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Testimony for HB1528, “An Act Relative to Down Syndrome Genetic Test Results”
Joint Committee on Public Health
May 10, 2011

My name is Dr. Brian Skotko, and I am a physician and a researcher in the Down Syndrome Program at Children’s Hospital Boston. We have an urgent issue that we can solve together: new and expectant parents are receiving diagnoses of Down syndrome every day, but research has shown that they are receiving little to no information about the condition. When they do, the information is oftentimes inaccurate, incomplete, and sometimes offensive. This also comes as groundbreaking news highlights that a new prenatal test for Down syndrome will be released by biotech companies this fall. This means that nearly all pregnant women will be offered a noninvasive test for Down syndrome during their pregnancies, and the number of identified prenatal diagnoses will dramatically increase.

And, this legislation is very clear: The bill does not discuss, decide, nor influence what choices couples can and do choose with their pregnancies. This bill was instead is considered because couples around the Commonwealth are routinely and consistently saying that they are not receiving the up-to-date information that they need to even begin to make informed decisions.

Let there be no doubt that there is an urgent need. Research has proven this over and over again. Last year, a team of 26 exceptional healthcare professionals joined me in reviewing the evidence from our research journals. We asked: Are new and expectant couples receiving sufficient information at the time of their diagnosis? When, where, and how should this information be conveyed? The team consisted of obstetricians, pediatricians, geneticists, genetic counselors, family practitioners, maternal-fetal medicine practitioners, among others. Our results have been published in the top-tier medical journals called *Pediatrics* and *American Journal of Medical Genetics*, and I have provided copies of this research with my written testimony (attached). We learned that the vast majority of women across the country, including our own state, did not feel that they received sufficient and complete information.

When couples receive a diagnosis of Down syndrome—either before or after birth—the moment is one that is almost uniformly filled with anxiety, fear, and shock. And, prenatally, couples typically receive the results by phone. During those days after receiving a diagnosis, couples go searching for information—any information—that they can find. Unfortunately, not all information on the Internet is credible or even accurate. Couples have said over and over again in the research literature that they are looking to their healthcare providers to provide or direct them to quality information.

Fortunately, we do not need to debate what information should be given. Five of our major medical and advocacy organizations have spent the past two years collaborating and creating a booklet that is credible, balanced, accurate, and informative about the prenatal diagnosis of Down syndrome. The booklet is titled, “Understanding a Prenatal Diagnosis of

Down Syndrome,” and this must-have information was created with input from the American College of Medical Genetics, the American College of Obstetricians and Gynecologist, the National Society of Genetic Counselors, the National Down Syndrome Society, and the National Down Syndrome Congress. The book is available now and ready to be distributed. I, too, have reviewed this booklet and find the content and style to be exceptionally balanced. In addition, the booklet is available in both Spanish and English. The reality is, however, that very few medical professionals are distributing it—due simply to lack of awareness and education about the booklet. This bill will change that and will ensure that all expectant couples are offered this information in the Commonwealth.

The research literature, however, also highlights over and over again another important message from expectant parents: Oftentimes, the most valued information comes from mothers and fathers who already have a child with Down syndrome—couples who can *really* explain what life is like with someone with Down syndrome. Not all new and expectant couples want to connect with an existing parent; however, all new and expectant couples want to have the contact information should they choose to use it. There is an important difference between providing the information and forcing a connection. This bill distinguishes this essential point.

And, we need not search any further on what organizational information that should be provided. The Massachusetts Down Syndrome Congress (MDSC) is a non-profit organization that is, very importantly, not pro-life, nor pro-choice. The organization is decidedly pro-information, and they have established a Parents’ First Call Program, which means that existing parents are available by phone, e-mail, text, Facebook, or Twitter 24 hours a day, 7 days a week. Their parent volunteers are also trained, knowing that their responsibility is to share their personal family stories, without enforcing any political or religious agenda. This non-profit has my full support, and their First Call Program has become a model program for similar organizations around the country.

So, in conclusion, we have an urgent need with an easy solution. New and expectant couples can and do deserve accurate and up-to-date information after receiving a diagnosis of Down syndrome. The credible information already exists; the credible referral organization is already established. This bill—through your leadership—will make sure that these essential connections take place at a crucial time for so many families.

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How to Deliver the News? New Advice for Doctors Diagnosing Prenatal Down Syndrome

BOSTON —September 28th, 2009 New prenatal tests for Down syndrome are soon to be offered to all pregnant women across the United States, yet telling an expectant couple that their child will be born with Down syndrome is a task very few physicians are trained for, claims research published in the *American Journal of Medical Genetics*. The study, which reviewed decades of surveys and interviews, offers several recommendations for how physicians can best deliver the news.

A 29-member research team, led by Dr. Brian Skotko from Children's Hospital Boston, supported by the National Down Syndrome Society and informed by experts from across the field reviewed surveys and research ranging from 1960 to present day to consider how prepared physicians felt they are to deliver a diagnosis. They also studied the opinions of couples who had received the diagnosis to determine the best way of delivering the news.

"Down syndrome (DS) remains the most common chromosomal condition. It occurs in one out of every 733 live births," said Skotko. "Nearly every obstetrician can expect to have a conversation with expectant parents about the realities of life with DS, but very little research has been dedicated to understanding how physicians should communicate the news."

The team found that in a 2004 survey approximately 45% of obstetric fellows rated their training as "barely adequate or nonexistent"; a similar survey four years later found little change as 40% thought their training was "less than adequate." In 2005 a survey of 2,500 medical students showed that 81% believed they were "not getting any clinical training regarding individuals with intellectual disabilities."

To improve this scenario the team set out to answer five critical questions which every physician should consider before delivering a diagnosis: Who is the best person to communicate the news? When is the best time to share the news? Where should the news be delivered? What information should be offered? How should the diagnosis be communicated?

The team found that while many sources are available, from trained counsellors to midwives, expectant couples prefer to receive the news from the health care professional with the most knowledge, the physician.

Also, women who decided to undergo definitive prenatal testing for DS prefer to receive the diagnosis as soon as possible in the company of their husbands or partner, while women who had arranged for the diagnosis to be delivered by a phone call were better prepared for the news than those who received the news from an unarranged call. Women who received the diagnosis through an unscheduled call expressed intense resentment towards their obstetricians and counsellors.

Regarding the amount of information a couple should be given, mothers emphasised that they should be provided with up-to-date information about DS, its causes and the expectations for a child living with DS today. This information should include descriptions of common or anticipated health conditions seen in infants and young children.

On top of this, parents found that they benefited from personal stories that demonstrate the potential and possibilities for children with DS and if possible if possible contact information for other parents who have children with DS should be made available.

Mothers emphasized that at the time of a diagnosis, physicians should discuss all options available to them, including continuing the pregnancy, offering the baby up for adoption after birth, or pursuing termination. In a survey of 71 women from the Netherlands who terminated their pregnancy after a diagnosis of DS, 34% indicated that the option of continuation was not raised.

Finally physicians should be mindful of how they communicate the news. In the largest study most mothers requested that physicians should not begin a conversation by saying "I'm sorry" or "unfortunately I have some bad news," instead they should use neutral and nondirective language.

Unsurprisingly the team also suggests that outdated and offensive language such as "mongolism" should be avoided and instead phrases such as "a fetus with Down syndrome" should be adopted.

Ultimately the research review found that mothers who received the diagnosis prenatally and continued their pregnancy were happier with the birth of their child than those who received the diagnosis after the baby had been born. Receiving the diagnosis in advance seems to allow parents the needed time to overcome the shock and initial grief of the diagnosis and begin preparing and celebrating the upcoming birth of a child.

"Of the studies reviewed nearly all mothers reported feelings of initial shock, anger and fear following the diagnosis," concluded Skotko. "Yet, these same mothers indicated that if physicians were to implement a few simple measures, as research suggests, the experience could be more sensitive to their emotions and needs."

This paper is published in the *American Journal of Medical Genetics: Part A*. To request a copy of this paper or for other media enquiries, please contact Ben Norman on Benorman@wiley.com +44(0) 1243 770 375

A companion paper by Dr Skotko has been published today in the Journal *Pediatrics*. For more information on this paper or to request a copy of this paper please contact Benorman@wiley.com

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9 New Steps Recommended in Delivering a Prenatal Diagnosis of Down syndrome

The 29-member Down Syndrome Diagnosis Study Group recommends the following steps for all physicians making a prenatal diagnosis of Down syndrome

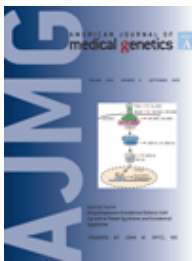
- Obstetricians should clearly outline the differences between prenatal *screening* tests and prenatal *diagnostic* test
- Prior to undergoing prenatal diagnostic tests, obstetricians should ask pregnant women if they have already formed a definitive personal decision on how they would proceed with the pregnancy. If they have not, they should mention all three options, depending on timing: continuing the pregnancy; placing the baby up for adoption after birth, terminating the pregnancy
- Obstetricians should work jointly with a health care professional knowledgeable about Down syndrome (e.g., geneticist) to share the diagnosis.
- This joint presentation should happen ideally in person or at a pre-scheduled time over the phone.
- During the conversation, the health care professionals should answer: What is Down syndrome? What causes the condition? What health care conditions go along with the condition?

- During the discussion, the health care professions should also answer: What are realistic expectations for a child with Down syndrome living today? Offering contact with other parents and support groups is of paramount importance.
- The health care professionals should use nondirective language.
- The health care professionals should offer an up-to-date bibliography of Down syndrome resources.
- The health care professionals should arrange for a follow-up appointment with the parents, including any desired meetings with subspecialists.

About the Author

The corresponding author for this paper is Brian Skotko, M.D., M.P.P. Dr. Skotko is a genetics fellow at Children's Hospital Boston, Massachusetts General Hospital, and Brigham & Women's Hospital. He has focussed his research on children with cognitive and development disabilities. Dr. Skotko is a prolific author and has co-authored the books, *Common Threads: Celebrating Life with Down Syndrome* and *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*. He currently serves on the Board of Directors for the Massachusetts Down Syndrome Congress, the National Down Syndrome Society, the Band of Angels Foundation and the Professional Advisory Council to the National Down Syndrome Congress. He has a 29-year-old sister, Kristin, who has Down syndrome. Dr. Skotko's web page is www.brianskotko.com.

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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 health-care 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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Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News

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KEY WORDS

postnatal diagnosis, Down syndrome, trisomy 21, postnatal care, delivery of healthcare, disclosure

ABBREVIATIONS

ACOG—American College of Obstetrics and Gynecology
DS—Down syndrome

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WHAT'S KNOWN ON THIS SUBJECT: Many parents of children with DS have expressed dissatisfaction with how their medical providers informed them of their child's diagnosis. As the most common chromosomal condition, DS is most often diagnosed postnatally.



WHAT THIS STUDY ADDS: This review provides physicians with an up-to-date, evidence-based structure on who should deliver a postnatal diagnosis of DS, when it should be given, where the discussion should occur, what should be said, and how best to say it.

abstract

CONTEXT: Many parents of children with Down syndrome (DS) have expressed dissatisfaction with how they learned about their child's diagnosis. DS remains the most common chromosomal condition, occurring in 1 of every 733 births, with the majority of children still diagnosed postnatally.

OBJECTIVE: Our goal was to review systematically all available evidence regarding how physicians should approach the conversation in which they explain DS for the first time to new parents.

METHODS: We searched online databases from 1960 to 2008, including Medline and PsychInfo, as well as Web sites maintained by academic organizations (eg, American Academy of Pediatrics) and other nonprofit or private organizations (eg, the National Down Syndrome Society), by using the terms "Down syndrome," "trisomy 21," "mongolism," "prenatal diagnosis," "postnatal care," and "delivery of health care." Articles were selected that answered ≥ 1 research question, 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? and (5) How should the news be communicated? All studies were evaluated for quality according to the method outlined by the US Preventative Services Task Force. Final recommendations were based on the strength of evidence.

RESULTS: Parents prefer to receive the diagnosis together in a joint meeting with their obstetrician and pediatrician. The conversation should take place in a private setting as soon as a physician suspects a diagnosis of DS. Accurate and up-to-date information should be conveyed, including information about local support groups and resources.

CONCLUSION: By implementing a few cost-neutral measures, physicians can deliver a postnatal diagnosis of DS in a manner that will be deemed by new parents as sensitive and appropriate. *Pediatrics* 2009; 124:e751–e758

Although the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics now recommends that all pregnant women, regardless of age, be offered prenatal testing for Down syndrome (DS),^{1–3} studies suggest that >85% of mothers who have children with DS first received the diagnosis postnatally.^{4,5} DS is the most common chromosomal condition, occurring in 1 of every 733 live births with >400 000 persons with DS estimated to be living in the United States.⁶ As such, today's neonatologists, geneticists, family practitioners, hospitalists, and general pediatricians can expect to join obstetricians in being among the first physicians who will share the diagnosis with the new parents.

Delivering and receiving that news is not simple. Even the most seasoned physicians admit that they have little, if any, training on how to discuss a new diagnosis of DS in a sensitive manner.⁷ In a survey of 2500 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.⁸ Mothers and fathers report feeling "shocked," "angry," "devastated," "overwhelmed," "depressed," "stunned," and/or "helpless" when they first learn of the diagnosis.⁴ In addition, parents from many parts of the world such as England, Scotland, Ireland, Spain, Sweden, Australia, and the United States have reported strong dissatisfaction with the way in which the diagnosis was conveyed and the amount of support provided during the immediate postnatal period.⁴

Our primary goal for this report was to review the current evidence on how physicians should best deliver a postnatal diagnosis of DS to new parents.

Specifically, we asked the following questions: Who is the best person to communicate the news? When is the best time to share the news? Where is the best place/setting to deliver the news? What information should be given? How should the news be communicated? We further graded the evidence in an effort to provide today's physicians with evidence-based recommendations on how to approach these sensitive encounters.

METHODS

Sources

This study was part of a larger literature review where we searched online databases including Medline and PsychInfo for all studies published in English from 1960 through February 2008, by using the following 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 further supplemented the primary literature search by searching the Web sites of the following entities: Google Scholars; the American Academy of Pediatrics; the ACOG; the American College of Medical Genetics; the National Newborn Screening and Genetics Resource Center; the National Down Syndrome Society; the National Down Syndrome Congress; March of Dimes; the National Federation of Voluntary Bodies; the International Mosaic Down Syndrome Association; the Down Syndrome Educational Trust; the Canadian Down Syndrome Society; and the United Kingdom Down Syndrome Association. Additional studies were identified by reviewing references of previously screened articles.

Study Selection and Data Extraction

Before the start of this review, we solicited input and advice 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 seemed to answer ≥ 1 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 prenatal period were reported elsewhere.⁹

After independently reading, in full, all of the articles meeting the initial criteria, the primary authors discussed and then eliminated those articles that (1) on full read 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 of <10 persons. No discordant opinion among the authors occurred with this criteria. In total, we identified 19 articles, collectively sampling 3359 parents who received a postnatal diagnosis of DS for their child.^{4,10–27} The studies came from a variety of countries, were performed from 1964 to 2005, and sampled primarily mothers (Table 1). All studies were evaluated for quality by 1996 US Preventative Services Task Force guidelines (levels of evidence are indicated in Table 1).²⁸ Final recommendations for practitioners were based on the strength of evidence, as assessed by both the 3-tiered levels used by the

TABLE 1 Details of Articles Included in the Literature Review, Listed in Chronological Order

Reference No.	Year of Publication	Location of Study	Participants, <i>n</i>	Study Design	Level of Evidence ^a
14	1964	United Kingdom	71 mothers	Retrospective case study with in-person interviews	II-3
10	1969	United Kingdom	95 mothers	Retrospective case study with in-person interviews	II-3
11	1970	United Kingdom	46 mothers	Retrospective case study with in-person interviews	II-3
24	1973	Scotland	31 mothers and 31 fathers	Retrospective case study with in-person interviews	II-3
16	1974	United States	85 mothers and 85 fathers	Retrospective case study with telephone interviews	II-3
21	1976	United States	414 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
13	1977	United Kingdom	30 mothers and 21 fathers	Retrospective case study with in-person interviews	II-3
22	1978	United Kingdom	54 mothers and fathers combined	Retrospective case study with in-person interviews	II-3
23	1980	United States	35 mothers and 3 fathers	Retrospective case study with telephone interviews	II-3
18	1980	Ireland	79 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
19	1983	Ireland	123 mothers	Retrospective case study with mailed questionnaires	II-3
12	1984	United Kingdom	59 mothers and 58 fathers	Prospective nonrandomized control trial between mothers of children with DS who received the diagnosis by a "model service" and those who received customary disclosure; evaluated with in-person interviews	II-1
20	1985	United States	285 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
25	1986	United Kingdom	63 mothers and fathers combined	Retrospective case study with in-person interviews	II-3
26	1994	United Kingdom	56 mothers and fathers	Retrospective case study with in-person interviews	II-3
15	1995	United States	18 mothers and fathers	Retrospective case study with in-person interviews	II-3
17	2002	Sweden	165 mothers and fathers	Retrospective case study with mailed questionnaires	II-3
27	2005	Spain	467 mothers	Retrospective case study with mailed questionnaires	II-3
4	2005	United States	985 mothers	Retrospective case study with mailed questionnaires	II-3

^a Levels of evidence as established by 1996 US Preventative Services Task Force guidelines.²⁸

ACOG^{1,2} (with which level A recommendations are based on "good and consistent scientific evidence," level B recommendations are based on "limited or inconsistent scientific evidence," and level C recommendations are primarily based on "consensus and expert opinion") and the 2-tiered levels used by the Grading of Recommendations, Assessment, Development, and Evaluations (GRADE) system²⁹ (with which a "strong recommendation" means that "most informed patients would choose the recommended management and that clinicians can structure their interactions with patients accordingly" and a "weak recommendation" means that "patients' choices will vary according to their values and preferences, and clinicians must ensure that patients' care is in keeping with their values and preferences").

RESULTS

Who Is the Best Person to Communicate the News?

Mothers first learned that their infants had DS from a variety of people:

"ward sisters"^{11,12,19}; nurses^{4,11,19,21,24}; spouses^{16,19–21,23,24}; lactation specialists⁴; hospital volunteers⁴; genetic counselors⁴; midwives¹²; medical students²¹; medical residents¹⁹; primary care practitioners^{12,16,19–21,23}; pediatricians (including geneticists and neonatologists)^{4,12,16,17,20,21,23,24}; and obstetricians.^{4,12,16,20,21,23} Since 1964 when researchers began tracking maternal reactions, however, mothers have offered consistent advice on from whom among this list they would prefer—and not prefer—to hear.

The person to first deliver the news should be a physician knowledgeable about DS, according to most mothers.^{4,11,13,16,18,19,21,27} In some cases, mothers preferred this clinician to be their obstetrician^{16,21} because a "trust relationship with the obstetrician had been established during the antenatal period."²¹ Other mothers valued a pediatrician or pediatric subspecialist given their expertise and training in childhood conditions.¹⁶ Recommendations from the largest studies, to date,

suggest that a "combined counseling effort of obstetrician and pediatrician might relieve some of the parental anxieties,"²¹ and "all hospitals should have a plan in place so that all relevant physicians know how best to coordinate their messages."⁴

Mothers have been clear from even the earliest studies that they do not want to hear the news first from their husbands or partners.^{4,16,23,24} Doing so, they argue, creates an unfair burden on a person who is experiencing the same initial feelings of shock, grief, and sadness. Nor should the person who delivers the news be a health care professional who is not fully trained to explain DS, such as lactation specialists, hospital volunteers, medical students, medical residents, and most midwives and nurses.^{4,21} Although these persons might not be the first communicators, the largest study on this topic, to date, emphasizes that "it takes a committed team to make the experience of having a child with DS a positive one."⁴

When Is the Best Time to Share the News?

In general, mothers prefer learning about DS as soon as possible even when the diagnosis is not confirmed^{16,24,27}; however, physicians must also use their best judgment and delay informing the mother if she seems ill¹⁰ or is still recovering from the delivery.^{4,10,17} Across time, mothers most often complained about a delay in receiving information, feeling that health care professionals were reluctant to disclose information regarding their child's condition.²⁴ The percentage of mothers who were notified of the diagnosis within 24 hours of their child's birth has been improving: 35% ($n = 31$) in 1973,²⁴ 28% ($n = 406$) in 1976,¹² 56% ($n = 54$) in 1978,²² 37% ($n = 78$) in 1980,¹⁸ 37% ($n = 123$) in 1983,¹⁹ 29% ($n = 59$) in 1984,¹² 68% ($n = 139$) in 1985,²⁰ 63% ($n = 63$) in 1986,²⁵ 63% ($n = 56$) in 1994,²⁶ and 75% ($n = 81$) in 2002.¹⁷

In most instances, a diagnosis of DS is based on distinctive physical features, quite apparent at birth, so parents are notified on the day of delivery or on the following day.^{19,20} Even when a physician has not reached a definitive diagnosis, he should report his or her suspicions to the parents as soon as possible.^{13,20,27} Most mothers recognize that something is different about their infant before ever having a conversation with their physician.^{19,22} By delaying the conversation or waiting for a confirmatory karyotype result, physicians cause unnecessary anxiety in parents.^{19,22,27} Parents further report that by having an early discussion, they can prepare themselves for more intense subsequent discussions, during which more of their questions can be answered.^{13,21}

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

Across time, parents have received the diagnosis in a variety of locations, including private hospital rooms,^{18,19} shared inpatient rooms,^{19,24} surgical

suites,¹⁹ and at home.^{11,19} Overwhelmingly, parents prefer receiving the diagnosis in a private place, where no other medical personnel are present.^{12,15,19} Parents have expressed intense dissatisfaction when they have received the diagnosis in the presence of hospital roommates and visitors.^{4,15,27} Mothers most often desired the company of their husband or partner when hearing the diagnosis, while being secluded from all others.^{12,13,15,16,23,24,27} The percentage of mothers who have received the diagnosis with their partner present has been improving over time: 33% ($n = 36$) in 1970,¹¹ 24% ($n = 170$) in 1974,¹⁶ 20% ($n = 417$) in 1976,²¹ 30% ($n = 30$) in 1977,¹³ 27% ($n = 37$) in 1980,²³ 17% ($n = 78$) in 1980,¹⁸ 47% ($n = 123$) in 1983,¹⁹ 75% ($n = 59$) in 1984,¹² 33% ($n = 139$) in 1985,²⁰ and 79% ($n = 77$) in 2002.¹⁷ Whenever possible, health care professionals should provide parents with a private place to talk with each other without interruption immediately after disclosure.^{4,12,13}

What Information Should Be Given?

Across time, parents have been as consistent about information that they would not like to receive as they have been about the details that should be discussed. They frequently resented information perceived as vague, inaccurate, or outdated.^{10–12,18,22,25} An emphasis on overly pessimistic or offensive terminology (eg, "mongolism") was commonly regarded as both unbalanced and hurtful.^{15–17,20,21,23,24,27} Too much information about possible medical conditions occurring later in life, such as obesity, leukemia, and Alzheimer disease, was also felt by parents to be overwhelming for the first conversation.^{17,23,27}

By contrast, parents generally expressed 3 desires about the information that they would like to receive. First, parents want to have access to

complete and accurate information for the following questions: What is DS? What is its cause? And, what does it mean for a family to have a member with DS, in practical terms?^{213,16,18,26} This should include providing parents with the contact information for local parent support groups and community resources.^{4,16,18,21} Second, parents want the information to be balanced, realistic, and contemporary given the current possibilities for people with DS in today's society. This should include providing the parents with an up-to-date bibliography of resource books on DS.^{4,13,20,22,23,27} Third, the information provided during this initial discussion should be limited to the most immediate or common medical conditions.^{12,15,16,18,21} The percentage of mothers who have felt that they received complete, accurate information has varied over time, with a trend for less satisfaction in more recent years: 60% ($n = 43$) in 1970,¹¹ 61% ($n = 85$) in 1974,¹⁶ 84% ($n = 414$) in 1976,²¹ 55% ($n = 33$) in 1977,¹³ 53% ($n = 79$) in 1980,²³ 65% ($n = 139$) in 1985,²⁰ 11% ($n = 27$) in 1986,²⁵ and 30% ($n = 165$) in 2002.¹⁷

How Should the News Be Communicated?

Again, parents were equally clear about the ways in which the diagnosis should not be communicated as they were about how the news should be conveyed. Parents resented when the information was delivered in a manner perceived to be insensitive, unkind, or unconcerned with the welfare of the mother.^{12,20,22,25,26} They further thought the delivery was unprofessional when news was provided to 1 parent before the other.^{15,25} The use of language conveying pity (eg, "I am sorry to have to tell you this, but . . ."), personal tragedy (eg, "Unfortunately, I have some bad news to share . . ."), or extreme sorrow (eg, "I know this might seem like a devastating loss . . .") was considered un-

necessary and not always reflective of mothers' emotional states.^{4,24} The percentage of mothers who felt that their physicians used respectful, nonjudgmental language varied over time, with no particular trends: 30% ($n = 95$) in 1969,¹⁰ 24% ($n = 79$) in 1980,¹⁸ 74% ($n = 59$) in 1984,¹² 65% ($n = 139$) in 1985,²⁰ 44% ($n = 62$) in 1986,²⁵ and 44% ($n = 65$) in 2002.¹⁷

Research has shown that mothers forever remember the first words that their physicians use.³⁰ They expressed the most satisfaction when their physicians offered congratulations over the fact that they had just had an infant.⁴ Parents further indicated a desire to be told the news with both partners and the infant present, whenever feasible.^{12,31} They also request sufficient time to receive a thorough explanation, with the opportunity to have all of their questions answered. At the end of the initial discussion, a follow-up appointment should be arranged within several weeks.^{11,12,14,18,19,24,26}

CONCLUSIONS

Although delivering a diagnosis of DS for the first time to new parents is not a comfortable situation for most physicians and families, researchers have been offering evidence-based suggestions since 1964 on how to make these encounters more sensitive. In fact, despite their commonly shared feelings of shock, anger, and fear after receiving such a diagnosis, parents can and do rate their physicians positively when some simple measures are followed.^{4,12}

Recommendations

The following recommendations are based on consistent evidence from the articles that were reviewed and are rated according to the criteria established both by the ACOG^{1,2} and the GRADE system.²⁹ 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 does 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 health care community. Nevertheless, the evidence suggests that most parents receiving a postnatal diagnosis of DS would want the following measures implemented:

- Obstetricians and pediatricians (or pediatric subspecialists) should coordinate their messaging and be the persons who first deliver the news to the parents. In ideal circumstances, the parents' obstetrician and pediatrician would meet jointly with the couple to explain DS. If this is not possible because of limited resources or if this would cause a prolonged delay to coordinate efforts, the obstetrician and pediatrician should connect with each other to ensure that a consistent message is conveyed (level A; strong recommendation).
- Physicians should inform parents of their suspicion for DS immediately, even if the diagnosis has not been confirmed by a karyotype result. Physicians can usually recognize DS immediately after the birth of a child, and often, parents can also discern that something is different about their infant before ever speaking with the doctor. Physicians must use their best judgment, however, in determining the precise timing of disclosure. For example, if the mother is ill after the childbirth, the physician might wait until the mother has recovered (level A; strong recommendation).
- Physicians should deliver the diagnosis in a private hospital room, away from other medical personnel, patients, and visitors (including other family members). Whenever possible, physicians should also offer the parents a private place to talk without interruption immediately after they have received the diagnosis (level A; strong recommendation).
- Parents should be informed together. Exceptions to this include when availability of the father or partner would significantly delay the conversation or in circumstances where the mother does not wish the child's father to be present. In the case where a mother is alone and preemptively asks her physician if "anything is wrong with her infant," the physician should ask if she would like to wait for her husband or partner to be present to talk about some observations. If not, the physician should plan to re-explain the diagnosis to the husband or partner when they arrive, if desired by the mother (level A; strong recommendation).
- The infant with DS should be present during the conversation and referred to by name by the physician (level C; weak recommendation).
- Physicians should begin their conversations with positive words, such as congratulating the parents on the birth of their child. They should avoid language conveying pity, personal tragedy, or extreme sorrow; moreover, they should avoid offering unsolicited personal opinions. The first few words that doctors use have been shown to set the tone for the remainder of the conversation. Moreover, mothers remember the first words >20 years after the initial discussion (level A; strong recommendation).
- Physicians should provide parents with accurate information that emphasizes in very practical terms, what DS is, what causes the condi-

tion, and what it means to live with DS in today's society. As part of this explanation, physicians should hand parents an up-to-date bibliography list of DS resources, such as the reference lists available from the National Down Syndrome Society (www.ndss.org) or the National Down Syndrome Congress (www.ndsccenter.org) (level A; strong recommendation).

- For the first conversation, physicians should limit their discussions on possible medical conditions to those that the infant is suspected of having (or has) and those conditions seen in young children with DS under the age of 1 year. Physicians should convey the frequency of developing any particular condition as well as available treatments or therapies, such as speech, occupational, or physical therapy. Discussion about transient myeloproliferative disorder, acute myelogenous leukemia, or acute lymphoblastic leukemia should be saved for subsequent conversations. The health care guidelines for children with DS in the United States^{30–32} are available at www.ndss.org and www.ndsccenter.org (level C; weak recommendation).
- The physician should offer contact information for local support groups and community resources to the new parents. If the family is interested and consents, the physician might even proactively contact the local support group and forward the contact information for the new family. Connecting the new parent with another parent has been shown to be among the most helpful measures a physician can do during the first conversation. Local DS support groups in the United States can be quickly located through the National Down Syndrome Society (www.ndss.org) and the National Down Syndrome Con-

gress (www.ndsccenter.org) (level A; strong recommendation).

- Follow-up appointments should be arranged, as desired by the parents, with the medical professionals who have an expertise in DS (eg, geneticists, genetic counselors, developmental-behavioral pediatricians). A directory of DS specialty clinics within the United States is available at www.ndss.org (level C; weak recommendation).

Future Research

Despite the enormous body of literature that exists on this subject, several crucial questions remain without evidence-based answers. First, how much accurate and up-to-date information should be divulged to parents during the first conversation? And, in how much detail should physicians describe the medical conditions associated with infants with DS? Research clearly shows that mothers retain with great accuracy the first words that physicians use⁴; other studies demonstrate that they can recall with nearly 82% accuracy most of the conversation ~20 years later.³³ Yet, how much is too much? And, what information might be better conveyed through books and handouts distributed after this first conversation?

Although this review focuses exclusively on the first conversation with new 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, and many others, regarding continued outreach to parents.

In addition, our research incorporates only those studies printed in English, and most of the participants were white. Future research should seek to incorporate parents with more socio-

economic and cultural diversities from the United States and other countries so that support and outreach could target unique needs.

Implications

The recommendations offered in this review article do not require many financial resources, if any, to be implemented by physicians. In many ways, the suggestions might even seem obvious to some. Yet, if these suggestions are cost-efficient and commonsensical, then why have thousands of mothers over decades of research indicated that their physicians have not incorporated these measures? Part of the explanation can likely be attributed to physicians' lack of training.⁸ For the recommendations in this review article to be implemented, medical schools, nursing schools, genetic counseling schools, pediatric residency programs, obstetrician/gynecologist residency programs, family medicine residency programs, and associated fellowship programs need to work collaboratively with leaders in the DS community on proper training. Educational opportunities include lecture series, grand rounds presentations, clinical experiences, and online simulation.³⁴ After nearly 50 years of research on how physicians communicate a diagnosis of DS, the time has long come for progress.

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Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News

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