enough, a Down's syndrome life is substantially limited in potential; some degree of mental retardation is almost always a symptom, and there are often physical disabilities with varying levels of severity. Still, children stricken with this condition often appear to be quite happy. They frequently take pride in their accomplishments (witness the Special Olympics), experience pleasure in bringing joy to others, and form lasting relationships. Given these facts, it would be implausible to suggest that for a typical Down's syndrome child life is not worth living, and thus difficult to construe an abortion of a Down's fetus as an informed expression of parental care.

[13] Finally, there will be borderline cases (neural tube afflictions) where there is enormous variation in degree of severity and where the exact degree in a given case cannot be projected by the appropriate prenatal tests. Given a moral presumption against fetal destruction, perhaps the reasonable choice under such conditions of uncertainty is to forego the abortion option in the hope that a tolerable existence will be achieved. However one assesses the matter, there can be no denying that abortion decisions in these contexts will be extremely difficult. The point to stress here is that morally responsible judgments in such cases will be those which keep in full view the fetus's well-being. That is, as far as can be determined, would abortion be in the interests of this child?

Mass Prenatal Screening Programs

[14] Beyond the moral issues related to individual parental choice about prenatal

diagnosis and genetic abortion, we are faced with a series of important ethical questions generated by mass prenatal screening proposals. In this section I shall address some of these questions, though it should be noted at the outset that the issues are enormous and warrant more detailed treatment than can be given them here. For the sake of convenience I shall focus on one of the more commonly discussed proposals, namely, advocacy of a mass prenatal screening program designed to detect neural tube defects (e. g. , anencephaly, spina bifida) in the general population. Needless to say, I proceed with the same moral reservations expressed earlier about aborting fetuses when such defects are indicated prenatally. Yet given the current situation, in which abortions are legally permissible, it becomes important to consider the proposal with an eye to other ethical questions that have structured the policy debate.

[15] A mass screening program for neural tube defect would involve routine implementation of a series of maternal tests currently provided by a number of medical centers in this country and others. The first of the series is given between the fifteenth and twentieth weeks of pregnancy, and is designed to measure AFP (alpha-fetoprotein) levels in the maternal blood. Excess levels of AFP in the blood at this stage of pregnancy signify any of the following conditions: (1) a twin pregnancy (2) a pregnancy further advanced than originally thought, or (3) a fetus suffering from neural tube defect. If this first test is positive, then the step is repeated, and often the result will be negative. If the second test is positive, then ultrasound is employed to determine whether condition (1) or (2) obtains. If ultrasound fails to detect either condition, then amniocentesis is performed to assess the levels of AFP in the amniotic fluid. An abnormally high level is a fairly reliable indicator of tubal defect, though there is still the slight possibility of misdiagnosis. At this juncture, either of two confirmatory tests is occasionally employed: an advanced ultrasound technique, which under optimal conditions can detect spina bifida lesions, or a chemical test for a particular enzyme (acetylcholinesterase) whose presence is associated with tubal defect. Given positive results at this point, the odds are very strong that the mother is carrying an affected fetus. On the other hand, for those women who have received negative results at any stage of the process, the likelihood is high that the pregnancy is unaffected.

[16] Such a screening program gives rise to a number of difficult moral questions related to the mutual expectations and responsibilities of individual physician and client participants.[8] How should a physician respond to a woman who wishes to undergo the screening process but who also announces her intention not to abort her child under any circumstances? May the physician refuse to administer the test on the grounds that the procedure would simply be an imprudent use of valuable resources or, perhaps more important, an unnecessary risk? Suppose a woman who undergoes part of the AFP process with provisionally positive results chooses to abort without submitting to follow-up tests. Should the physician refuse to perform the abortion? Should the physician refer the woman to other physicians who will perform the desired services? Naturally, these sorts of questions will be especially wrenching for any physician who believes that the moral point of participating in screening

programs is to serve parents in the process of caring for their children.

[17] There is also the larger issue of who precisely ought to have access to an AFP screening program. If it is correct to assume that the moral purpose of such a program should be to provide support for parental care, then theoretically the only acceptable condition of access to the system will be the requirement that the applicant be a parent at risk, and thus any restriction based on factors such as wealth or geographical location will be unjust. What this conclusion will mean in practical terms for an AFP program is more difficult to say. In ideal circumstances all who are in need and who give informed consent should be served; yet realistically matters are never so simple. Indeed, under certain conditions of scarcity perhaps we would have to settle for a program with limited outreach, where all parents at risk submitted to some just (e.g., random) preliminary selection process. At any rate, whether resources are scarce or abundant, a prenatal screening program which denies those in need an equal opportunity for entry will be a deficient system from the point of view of justice.

[18] Of course, any moral demand for equal access to an AFP screening system will be fully satisfactory only on the assumption that the overall goals of the program are morally acceptable. As I intimated earlier, there ought to be reservations about such goals because there ought to be reservations about the abortions which the program inevitably encourages. The painful result for someone sharing the fundamental values expressed in this essay will be a divided conscience on many of the policy issues related to the institution and administration of an AFP system. For, while there is a responsibility to raise questions in the public arena about a system whose goals are morally problematic, there is also an obligation within limits to maximize the justice of such a system already in place. The irony is that in seeking this justice one will actually be seeking wider opportunity for participation in an activity one regards as morally dubious. Indeed, I suspect that this sort of tension will inform ethical reflection on many (though not all) of the genetic screening proposals likely to be offered in the future. At the very least, such moral ambivalence will be experienced by those who believe both that fairness requires equal access to the services society affords, and that caring for children, prenatally or postnatally, is an activity which signifies in the created order the divine providential economy at the heart ofthe Christian story.[9]

End Notes

1. John Fletcher, *Coping with Genetic Disorders: A Guide for Clergy and Parents* (San Francisco: Harper & Row, 1982), 184-89.
2. Recently, however, questions have been raised about the safety of ultrasound. See Barbara Bolson, "Question of Risk Still Hovers over Routine Prenatal Use of Ultrasound," *Journal of the American Medical Association* 247 (April 1982), 2195-97.
3. A relatively new technique involving chorion sampling is now beginning to provide diagnostic results much earlier in the pregnancy's term.

Indications are, however, that the risks of the technique are comparable to those of amniocentesis described below. For a general account of the chorion sampling process see A.V. Cadkin, et al., "Chorionic Villi Sampling: A New Technique for Detection of Abnormalities in the First Trimester," *Radiology* 151 (April, 1984), 159-62.

4. Duane Alexander, M.D., "Workgroup Paper: Risks of Amniocentesis in Maternal Serum Alpha-Fetoprotein: Issues in the Prenatal Screening and Diagnosis Neural Tube Defects, ed. by Barbara Gastel, et al. (Washington. D.C.: U.S. Government Printing Office, 1980), 20-24. Most of the information on risks has been taken from this paper. See also the landmark American, Canadian and British studies: NICHD National Registry for Amniocentesis Study Group, "Midtrimester Amniocentesis for Prenatal Diagnosis: Safety and Accuracy," *Journal of the American Medical Association* 236 (1976), 1471-76; Nancy E. Simpson, et al., "Prenatal Diagnosis of Genetic Disease in Canada: Report of a Collaborative Study," *Canadian Medical Association Journal* 115 (1976), 739-48; MRC Working Party on Amniocentesis, "An Assessment of the. Hazards of Amniocentesis," *British Journal of Obstetrics and Gynecology* 85: Supplement no. 2 (1978),1-41.
5. Department of Health and Human Services, "Amniocentesis for Prenatal Chromosomal Disorders" (Atlanta: Public Health Service, Center for Disease Control, 1980), 15.
6. See David E and Victoria S. Allen, *Ethical issues in Mental Retardation* (Nashville: Abingdon, 1979), 51.
7. Of course, this view of the fetus's moral status and the general stance on abortion are both controversial, but neither can be defended at length here. For an extended exploration of the notion that the fetus is an unborn child see William Werpehowski, "The Pathos and Promise of Christian Ethics: A Study of the Abortion Debate," (unpublished paper).
8. Natalie Abrams, "Workgroup Paper: Ethical issues in MSAFP Screening Programs," in *Maternal Serum Alpha-Fetoprotein*, ed. by Gastel, et al.
9. I am indebted to Edward Langerak, Paul Nelson, and Rache D. Santurri for helpful conversations bearing on the contents of this pamphlet.