CASE STUDY #5

MOSAIC TRISOMY 21

firststep Dx BUCCAL PLUS

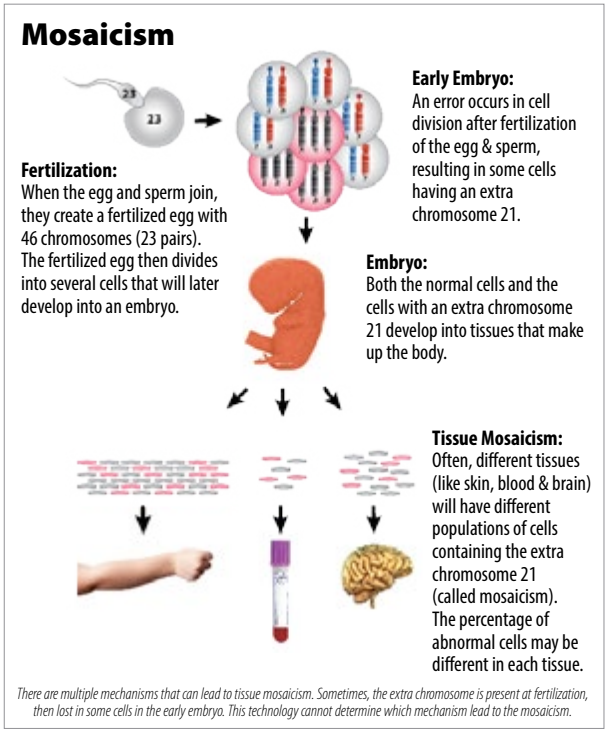
For individuals with autism and other disorders of childhood development

Patient and Clinical Presentation

Pediatrician ordered FirstStep[Dx] testing for a 12-year-old male with multiple challenges, including autism spectrum disorder, hypotonia, and intellectual disability

FirstStep[Dx] Results

Mosaicism for trisomy 21 - an extra chromosome 21 was found in 10-20% of buccal cells and 4% of blood cells



There are multiple mechanisms that can lead to tissue mosaicism. Sometimes, the extra chromosome is present at fertilization, then lost in some cells in the early embryo. This technology cannot determine which mechanism lead to the mosaicism.

Mosaic Trisomy 21 Characteristics

- Trisomy 21: incidence of 1-in-800 births
  - 2% of these have mosaic trisomy 21
- Heart defects
- Vision, thyroid, and hearing problems
- Unique facial features and other congenital anomalies
- Intellectual disabilities

AAP Management Guidance*

CLINICAL AND LABORATORY EXAMS

- Cardiology evaluation
- Annual thyroid function
- Complete blood count (leukemia risk)
- Down syndrome specific growth charts
- Developmental exam
- Feeding problems
- Duodenal atresia
- Constipation
- Otitis media
- Hearing and vision
- Risk for subluxation/atlantoaxial instability
- Obstructive sleep apnea
- Skin problems

TREATMENT

- Physical, speech and language, occupational therapies
- Obesity and weight management
- Discuss therapy options as needed for hypothyroidism
- Discuss therapy options as needed for leukemia
- Education placement assessment and planning

* American Academy of Pediatrics Committee on Genetics. 2001. Health Care Supervision for Children with Down Syndrome. Pediatrics 107: 442-449.

Case Study Summary

- Diagnosis would most likely have been missed by testing DNA collected from a blood draw alone
- Buccal samples deliver a higher diagnostic yield in head-to-head comparison with blood-derived DNA*
- Identification of genetic diagnosis led to significant changes in clinical management

* Sdano, et al. 2014. Clinical Utility of Chromosomal Microarray Analysis of DNA from Buccal Cells: Detection of Mosaicism in Three Patients. http://www.ncbi.nlm.nih.gov/pubmed/25120037

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