CHALLENGING CASE: FAMILY RELATIONSHIPS AND ISSUES

Responding to Parental Concerns After a Prenatal Diagnosis of Trisomy 21*

CASE

Jane and Luke were excited after hearing that Jane was pregnant. They had been married for ten years and had a healthy six year old daughter. Because of her age (36 years old), they chose to have a genetic amniocentesis at 16 weeks gestation. A trisomy 21 fetus was discovered through standard karyotyping.

They described their marriage as strong. Luke managed a small shoe store and Jane was an elementary school teacher. Their daughter's growth and development was uncomplicated; she adapted to kindergarten very well. Social support, both family and friends, was an important part of family life. Jane's younger sister had a significant learning disability and was always in special education.

When informed about the genetic diagnosis of a trisomy 21 fetus and presented with the options of either continuing the pregnancy or elective termination, the parents asked the following questions: "What is it like to raise a child who is not normal, especially a child with Down syndrome?" "How will the child do in school?""How will our six year old daughter respond to her?""Can we still keep our jobs and raise such a child?"

Dr. Martin T. Stein

Advances in genetic amniocentesis, chorionic villous sampling, fetal karyotype analysis, molecular diagnosis and fetal ultrasonography have brought precision and reliability to the prenatal diagnosis for many disorders. When a condition that is associated with a significant disability is diagnosed prenatally, the manner in which a clinician delivers the "bad news" impacts the parents' understanding of the disorder, initial coping strategies, and reproductive decision making. The provision of accurate and current information about the cause, natural history, and prognosis of the disorder is foremost. The next critical step to assist parents in the process of integrating that information base into their individual and family values and social support network. The medical and psychosocial components of prenatal counseling cannot be separated. They are linked intimately; the latter will guide the way clinical information is processed, interpreted and acted upon.

Jane and Luke's situation is not unusual. A prenatal diagnosis of a serious disorder occurs in approximately 1-2% of pregnancies. The diagnosis of a Trisomy 21 Down syndrome fetus was chosen for this discussion because it is relatively common (1/600 newborns) in pediatric practice. This particular family history was chosen because the parents raised issues about cognitive development, sibling responses, and the effect a disabled child would have on their family and careers.

Responses to the case were solicited from four different perspectives: **Dr. Angela Scioscia** is a perinatologist who specializes in prenatal diagnosis and genetic disorders. Dr. Scioscia has an active prenatal diagnosis practice in an academic center. **Dr. Kenneth Lyons Jones,** a pediatrician and dysmorphologist, works in the area of diagnosis and the elaboration of mechanisms of genetic and teratogenic disorders. **Dr. William Cohen** is a developmental-behavioral pediatrician who directs a regional center that cares for children and families with Down syndrome. Dr. Cohen's work emphasizes the importance of participating family members in the care of children with disabilities. **Christopher and Renee Glass** are parents of four children, one of whom has Down syndrome. They have written about their family's experience with raising a child with a significant disability.

Dr. Angela Scioscia

The discovery of a serious fetal abnormality is one of the most traumatic experiences a couple can face. They will be flooded with strong emotions, including sadness orgrief over the loss of their "normal child", hopelessness and frustrations over their inability or that of health professionals, to correct the problem, as well as anger ("why me?"). Theremay even be denial or disbelief of the diagnosis. Ultimately, they will need to decide on further management. As health professionals, though, we may be able to help guide them through the process in a way which may be less painful and which maximizes the information base that assists them in their decision.

Recent studies employing molecular DNA techniques indicate that 95% of trisomy 21 is maternal in origin, only 5% is paternal.[1] I think it is very important to emphasize that one has no control over the chromosomes in one's gametes, and while we know of the maternal age related risk we do not understand what triggers a nondisjunction event. Since maternal guilt is often irrationally present, it is worth raising this issue to dispel any misconceptions and offer the opportunity to discuss these feelings.

The decision to terminate a wanted pregnancy is excruciatingly difficult; it is counter to all nurturing parental instincts. On the other hand, the decision to accept the life long responsibility for a child/adult

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with significant limitations in development and independent living is an onerous one. Personal, family, cultural and religious values regarding abortion will weigh heavily in the decision. For some individuals, the answer will come quickly; others will require more time. It is not uncommon for the parents to arrive at their decisions indifferent fashions and at different paces. Parenting is a shared venture and it is critical for their relationship that they work through this process both as individuals and together. I emphasize the need to recognize differences and yet maintain communication. It is important to take the time needed to work through a decision with lifelong implications.

While parents may benefit from support and discussion with close family members,friends, and clergy, because of the intense feeling regarding this issue, I suggest they carefully consider with whom they wish to share information. In the case of trisomy 21, the diagnosis will be readily evident at birth; when dealing with chromosomal abnormalities without obvious physical manifestations, I strongly suggest parents limit the number of individuals who know of the prenatal diagnosis to prevent stigmatizing or excessive scrutiny of the child.

The risk for recurrence of a chromosomal abnormality after a child with trisomy 21 is 1.2–1.5%;[2] the second pregnancy may have trisomy 21 or a different aneuploidy. The actual risk Jane would face in a future pregnancy, therefore, would be her age related risk, or 1.5%, whichever is greater. In a future pregnancy, prenatal diagnosis with chorionic villus sampling after 10 weeks of gestation, or an earlier amniocentesis, might provide karyotypic information earlier in gestation.

If Jane and Luke choose to continue the pregnancy they will begin the process of modifying their perceptions of and expectations for their child prior to its birth. At birth, one can anticipate a more natural bonding, which frequently is impaired when the diagnosis is not known ahead of time. In continuing a pregnancy with a trisomy 21 fetus, I recommend a fetal cardiac echo to look for serious cardiac abnormalities; usually this is performed at 20–21 weeks. I also suggest an ultrasound for growth between 28–32 weeks; this scan will also detect duodenal atresia,which may not be evident on earlier studies. The stillbirth rate after midtrimester amniocentesis with trisomy 21 is approximately 30%.[3] I inform patients of this now, so that if a loss occurs, they are not left with a sense of guilt,which may be exaggerated because of initially ambivalent feelings for the pregnancy.

The importance of careful thought and communication in this process cannot be overemphasized. By the nature of Jane and Luke's questions, it is clear that this family is asking appropriate questions and carefully considering their options. In the beginning, no decision will "feel good;" there is no immediately simple or happy answer. Hopefully, working together, they will reach the decision which is right for their family.

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Dr. Kenneth Lyons Jones

Because most parents have had at least some experience with Down syndrome in the past, recognition that their unborn baby has the Down syndrome is perhaps the most traumatic diagnosis that a family has to face. In counseling a couple such as Jane and Luke, it is important to explain that many of their preconceived ideas may not be completely true; that as in the case of their healthy 6 year old daughter, it is impossible at 18 to 20 weeks gestation to predict exactly how their unborn baby with Down syndrome will perform as a first grader; that there is a broad range of outcome in children with Down syndrome; and that there is nothing that can be done at this point to determine where their child is going to fit within that broad range.

Despite the fact that children with Down syndrome have a very specific pattern of minor malformations and many have significant major malformations that may require surgery, problems with behavior and intellectual performance represent the primary long term issue.[1]

Although brain development is affected in all children, the majority of those born in the last 5 to 10 years are doing much better than those born earlier. The reason for this is not completely clear. However, early intervention and special education programs are available for most children with Down syndrome, providing them with the opportunity to overcome many of the developmental handicaps resulting from the poor muscle tone which is so characteristic throughout the first year of life. Although it is realistic for Jane and Luke to expect that their child will require special education programs throughout school and will need assistance with respect to vocational counseling, it is important to recognize that the potential of most individuals with Down syndrome is far greater than has been previously considered. Furthermore, most of the educational and vocational assistance programs are publicly funded.[2] Therefore one does not have to be wealthy to help a child with Down syndrome reach that potential. It does, however, take energy and commitment. This is not to suggest that advocating for a child requires a parent to quit a job. In fact, during the first few years of life, in many communities there are more resources available for children with Down syndrome and their families than is the case for unaffected children.

Certainly one of the critical issues to consider is how their six year old will relate to a sibling with Down syndrome. The answer to this question is intimately tied to the parents' relationship with the baby. There is no question that siblings are affected by having a brother or sister with Down syndrome, but the effect doesn't have to be bad.[3] If parents are able to treat the baby first and foremost as a member of the family, to be loved and nurtured like anyone else, siblings will incorporate those feelings and develop the same pride, loyalty and love that they would for any newborn sibling.

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Dr. William I Cohen

Jane and Luke's questions to their primary care physician (PCP) are reasonable ones. Having learned that the child that Jane carries will be born with Down syndrome (DS), they wonder how this child's needs will impact on all members of the family, and they wonder how she will be affected by this disorder.

While many PCP's may have a number of children of different ages with DS in their practice, I would caution against using that anecdotal experience as the sole or even primary source of information to respond to Jan and Luke's concerns. Rather, I would like to suggest a process of assessing the family's situation, followed by a wide range of information about the lives of children with DS.

This situation seems potentially delicate: if the answers they hear paint a particularly gloomy picture, they may then choose to terminate the pregnancy, since these questions are presented in the context of needing to decide whether to continue the pregnancy. This can place an enormous burden on the PCP to provide as broad and complete a picture as possible. More than one family has decided not to have more children after learning about the "tragedy" of raising a child with DS.

Consequently, I would suggest that we use the same principles in exploring this matter that we would in any complaint or concern that comes our way. We need to understand the concerns behind the questions and define the family environment: its beliefs and resources, its strengths and vulnerabilities. Jane and Luke are looking for some factual information about Down syndrome, and for some opinion on the part of the PCP as to their ability to handle the challenges involved. I would recommend an exploration of this couple's concerns and especially their fears, followed by a review of their social situation to include current job and home responsibilities, financial matters, and the presence of support from the social network and extended family.

This kind of counseling requires that the physician examine his or her own beliefs about raising children with disabilities, so that the information presented is as free of any bias as possible. Current information about the medical and developmental needs of children with DS will extend the PCP's own experience.[1] In addition to the knowledge that termination is a legitimate choice, parents who do not wish to raise the child with DS should know that adoption is also an option.

The PCP need not feel alone in addressing these questions. He or she can seek consultation from a local Down syndrome program or center, and, of course, can refer the family to the program. At our center, parents can speak with our coordinator, herself the parent of a child with DS and they can choose to meet with the Center director, a developmental/behavioral pediatrician. We would give these parents the option to speak with other parents: parents who chose to terminate a pregnancy, parents who found out via amniocentesis and decided to continue the pregnancy, parents who considered adoption, adoptive parents, and parents who have children with DS of various ages.

In a similar fashion, books[2] and videos[3] can be helpful to provide more specific information. Two national organizations, the National Down Syndrome Society (1-800-221-4602) and the National Down Syndrome Congress are also sources of information. The Down Syndrome Medial Interest Group (412-692-7963) can also provide information about DS centers.

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Christopher K. Glass and Renee F. Glass

We have been asked to comment on this case from the point of view of parents of a child with Down syndrome. Our first thoughts on the concerns raised by this case were that the challenges and rewards of raising our child with trisomy 21 have been, for the most part, variations of the same challenges and rewards that we have experienced in raising our other three children. Our son, Bryan, who is now eleven, is an integral and dearly loved member of our family. He is otherwise healthy, and participates fully in all of our activities. Our other children have grown to understand his limitations and, aside from

some special considerations related to his disability, they treat him just like their other siblings. While our expectations of Bryan are somewhat different than for our other children, a common goal in raising Bryan is to help him make the most of his abilities.

We were not aware of Bryan's condition before birth, so the diagnosis of Down syndrome came as a shock to us. Once we got over the initial phase of bewilderment and anguish, however, we discovered the existence of an extensive network of services and support groups that were very helpful in dealing with the special issues of raising a child with disabilities. On the basis of advice from our pediatrician and contacts with these groups, we enrolled Bryan in early intervention programs that helped him acquire various developmental skills at a faster rate than he would have otherwise. These activities certainly required an investment of time, but they were not so demanding that they influenced our respective career plans any more than the responsibilities of raising our other children. As Bryan got older, he participated in a variety of other activities, including soccer, basketball, baseball and piano lessons. In some cases these activities were sponsored by organizations that focused on providing these opportunities to children with disabilities, while in others he was mainstreamed with normal children. Bryan takes tremendous pride in his participation in these activities, which have also provided important opportunities for developing social skills.

Dr. Martin T. Stein

A prenatal consultation with a focus on a disorder associated with medical, developmental and behavioral disabilities may occur in a variety of settings that are dependent on resource availability. In addition to the perinatologist, dysmorphologist and developmental-behavioral specialist represented in this commentary, other providers of care might include a genetic counselor, a medical geneticist, a child psychologist or a general pediatrician. Although the knowledge level about detailed scientific aspects of a disorder may vary among professionals, several fundamental principles of counseling parents who have been given a potentially disabling prenatal diagnosis have been clarified by the commentaries:

Medical information about the condition should be accurate and conveyed to parents in language that is understandable. Written material to be read at home complements the office visit.

Some parents have incorrect prior knowledge or preconceived ideas about the condition; the visit should discover what they know and determine its accuracy.

Parents such as Jane and Luke enter into a prenatal visit as they are experiencing the loss of a "normal child." Feelings of sadness alternate with anger. These are expected emotional responses but may vary by intensity, setting and rate.

Assess the individual family by ascertaining what the parents expect from the visit. Jane and Luke were able to state that they wanted factual information about Down syndrome and an opinion about their ability to handle this challenge. Other parents require probing to establish their concerns or agenda for the visit.

Define the family in terms of their concerns and fears, economic and social support resources, psychosocial strengths and vulnerabilities.

Be cautious about specific clinical pitfalls:

a) Anecdotal experiences from clinical practice may not reflect a spectrum of outcomes for a disability. Down syndrome, with its broad developmental spectrum, is a good example.

b) An overemphasis on negative outcomes may influence reproductive decision making.

c) A physician bias about raising a child with disabilities may influence the discussion; clinicians should examine their own beliefs about families with a disabled child.

Discuss with the parents their plans to share the prenatal diagnosis with family members and friends.

The medical aspects and predictable disabilities following a prenatal diagnosis will be comprehended by parents at different rates. Some are quick to understand medical data; others will comprehend selective information and either discard or deny other information. Respect for individual variations in the quality and quantity of information retained is important. Dr. Scioscia noted that two parents in the same family will comprehend the information and develop conclusions in a different fashion and at different pace. This observation is not only relevant to reproductive decision making but to all aspects of prenatal counseling. Although a clinician may hope for full comprehension after an initial visit, for many parents time is required to assimilate knowledge about the condition and its implications for their particular family. Luke and Jane asked direct questions about the effect on their daughter and their careers. Attention to a parent's agenda for the visit is a good starting point to help focus the information to be conveyed.[1]

The pediatrician who counsels parents like Jane and Luke about a disabling condition is uniquely qualified. With knowledge about the family as a result of caring for another child, their pediatrician can utilize that knowledge to shape the visit to the individual needs of the family. In most cases, a therapeutic alliance between the pediatrician and parents as a result of past experience will enhance the dialogue, maximize information exchange, and encourage the parents to ask questions about and be receptive to discussions surrounding painful issues. However, continuity of care for a family does not ensure effective communication. Pediatricians will maximize their effectiveness in these challenging encounters if they practice fundamental principles of medical interviewing when an outcome is uncertain or a decision about a procedure is conflictual: listen to the concerns of each parent, address those concerns where possible, acknowledge uncertainty, and validate the feelings of both parents. [2]

Primary care and developmental-behavioral pediatricians can mobilize aspects of their training and experience to assist parents like Jane and Luke. Pediatric practice teaches us about the broad range of many childhood disabilities. Down syndrome is not

unique among children with multiple disabilities in the spectrum of cognitive, sensory and endocrine deficits that combine to affect educational and social functions. Experience and appreciation for the whole range of developmental outcomes for a particular disorder enables the clinician to be a more effective counselor—to listen to parental concern without feeling the need to provide definitive answers when they are not available. Referral to other parents in the practice with a child with a similar condition may be very useful as suggested in the commentaries by the Glass family and Dr. Coleman. Finally, a pediatrician's knowledge and assessment of available community resources emphasizes contemporary societal support for children with disabilities at a critical time for the parents.

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