Complete Non-Mosaic Trisomy 22 in a Term Infant
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Case Presentation

- 37.2 week gestation female infant born to 37 yo G5P4004
- Initial prenatal u/s revealed thickened nuchal fold & possible cardiac defect
- Repeat u/s was normal
- NICU team present at birth secondary to FHR abnormalities
- Initial physical exam revealed:
  - Cleft Palate
  - Left sided microtia
  - Micronagnathia
  - Frontal bossing
  - Hypertelorism
  - Flattened nasal bridge
  - 3/6 systolic heart murmur loudest at LLSB
Physical Exam Findings
Genetics

- Serum karyotype analysis revealed complete non-mosaic trisomy 22
- Parental genetic test normal
- Review of first, second, and third degree relatives was negative for:
  - Physical birth defects
  - Intellectual disabilities
  - Multiple miscarriages
  - Stillbirths
NICU Course

- Hirschsprung’s disease confirmed on barium enema and rectal biopsy
- Significant congenital heart disease confirmed on echocardiogram
- Agenesis of corpus callosum and absence of septum pellucidum confirmed on brain MRI
- ABR test failed bilaterally. Partial atresia of left external auditory canal and soft tissues confirmed on orbital CT
- Bilateral dilated eye exam revealed micro-cornea, shallow inferior chamber, and inferior iris coloboma of both eyes
- Discharged home on DOL 30 with proper follow-up scheduled
Radiology

- Barium enema with transition zone
- MRI with agenesis of corpus callosum
Trisomy 22

- Occurs in estimated 1 in 30,000-50,000 live-born neonates

- Second most common chromosomal anomaly found in early spontaneous abortion

- Common minor physical findings
  - Low set/dysplastic ears
  - Epicanthic folds
  - Depressed nasal bridge
  - Micronagnathia
  - Webbed neck
  - Finger/toe positional abnormalities
  - Cleft palate

- Major physical anomalies
  - 50% will have complex CHD
  - 50% will have renal agenesis/dysplasia
  - Also can have coloboma, lung hypoplasia, anal atresia/stenosis, ambiguous genitalia
Diagnosis

- Serum karyotype analysis and fluorescent in situ hybridization together diagnose partial, mosaic, and complete trisomy 22

- Stressig et al found several reliable u/s findings useful in making prenatal diagnosis
  - IUGR
  - Hypoplastic femurs
  - Nuchal thickening
  - Cerebellar defects
  - Oligohydramnios
Prognosis

- Life expectancy for complete trisomy 22 remains quite limited.

- Tinkle et al's case review of 30 cases complete trisomy 22 documented over 20 years revealed 22 live-born infants.

- Average life expectancy of those 22 infants ranged from a few minutes to 3 months.

- Our patient is currently 9 months old and doing well.
References


