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MINISYMPOSIUM: FETAL/NEONATAL IMAGING

Team counseling in prenatal evaluation: the partnership of the radiologist and genetic counselor

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Abstract Fetal medicine programs within children's hospitals have been developed to ensure access to pediatric specialists across multiple disciplines. The cases that present to these programs are usually complex and require involvement of a multidisciplinary care team. Although some providers on the team limit their focus to their pediatric specialty when counseling patients, the radiologist and genetic counselor have a distinct perspective allowing them to take the larger picture into account in the evaluation of the fetus. As first responders, they come together to review images and identify which consultants are most appropriate to counsel the families, and they can help guide patient discussions. In this paper we demonstrate how the combined expertise of the genetic counselor and pediatric radiologist can facilitate more accurate diagnoses and guide the appropriate management of complex fetal anomalies.

Keywords Anomalies · Communication · Counseling · Fetus · Genetics · Multidisciplinary team · Prenatal diagnosis

Introduction

Fetal medicine programs within children's hospitals have been developed to ensure access to pediatric specialists across multiple disciplines. Cases presenting to these programs are usually complex and require involvement with a multidisciplinary care team. The team includes a cooperative effort among the family, obstetrician, radiologist, genetic counselor, geneticist, neonatologist and pediatric subspecialists. Although some providers on the team might limit their counseling to their specific pediatric specialty, the radiologist and genetic counselor have a distinct perspective allowing them to take a larger view in the evaluation of the fetus. As first responders, they meet to review images and select the appropriate experts for consultation, and they guide patient discussions. Team counseling with the radiologist and genetic counselor can offer the family not only a description of the primary abnormality, be it the brain, kidney or bowel, but also can provide a more comprehensive discussion of associated anomalies and their potential impact on fetal growth, delivery management and long-term outcome. Having the radiologist review the fetal MR and US images with the genetic counselors, subspecialists and family brings the team together and helps to unify the diagnosis; this in turn helps decrease confusion and optimizes a supportive environment for the family in crisis [1–5].

In this paper we demonstrate how combining the expertise of the genetic counselor and pediatric radiologist can help guide diagnosis and management of complex fetal anomalies.

Role of the genetic counselor in the fetal center

Genetic counselors are master's-degree board-certified professionals who have specialized education in genetics and counseling. In a prenatal setting, one of the main roles of the genetic counselor is to help couples make informed choices about their pregnancies. This involves reviewing the benefits and limitations of genetic testing

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options, interpreting genetic test results, and guiding patients seeking more information about how certain diagnoses might impact them and their families [6].

A genetic counselor can be of great value to the radiologist in the fetal center. Ideally, the genetic counselor elicits and reviews the family and medical history, prior genetic screening, prior imaging, and patient understanding of the reason for referral. This information is then shared with the fetal center radiologist prior to the final interpretation of imaging.

Family and medical history

A known family history of genetic disease is relevant because the fetus might be at increased risk for the same condition and the family needs to understand the screening and testing options. If the condition is associated with fetal anomalies, this is useful information for the radiologist. For example, a mother or father with Stickler syndrome, an autosomal-dominant connective tissue disorder, would put a fetus at 50% risk for Stickler syndrome. Prenatally, Stickler syndrome can present with Pierre Robin sequence. Having the counselor alert the radiologists to the family history and 50% risk could help guide appropriate imaging for micrognathia. Conversely, in one case where micrognathia was identified by ultrasound, upon interviewing the family the counselor noticed that the father of the fetus had clinical features of Stickler syndrome. He was unaware that he carried this diagnosis, which genetic testing subsequently confirmed, allowing for appropriate medical management for him and his offspring, in addition to recurrence risk counseling.

Obtaining a thorough maternal medical history can help guide imaging as well. For example, fetuses of mothers with lupus are at increased risk for Ebstein anomaly and chondrodysplasia punctata. A fetal echocardiogram and consultation with a pediatric cardiologist would be appropriate to recommend in such a case. Additionally, the genetic counselor could alert the radiologist to look for potential subtle findings of chondrodysplasia punctata such as a small nose (Binder type), and extra-cartilaginous ossifications that could be easily missed if not searched for specifically.

Genetic screening and testing interpretation

If prenatal screening or diagnostic testing has resulted in an increased risk for Down syndrome or another chromosome abnormality, the radiologist should have this information to help search for associated features. Knowing about an elevated maternal serum alpha-fetoprotein (AFP) could prompt careful assessment of the umbilical cord insertion, spine and brain. Genetic counselors can help radiologists navigate the growing pool of available fetal testing in regard to the significance of false-positive and false-negative results. Recent advances in prenatal screening such as noninvasive prenatal screening and the associated potential for false positives and negatives highlight the importance of the expertise of a genetic counselor for interpretation. A woman who has had noninvasive prenatal screening positive for a *22q11* deletion, for example, might still have a relatively low risk for carrying a fetus with this condition given the low positive predictive value of the test for this condition. Alternatively, a 40-year-old woman with a positive noninvasive prenatal screening for trisomy 21 has a greater than 99% chance of carrying a pregnancy with trisomy 21. Interpretation of prenatal test results can be complex and might be affected by multiple factors such as the specific disease, the age of the mother and the performing laboratory. An accurate understanding of these factors is crucial information that the genetic counselor can relay to the radiologist.

Coordinating findings

The genetic counselor can discuss the need for additional screening or testing options with the family, depending on the information gathered and imaging findings. For example, a woman with a low estriol on a maternal serum screen has an increased risk of carrying a fetus with Smith–Lemli–Opitz syndrome. Relaying this information to the radiologist can help guide closer evaluation of the heart, lungs, kidneys and genitalia. Conversely, if the radiologist identifies ambiguous genitalia, this could alert the genetic counselor to review the estriol levels and recommend additional testing.

In a recent case at our fetal center, a fetal patient presented with multiple anomalies including a cystic hygroma, cardiovascular defect, polyhydramnios, hydrocephalus and microphthalmia. Karyotype analysis was normal. The genetic counselor and radiologist reviewed potential diagnoses including a microarray abnormality, a dystroglycanopathy such as Walker–Warburg syndrome given the microphthalmia, and Fryns syndrome given the cystic hygroma, microphthalmia, cardiac abnormality, central nervous system abnormality and polyhydramnios. Because the possibility of Fryns syndrome was raised, the radiologist looked more closely at the fetus and was able to identify a subtle diaphragmatic hernia as well as digit abnormalities. Although there is no genetic testing available for Fryns syndrome, this additional information moved Fryns syndrome to the top of the list in regard to differential diagnosis and allowed for a more focused discussion regarding delivery planning and management, postnatal evaluation by genetics and the possibility of a 25% recurrence risk in future pregnancies. Without this teamwork, the diaphragmatic

hernia and digit abnormalities would likely have not been identified on prenatal imaging.

Identification of psychosocial support and resources

Each woman and family is unique in how they process a referral to a fetal medicine center and the information they receive during the visit. A genetic counselor can help prepare the radiologist and other members of the team around myriad factors such as patient education, language barriers, stress level, and family dynamic prior to meeting with them. The genetic counselor can also help identify appropriate counseling and support resources for the family. Patient education and prior knowledge of the diagnosis are important to know before counseling families. For women with significant learning disabilities, for example, the words used and pictures shown need to be tailored to their ability to process the information. It is also important to be aware of high levels of stress and anxiety. For patients who are very anxious, it may be helpful to include a social worker or psychologist during the counseling. Family dynamics can also affect the counseling. The genetic counselor can present the patient with the option of receiving diagnostic information or counseling without other family members in the room. All of these scenarios can be addressed appropriately when the genetic counselor and radiologist work together to share insight regarding observed family dynamics prior to counseling.

Radiologist and genetic counselor as a team in diagnosis and counseling

Counseling in the prenatal setting requires distinct skills and close collaboration between providers [7]. After completing an initial intake with the family, the genetic counselor and radiologist can work as a team developing reasonable differential diagnoses and an approach to counseling.

Although the radiologist might be the most able to explain to the family what the fetal MR and US images show, training in communication of difficult news is relatively limited in radiology residency and fellowship programs. The counseling needs to be unbiased and respectful of the patient's choices, culture, religion and beliefs. Families should receive information regarding the abnormal imaging findings in a clear, sympathetic, timely fashion, and in a supportive environment that ensures privacy. Parents want realistic medical information specific to their situation, provided in an empathetic manner, and want to be allowed to hope for the best possible outcome [5]. Counselors are trained to deliver difficult news and can help the radiologist present findings to the family appropriately.

The discussion between the radiologist and genetic counselor should include the following prior to meeting with the family:

- maternal medical history including illnesses and exposures,
- family and pregnancy history,
- genetic screening and testing results,
- previous imaging results,
- patient knowledge, family dynamic and emotional state,
- results of the imaging from the studies performed at your center,
- possible differential diagnoses, and
- need for additional imaging and/or inclusion of other specialists.

Once in the room with the family, the counselor and radiologists should be clear as to the role each plays in the conversation.

- They should make an introduction and give a clear description of their roles.
- The radiologist should review the indication for referral and the results of the images obtained at the visit.
- The genetic counselor should provide input based on family history, genetic testing results and imaging results.
- When appropriate, the genetic counselor and radiologist might review the case as a team with other specialists.
- The team should make the recommendation for further imaging and pregnancy/delivery planning.

Follow-up: keeping the communication going

The unique cases that present in fetal medicine provide ongoing learning opportunities and areas for research collaboration. The genetic counselor remains in contact with families until genetic testing is complete. When a diagnosis is confirmed, the counselor can update the fetal team with this valuable information. Diagnostic information might be obtained after further genetic testing or after autopsy following a termination. In pregnancies that are continued, it could be several months after the prenatal consult when postnatal evaluation and genetic testing are performed. Continued follow-up allows for ongoing communication with the family regarding recurrence risks and options for future pregnancies. Patients might elect to return to the fetal medicine center for imaging in a subsequent pregnancy. In such cases the genetic counselor can notify the radiologist previously involved to ensure continuity of care.

One woman returned to our center for a subsequent pregnancy after multiple abnormalities were identified in a prior

pregnancy with negative routine genetic testing. Similar findings were observed in the subsequent pregnancy. After thorough pre-test counseling, the couple opted to pursue whole-exome sequencing, which confirmed the fetus had Peters plus syndrome, a rare autosomal-recessive disease characterized by multiple anomalies. The parents were both confirmed to be carriers and went on to pursue in vitro fertilization with pre-implantation genetic diagnosis.

The radiologist provides invaluable feedback when new information is obtained by postnatal imaging. The postnatal imaging findings might direct further genetic testing in cases where the etiology for multiple congenital anomalies was uncertain prenatally. For example, in cases of suspected skeletal dysplasia, the postnatal skeletal survey often confirms a clinical diagnosis or narrows an extensive differential diagnosis list.

Ongoing communication among team members in fetal medicine is crucial for program advancement. Comparing previous cases where the diagnosis was confirmed to new cases enhances the interpretation and counseling. Publication and research opportunities also arise through follow-up discussions.

Conclusion

Multidisciplinary team counseling has been shown to be an effective method in helping families faced with fetal anomalies [1, 8]. Team counseling, however, can be challenging to coordinate and schedule, requiring flexibility and time. While radiologists have typically not been involved in primary counseling, the fetal imager's expertise in identifying multiple fetal anomalies, understanding the big picture and familiarity with long-term outcomes can be of tremendous benefit to the family. The partnership between the genetic counselor and radiologist provides a distinct opportunity to improve diagnostic accuracy, management planning and family counseling,

Compliance with ethical standards

Conflicts of interest None

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