## Case Study #5

### Mosaic Trisomy 21

**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


### Patient and Clinical Presentation

Pediatrician ordered FirstStepDx testing for a 12-year-old male with multiple challenges, including autism spectrum disorder, hypotonia, and intellectual disability.

### FirstStepDx Results

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

### 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


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**Mosaicism**

**Early Embryo:**

An error occurs in cell division after fertilization of the egg & sperm, resulting in some cells having an extra chromosome 21.

**Embryo:**

Both the normal cells and the cells with an extra chromosome 21 develop into tissues that make up the body.

**Tissue Mosaicism:**

Often, different tissues (like skin, blood & brain) will have different populations of cells containing the extra chromosome 21 (called mosaicism). The percentage of abnormal cells may be different in each tissue.

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**Fertilization:**

When the egg and sperm join, they create a fertilized egg with 46 chromosomes (23 pairs). The fertilized egg then divides into several cells that will later develop into an embryo.

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