
Genetic Services Policy Project

Multiple Congenital/Fetal Anomalies: A Vignette

Annie Klein is a genetic counselor in Alaska who focuses primarily on prenatal genetics. She works closely with Dr. James, the only perinatologist in the state.

Dr. James referred Elizabeth and Tom Bronson to Annie for genetic counseling after a series of tests indicated that their fetus was affected by multiple anomalies. Elizabeth and Tom were both 27 years old. They had been married for 3 years and lived in Sitka where they ran a small family business. This was their first pregnancy and it had been uneventful up to that point. Neither had a family history of birth defects or developmental delay. Elizabeth had no medical problems or any history of alcohol, tobacco or other substance use. At 17 weeks, Elizabeth's doctor in Sitka (a family physician with OB training) recommended a routine maternal serum screening test to check for Down syndrome and other defects such as spina bifida. Though Elizabeth and Tom were not at high risk for any problems, offering screening to all women is now standard practice.

The screening test came back two days later showing that there was an increased chance of a problem. The test was repeated for confirmation. The second test also showed an increased risk. Because there is no perinatologist in Sitka, Elizabeth and Tom were referred to Anchorage for a diagnostic ultrasound and set up an appointment for two days later. Elizabeth and Tom booked the 3-hour flight, paying the higher rate associated with last minute travel.

Once in Anchorage, the ultrasound examination revealed a female fetus that was small for gestational age (estimated at 18 weeks), with an omphalocele (abdominal wall defect), a ventricular septal defect (hole) in the heart, and a choroid plexus cyst. After discussing these results with Elizabeth and Tom and expressing his concerns that there might be a chromosomal abnormality, Dr. James recommended an amniocentesis, which was performed later that day. Amniotic fluid samples were obtained and shipped overnight to a commercial laboratory in Seattle. Chromosomal analyses by karyotyping (structural analysis of all chromosomes) and FISH analysis (rapid analysis of chromosomes 13, 18, 21, X and Y) were requested. Additional amniotic fluid and blood samples were collected for possible high-resolution chromosomal microarray testing, a new diagnostic process that allows identification of a large number of conditions, many not detected in traditional studies. At this point, Dr. James referred Elizabeth and Tom to Annie for genetic counseling. He gave the Bronsons a contact number since Annie was currently out-of-town, and he also called Annie to alert her to the case.

Annie and the Bronsons were able to connect the next day by phone after the Bronsons returned home to Sitka. Annie could tell Elizabeth and Tom were quite distraught and trying to process all the events of the past two weeks. Her role was to listen to their concerns and answer as many questions as she could with accurate information that could help them make informed decisions about this pregnancy and future pregnancies. She explained that while their histories did not put them at increased risk for having a baby with birth defects, approximately 3 percent (1 out of 33)

of all babies are born with a birth defect. Annie reviewed again the possible causes of the ultrasound findings and the fact that in many cases, the cause is unknown.

Annie discussed the various tests that were currently being done on the amniotic fluid. She also discussed possible scenarios and options for this pregnancy. If they chose to continue the pregnancy, the pregnancy would be deemed high risk and additional fetal monitoring would be advised. If the chromosome studies were normal, the large omphalocele and heart defect would suggest that ideally the baby would be delivered by caesarean section in a tertiary hospital (e.g., Seattle) where there would be immediate access to intensive treatment and surgery for the newborn. If they chose to deliver in Sitka, the baby would be flown to Anchorage or Seattle immediately after birth for treatment in a tertiary setting. If, on the other hand, the chromosomes demonstrated a typically lethal condition such as Trisomy 13 or 18, they could choose to continue the pregnancy but it was unlikely that the doctors would want to perform a caesarean section given the risks to the mother from a surgical procedure. The other option was to terminate the pregnancy.

Annie and the Bronsons discussed the pros and cons of each of these options. Tom had some concerns about termination given his religious upbringing but felt this might be in the best interest of the baby if it was likely the baby would suffer. Elizabeth worried about how they would take care of a child with complex special needs, especially in a remote area like Sitka. Annie told them about the availability of services for children with special health needs, such as public health nursing and educational programs. Tom and Elizabeth also had concerns about cost—the couple did have health insurance, but because they were small business owners, it was a high-deductible individual plan with a maximum maternity benefit. While they wanted to make the best decisions for their baby, the financial impact was very concerning to them. Already, the perinatology consultation, the diagnostic ultrasound, the amniocentesis and associated tests would add up to a significant cost. They also needed to factor in costs for travel, time away from work, etc. Annie talked with the Bronsons for over an hour and reassured them that her costs were covered as a component of the perinatal consultation. Annie informed Elizabeth and Tom that she would call back as soon as results were available.

The FISH test results came back the next day and were positive for a Trisomy 18. After talking with Dr. James, Annie notified Elizabeth and Tom of the results and discussed the importance of waiting until the karyotype came back to make any final decisions about the pregnancy. The karyotype results came back several days later and revealed a Trisomy 18, translocation type. This is a rare cause of Trisomy 18, and may result from either a balanced translocation in a parent or a “de novo” occurrence in the baby.

Annie called Elizabeth and Tom with the confirmatory results and reviewed with them information about how translocations occur, the features associated with Trisomy 18, and the fact that while 90 percent of infants die within the first year, sometimes an individual may live longer. Not having her usual visual aids was challenging, but she did her best to describe the condition over the phone. Mental retardation is a common feature in survivors. She emphasized that Elizabeth and Tom had not caused this condition by anything that they had done. Given that this was a translocation, there was a possibility that either Elizabeth or Tom could be a carrier of

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a balanced translocation (a silent abnormality). If one of them was a carrier, the risk for recurrence in future pregnancies was high (1 in 4). Annie reviewed the options and offered to refer the couple to speak with others who have faced the same diagnosis. She also gave them the website information for a trisomy support group.

Elizabeth expressed relief at knowing how this happened but was very sad it happened at all, and she was concerned about the possibility that it could happen again. Tom and Elizabeth declined to speak with others since they felt they had already determined what they wanted to do: they decided to terminate the pregnancy. Annie explored this decision with them to make sure they were not feeling coerced. She also reassured them that she would share the information and their decision with Elizabeth's doctor in Sitka and he would be in touch with them about scheduling the termination procedure. She provided them with anticipatory grief counseling, recommended additional resources in the Sitka area and on the Internet, and suggested that when they were ready, she would like to meet with them again (ideally in person and before another pregnancy) to discuss further testing and recurrence risk.

Though each situation is unique, Annie finds that many of the issues, concerns, and questions that people have are the same. Being able to help people like the Bronsons sort through all the complex information and make informed decisions is very rewarding. The strength and courage of the individuals and couples that she works with continually inspire her.

Case Issues for Discussion

1. Alaska is a frontier state with a number of unique challenges for health services delivery, including genetic services delivery.
 - a. What mechanisms are in place to address these challenges?
 - b. What additional services would be beneficial?

2. Multiple congenital anomalies (MCA)—the presence of two or more major abnormalities—are complex conditions. Chromosomal changes are a common cause of MCA, though often the underlying etiology (cause of disease) cannot be identified.
 - a. How does uncertainty surrounding etiology and disorder severity affect the role of screening, testing, and counseling for families?
 - b. In this case, the diagnosis of Trisomy 18 is associated with a poor prognosis. How might the case have been different if the diagnosis was Trisomy 21 (Down syndrome), for which the prognosis is less certain?
 - c. How might the case have been different if the cause of MCA was not identified?

3. As highlighted in this case, the identification of MCA has a significant emotional impact on the family.
 - a. What services and supports are available to assist families with these issues?
 - b. What additional services might be beneficial?

4. In this scenario, the presence of MCA was identified prenatally. However, in the majority of cases, anomalies are not identified until birth.
 - a. What factors contribute to delay in identification?
 - b. What additional steps, if any, could be taken to improve prenatal identification?
 - c. What challenges would Elizabeth and Tom have faced if the anomalies had not been identified prenatally?
 - d. What additional resources would they have used?

5. Currently, there is considerable activity in the area of prenatal screening and diagnostic testing on a number of fronts (e.g., clinical policy, research, emerging technology). Recent recommendations from the American College of Obstetrics and Gynecology (2007) suggest that screening for Down syndrome and other birth defects should be offered to all prenatal patients regardless of age or risk factors, a change from the previous emphasis on women with risk factors or advanced maternal age (>35 years). There is also a move toward earlier screening (first trimester) and newer techniques (nuchal translucency, quad screening, etc.).
 - a. What are the potential benefits of earlier and more universal screening?
 - b. What are the potential harms?
 - c. What ethical concerns do the recommendations raise?
 - d. As technology becomes available to screen for an increasing number of disorders quickly and reliably with minimal risk to mother or fetus, how will appropriate use of this technology be assured?
 - e. Who will, or who should, decide what constitutes “appropriate use”?

6. Increasingly, consumers are enrolling in health plans with high deductibles and limitations on coverage, or are unable to afford or choose not to purchase health insurance.

- a. What are the implications of this trend on consumer behavior and choices?
- b. What are the implications for demand and utilization of genetic services, including new technology?

7. Reimbursement has been a significant issue for genetic service providers, especially genetic counselors who provide time-intensive cognitive services and may be unable to bill insurance companies because they are not covered providers. In this case, Annie spends several hours on the phone with the Bronsons, but is unable to bill directly for her time. Her salary is covered by the perinatology clinic.

- a. What are the advantages and disadvantages of this reimbursement system?
- b. What alternative reimbursement systems might be considered and what would be their impact?
- c. What barriers exist to implementing alternative reimbursement systems?

What service delivery issues does this scenario raise?

- Lack of genetics professionals and other specialty services in frontier area
- Significant distance from community to needed resources
- Need for alternative service models (phone, telemedicine, electronic, etc.)
- Need for enabling services (transportation, etc.)
- Need for support resources (grief counseling, etc.)

What provider issues are identified?

- Coordination of care between providers in Sitka, Anchorage, Seattle, etc.
- Criteria for use of new technology
- Balancing patient needs and desires with available technology
- Reimbursement mechanisms for time-intensive services

What consumer issues are identified?

- Educational needs (dealing with complex information)
- Difficult choices/ethical dilemmas related to choices
- Grief and other emotional issues (potentially long-lasting)
- Need for support services (grief counseling)
- Financial concerns: cost-sharing and out-of-pocket expenses

What payer or coverage issues are identified?

- Consumer cost sharing
- Complicated health insurance plans
- Coverage of new technologies
- Reimbursement of counseling services and complex cases

What industry issues are identified?

- Development and role of new technologies (e.g., microarray testing)

What policy issues are raised?

- Ethical and societal implications related to recommendations for screening all pregnant women for Down syndrome and other birth defects