

Preamble: The Genetic Counseling Program at the University of South Carolina (GCP-USC) from its inception has recognized the strengths and rights of persons with disabilities. We also teach the importance of reaching out to the community to promote understanding of genetic services. Recently, concerns from the Down syndrome advocacy community about prenatal screening and diagnosis have been heightened, particularly in response to the American College of Obstetricians and Gynecologists (ACOG) Practice Bulletins 77¹ and 88². The GCP-USC decided that the time had come to bring representatives of stakeholder organizations together to discuss openly perceptions and misconceptions about prenatal testing as it relates to Down syndrome for the purpose of identifying areas of consensus that could be built upon. The accompanying document is a result of two days of conversations and a testimony to what can happen when the goal is to seek common ground for the greater good.

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Toward Concurrence: Understanding Prenatal Screening and Diagnosis of Down Syndrome from the Health Professional and Advocacy Community Perspectives

The University of South Carolina's Genetic Counseling Program and the University's Center for Disability Resources hosted a meeting of representatives of the National Down Syndrome Society (NDSS), National Down Syndrome Congress (NDSC), American College of Obstetricians and Gynecologists (ACOG), American College of Medical Genetics (ACMG), and National Society of Genetic Counselors (NSGC) on November 16 and 17, 2008, in Columbia, South Carolina. The goal of the "Consensus Conversation" was to promote greater understanding of each organization's perspective on prenatal screening and diagnosis as it relates to Down syndrome. Roundtable discussions over two days allowed for elucidation of misperceptions and identification of areas of consensus. Several potential collaborations were recognized and the representatives affirmed the value of continued communication between the organizations.

Gaining Perspective: Clarifying Misperceptions

Representatives from the five organizations identified several apparent misperceptions among the public, the advocacy community and health care professionals. Simply stated in italics, each misperception listed below is explicated toward the goal of replacing misunderstanding with accurate information.

Misperception 1: Obstetricians recommend prenatal tests to reduce the number of individuals in society who have birth defects and genetic conditions. Prenatal genetic screening and diagnostic tests are available options for women. All parents desire current, complete and unbiased information from their health care providers. Obstetricians inform pregnant women of their options without specific interest in women's choices. Prenatal diagnosis allows for informed reproductive decision making. However, women may decide not to participate in screening or testing. In cases of identified genetic conditions, information available to the patient and her partner may lead to decisions that include further information gathering, emotional preparation for an unexpected outcome, in utero intervention, termination of the pregnancy, personal and family preparation for the birth of the child with a genetic condition, guidance about delivering the child at an appropriate facility, as well as health care provider preparedness to administer to the needs of the neonate and family at the time of delivery.

Misperception 2: The purpose of offering prenatal diagnosis to all women (ACOG Practice Bulletins Nos. 77¹ and 88²) is to decrease the number of births of children with Down syndrome.

The practice of offering amniocentesis to women age 35 and older was set in the mid 1970s.

At that time, the chance at age 35 to have a child with Down syndrome was approximately

the same as the statistical chance of having a procedure-related miscarriage. In the intervening years, the improved safety of amniocentesis and chorionic villus sampling has significantly reduced the risk of a procedure-related miscarriage. Therefore, it is appropriate that women of all ages have the opportunity to consider prenatal diagnosis, and to decide for themselves if they desire the information provided. The January and December 2007 ACOG bulletins cited above lay out current data on screening and testing options to be offered (not necessarily recommended) to pregnant women who might find the information valuable. Importantly, these bulletins do not offer pregnancy termination as the target outcome for prenatally diagnosed conditions, recognizing women's choices are influenced by many personal factors. The value of prenatal test information for preparation toward the birth of a child with a genetic condition is underscored.

Misperception 3: Ninety percent of pregnancies prenatally diagnosed with Down syndrome are terminated. No current, comprehensive estimate of the number of pregnancy terminations following prenatal diagnosis exists. Several studies reporting older data, studies from single centers, and studies from other countries have reflected variation in the number of pregnancies terminated. These studies are frequently cited, but given their limitations, are difficult to generalize to the current population of pregnant women in the United States. Undocumented observations from prenatal genetic counselors in the United States suggest that the rate of termination for prenatally diagnosed Down syndrome may vary across the country. New research is called for to comprehensively explore the uptake of prenatal testing and the outcomes of prenatally diagnosed pregnancies in order to more accurately define how women currently incorporate prenatal testing into their lives.

Misperception 4: NDSS and NDSC are pro-life organizations. These organizations represent thousands of families and focus on advocating for the value, acceptance, and inclusion of people with Down syndrome at the grass roots and public policy levels. Through public education, and promulgation of current information, the advocacy movement strives to ensure that children and adults with Down syndrome are leading healthy, productive lives. The organizations also seek to shift public perceptions of Down syndrome as a medical diagnosis to a larger perspective that recognizes the full potential of people with Down syndrome and underscores their contributions to families and communities.

Misperception 5: Genetic counselors with master's degrees influence parents to accept pregnancy termination. Genetic counselors serve parents who are considering their prenatal diagnosis results in a number of ways, such as providing information and support and facilitating connections to resources. They are encouraged by their professional code of ethics to, “enable clients to make informed decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences”.³ All parents desire current, complete and unbiased information from their health care providers. Genetic counselors therefore strive to present accurate, up-to-date, unbiased information, facilitate decision making in a non-coercive manner and provide support to parents whether they choose to continue a pregnancy, terminate a pregnancy, or place the infant for adoption.

Misperception 6: Prenatal screening and testing are performed exclusively to detect Down syndrome. Down syndrome is among the most common chromosome conditions detected through prenatal screening and diagnosis; hence, these tests, in the aggregate, are often

erroneously referred to as “prenatal tests for Down syndrome.” Prenatal screening and diagnostic testing detect many other conditions. This mislabeling singles out Down syndrome as an implicitly undesirable outcome. In addition, it often confuses screening and testing. Prenatal screening is population-based while prenatal diagnosis is offered based on the chance of a birth defect or genetic condition associated with family or medical history. Health professionals must be cognizant of the language used in all activities—from the publication of practice guidelines to conversations with the public (patients as well as the media)—to refer to these tests as detecting pregnancies with, or at an increased chance for, genetic conditions or birth defects. Likewise, when language is misused, all efforts should be made to correct it.

Common Ground: Recognizing Opportunity in Consensus

As misperceptions were clarified, areas of consensus among the five national organizations were identified. Themes reflecting agreement are listed in bold below:

Public education elucidating the lives and value of individuals with Down syndrome in today's society is necessary. Information presented should be balanced and accurately reflect aspects of the medical, educational and social realms of individuals with Down syndrome and the challenges and opportunities they and their families face.

Health professional education about Down syndrome based on the most up-to-date information is necessary. Disability awareness within primary and continuing medical education that includes current information about Down syndrome is important. Health care provider knowledge base, communication skills, behaviors and attitudes should reflect current understanding of Down syndrome.

Education for expectant parents regarding prenatal genetic screening and prenatal diagnosis should be consistent. Currently there is much variability in the settings and manner in which health care professionals provide this information to parents and the amount of information provided both before and after testing. In recognizing that prenatal testing is a process, there is a need to define who should educate parents along with a minimum level of training of those individuals, when in the preconception or prenatal period this education should ideally be delivered, and how much information, the quality of the information, and in what format(s), it should be presented. In addition, educational materials need to be culturally sensitive and available in the languages necessary to meet the needs of any given community.

Information and counseling provided to parents regarding a prenatal or postnatal genetic diagnosis should be complete, consistent, non-judgmental, and non-coercive. The information should not be solely medically driven but reflective of medical and psychosocial implications as well as inter-related health issues affecting the individual with Down syndrome and his/her family. Care must be taken to make sure that an accurate, up-to-date picture of the opportunities and challenges associated with Down syndrome is provided.

Prenatal testing is a process that provides valuable information in and of itself. For parents who receive a prenatal diagnosis, continuing the pregnancy with plans to raise the newborn, placing the newborn for adoption, and termination of the affected pregnancy are possible options. Parents should be allowed to make their personal choice based on complete, up-to-date information without coercion from health care professionals or the advocacy community.

Moving Forward: Collaborative Potential

This “Consensus Conversation” represents an initial step toward building understanding among health care providers and the Down syndrome advocacy community. Representatives affirmed the value of clarifying understanding and identified several areas where their efforts could be strengthened by working in partnership. Representatives from the NDSS, NDSC, NSGC, ACMG and ACOG agreed that continued communication between the organizations would be valuable. In addition, other stakeholders involved in prenatal testing such as from the American Academy of Family Physicians and the American Academy of Pediatrics should be included in future cooperative activity.

Several potential collaborations were identified as next steps. First, the NDSS and NDSC have received a grant to develop a “gold standard” packet of information to be given to parents receiving a new diagnosis of Down syndrome, and a training manual for parent-to-parent support. These efforts will strengthen NDSS and NDSC affiliate programs for new and expectant parents throughout the United States. The NSGC, ACMG, and ACOG representatives offered to provide input to the materials developed from this project. All of the organizations recognized the need to develop materials and methods that will transcend barriers to reach families who are underserved, are from diverse cultures or have other barriers, such as language, that might impede understanding.

Representatives from the ACMG, ACOG, NDSC, NDSS, and NSGC agreed that families deserve current, comprehensive and unbiased information about Down syndrome. Practice guidelines and patient education materials around prenatal and postnatal diagnosis of Down syndrome afford another important opportunity for collaboration among health care and

advocacy organizations. The organizations offered to provide input on guidelines and patient education materials being developed by the other organizations in attendance as a method of enhancing the effort toward current, comprehensive information about Down syndrome. Shared input among national organizations would promote prevention of misperceptions before new guidelines and patient education materials are published or disseminated to practitioners for implementation.

Research to define models for best practice is encouraged. The consensus included acknowledgement that the health care community as well as the Down syndrome advocacy community have much to explore through research, as we seek to define the kind(s) of information that should be presented to parents with a new diagnosis, and the most appropriate provider(s) to deliver the information, and as we broaden our understanding of how parents utilize this information in decision-making. The call for evidence-based medical practice demands focus on these important aspects of prenatal screening and diagnosis.

Finding common ground has laid a foundation for future work together as multidisciplinary organizations. Much was gained from thoughtful, personal exchange and participants departed with sincere intention for continuing conversations and collaborations. This first endeavor into substantial dialogue has moved the National Down Syndrome Society, the National Down Syndrome Congress, the American College of Obstetricians and Gynecologists, the American College of Medical Genetics and the National Society of Genetic Counselors toward greater mutual understanding.

Consensus Conversation Co-Chairs: Janice Edwards, Richard Ferrante

ACOG Representative: Anthony Gregg

ACMG Representatives: Robert Saul, Judith Benkendorf

NDSC Representatives: Laura Cifra-Bean, Rich Robison

NDSS Representatives: George Capone, David Hoppe, Madeleine Will

NSGC Representatives: Angela Trepanier, Meghan Carey, Cam Brasington

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References

¹Screening for fetal chromosomal abnormalities. ACOG Practice Bulletin No. 77. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007;109:207-217.

²Invasive prenatal testing for aneuploidy. ACOG Practice Bulletin No. 88. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2007;110:1459-67.

³National Society of Genetic Counselors (1992) The NSGC Code of Ethics. *Journal of Genetic Counseling* 1(1):41-44.