

Prenatal Diagnosis of Down Syndrome: How Best to Deliver the News

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We sought to provide evidence-based recommendations to physicians on how to best deliver a prenatal diagnosis of DS to expectant parents. Our study design consisted of searching Medline and PsychInfo from 1960 to 2008, as well as Web sites from academic organizations and other nonprofit or private organizations, using the terms “Down syndrome,” “Trisomy 21,” “mongolism,” “prenatal diagnosis,” “postnatal care,” and “delivery of health care.” Our results showed that a health care professional knowledgeable about DS with specific training in the delivery of sensitive diagnoses should be part of the first conversation. A prenatal diagnosis of DS should be presented in person or at a pre-established time by phone. Physicians should provide accurate information about medical conditions associated with DS and connect parents to local DS support groups and other resources. We conclude that physicians can deliver prenatal diagnoses of DS in a sensitive manner that can be appreciated by expectant parents. © 2009 Wiley-Liss, Inc.

Key words: delivery of health care; Down syndrome; prenatal diagnosis; postnatal care; mongolism; Trisomy 21

INTRODUCTION

Delivering a prenatal diagnosis of Down syndrome (DS) to expectant parents is a challenging medical encounter for even the most seasoned physicians. Traditionally, formal training on how to deliver a diagnosis has been limited to genetic counselors or geneticists, but as more prenatal testing options become available, obstetricians will increasingly find themselves in need of this education. Recently, the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics (ACMG) recommended that all pregnant women, regardless of age, be offered prenatal screening and diagnostic testing for DS by their obstetrician [ACOG Committee on Practice Bulletins, 2007; American College of Obstetricians and Gynecologists, 2007; Driscoll et al., 2008].

A menu of prenatal screening tests exists—the triple screen, quadruple screen, first-trimester combined screen, sequential

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screen, and integrated screen—which can provide between 69% and 96% detection rates for DS, using a 5% false positive screen rate with cut-off risk ratios around 1:270 [ACOG Committee on Practice Bulletins, 2007]. DS, however, can only be definitively diagnosed by karyotyping or chromosome analysis using chorionic villus sampling (CVS) in the first trimester or amniocentesis in the second or third trimester, with no statistical increase in procedure-related fetal loss rate now being reported at some centers [Odibo et al., 2008]. In the near future, noninvasive serum testing involving cell-free fetal DNA or RNA might also provide a definitive diagnosis of DS in the first trimester at no risk to the fetus [Lo et al., 2007; Fan et al., 2008; Lo and Chiu, 2008; Puszyc et al., 2008].

Yet, with these new guidelines and scientific tests comes a central question: Are today's physicians adequately trained in explaining a prenatal diagnosis of DS to expectant parents? In a survey conducted in 2005 of 2,500 medical school deans, students, and residency directors, 81% of medical students report that they “are not getting any clinical training regarding individuals with intellectual disabilities,” and 58% of medical school deans say such training is not a high priority [Special Olympics, 2007]. In a

The members of the Down Syndrome Diagnosis Study Group are listed in the Appendix.

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questionnaire completed by 532 ACOG fellows and junior fellows in 2004, 45% rated their training regarding prenatal diagnosis as “barely adequate or nonexistent,” and only 28% felt “well qualified” in general prenatal genetic counseling [Cleary-Goldman et al., 2006]. A survey of 507 ACOG fellows and junior fellows conducted 4 years later found little change—approximately 40% thought their training was “less than adequate,” and only 36% felt “well qualified” in counseling an expectant mother whose prenatal screen suggests a high chance for Down syndrome [Driscoll et al., 2009]. Not surprising, then, is the fact that today’s obstetricians have been deemed “incomplete” and “inaccurate” in delivering a diagnosis by mothers who had children with DS diagnosed prenatally [Skotko, 2005]. Further, in anonymous surveys completed by 499 physicians who deliver prenatal diagnoses, only 63% of them “tried to be as unbiased as possible when delivering a prenatal diagnosis.” Thirteen percent reported that they “emphasize” the negative aspects of DS so that parents would favor a termination; 10% actively “urge” parents to terminate; 10% “emphasize” the positive aspects of DS so that parents favor continuation; and 4% actively “urge” parents to continue the pregnancy [Wertz, 2000].

The primary goal of this report is to review the current evidence on how physicians should best deliver a prenatal diagnosis of DS to expectant parents. As DS remains the most common chromosomal condition, occurring in 1 out of every 733 live births [Canfield et al., 2006], with the average life expectancy now approaching 55 years, nearly every obstetrician can expect to have a conversation with expectant parents about the realities of life with DS. We seek to provide today’s obstetricians with evidence-based recommendations based on the current body of published literature on how to approach these sensitive encounters.

MATERIALS AND METHODS

Sources

We searched online databases including Medline and PsychInfo for all studies published in English from 1960 through February, 2008, using the National Library of Medicine Medical Subject Headings terms: “Down syndrome,” “Trisomy 21,” “mongolism,” “prenatal diagnosis,” “postnatal care,” and “delivery of health care.” To ensure completeness of the literature search, we reviewed reference lists and articles from the authors’ libraries. We supplemented the primary literature search by searching the Web sites of following entities: Google Scholars, American Academy of Pediatrics (AAP), ACOG, ACMG, the National Newborn Screening and Genetics Resource Center, the National Down Syndrome Society, the National Down Syndrome Congress, March of Dimes, National Federation of Voluntary Bodies, International Mosaic Down Syndrome Association, Down Syndrome Educational Trust, Canadian Down Syndrome Society, UK Down Syndrome Association. Additional studies were located by reviewing references of previously identified articles. We excluded unpublished data or studies that were not submitted to peer-reviewed journals.

Study Selection

Prior to the start of this review, we solicited input from an expert in study design and public health policy. To keep our initial search as

unbiased and comprehensive as possible, we chose to review a wide range of study designs, selecting to analyze, on a consistent basis, the titles and abstracts of the articles that appeared to answer one or more of our research questions, established a priori: (1) Who is the best person to communicate the news? (2) When is the best time to share the news? (3) Where is the best place or setting to deliver the news? (4) What information should be delivered? (5) How should the news be communicated? Our research questions for the post-natal period are reported elsewhere [Skotko et al., in press].

After independently reading, in full, all of the articles meeting the initial criteria, the primary authors discussed and then eliminated those articles that (1) did not answer any of the research questions established a priori, (2) did not have results that were specific to DS, (3) contained only duplicative and not original data, (4) contained only opinion based on clinical or personal experience, or (5) had a participant pool <10 persons. No discordant opinion among the authors occurred with this criteria. In total, we identified five articles, with a composite sample size of 232 parents who had received a definitive prenatal diagnosis of Down syndrome and 70 professionals who participated in delivering such a diagnosis [Helm et al., 1998; Williams et al., 2002; Tymstra et al., 2004; Skotko, 2005; Korenromp et al., 2007]. These studies came from a variety of countries from 1998 to 2007. Four of the five articles surveyed mothers who chose to continue their pregnancies after receiving a definitive prenatal diagnosis of DS for their fetus; one article questioned mothers who chose to terminate a pregnancy after receiving a definitive prenatal diagnosis of DS (Table I). All studies meeting final criteria were evaluated for quality by 1996 USPSTF guidelines [Agency for Healthcare Research and Quality, 2008]. Levels of evidence are indicated in Table I.

RESULTS

Who Is the Best Person to Communicate the News?

Pregnant women first learn about a prenatal diagnosis of DS from a variety of people: genetic counselors [Skotko, 2005], midwives [Williams et al., 2002], nurses [Helm et al., 1998], pediatricians and pediatric subspecialists [Williams et al., 2002], and obstetricians [Williams et al., 2002; Skotko, 2005]. Over time, pregnant women have consistently preferred to receive the news from the health care professional that is most knowledgeable about DS [Williams et al., 2002; Skotko, 2005]. However, this is not always the obstetrician, so collaboration among health care professionals is essential. A study of 141 women who had received a prenatal diagnosis of DS cautions that “if obstetricians rely on genetic counselors or other specialists to convey DS, sensitive, accurate, and consistent messages must be conveyed” [Skotko, 2005].

When Is the Best Time to Share the News?

In general, pregnant women who choose to undergo definitive testing prefer to receive the diagnosis as soon as possible in the company of their husbands or partners [Skotko, 2005; Helm et al., 1998]. In a survey of 141 women who had received a prenatal diagnosis of DS, 71% of them had learned of the diagnosis with their partners present [Skotko, 2005]. Although pregnant women generally prefer to receive the diagnosis in person, as opposed to over

TABLE I. Details of Articles Included in the Literature Review, Listed in Chronological Order

References	Location of study	Participants	Study design	Level of evidence ^a
Helm et al. [1998]	United States	N = 10, mothers who continued pregnancy with fetus with DS	Retrospective case study with in-person interviews	II-3
Williams et al. [2002]	United Kingdom	N = 70, practitioners involved in perinatal care	Retrospective case study with in-person interviews	II-3
Tymstra et al. [2004]	Netherlands	N = 10, mothers who continued pregnancy with fetus with DS	Retrospective case study with in-person interviews	II-3
Skotko [2005]	United States	N = 141, mothers who continued pregnancy with fetus with DS	Retrospective case study with mailed questionnaires	II-3
Korenromp et al. [2007]	Netherlands	N = 71, women who terminated pregnancy with fetus with DS	Prospective cohort study with mailed questionnaires	II-2

DS, Down syndrome.

^aLevels of evidence as established by 1996 USPSTF guidelines [Agency for Healthcare Research and Quality, 2008].

the telephone, pregnant women who had arranged a phone call with their physicians at a pre-established time to learn the test results were better able to prepare themselves [Skotko, 2005; Helm et al., 1998]. Mothers who received the diagnosis prenatally were often happier with the birth of their child than mothers who had received the diagnosis postnatally [Skotko, 2005]. This pattern can be attributed to the fact that mothers who receive the diagnosis prenatally have chosen to have a child with DS and have more time to come to terms with the diagnosis [Skotko, 2005].

General information about DS, however, should not be saved until a definitive diagnosis is made [Williams et al., 2002]. Many pregnant women choose some form of prenatal screening prior to a more invasive test such as CVS or amniocentesis. Physicians should acknowledge that *screening* is an optional procedure and that having knowledge about possible fetal anomalies has an ethical dimension for some persons, which should be considered a priori [Williams et al., 2002]. Further, some pregnant women misunderstand the screening tests as diagnostic options [Skotko, 2005]. Physicians should spend time explaining the difference between screening and diagnostic testing, being careful to indicate that the results of prenatal screening will come as a risk assessment and not as a “positive” or “negative” result [Skotko, 2005].

Where Is the Best Place or Setting to Deliver the News?

Pregnant women generally prefer to receive the prenatal diagnosis during a personal visit with their physician, as opposed to receiving the diagnosis at home over the telephone [Helm et al., 1998; Skotko, 2005]. The percentage of pregnant women who actually learned of the diagnosis at an in-person visit with their physician varied by study—20% (N = 10) in 1998 [Helm et al., 1998], 40% (N = 10) in 2004 [Tymstra et al., 2004], and 27% (N = 141) in 2005 [Skotko, 2005]. Some pregnant women who had learned of the diagnosis by an unscheduled call from their physician or physician’s office expressed intense resentment toward their obstetricians and genetic counselors [Tymstra et al., 2004; Skotko, 2005]. By contrast, pregnant women who had received the diagnosis by telephone at a pre-established time were able to ensure that they were in a setting

that was most supportive for their needs. If the mother chooses to receive the diagnosis over the phone, the physician should arrange for a follow-up office visit as soon as possible [Skotko, 2005]. Pregnant women from the Netherlands who were informed by a home-visit from a general practitioner or midwife were satisfied with the way in which they were given the diagnosis [Tymstra et al., 2004].

What Information Should Be Given?

Mothers emphasized that at the time of receiving a definitive prenatal diagnosis, they should be provided with up-to-date information about what is DS, what causes DS, and what are the expectations for a child with DS living today [Helm et al., 1998; Skotko, 2005]. Current information should include descriptions of common or anticipated health conditions seen in infants and young children with DS. However, the fact that the medical and neurodevelopmental outcomes associated with DS cannot be predicted prenatally should be discussed explicitly [Korenromp et al., 2007]. Pregnant women who had received an up-to-date bibliography of DS resources expressed satisfaction with their physicians [Skotko, 2005].

Personal stories that demonstrate “the potential and possibilities for children with DS” should also be included [Skotko, 2005]. Pregnant women who expressed highest satisfaction with their physicians were further offered contact information to other parents who have children with DS [Helm et al., 1998; Skotko, 2005]. By contrast, information that should not be provided includes outdated information, unsolicited personal opinions, or any comments which appear to question parents’ decisions [Helm et al., 1998].

Mothers emphasized that at the time of providing a definitive prenatal diagnosis, physicians should discuss all options available to them regarding the disposition of their pregnancy [Skotko, 2005]. These include continuing the pregnancy, terminating the pregnancy, or placing the baby up for adoption after birth. In a survey of 71 women from the Netherlands who terminated their wanted pregnancies after learning their fetus had DS, 34% of them indicated that the option of continuation was not raised [Korenromp et al., 2007].

Physicians should not assume that the exclusive decision made by women is termination [Skotko, 2005]. They may also need to emphasize that both parents might receive negative comments from people around them no matter what decision they make and that feelings of guilt are common [Korenromp et al., 2007].

How Should the News Be Communicated?

Parents have expressed a desire to receive information in a manner respectful of their feelings and discussed in a nonjudgmental fashion which supports their own personal decisions [Helm et al., 1998; Skotko, 2005]. Sensitive language should also be used. In the largest study to date, most of the mothers requested that physicians not begin the conversation by saying, “I’m sorry,” or “Unfortunately, I have some bad news to share” [Skotko, 2005]. Instead, physicians should use neutral and nondirective language. Outdated and offensive terminology (e.g., “mongolism”) should not be part of the discussion. The most appropriate descriptor is “a fetus with Down syndrome” or “a fetus with Trisomy 21,” if applicable. Mothers further advise against physicians making them feel hurried in their decision-making, sharing unsolicited personal opinions, or trying to change parents’ decisions [Helm et al., 1998; Williams et al., 2002; Skotko, 2005].

DISCUSSION

While the number of research articles on advances in prenatal testing for DS continue to multiply, few are dedicated to understanding how physicians communicate a test result to expectant mothers. Of the studies reviewed here over the past decade, nearly all mothers reported initial feelings of shock, anger, and fear after receiving such a diagnosis [Helm et al., 1998; Skotko, 2005]. Yet, these same mothers indicate that if physicians were to implement a few simple measures, the experience could be much more sensitive to their emotions and needs.

Recommendations

The following recommendations are based on consistent evidence from the articles that were reviewed. These suggestions are meant to serve as helpful guideposts for today’s physicians but should not be considered inclusive of all possible recommendations. Likewise, adherence to these suggestions do not necessarily ensure a satisfactory experience for both the physician and patient. Recommendations are offered for the ideal situations, with the understanding that some measures might need to be adapted to fit the resources available within a particular healthcare community. Nevertheless, the evidence suggests that most parents receiving a prenatal diagnosis of DS would want the following measures implemented:

- Obstetricians should clearly outline the differences between prenatal screening and definitive testing so that parents can understand what the results will mean and make an a priori informed decision on how best to proceed with DS testing. Many women, especially those reluctant to undergo CVS or amniocentesis, regret receiving the results of prenatal screening if they had incorrectly understood them to be definitive tests. The

results of prenatal screening tests should always be conveyed as risk assessments and never as “positive” or “negative” results. The “positive” and “negative” interpretations are based on arbitrary risk cut-offs established by physicians and researchers; pregnant women have asked that they be the ones to determine their own personal risk cut-off value.

- Prior to undergoing CVS or amniocentesis, obstetricians should ask pregnant women if they have already formed a definitive personal decision on how they would proceed with the pregnancy if their fetus were to be identified as having DS. If the pregnant women have already come to a conclusive personal decision, obstetricians should respect those wishes. If they have not, the obstetrician should mention that, dependent on timing, the options include terminating the pregnancy, continuing the pregnancy and raising the baby, or placing the baby up for adoption after birth.
- Once a definitive prenatal result for DS comes back, the person to deliver the news should be the health care professional most knowledgeable about DS who has also received specific training on how to deliver sensitive diagnoses to parents. In some cases, this is the obstetrician; but most often, the obstetrician will need to work jointly with the local health care professional who has the most expertise in DS (such as a geneticist, genetic counselor, developmental-behavioral pediatrician, or neonatologist). A health care professional who can speak knowledgeably about DS *should be available for the first conversation and not simply by referral on subsequent visit.*
- Ideally, these health care professionals should inform parents of the diagnosis during a personal visit. In cases where a personal visit is not feasible or practical, the obstetrician should preemptively identify a time with the mother when the results—whatever they might be—can be discussed by phone. The obstetrician should also mention that if the results indicate that the fetus has DS, he or she might invite a DS expert to participate in the telephone call. By establishing, in advance, a time and setting in which to receive the diagnosis, physicians allow pregnant women to ensure that any desired people or support systems are in place.
- During this discussion the physician needs to answer: What is DS, and what causes the condition? As part of the explanation, physicians should include descriptions—and the probabilities—of common or anticipated health conditions seen in infants and young children with DS <1 year old. Also, included should be the availability and success of medical and surgical treatments for these conditions. (The healthcare guidelines for DS can be accessed through the National Down Syndrome Society, www.ndss.org, and the National Down Syndrome Congress, www.ndscenter.org.) Parents should be counseled that the level of neurodevelopmental function for their fetus with DS cannot be predicted prenatally. While they should be told to anticipate delays in reaching developmental milestones, every child with DS is expected to make developmental progress during the early years at his or her own pace. Early Intervention, including speech, occupational, and physical therapies, is available to help children with DS reach their full potential. This recommendation is consistent with the healthcare guidelines established by the AAP [American Academy of Pediatrics. Committee on Genetics, 2001].

- During the discussion, physicians must also answer: What are realistic expectations for a child with DS living today? Until more epidemiological family research is done on DS, physicians should use representative stories that demonstrate the possibilities available for people with DS today (examples available at www.ndss.org and www.ndscenter.org). Further, physicians should be certain to offer contact information for local support groups and community resources to all expectant parents who have not reached an unequivocal decision on how to proceed with their pregnancy or who have definitively chosen to continue the pregnancy. Physicians should explain that DS-specific support groups are informed primarily by parents who chose to continue their pregnancies and are willing to offer their perspectives on having a son or daughter with DS. Many of these DS support groups can also offer up-to-date and accurate information about DS, helping expectant parents to make informed decisions. If the expectant mother is interested and consents, the physician might even proactively contact the local support group and forward the contact information for the expectant parent(s). Connecting the expectant parent(s) with another parent(s) has been shown to be among the most helpful measures a physician can do during this first conversation. Other parents are able to share real-life experiences that physicians most often cannot. Local DS support groups can be quickly located at www.ndss.org and www.ndscenter.org.
- Physicians should use nondirective language during their counseling. Instead of saying “I’m sorry . . .” or “Unfortunately, have some bad news to share . . .,” physicians should be careful to use sensitive language that does not proscribe value on people with DS. Offensive language (e.g., “mongolism”) should never be used in the discussion.
- At the end of the visit, the physician should offer an up-to-date bibliography of DS resources such as those available from the National Down Syndrome Society (www.ndss.org) or the National Down Syndrome Congress (www.ndscenter.org) for those parents have not reached an unequivocal decision on how to proceed with their pregnancy or who have definitively chosen to continue the pregnancy. A study of 507 ACOG fellows and junior fellows, conducted in 2008, indicated that only 29% of physicians provide educational materials when making a prenatal diagnosis [Driscoll et al., 2009].
- The physician should make arrangements for a follow-up appointment with the parent(s), including any desired meetings with pediatric subspecialists (e.g., geneticists, genetic counselors, or developmental-behavioral pediatricians). If the fetus with DS has a known structural cardiac defect, a consultation with a pediatric cardiologist should be arranged, and the delivery may need to be performed at a hospital where a pediatric cardiac surgical team is available.

Future Research

With the rapid advances in prenatal testing, there is a real potential that nearly all women in the future will have the opportunity in the first trimester to know whether or not their fetus has DS from a definitive, noninvasive test through the detection of fetal DNA or RNA in maternal serum [Lo et al., 2007; Fan et al., 2008; Lo and

Chiu, 2008; Puszyk et al., 2008]. A paucity of literature exists, however, in how physicians will convey these diagnoses, and crucial to this process will be the answers to several pressing questions. While the literature is clear that accurate, up-to-date information about DS should be conveyed, what exactly should be communicated—and to what detail? Further, what knowledge is best conveyed orally, and what information is best relayed in print or alternative media? Research clearly shows that mothers retain with great accuracy the first words that physicians use [Skotko, 2005]; other studies demonstrate that they can recall with nearly 82% accuracy most of the conversation some 20 years later [Carr, 1988]. Focus on the right balance of information should be a priority for further investigation.

The recommendations offered here are predominantly based on research surveying mothers who received a prenatal diagnosis for DS and chose to continue their pregnancies. We could find only one article surveying mothers who chose to terminate their pregnancies after receiving a definitive diagnosis. This research, conducted in the Netherlands, suggests that these mothers’ decisions are based on an understanding that DS was “an abnormality too severe” and a “burden” that was “too heavy” for the child [Korenromp et al., 2007]. Similar research should ask pregnant women in the U.S. who have terminated their pregnancies: How was the experience for you? What understanding did you have of DS, and what information was provided to you from your physicians? Mothers who choose to terminate their pregnancies after receiving a prenatal diagnosis of DS might have different perceptions of their medical providers in comparison to those mothers who continue their pregnancies. Nonetheless, the evidence-based recommendations for improved counseling and better information should benefit all patients who receive a prenatal diagnosis of DS, regardless of the personal decision that they make with the information.

Further, the majority of research has surveyed mothers who are white and from middle- to upper-economic brackets. Also, our review was limited only to those studies published in English. Future research should seek to incorporate parents with more socioeconomic, cultural, and religious diversities from the U.S. and other countries so that support and outreach could target unique needs.

While this review focuses exclusively on the first conversation with expectant parents, equally important are the dynamics of the subsequent conversations. Who should meet with the parents next? When and where should this meeting take place? What information should be introduced and discussed then? Research is noticeably absent in addressing these questions.

Implications

Pregnant women who receive a prenatal diagnosis of DS and continue their pregnancies are able to experience the birthing process in more celebratory ways in comparison to their counterparts who learn about the diagnosis for the first time during the postnatal period [Skotko, 2005]. Receiving the diagnosis in advance seems to allow parents the needed time to reconcile their own emotions and prepare for the child, should they choose to carry the pregnancy to term. As more noninvasive definitive serum testing becomes commercially available to women, a likely hypothesis is

that more women will receive prenatal diagnoses of DS. How they deal with the news and what personal decisions are ultimately made is dependent, to a certain extent, on the accuracy of the information being conveyed.

Most, if not all, of these recommendations from surveyed parents are reasonable and thoughtful. Yet, many mothers continue to report that medical professionals do not yet incorporate these measures. Part of the explanation can likely be attributed to physicians' lack of training [Skotko, 2005; Cleary-Goldman et al., 2006; Driscoll et al., 2009]. Training should become a priority for obstetricians, geneticists, genetic counselors, family medicine physicians, midwives, and other medical professionals associated with the delivery of a prenatal diagnosis of DS. Educational opportunities include lecture series, grand rounds presentations, clinical experiences, and online simulation [Ferguson et al., 2006]. Until such training is put in place, pregnant women will continue to base "informed" decisions on sometimes incomplete and inaccurate information.

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APPENDIX

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