J Genet Counsel
DOI 10.1007/s10897-011-9375-8

PROFESSIONAL ISSUES

Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome: Recommendations of the National Society of Genetic Counselors

Kathryn B. Sheets · Blythe G. Crissman · Cori D. Feist · Susan L. Sell · Lisa R. Johnson · Kelly C. Donahue · Diane Masser-Frye · Gail S. Brookshire · Amanda M. Carre · Danielle LaGrave · Campbell K. Brasington

Received: 8 April 2011 / Accepted: 12 May 2011

Abstract Down syndrome is one of the most common conditions encountered in the genetics clinic. Due to improvements in healthcare, educational opportunities, and community inclusion over the past 30 years, the life expectancy and quality of life for individuals with Down syndrome have significantly improved. As prenatal screening and diagnostic techniques have become more enhanced and widely available, genetic counselors can expect to frequently provide information and support following a new diagnosis of Down syndrome. This guideline was written for genetic counselors and other healthcare providers regarding the communication of a diagnosis of Down syndrome to ensure that families are consistently given up-to-date and balanced information about the condition, delivered in a supportive and respectful manner.

K. B. Sheets (✉) · B. G. Crissman
Division of Medical Genetics, Duke University Medical Center,
595 LaSalle Street, GSRB1, Box 103857, Durham, NC 27710,
USA
e-mail: Katie.Sheets@duke.edu

C. D. Feist
Division of Maternal Fetal Medicine,
Oregon Health and Science University,
Portland, OR, USA

S. L. Sell
Department of Genetics, Penn State Hershey Children's Hospital,
Hershey, PA, USA

L. R. Johnson
Center for Maternal-Fetal Medicine,
Dayton, OH, USA

K. C. Donahue
Division of Maternal Fetal Medicine,
West Penn Allegheny Health System,
Pittsburgh, PA, USA

D. Masser-Frye
Department of Genetics and Dysmorphology,
Rady Children's Hospital,
San Diego, CA, USA

G. S. Brookshire
Department of Genetics and Metabolism,
Children's Medical Center,
Dallas, TX, USA

A. M. Carre
Department of Obstetrics and Gynecology,
Drexel University College of Medicine,
Philadelphia, PA, USA

D. LaGrave
Genetics Division, ARUP Laboratories,
Salt Lake City, UT, USA

C. K. Brasington
Department of Clinical Genetics, Carolinas Medical Center,
Charlotte, NC, USA

Published online: 27 May 2011

Keywords Down syndrome · Trisomy 21 · Genetic counseling · National Society of Genetic Counselors · Practice guidelines

PURPOSE

Down syndrome is one of the most common conditions a genetic counselor is likely to encounter in the genetics clinic. Given the improved outlook for individuals with Down syndrome, it is crucial that genetic counselors and other healthcare providers are aware of the key elements in communicating this diagnosis to ensure that families receive up-to-date and balanced information, delivered in a supportive and respectful manner. This guideline serves to 1) summarize the various etiologies of Down syndrome and the associated recurrence risks; 2) review the key components of disclosing a diagnosis of Down syndrome in both the prenatal and the postnatal settings; and 3) provide references for both professional and patient resources.

DISCLAIMER

The practice guidelines of the National Society of Genetic Counselors (NSGC) are developed by members of the NSGC to assist genetic counselors and other health care providers in making decisions about appropriate management of genetic concerns; including access to and/or delivery of services. Each practice guideline focuses on a clinical or practice-based issue, and is the result of a review and analysis of current professional literature believed to be reliable. As such, information and recommendations within the NSGC practice guidelines reflect the current scientific and clinical knowledge at the time of publication, are only current as of their publication date, and are subject to change without notice as advances emerge.

In addition, variations in practice, which take into account the needs of the individual patient and the resources and limitations unique to the institution or type of practice, may warrant approaches, treatments and/or procedures that differ from the recommendations outlined in this guideline. Therefore, these recommendations should not be construed as dictating an exclusive course of management, nor does the use of such recommendations guarantee a particular outcome. Genetic counseling practice guidelines are never intended to displace a healthcare provider's best medical judgment based on the clinical circumstances of a particular patient or patient population. Practice guidelines are published by NSGC for educational and informational purposes only, and NSGC does not "approve" or "endorse" any specific methods, practices, or sources of information.

BACKGROUND

Clinical Description and Management

Dr. John Langdon Down provided the first clinical description of Down syndrome in 1866. Down syndrome is characterized by intellectual disability, short stature, distinctive facial features and various congenital anomalies (Jones 2006). While intellectual disability is universal, the degree is variable, most often within the mild-to-moderate range (IQ 35–70), and only rarely in the severe range (IQ <35) (American Academy of Pediatrics Committee on Genetics 2001). There are also a number of co-morbidities that occur more frequently in individuals with Down syndrome, including cardiac and digestive issues, thyroid conditions, childhood cancer, and hearing impairment (Cohen 1999; Leshin 2002; Patterson 2002)).

Healthcare guidelines for individuals with Down syndrome were first developed in 1981 and later revised by the Down Syndrome Medical Interest Group (Cohen 1999). The American Academy of Pediatrics' Committee on Genetics published guidelines for pediatricians in 2001 and a statement of reaffirmation of these guidelines was published in 2007 (American Academy of Pediatrics Committee on Genetics 2001, 2007). As healthcare has improved for individuals with Down syndrome, the average lifespan has increased by more than 30 years, from an average of 25 years of age in 1983 to almost 60 years of age now. (Glasson et al. 2002).

Because individuals with Down syndrome often experience delays in reaching various developmental milestones, early intervention with speech therapy, occupational therapy, and physical therapy is recommended as it maximizes long-term outcomes (Davidson 2008; Rihtman et al. 2010). After age three, children with Down syndrome have individualized education plans tailored to their needs, which typically involve a special education classroom setting. Inclusion is encouraged whenever possible as there is evidence of progress in many domains, including speech and language, literacy, socialization, daily living skills and behavior (Buckley et al. 2006).

There is a lack of published data regarding the long-term natural history for adults with Down syndrome. There is a wide spectrum of ability observed in individuals with Down syndrome, ranging from nonverbal individuals who require fulltime supervision to high-functioning individuals who live semi-independently. It is anticipated that most individuals with Down syndrome will require some supervision as adults, particularly with finances and emergency situations.

Higher-functioning adults with Down syndrome can successfully participate in a wide range of activities in social, physical, educational, and vocational contexts (Brown et al. 2001). At the highest end of the spectrum,

some young adults are able to live outside of the primary household, obtain driver's licenses, get married, and maintain gainful employment. Individuals with milder intellectual disability may go on to attend post-secondary schooling. Today, approximately 250 post-secondary educational programs are available to students with intellectual disabilities (Hart et al. 2006).

Prevalence of Down Syndrome

Down syndrome is the most common chromosomal trisomy in newborns as well as the most common genetic cause of intellectual disability. A recent study by Parker et al. (2010) estimates 6,037 of the more than 4 million live births in the United States each year will have Down syndrome, representing a national prevalence of 1 in 691 births. While 61% of live births with Down syndrome are conceived by women under the age of 35 (Egan et al. 2011) with an average maternal age of 31.5 (Hassold and Sherman 2000), advanced maternal age is the most significant risk factor for Down syndrome. As women age, the cell cycle proteins involved in meiotic segregation in the oocytes begin to deteriorate, leading to an increasing chance of a nondisjunction error during the formation of the ovum (Snijders et al. 1999).

Genetics of Down Syndrome and Recurrence Risks

The genetic basis of Down syndrome was first described by Jerome Lejeune in 1959 (Lejeune et al. 1959). Down syndrome occurs as the result of a partial or complete third copy of chromosome 21. Table 1 summarizes the various etiologies of Down syndrome and the associated recurrence risks.

For second-degree or more distant relatives of an individual with nondisjunction trisomy 21, there is no increased chance of having a child with trisomy 21. However, it is not possible to determine whether an individual has Down syndrome as a result of nondisjunction trisomy 21, an unbalanced chromosome translocation, or mosacism based on clinical features alone. Accurate recurrence risk estimates depend upon the verification of the individual's karyotype. If the chromosome status of the person with Down syndrome is

Table 1 Genetic causes of Down syndrome and associated recurrence risks

	Nondisjunction Down syndrome	Translocation Down syndrome	Mosaic Down syndrome
Frequency (% of cases)	95%	3–4%	1–2%
Cytogenetic findings	An extra copy of chromosome 21 (free trisomy) as the result of meiotic nondisjunction (leading to 47 chromosomes)	An unbalanced structural chromosome rearrangement involving chromosome 21	A mixture of cells that contain 46 chromosomes and cells that contain an extra copy of chromosome 21 (a total of 47 chromosomes)
Etiology	Maternal nondisjunction is causative in 90% of cases, and paternal nondisjunction is in the remaining 10% of cases.	Robertsonian translocations between 21q and another acrocentric chromosome (13, 14, 15, 21 or 22) account for most familial cases. Approximately 1/3 of these cases are due to the inheritance of an unbalanced chromosomal segment from a balanced translocation carrier parent.	It may occur as the result of an early "trisomy rescue" or early somatic nondisjunction error. It is difficult to predict phenotype due to variable percentage of mosaicism in different tissues.
Recurrence risk	For trisomy 21: Maternal age <35 at previous trisomy 21, the revised risk is the age-related risk times 3.5. Maternal age ≥35 at previous trisomy 21, the revised risk is the age-related risk times 1.7. For any trisomy: Maternal age <35 at previous trisomy 21, the revised risk is the age-related risk times 1.3. Maternal age ≥35 at previous trisomy 21, the revised risk is the age-related risk times 1.5.	If neither parent carries a balanced translocation, the Down syndrome recurrence risk is low, probably similar to that of nondisjunction trisomy 21. Recurrence risk for translocation carriers is dependent on the type of translocation and the sex of the carrier parent. See Table 2.	For a couple whose child has mosaic Down syndrome, the recurrence risk is similar to that of nondisjunction trisomy 21, although this may be an overestimate for some families.

Sources: De Souza et al. 2009; Gardner and Sutherland 2004; Harper 2004; Jones 2006; Warburton et al. 2004

unknown or unavailable, it is appropriate to offer karyotyping for the at-risk relative, since individuals with Robertsonian translocations involving chromosome 21 have an increased chance of having a child with Down syndrome (see Table 2).

The theoretical chance for an individual with Down syndrome to have a child with Down syndrome is 50%, and 66% when both partners have Down syndrome. However, empiric risks are difficult to estimate, as reproduction rates are low. Males with Down syndrome are typically infertile, although there have been reports of men with Down syndrome who have fathered pregnancies (Pradhan et al. 2006). Females with Down syndrome are typically fertile and empiric data indicate a 30–50% chance of having a child with Down syndrome (Gardner and Sutherland 2004; Harper 2004). For individuals with mosaic Down syndrome, the theoretical recurrence risk is as high as 50%, but is dependent upon the proportion of trisomic gonadal cells (Gardner and Sutherland 2004) and whether the other partner has Down syndrome as well.

Diagnostic Testing Options

Prenatal Diagnosis

Although there are a number of prenatal screening options available, the prenatal diagnosis of Down syndrome continues to rely upon invasive testing by amniocentesis or chorionic villus sampling to evaluate the fetal karyotype. Interphase fluorescence in situ hybridization (FISH) can also be performed for a more rapid turnaround time to detect aneuploidy of chromosomes 13, 18, 21, X and Y. A diagnosis of Down syndrome by interphase FISH should always be confirmed by traditional karyotyping (American College of Medical Genetics 2010).

Table 2 Chromosome 21 translocations and associated recurrence risks

Type of translocation	Risk for unbalanced translocation at time of amniocentesis	Other concerns
rob(13q21q)	10–17% if mother is carrier <0.5% father is carrier	risk for translocation trisomy 13
rob(14q21q)	15% if mother is carrier 1.4% if father is carrier	risk for uniparental disomy (UPD) 14
rob(15q21q)	0–11% if mother is carrier <0.5% if father is carrier	risk for UPD 15
rob(21q21q)	100% if either parent is carrier	____
rob(21q22q)	13% if mother is carrier 1.4% if father is carrier	____

Source: Gardner and Sutherland 2004

Postnatal Diagnosis

While the postnatal diagnosis of Down syndrome can be made based on clinical features, confirmatory genetic testing via karyotyping is necessary for accurate recurrence risk estimates. This testing can be performed during the newborn period or at a subsequent follow-up visit. In the majority of cases, a karyotype of the peripheral blood is sufficient to confirm the diagnosis. FISH analysis is an option for rapid turnaround time, but should be confirmed by traditional karyotyping (American College of Medical Genetics 2010). If there is concern about the possibility of mosaic Down syndrome, an additional tissue sample, such as a skin biopsy, may be helpful in confirming the diagnosis.

GENETIC COUNSELING ABOUT DOWN SYNDROME

Genetic counseling is a communication process that incorporates education about genetic conditions with counseling to promote autonomy and adaptation to the diagnosis. Non-directiveness is a central tenet, requiring the counselor to maintain a neutral stance in order to support and respect the patient's personal values and decisions. Facilitating informed decisions requires balance in the information provided (Williams et al. 2002). Genetic counselors should balance the negative aspects of Down syndrome, such as birth defects, medical complications, and developmental delay, with positive aspects like available treatments, therapies, and the ability for people with Down syndrome and their families to enjoy a high quality of life (Bryant et al. 2001).

Delivering a Diagnosis of Down Syndrome

Many articles have published recommendations for informing parents of a new diagnosis of Down syndrome. Most studies are surveys of women who are asked to reflect on the way their healthcare providers delivered the diagnosis of Down syndrome. These recommendations provide examples of the ideal manner in which a new diagnosis of Down syndrome should be delivered. A summary of these recommendations is presented in Table 3.

Providing Information about Down Syndrome

The information provided about Down syndrome must be tailored to a family's knowledge base and emotional needs. Assessing the family's prior knowledge about Down syndrome and previous experiences with Down syndrome should be part of the conversation that continues from the initial counseling session as it may provide valuable insight and help guide the discussion. Healthcare providers should

Table 3 Recommendations for delivering a diagnosis of Down syndrome

- Tell the parents about the diagnosis as soon as possible, even if the diagnosis is suspected but not yet confirmed. If the diagnosis has not been confirmed by karyotype, explain what physical features or medical concerns are suggestive of the diagnosis.
- Ideally, the diagnosis should be delivered in person, by a healthcare professional with sufficient knowledge of the condition. Healthcare providers should coordinate the message to ensure consistency in the information provided to the family.
- Whenever possible, meet with both parents together, or arrange a telephone call at a time when both partners will be present. If only the mother is available, ask to make arrangements to speak with her partner at a later time. If an initial face-to-face visit is not possible, the couple should be offered an office visit as soon as possible. The plan for discussing results should be agreed upon between patient and counselor during the pretest counseling session.
- The family should be informed of the diagnosis in their preferred language. If possible, a professional medical interpreter should be present at the time of disclosure.
- Discuss the diagnosis in a private, comfortable setting, free from interruptions. Allow time for questions and make plans for a follow-up conversation.
- Parents should be provided with accurate and up-to-date information. Information should be given with a balanced perspective, including both positive aspects and challenges related to Down syndrome.
- Provide the information in a sensitive and caring, yet confident and straightforward manner, using understandable language that is clear and concise.
- Use neutral language and avoid using value judgments when starting the conversation, such as "I'm sorry" or "Unfortunately, I have bad news".
- Use sensitive language and avoid outdated or offensive terminology. In the newborn setting, the baby should be present, and should be referred to by name. Use person-centric language, emphasizing that this is a baby who has Down syndrome, rather than a "Downs baby" or a "Down syndrome child."
- Allow time for silence and time for tears. Do not feel that you need to talk to "fill the silence". Offer the family time alone.
- Assess the emotional reactions of the parents, and validate these feelings. Use active listening and empathic responses to support the parents.
- Informational resources should be provided, including contact information for local and national support groups, up-to-date printed information or fact sheets, and books. The opportunity to meet with families who are raising a child with Down syndrome, those who have chosen to create an adoption plan, and/or those who have terminated a pregnancy should be offered. When appropriate, referrals to other specialists may also be helpful (e.g., medical geneticists, genetic counselors, cardiologists, neonatologists, etc.).

Sources: Cooley 1993; Dent and Carey 2006; Helm et al. 1998; Krahn et al. 1993; Powell 1991; Skotko 2005a, b, c; Skotko and Bedia 2005; Skotko et al. 2009; Skotko et al. 2009

coordinate the message to ensure consistency in the information provided to the family.

The Prenatally and Postnatally Diagnosed Conditions Awareness Act of 2008 requires the availability of "up-to-date, evidence-based, written information concerning the range of outcomes for individuals living with the diagnosed condition…" as well as "contact information regarding support services…support groups, and other education and support programs…".

The amount of information provided at the initial consultation will depend on the parents' informational and emotional needs. Communicating a new diagnosis is not a discrete event, but a continuous one, requiring further contact and information provided at a later time (Cooley 1993; Pueschel and Murphy 1976). Therefore, genetic counselors and other members of the healthcare team should strive to address the family's immediate concerns and facilitate transition from the emotional first days post-diagnosis by providing information, access to resources, and caring support. Information about pregnancy termination, adoption or foster care should be provided to families who feel they cannot raise a child with special needs.

Table 4 summarizes the information that is essential to discuss with parents receiving either a prenatal diagnosis for their unborn child or a postnatal diagnosis for their newborn child. Parents and genetic counselors rated 100 clinical features, prognostications, and informational resources related to the condition to determine a bare minimum of informational content for the initial conversation (Sheets et al. 2011). Diagnostic conversations should not be limited to this list of features.

Additional Counseling Issues in the Prenatal Setting

Pregnant women of all ages should be offered prenatal screening and diagnostic testing for Down syndrome (American College of Obstetricians and Gynecologists Committee on Practice Bulletins No. 77 and No. 88, 2007a, b). The primary goal of genetic counseling in the prenatal setting is to uphold patient autonomy regarding reproductive choices by providing personalized genetic information, exploring what the information means to the patient, explaining all options, and preparing the patient for the outcomes of their decision (Biesecker 2001). Couples require balanced, accurate, and consistent information

Table 4 Essential information for the initial discussion of a diagnosis of Down syndrome

- Down syndrome (DS) is caused by extra genetic material from chromosome 21. DS may be suspected based on physical findings, but the diagnosis is confirmed by chromosome analysis.
- Individuals with DS have a variable range of intellectual disability from mild to moderate.
- Babies with DS have delays in achieving developmental milestones and benefit from early intervention including physical, occupational and speech therapy.
- 80% of babies with DS will have hypotonia.
- 50% of babies with DS have one or more congenital abnormality: 40–60% of babies with DS have a heart defect and 12% have a gastrointestinal defect that may require surgery. Assistance with referrals to specialists is appropriate for identified complications.
- Children with DS are more like other children than they are different.
- Raising a child with DS may involve more time commitment than typical children.
- Individuals with DS can participate in community sports, activities, and leagues.
- Individuals with DS can learn in a special education class or may be included in regular classes, and most can complete high school.
- Individuals with DS can be employed competitively or in a workshop setting.
- Individuals with DS can live independently or in a group home.
- Individuals with DS have friends and intimate relationships.
- Life expectancy extends into the 50s or 60s.
- Information on local support groups, advocacy organizations, early intervention centers, printed material, fact sheets, books, specialist referral(s) as needed, and the option to contact a family raising a child with DS should be offered.
- A personalized recurrence risk for future pregnancies should be offered.

Source: Sheets et al. 2011

about alternatives and anticipated consequences from genetic counselors, before and after diagnostic testing, in order to facilitate informed decision-making about their pregnancies (Hodgson and Spriggs 2005; National Society of Genetic Counselors 1992). Information should be nondirective and sufficient in content, provided in an unbiased, sensitive, and nonjudgmental manner.

The initial discussion should include not only common health concerns, but also treatments and therapies that are available. Any known medical issues that would require immediate attention should be addressed, as well as anticipated pregnancy outcomes, such as miscarriage and stillbirth. The overall fetal loss rate for Down syndrome between a diagnosis by chorionic villus sampling and term

Table 5 Discussion of options for pregnancy management after a prenatal diagnosis

Continuing the pregnancy and raising the child	Continuing the pregnancy and creating an adoption plan	Pregnancy termination
Discuss anticipated pregnancy outcomes including an increased risk for miscarriage or stillbirth, and general post-delivery statistics.	Discuss anticipated pregnancy outcomes and options for high-risk obstetrical care.	Explain the various termination procedures, related obstetric issues, and availability based on gestational age, as well as local and national services.
Discuss options for obstetrical care with a high-risk provider. Assist with referrals to other specialists based on ultrasound findings or the needs of the family.	Discuss various private and public adoption agencies that place children regardless of medical diagnosis as well as those which specialize in the adoption of children with Down syndrome.	Discuss options for memory making as appropriate, which can include taking time to hold the baby after delivery, taking photographs, making hand and footprints, etc.
Discuss delivery options including a tertiary care center, community hospital or home birth, as well as the availability of NICU or pediatric specialty team.	Mention that there is a waiting list for people interested in adopting children with Down syndrome. Refer to adoption resources in Table 6.	Address parental coping skills. Assess the need for additional counseling or support services. Discuss and assist with appropriate referrals as needed.
Parents may wish to begin their search for a primary care provider experienced in the care of children with Down syndrome.	Address parental coping skills. Assess the need for additional counseling or support services. Discuss and assist with appropriate referrals as needed.	A follow up telephone call and/or letter from the genetic counselor as indicated.
Address parental coping skills. Assess the need for additional counseling or support services.	A follow up telephone call and/or letter from the genetic counselor as indicated.	
A follow up telephone call and/or letter from the genetic counselor as indicated.		

is approximately 32%, and 25% between amniocentesis and term (Savva et al. 2006).

Each baby with Down syndrome is different and physical and cognitive development cannot be predicted prenatally. Provide a range of possible outcomes to illustrate what life is like for individuals with Down syndrome and their families (Sheets et al. 2011). Specifically, discuss how having a child with Down syndrome might affect the family as a unit, the parents' relationship as a couple, and other present or future children. Families should be directed to local and national support groups for Down syndrome. Offer the opportunity to contact other families raising a child with Down syndrome, those who have chosen to create an adoption plan or ended a pregnancy with Down syndrome who are willing to share their experiences with the patient.

After evaluating the patient or couple's understanding of the diagnosis, let their feelings about having a child with Down syndrome dictate the conversation that follows regarding options for pregnancy management. In a neutral, nondirective, and nonjudgmental manner, discuss all available reproductive options, which include continuing the pregnancy and either raising the child or creating an adoption plan for the child, or terminating the

Table 6 Down syndrome resources

Organizations		
Christian Homes and Special Kids	1-800-266-9837	www.chask.org
International Mosaic Down Syndrome Association	1-888-MDS-LINK	www.IMDSA.org
National Down Syndrome Adoption Network	(513) 213-9615 [a](513) 490-2834	www.ndsan.org
National Down Syndrome Congress	1-800-232-6372	www.NDSCCenter.org [a]www.NDSCcenter.org/Espanol
National Down Syndrome Society	1-800-221-4602 [a]www.esp.NDSS.org	www.NDSS.org
Reece's Rainbow Down Syndrome Adoption Ministry		www.reecesrainbow.org
Sibshops		www.SiblingSupport.org
Online Resources		
A Heartbreaking Choice		www.Aheartbreakingchoice.com
Brighter Tomorrows—family edition		www.BrighterTomorrows.org
Brighter Tomorrows—professional edition		www.Brighter-Tomorrows.org
Diagnosis to Delivery: A Pregnant Mother's Guide to Down syndrome		www.downsyndromepregnancy.org
Light at the End of the Tunnel		www.ndsccenter.org/resources/light.php
Understanding a Down Syndrome Diagnosis		www.lettercase.org
Publications		
A Mother's Dilemma, by Wendy Lyon, Molly A. Minnick. Pineapple Press, 1993.		
A Time to Decide, a Time to Heal: For Parents Making Difficult Decisions About Babies They Love, by Molly A. Minnick, Kathleen J. Delp, edited by Mary C. Ciotti. Pineapple Press, 1992.		
American Academy of Pediatrics Committee on Genetics: Health supervision for children with Down syndrome. *Pediatrics,* 2001.		
Babies with Down Syndrome: A New Parent's Guide, edited by Susan J Skallerup. Woodbine House, 2008.		
[a]Bebés con síndrome de Down: Nueva guía para padres, compilado por Susan J Skallerup. Woodbine House, 2008.		
Choosing Naia: A Family's Journey, by Mitchell Zuckoff. Beacon Press, 2002.		
Difficult Decisions: For Families Whose Unborn Baby Has a Serious Problem, by Patricia Fertel. Centering Corporation, 2004.		
Down Syndrome: Visions for the 21st Century, edited by William Cohen, Lynn Nadel, Myra Madnick. Wiley-Liss, Inc., 2002.		
Expecting Adam: A True Story of Birth, Rebirth and Everyday Magic, by Martha Beck. Random House, 1999.		
Gifts: Mothers Reflect on How Children with Down Syndrome Enrich Their Lives, edited by Kathryn Lynard Soper. Segullah Group, Inc., 2008.		
Our Heartbreaking Choices: 46 Women Share Their Stories of Interrupting a Much-Wanted Pregnancy, by Christie Brooks. IUniverse, 2008.		
Precious Lives, Painful Choices: Prenatal Decision-Making Guide, by Sherokee Ilse. Wintergreen Press, 1993.		
Road Map to Holland: How I Found My Way Through My Son's First 2 years With Down Syndrome, by Jennifer Graf Groneberg. Penguin Books Ltd., 2008.		
Shattered Dreams—Lonely Choices: Birthparents of Babies with Disabilities Talk About Adoption, by Joanne Finnegan, 1993.		
What Parents Wish They'd Known: Reflections on Parenting a Child with Down Syndrome, by Kathryn Lynard Soper, 2008.		

[a] En Español

pregnancy (Helm et al. 1998; Hodgson and Spriggs 2005; Prenatally and Postnatally Diagnosed Conditions Awareness Act of 2008). Referrals to clergy, spiritual leaders, religious groups, and trusted community members may be helpful, especially if cross-cultural issues exist. Regardless of their decision, offer the parents unbiased support and direct them to appropriate resources. Once a decision has been articulated, consider documenting the couple's decision in the medical record to avoid subsequently revisiting the conversation about options for pregnancy management. See Table 5 below for additional discussion points.

Additional Counseling Issues in the Postnatal Setting

The primary goal of genetic counseling after the birth of a child with Down syndrome is to facilitate understanding and acceptance as well as to promote the parents' perceived personal control by explaining the scientific cause and exploring the parents' personal beliefs (Biesecker 2001). New parents often experience a variety of emotions following an unexpected diagnosis of Down syndrome. In the first encounter, families may not need to know every medical detail and may be unable to digest much of the information provided. As in the prenatal setting, genetic counselors and other members of the healthcare team should remain cognizant of the parents' educational and psychological needs and tailor the discussion accordingly.

Congratulations are in order upon the successful delivery of a new baby; however, the demeanor of the medical professional should mirror the parents' current state of mind. Give the parents' permission to grieve the loss of the child the family expected while reassuring them with the hope that they can be wonderful parents to this baby. Be empathic and address potential guilt issues. Compliment the parents on their baby and refer to the baby by name.

Genetic counselors should help families focus on what their child *can* do (Brasington 2007). Discuss the fact that each baby with Down syndrome is unique and should demonstrate characteristics inherited from both "mom" and "dad", but also, that people with Down syndrome are more like their typically developing peers than they are different. Medical complications do not usually dominate medical care, and that barring major medical issues (i.e., heart or gastrointestinal defects), caring for an infant with Down syndrome is not much different than caring for any other baby. Babies with Down syndrome need love and care like any other baby. A balanced perspective, with hope and encouragement, and discussion of positive aspects can promote parents' adaptation to the diagnosis (Cooley 1993; Pueschel and Murphy 1975; Skotko 2005a, b, c).

Ensure awareness of anticipated medical problems and recommended therapies. Focus on the baby you are seeing and any known medical conditions that require immediate attention (i.e., hypotonia, feeding difficulties, heart defect, and referral for early intervention). Other co-morbidities that manifest beyond the pediatric period may be discussed at a later time unless parents are interested in discussing these health concerns immediately. For ongoing surveillance, discuss availability of a medical genetics or Down syndrome specialty clinic.

Families are often interested in how this child will affect the family unit, the parents' relationship as a couple, and other children in the family. While some families may struggle initially, others are able to adapt and can even thrive. Direct patients to local and national support groups, as well as other families raising a child with Down syndrome who are willing to share their experiences with the family. Assist with additional referrals as needed, including clergy, spiritual leaders, religious groups, and trusted community members if cross-cultural issues exist. Information regarding adoption or foster care should be provided if parents wish to explore these options.

SUMMARY

These recommendations for communicating a diagnosis of Down syndrome provide an overview of the current literature and practical recommendations for delivering the diagnosis in a sensitive and supportive manner as well as providing accurate, up-to-date information. Disclosure is an ongoing rather than a discrete event, requiring thorough consideration of medical, psychosocial, and family circumstances. Routinely, genetic counselors are responsible for delivering a diagnosis of Down syndrome to new or expectant parents, and must serve as an educational and emotional resource. Genetic counselors have the expertise to provide accurate and relevant medical information, empathy, anticipatory guidance, and support materials as well as assist with appropriate referrals for their patients. Offering the opportunity to contact families who have experienced a similar situation can be invaluable. Informational resources for families and professionals are provided in Table 6.

REFERENCES

American Academy of Pediatrics Committee on Genetics. (2001). Health supervision for children with Down syndrome. *Pediatrics, 107*(2), 442–449.

American Academy of Pediatrics (AAP) Committee on Genetics. (2007). AAP publications reaffirmed and retired. *Pediatrics, 120* (3), 683–684.

American College of Medical Genetics (2010). Standards and guidelines for clinical genetics laboratories: Section E, Clinical Cytogenetics (E11.1). Retrieved from http://www.acmg.net/StaticContent/SGs/Section_E_2010.pdf

American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins. (2007a). ACOG practice bulletin no. 77. Screening for fetal chromosomal abnormalities. *Obstetrics and Gynecology, 109*(1), 217–227.

American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins. (2007b). ACOG practice bulletin no. 88. Invasive prenatal testing for aneuploidy. *Obstetrics and Gynecology, 110*(6), 1459–1467.

Biesecker, B. B. (2001). Goals of genetic counseling. *Clinical Genetics, 60*(5), 323–330.

Brasington, C. K. (2007). What I wish I knew then…Reflections from personal experiences in counseling about Down syndrome. *Journal of Genetic Counseling, 16*(6), 731–734.

Brown, R., Taylor, J., & Matthews, B. (2001). Quality of life—ageing and Down syndrome. *Down Syndrome Research and Practice, 6*(3), 111–116.

Bryant, L. D., Murray, J., Green, J. M., Hewison, J., Sehmi, I., & Ellis, A. (2001). Descriptive information about Down syndrome: A content analysis of serum screening leaflets. *Prenatal Diagnosis, 21*, 1057–1063.

Buckley, S., Bird, G., Sacks, B., & Archer, T. (2006). A comparison of mainstream and special education for teenagers with Down's syndrome: Implications for parents and teachers. *Down Syndrome Research and Practice, 9*(3), 54–67.

Cohen, W. I. (1999). Health care guidelines for individuals with Down syndrome: 1999 revision. *Down Syndrome Quarterly, 4*(3), 1–15.

Cooley, W. C. (1993). Supporting the family of the newborn with Down syndrome. *Comprehensive Therapy, 19*(3), 111–115.

Davidson, M. A. (2008). Primary care for children and adolescents with Down syndrome. *Pediatric Clinics of North America, 55*(5), 1099–1111.

De Souza, E., Halliday, J., Chan, A., Bower, C., & Morris, J. K. (2009). Recurrence risks for trisomies 13, 18, and 21. *American Journal of Medical Genetics. Part A, 149A*(12), 2716–2722.

Dent, K. M., & Carey, J. C. (2006). Breaking difficult news in a newborn setting: Down syndrome as a paradigm. *American Journal of Medical Genetics. Part C: Seminars in Medical Genetics, 142*(3), 173–179.

Egan, J. F., Smith, K., Timms, D., Bolnick, J. M., Campbell, W. A., & Benn, P. A. (2011). Demographic differences in Down syndrome livebirths in the US from 1989 to 2006. *Prenatal Diagnosis*. doi:10.1002/pd.2702.

Gardner, R. J. M., & Sutherland, G. R. (2004). *Chromosome abnormalities and genetic counseling* (3rd ed.). New York: Oxford University Press, Inc.

Glasson, E. J., Sullivan, S. G., Hussain, R., Petterson, B. A., Montgomery, P. D., & Bittles, A. H. (2002). The changing survival profile of people with Down's syndrome: Implications for genetic counselling. *Clinical Genetics, 62*(5), 390–393.

Harper, P. S. (2004). *Practical genetic counselling* (6th ed.). London: Edward Arnold, Ltd.

Hart, D., Grigal, M., Sax, C., Martinez, D., & Will, M. (2006). Postsecondary education options for students with intellectual disabilities. *Research to Practice, 45*, 1–4.

Hassold, T., & Sherman, S. (2000). Down syndrome: Genetic recombination and the origin of the extra chromosome 21. *Clinical Genetics, 57*(2), 95–100.

Helm, D. T., Miranda, S., & Chedd, N. A. (1998). Prenatal diagnosis of Down syndrome: Mother's reflections on supports needed from diagnosis to birth. *Mental Retardation, 36*(1), 55–61.

Hodgson, J., & Spriggs, M. (2005). A practical account of autonomy: Why genetic counseling is especially well suited to the facilitation of informed autonomous decision making. *Journal of Genetic Counseling, 14*(2), 89–97.

Jones, K. L. (Ed.). (2006). *Smith's recognizable patterns of human malformation* (6th ed.). Philadelphia: Elsevier Inc.

Krahn, G. L., Hallum, A., & Kime, C. (1993). Are there good ways to give 'bad news'? *Pediatrics, 91*(3), 578–582.

Lejeune, J., Gautier, M., & Turpin, R. (1959). Etude des chromosomes somatiques de neuf enfants mongoliens. *Les Comptes Rendus de l'Académie des sciences Paris, 248*(11), 1721–1722.

Leshin, L. (2002). Pediatric health update on Down syndrome. In W. I. Cohen, L. Nadel, & M. E. Madnick (Eds.), *Down syndrome: Visions for the 21st century*. New York: Wiley-Liss, Inc.

National Society of Genetic Counselors. (1992). Code of ethics. *Journal of Genetic Counseling, 1*(1), 41–43.

Parker, S. E., Mai, C. T., Canfield, M. A., Rickard, R., Wang, Y., Meyer, R. E., et al. (2010). Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. *Birth Defects Research. Part A: Clinical and Molecular Teratology, 88*(12), 1008–1016.

Patterson, B. (2002). Behavioral concerns in persons with Down syndrome. In W. I. Cohen, L. Nadel, & M. E. Madnick (Eds.), *Down syndrome: Visions for the 21st Century*. New York: Wiley-Liss, Inc.

Powell, N. (1991). Your baby has Down's syndrome. *Nursing Times, 87*, 60–61.

Pradhan, M., Dalal, A., Khan, F., & Agrawal, S. (2006). Fertility in men with Down syndrome: A case report. *Fertility and Sterility, 86*(6), 1765.e1–1765.e3.

Prenatally and Postnatally Diagnosed Conditions Awareness Act of 2008. Public Law 110–374, 122 Stat. 4051 (October 8, 2008)

Pueschel, S., & Murphy, A. (1975). Counseling parents of infants with Down's syndrome. *Postgraduate Medicine, 58*(7), 90–95.

Pueschel, S. M., & Murphy, A. (1976). Assessment of counseling practices at the birth of a child with Down's syndrome. *American Journal of Mental Deficiency, 81*(4), 325–330.

Rihtman, T., Tekuzner, E., Parush, S., Tenebaum, A., Bachrach, S. J., & Ornoy, A. (2010). Are the cognitive functions of children with Down syndrome related to their participation? *Developmental Medicine and Child Neurology, 52*(1), 72–78.

Savva, G. M., Morris, J. K., Mutton, D. E., & Alberman, E. (2006). Maternal age-specific fetal loss rates in Down syndrome pregnancies. *Prenatal Diagnosis, 26*(6), 499–504.

Sheets, K. B., Best, R. G., Brasington, C. K., & Will, M. C. (2011). Balanced information about Down syndrome: What is essential? *American Journal of Medical Genetics. Part A*. doi:10.1002/ajmg.a.34018

Skotko, B. (2005a). Mothers of children with Down syndrome reflect on their postnatal support. *Pediatrics, 115*(1), 64–77.

Skotko, B. G. (2005b). Communicating the postnatal diagnosis of Down's syndrome: An international call for change. *Italian Journal of Pediatrics, 31*, 237–243.

Skotko, B. G. (2005c). Prenatally diagnosed Down syndrome: Mothers who continued their pregnancies evaluate their health care providers. *American Journal of Obstetrics and Gynecology, 192*(3), 670–677.

Skotko, B., & Bedia, R. C. (2005). Postnatal support for mothers of children with Down syndrome. *Mental Retardation, 43*(3), 196–212.

Skotko, B. G., Capone, G. T., Kishnani, P. S., & for the Down Syndrome Diagnosis Study Group. (2009). Postnatal diagnosis of Down syndrome: Synthesis of the evidence on how to best deliver the news. *Pediatrics, 124*(4), e751–e758.

Skotko, B. G., Kishnani, P. S., Capone, G. T., & for the Down Syndrome Diagnosis Study Group. (2009). Prenatal diagnosis of

Down syndrome: How best to deliver the news. *American Journal of Medical Genetics. Part A, 149A*(11), 2361–2367.

Snijders, R. J. M., Sundberg, K., Holzgreve, W., Henry, G., & Nicolaides, K. H. (1999). Maternal age-and gestation-specific risk for trisomy 21. *Ultrasound in Obstetrics & Gynecology, 13* (3), 167–170.

Warburton, D., Dallaire, L., Thangavelu, M., Ross, L., Levin, B., & Kline, J. (2004). Trisomy recurrence: A reconsideration based on North American data. *American Journal of Human Genetics, 75*, 376–385.

Williams, C., Alderson, P., & Farsides, B. (2002). What constitutes balanced information in the practitioners' portrayals of Down's syndrome? *Midwifery, 18*, 230–237.