How genetic counselors Help Fetal Imaging Assessment

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TEAMWORK
Share Victory. Share Defeat.
Who needs to talk? Members of our team:

- Radiologists, Neuroradiologists and Sonographers
- Genetic Counselors
- Other specialists: Neurologist, Pediatric surgeon, Neurosurgeon, Infectious disease specialist, Orthopedic surgeon, Cardiologist, Neonatologist
First responders:
Radiologist and Genetic Counselor

- Radiologist: Board Certified Physician specializing in Pediatric and fetal Radiology

- Genetic Counselor: Masters degree and board certification in genetic counseling
“Er, so Mrs Trimble, the good news is we got your test results back in record time! ...”
Why a genetic counselor?
Expertise in:

• Evaluating risks for genetic disease, communicating complex medical information to families

• Providing psychosocial support and resources for families

• **Sharing information with other members of the medical team regarding the patient’s medical and psychosocial background**
Ways in which a genetic counselor can help you:

• Provide you with patient medical and family history

• Provide you with information regarding prior ultrasounds, genetic screening or testing and reason for referral

• Share with you first impression of patient: patient background knowledge, concerns, fears, family dynamic

• Provide guidance regarding what patient is looking for from the counseling session

• Follow up diagnoses /outcomes for knowledge and publications
Case example 1

• G2P0TAB1 22 –year-old referred at 30 weeks gestation for fetal MRI, ultrasound, genetic counseling, and Neurology consult.

• Reason for referral: multiple congenital anomalies

• Pregnancy history: patient noted she had been having brown discharge and pain

• Prenatal testing: Amniocentesis with normal karyotype
Case 1 continued

- Ultrasound history: 12 week ultrasound had detected ventriculomegaly

- Family history: patient with maternal ½ sister with mental retardation of unknown etiology

- Social history: patient alone; FOB involved but not present. Patient spanish speaking

- Delivery planning: patient has had insurance issues was unclear of delivery hospital
Initial impression of MRI and Ultrasound findings

- Severe hydrocephalus, diffuse cortical mantle thinning with macrocephaly
- Severe brainstem dysplasia, within pontine kink and thickened midbrain tectum. Poorly formed deep gray structures bilaterally, with unusual multicystic appearance.
- Bilateral microphthalmia.
- Small stomach and polyhydramnios.
Initial impression of MRI and ultrasound findings

- Diffuse skin thickening/cystic hygroma
- Right pleural effusion, Small chest - by MRI heterogeneous parenchyma
- ? Nutmeg lung
- Large kidneys
- Kyphosis lumbar lordosis
- Equinovarus deformity, flexed wrists, decreased tone.
Differential diagnoses

- Microarray abnormality (only karyotype was done)
- Walker Warburg or other muscle eye brain disease
- Fryns syndrome
Fryns syndrome clinical features

• *Diaphragmatic defects (diaphragmatic hernia, eventration, hypoplasia or agenesis).* *Distal digital hypoplasia of nails and/or phalanges*

• Characteristic facial appearance: coarse facies, ocular hypertelorism, broad and flat nasal bridge, thick nasal tip, long philtrum, low-set and poorly formed ears, macrostomia, micrognathia

• Pulmonary hypoplasia, polyhydramnios, cloudy corneas and/or microphthalmia, orofacial clefting, renal dysplasia/renal cortical cysts, IUGR, cystic hygroma

• Malformations involving the brain, cardiovascular system, gastrointestinal system and/or genitalia
Fryns syndrome

- Autosomal recessive genetic condition affecting 1 in 15,000 pregnancies
- No gene or loci have yet been identified or mapped so no genetic testing is available
- Prognosis very poor: survival beyond the neonatal period is unusual
Further evaluation of MRI and ultrasound

• By MRI likely right congenital diaphragmatic hernia without mediastinal shift.

• Possible digital abnormalities
Case example 2

- G4P2 SABx1, 35 year old patient

- Referred at 20 weeks gestation for multiple abnormalities – short long bones, small chest circumference, clubfoot, ambiguous genitalia

- Normal male by NIPT, amniocentesis results pending

- At the visit a demise was noted and the couple was counseled by a radiologist and genetic counselor

- Concern for underlying genetic syndrome emphasized and support resources provided to the couple

- Amniocentesis confirmed triploidy
Triploidy

- Extra set of chromosomes in every cell - 69 (vs. normal 46)
- 1-2 % of all pregnancies
- Detected by CVS, amniocentesis, and screening by *some* NIPT
- Sporadic
- Multiple congenital anomalies- IUGR, CNS, increased nuchal fold, micrognathia, heart defects, syndactyly, renal, and limb anomalies
- Placenta: XXY usually enlarged with cystic spaces
  XXX very small, cystic
- Typically results in early miscarriage or less commonly stillbirth or early neonatal death