EDITOR'S NOTE: The following five papers were presented as part of a panel discussion entitled "Beyond Easy Answers: Prenatal Diagnosis and Counseling During Pregnancy" at the 2000 annual meeting of The American Cleft Palate–Craniofacial Association. The series of papers explore how families and doctors respond when a prenatal diagnosis is made, and consider ethical and social issues around counseling, managing information, and making decisions.

Beyond Easy Answers: Prenatal Diagnosis and Counseling During Pregnancy

RONALD P. STRAUSS, D.M.D., PH.D.

Objective: **The advancing sophistication and availability of prenatal diagnostic technologies, such as transvaginal ultrasound, chorionic villus sampling, amniocentesis, and alpha feto-protein testing, have increased the medical capacity to detect genetic and congenital conditions during pregnancy. This paper raises many social and ethical questions about how families, craniofacial teams, and society respond when a prenatal diagnosis is made and considers the ethical and social issues around counseling, managing information, and making decisions.**

Design: **Ethical and sociological analysis.**

Setting: **Implications examined on the societal, health professional, and family level.**

Results: **Families and health professionals often manage ambiguity, uncertainty, and complex decision making in facing a prenatal diagnosis. Embedded in parental and clinical decisions are values about children with birth defects. Families are making decisions about whether to bear or abort an affected fetus on the basis of their perceptions of the impairment and on their expectation of the burden involved for the family and the child. On a broader, societal level, pressures to conform and minimize human differences are apparent in biomedical interventions, the Human Genome Project, advertising and media images, and social pressures to normalize disabilities. How society deals with prenatal diagnosis will impact upon social values; moral, legal, and ethical perspectives; and on health policy.**

Conclusion: **Prenatal diagnostic technologies raise complex ethical, family, policy, and legal issues that have broad implications for the lives of children born with special health care needs, including children with cleft lip and palate.**

KEY WORDS: *cleft lip, cleft palate, counseling, prenatal diagnosis*

The increasing sophistication and availability of prenatal diagnostic technologies, such as transvaginal ultrasound, chorionic villus sampling, amniocentesis, and alpha feto-protein testing, have increased the medical capacity to detect genetic and congenital conditions during pregnancy. The availability of this technology has made it possible for parents to learn a great deal about the health of a fetus while *in utero.* Such information clearly is desired by parents who seek to screen the unborn for birth defects and other health conditions that can be identified by prenatal testing. On the basis of prenatal diagnostic findings, parents may choose to continue the pregnancy with a new awareness and armed with information, may seek prenatal therapy if that is available, or they may choose to terminate the pregnancy. Prenatal diagnostic technology implies a high degree of parental choice regarding the fetus. However, little is known about how families and professionals deal with prenatally diagnosed conditions. How do they manage the inevitable ambiguity, uncertainty, and difficult decision making that happens with a prenatal diagnosis? Aside from the parental issues, there are societal implications of prenatal diagnosis (Bosk, 1992). One might seriously consider the soci-

Dr. Strauss is Professor and Chair, Department of Dental Ecology, University of North Carolina School of Dentistry, and Professor, Department of Social Medicine, University of North Carolina School of Medicine, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina.

This paper was presented as part of a panel entitled ''Beyond Easy Answers: Prenatal Diagnosis and Counseling During Pregnancy'' at the Annual Meeting of the American Cleft Palate–Craniofacial Association; April 12, 2000; Atlanta, Georgia.

Submitted September 2000; Accepted May 2001.

Reprint requests to: Dr. Ronald Strauss, Department of Dental Ecology, University of North Carolina School of Dentistry, University of North Carolina at Chapel Hill, C.B. #7450, Chapel Hill, NC 27599-7450. E-mail Ron_Strauss@unc.edu.

etal impact of prenatal diagnosis in terms of tolerance for human difference. This paper will examine the issues relating to prenatal diagnosis that are likely to arise in medical practice and in craniofacial centers.

Options for Families After Prenatal Diagnosis of an Affected Fetus

The success of prenatal diagnostic technologies occurs at a moment in medical history at which there is relative ease of access to termination of pregnancy by abortion in the United States. Although organized antiabortion efforts exist, most Americans who desire termination of pregnancy because of a prenatally diagnosed fetal condition will generally be able to access abortion if they so desire. The right to choose to terminate a pregnancy, although limited, has been protected by judicial decisions and social policy.

The parental decision about whether to terminate a pregnancy in the face of a birth defect is a personal and spiritual matter (Eng et al., 1997). However, embedded in these parental decisions are values about the social and familial worth of children with birth defects. Some families will perceive a prenatally diagnosed birth defect to be acceptable and will plan to provide care and treatment to the child in the postpartum period. Some families will perceive a child with a known birth defect as a potential burden, or they may fear that the child will ultimately not have a high quality of life. Some families weigh the risks and costs of treatment against the chance that they could ''try again'' to have an unaffected pregnancy. Some families plan to place a child for adoption after delivery if they feel incapable of providing parenting and care to the affected child.

Families are also making decisions about what defects are so serious as to make the birth of an affected fetus undesirable (Wexler, 1995). Although families are rarely aware that their decision about whether to continue the pregnancy has broader societal implications, for those working in craniofacial care, this is apparent.

Prenatal Diagnosis and Human Conformity

At the same time that prenatal diagnosis occurs more commonly, the United States is also investing ever more costly and scarce health care resources in the care of newborns with craniofacial and other birth defects. The neonatal intensive care unit and its remarkable capacity for helping seriously impaired children survive represents a major societal and fiscal investment in the preservation and future health of disabled newborns. The capacity to survive and thrive with a birth defect has never been greater. At this same moment, many affected fetuses, if prenatally diagnosed, are never born. Prenatal diagnosis has become a more routine part of pregnancy, and noninvasive tests increasingly allow for fetal screening without the risk of premature labor. Although these various trends might at first seem like contradictions or polarities, they may really represent a single position—that all biomedical efforts must be exerted to reduce and repair disfigurations and disabilities in newborns.

Prior to the advent of prenatal diagnosis, most parents had little choice but to accept the birth of a child with a birth defect. The ability to ''know'' in advance about a future child's special needs now allows for parents to anticipate future social pressures from the family or community regarding the integration and social roles for the child. If parents perceive that they or the child will be greeted with tolerance and that efforts will be made to accept the child, they will feel supported. If they perceive that they or the child will face a high level of stigmatization and prejudice, the family may feel isolated and marginalized. The pressures for conformity to a common societal standard of appearance or function are evident. The impetus to conformity is also possible in other biomedical interventions, such as in the Human Genome Project. Conformity is also seen in advertising and media images and in the social pressures placed on persons with disabilities to normalize by surgical and other treatment efforts (Lapham et al., 1996).

Is prenatal diagnosis a vehicle to normalize human differences and reduce variability in the human genome? Is there inherent, but unappreciated, value in the variation expressed in the human genome? Will the Human Genome Project and genetic research be used to define what is normatively human? Or will such research show that all humans have diversity in their phenotypic manifestations and that all people possess genes that are suboptimal and that encode their future illnesses? Are we as a society able to identify genetic differences among people but not to seek to normalize non–life-threatening genetic imperfections (Strauss, 1983)? Are we ready to manage genetic knowledge?

The Changing Emotional Landscape of Pregnancy With Prenatal Technology

For families, the ability to visualize the fetus or determine its developmental stage, health, or sex has changed the emotional and psychological landscape of pregnancy. Parents are bonding to and identifying with fetuses as children at a much earlier time. At one point the ''quickening'' or movement of a fetus was a landmark for parents. Now quickening is often preceded by hearing the heartbeat and getting a sonogram photograph of the unborn baby. Parents bond to their first baby picture months before the birth actually occurs. The use of prenatal visualization encourages families to personalize the fetus and invest in what has essentially become a new member of the family (Rothman, 1986). When the family has received a photograph of the fetus, knows its sex, and selects a name on the basis of prenatal diagnostic information, does the meaning of birth change?

Craniofacial centers report that parents sometimes seek prenatal consultation and referral in anticipation of their baby's needs or collect advance information. Activist parents may arrive at craniofacial centers with sheaves of Web-based printouts about their future child's possible condition, making it clear that there is a high level of family anxiety. The families

will sometimes feel as though they need to have a clear treatment plan and seek to understand the extent of their treatment choices in advance of their child's birth.

The ease and access to prenatal diagnostic technologies raises other important emotional and psychological questions. Is prior knowledge of a craniofacial defect an advantage to families? What are the implications of aborting fetuses that have conditions that are not life threatening and are largely repairable? Who should have access to prenatal diagnostic information (Murray and Botkin, 1995)? Should prenatal testing be limited to the detection of conditions apparent in infancy, or should diagnosis seek to predict adult health status? Should fetuses be tested for a predisposition to develop breast cancer, cardiovascular disease, or diabetes as adults?

As prenatal diagnosis becomes more accurate and available at ever-earlier times in pregnancy, other psychological and societal issues will arise.

Prenatal Diagnosis: Implications for Cleft/Craniofacial Team Clinicians

Craniofacial centers have reported ethical issues related to prenatal diagnosis and nonlethal birth defects. Several recent papers (Blumenfeld et al., 1999; Eiserman and Strauss, 1999; Jones, 1999) discussed an Israeli center's experience with prenatal diagnosis and weighed the Israeli parents' decisions to abort 23 of 24 fetuses identified with cleft lip at 13 to 16 weeks of gestation. The Israeli center's use of early prenatal diagnosis and the parents' subsequent decisions to terminate their pregnancies in response to prenatal diagnosis raised concerns. Some have concluded that the Israeli parents and clinicians in this study are weighing the value of living a life against the impact of having a cleft lip. Fundamental questions about the rationale for prenatal diagnosis are raised if a repairable condition such as cleft lip can be widely seen as the basis for the termination of pregnancy. Is prenatal diagnosis meant to detect seriously impairing or life-threatening conditions, or is it meant to identify genetic or congenital traits that are compatible with a high-quality life span? And who should make that determination?

In evaluating the social and ethical impacts of prenatal diagnosis, it is important to differentiate between fetuses with life-threatening and seriously impairing conditions and those with conditions that are compatible with a high quality of life. The line that lies between these two groups probably is culturally and socially determined (Wertz and Fletcher, 1989). What criteria do people use to judge a future child's quality of life? Who should participate in this dialogue?

For craniofacial health professionals who classically focus on the giving of care to individuals with cleft lip and other craniofacial conditions, other ethical quandaries arise. Advocates for persons with disabilities have asked whether a craniofacial team clinician has any reason to participate in efforts that might result in selectively preventing the birth of individuals, whom, if born, would have become their patients. They ask how craniofacial health professionals can treat their patients with oral clefts one minute and turn around the next minute and have a conversation in which a fetus with a cleft lip or palate is not seen as having the potential for a ''good life.'' Can parents trust in a doctor who might not fully believe their child's life to be worthy?

Should the prenatal diagnostic adviser be entirely separate from the treatment team? Does an overlap of treatment and prenatal diagnostic functions imply a conflict of interest? Some hold that craniofacial team professionals should focus on promoting the quality of life for all their patients and not become engaged in advising parents about prenatal diagnosis and about the possible termination of pregnancy. Others would say that craniofacial team members have unique knowledge and information that parents will need to guide their decisions about whether to bear the affected child.

Counseling and Conveying Optimism: A Role for Team Professionals?

The establishment of optimism and hope around a craniofacial diagnosis may be a critical step in starting the process of treatment and care, even during the prenatal period. It is their special vantage point that permits craniofacial health professionals to help families and individuals with cleft lip and palate to see their lives as rich with possibilities. They can help families envision good outcomes through treatment and prepare them for the course of therapy. They can help families to see that the unique and often difficult experiences faced by persons with cleft lip and palate may also afford them with special and worthwhile perspectives. To succeed in this role, craniofacial professionals need to deeply believe in and value the lives of persons with cleft and craniofacial conditions.

For the affected individual, even the discussion about the value of a life lived with a cleft raises the fear that somehow their life was not worth it, that somehow they have placed a burden on their parents that was just too heavy. As Eiserman and Strauss (1999) suggested, perhaps some affected persons wonder, ''What would my family's life have been like if they had been spared all the expense and heartache that I brought with me?''

How professionals present information regarding a prenatal diagnosis may determine much about how parents will respond. If the professionals ''hang crepe'' and describe every possible complication, every frightening ramification, no matter how rare, then it becomes more likely that parents will decide to terminate the pregnancy. What should be included in the professional's discussion with the parents about prenatal diagnosis? Where and with whom present should this discussion occur?

It is well accepted that professionals should not serve as opinionated advocates for or against the termination of pregnancy in the wake of a prenatal diagnosis (Marteau, 1998). Professionals must always respect the family's right to autonomously make such critical decisions, without having foregone conclusions about what the family should do. The decision about how to respond to a prenatal diagnosis is never easy for

parents, and perhaps it should always entail a difficult, personal, and weighty consideration for the family. Prenatal diagnosis must be seen as a process that includes the professional provision of information and caring support after a finding occurs. It is critical to evaluate how prenatal counseling occurs and in what manner the professional provides the family with needed information. Parents must be offered support during this period and must be given the full spectrum of decisions and choices. It is possible that termination of pregnancy should be offered, or discussed, only if the family raises the question. All parents should be provided information about the treatment and support available for their unborn child.

The prenatal diagnosis of a congenital defect marks the beginning of a period of high stress and uncertainty for many parents. When people receive bad news, they experience a loss of control and are in crisis. This may be a time when parents are vulnerable and are easily swayed by the opinions of professionals or other parents. Prospective parents should be carefully informed of all of their possible options and afforded time to make their decision. In the prenatal period, prospective parents should receive nonjudgmental and unbiased information, support, and advice from health professionals who are specifically trained to counsel and support decisions. Family physicians and other professionals with a continuous relationship with the family would be ideal counselors at this time of considerable parental stress. In a paper by Jones (1999), the presentation of information to families was described in a positive way, with good achievable outcomes.

Legal and Ethical Issues

Privacy and Availability of Testing and Information

The question of privacy and confidentiality and who is given prenatal diagnostic information has become an issue (Weaver, 1997). Insurance policies often do not cover persons who have pre-existing conditions. Will prenatal diagnostic information result in exclusions of such conditions as pre-existing? Could insurers avoid women known to be carrying an affected fetus, or could they selectively exclude those who choose to bear a child with a predictable health problem? Could insurers require mandatory prenatal diagnostic evaluations and demand to see the results? Could these studies be part of pre-employment physicals? Could prenatal genetic screening become mandatory, as has PKU screening? Other issues exist relative to the distribution of costly and scarce resources, such as prenatal diagnostic equipment. Will affluent societies commonly abort fetuses with genetic disorders, leaving those in impoverished nations with a greater relative burden of disease and disability than already exists?

Wrongful Birth

Legal issues have on rare occasion arisen as a result of elevated expectations regarding the precision and conduct of prenatal medical professional practice. Childbearing is increasingly perceived by the public as a process with controllable risks, in which health professionals are expected to assure the birth of healthy offspring. When physicians have not informed parents of the availability of a prenatal test, performed the test improperly, failed to refer, failed to act on a positive finding, or neglected to inform the parents about the risk to the child, the courts have sometimes intervened (Lambert, 1983). Such legal actions, called wrongful birth suits (Rogers, 1982), are based on the claim that parents have been deprived the right to make an informed decision about the pregnancy. Wrongful birth judgments have concluded that the physician may be at fault for effectively depriving parents of their right to make an informed decision about maintaining or terminating the pregnancy (Annas and Elias, 1985). The wrongful birth suits have sometimes resulted in large financial awards to cover the expenses of health care, personal supervision, and maintenance for handicapped persons, sometimes through their lifespan (*Phillips v U.S.*, 1981). The parents of the child generally receive such settlements, often with few guidelines for how the funds are to be spent.

Wrongful Life

The wrongful life suit differs from the wrongful birth suit. The wrongful life suit is the claim on the behalf of the child that it might have been better not to be born than to have been born disabled. This provides an affected child with the possibility of receiving compensation for the health and special care that he or she will require in life. These settlements are recognition that the child is a person with rights including rights to have been aborted or never conceived. The courts have had to weigh whether there are children who would be better off having never been born. They have considered whether it is ever parent negligence to permit a fetus's birth given the life risks that the newborn would face (i.e., crack babies, fetal alcohol exposure). This is an area of debate, and the legal awards that attempt to compensate for a specific condition are very controversial.

Impacts of Litigation

The ultimate issue raised by these wrongful birth/life lawsuits regards the determination of who is responsible for the costs of the care of children with special needs. If the medical professional might have taken action to prevent the child from being born, should he or she have done so? Is it a parental right to have information available on which to base a decision about a child's birth? Some have even argued that parents who do not choose to abort or protect a fetus should be liable to be sued for wrongful life by the child (Shaw, 1980).

The development of genetic screening methods implies not only their use but also the availability of accurate results and nonjudgmental follow-up counseling. When prenatal or genetic tests were unavailable, congenital anomaly risk was generally unknown, and there was less possibility of disagreement over professional responsibility or wrongful birth. With the advent

of readily available genetic testing, the expectations for professional skill and follow-up became high. Litigation is now seen as a means to create accountability.

Maternal-Fetal Conflicts

When prenatal diagnosis is performed, the fetus comes to be thought of as a ''patient,'' even though unborn. It raises basic questions about the rights and responsibilities of the mother, the physician, and the fetus. Do fetuses have rights? Past U.S. Supreme Court decisions have found that the mother's interests override the interests of the fetus up to the point of viability. Some (Robertson, 1986) have posed the question of whether fetuses that are going to be carried to term have rights that protect them from injury? It is possible for a parent or other person to knowingly injure a fetus, by neglect or substance abuse, and be liable for such injury.

In *Grodin v Grodin* (1981), a child brought suit against his mother for her negligence in taking tetracycline during pregnancy and causing his teeth to be discolored. Can such a liability be broadened to presume that a fetus has the right to demand consistent and caring maternal behavior? Should an alcoholic mother be subject to suit from her child with fetal alcohol syndrome? One wonders whether physicians or parents who choose not to pursue prenatal diagnosis might be subject to future liability.

What rights does the mother have to freedom and autonomy over herself? Does her right to self- determination override the rights of the fetus or the physician? Currently, it is widely held that mothers have the right to refuse prenatal diagnosis. This right may become problematic as prenatal diagnostic testing becomes routine medical practice, and it is assumed that parents will desire such information and screening.

In Closing—A Positive Vision of Future Craniofacial Practice

Craniofacial centers can be places that assist families in understanding and responding to prenatal information. They can help parents of yet-unborn children evaluate the condition of their fetus, and they can help to project the range of possibilities that might occur in their child's life. They can help lighten the load of parents bearing Internet printouts about their baby's condition by guiding their search, assuring their data are accurate, and interpreting information. They can help the many families for whom prenatal diagnostic data may just be too much information to handle on their own. They can constantly be aware that their medical advice in the delicate aftermath of prenatal diagnosis may easily sway a family's perspectives about a pregnancy. They can be especially careful to be neutral, nonjudgmental, and accurate. They can help a family assess whether a child with a less-than-perfect body or face could thrive in their home. They can help a family that has decided to bear an affected child to see the future in hopeful terms. The craniofacial team's ability to manage prenatal diagnostic and future genome technologies will depend on their insight, wisdom, and vision of what constitutes a good life.

References

Annas GJ, Elias S. Perspectives on fetal surgery. On the road from experimentation to therapy (and what to do when we arrive). In: Milunsky A, Annas GJ, eds. *Genetics and the Law, III.* New York: Plenum Press; 1985:347–363.

Bosk CL. *All God's Mistakes: Genetic Counseling in a Pediatric Hospital.* Chicago: University of Chicago Press; 1992.

Blumenfeld Z, Blumenfeld I, Bronshtein M. The early prenatal diagnosis of cleft lip and the decision-making process. *Cleft Palate Craniofac J.* 1999; 36:105–107.

Eiserman W, Strauss R. Commentary on The early prenatal diagnosis of cleft lip and the decision-making process [Letter to the editor.] *Cleft Palate Craniofac J.* 1999;36:542–544.

Eng C, Schechter C, Rabinowitz, Fulop G, Burgert T, Levy B, Zinberg R, Desnick RJ. Prenatal genetic carrier testing using triple disease screening. *JAMA.* 1997;278:1268–1272.

Grodin v Grodin, 301 NW 2d 869; (Mich App 1981).

Jones MC. Prenatal diagnosis of cleft lip and palate: experiences in Southern California. *Cleft Palate Craniofac J.* 1999;36:107–109.

Lambert T. Tort liability for inadequate genetic counseling. *Association of Trial Lawyers of America Law Reporter* 1983;106.

Lapham VE, Kozma C, Weiss JO. Genetic discrimination: perspectives of consumers. *Science.* 1996;274:621–624.

Marteau T. Revealed identity: a study of the process of genetic counseling. *Soc Sci Med.* 1998;47:1653–1658.

Murray TH, Botkin JR. Genetic testing and screening: ethical issues. In: Reich WT, ed. *Encyclopedia of Bioethics.* Vol. 2. 2nd ed. New York: Macmillan; 1995:174–178.

Phillips v U.S., 508 F. Supp. 544 (S.C. 1981).

Robertson JA. Legal issues in prenatal therapy. *Clin Obstet Gynecol.* 1986;29: 603–611.

Rogers TD. Wrongful life and wrongful birth: medical malpractice in genetic counseling and prenatal testing. *South Carolina Law Review* 1982;713:33.

Rothman BK. *The Tentative Pregnancy: Prenatal Diagnosis and the Future of Motherhood.* New York: Viking; 1986.

Shaw M. Preconception and prenatal torts. In: Milunsky A, Annas GJ, eds. *Genetics and the Law II.* New York: Plenum Press; 1980:225–232.

Strauss RP. Ethical and social concerns in facial surgical decision making. *Plast Reconstr Surg.* 1983;72:727–730.

Weaver KD. Genetic screening and the right not to know. *Issues Law Med.* 1997;13:243–281.

Wertz DC, Fletcher JC, eds. *Ethics and Human Genetics: A Cross Cultural Perspective.* Berlin: Springer-Verlag; 1989.

Wexler A. *Mapping Fate: A Memoir of Family, Risk, and Genetic Research.* New York: Random House; 1995.