CASE STUDY #4

firststep Dx BUCCAL PLUS

For individuals with autism and other disorders of childhood development

CHROMOSOME 20p DELETION

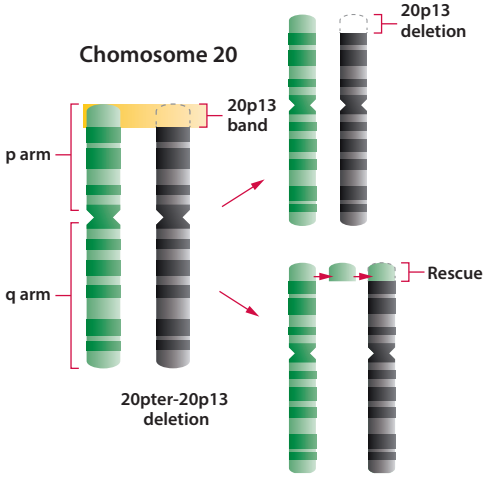
Patient and Clinical Presentation

Pediatrician ordered FirstStep[Dx] for 23-month-old female with developmental delay

FirstStep[Dx] Results

- Complex genetic mechanism
  - Mosaicism – refers to the presence of two cell populations with different genotypes in the same individual
    - Genotype 1: 1.64 Mb deletion of the p-terminus of chromosome 20
    - Genotype 2: long continuous stretch of homozygosity (LCSH) of the p-terminus of chromosome 20
  - Suggests that the normal copy of chromosome 20 tried to "rescue" the other copy, but this did not occur in all cells
- CNVs in this region have been previously reported in 2 other cases with similar features

Mosaic 20p Deletion - SNP Probes Indicate Mosaic 20p Rescue



There is one population of cells with the deletion and one population of cells without the deletion.

The cells without the deletion have a region of homozogosity in its place. This indicates that both regions on the end of 20p come from one parent.

Clinical Features

- Large fontanelles
- Ear abnormalities
- Developmental delay
- Seizures
- Intellectual disability
- Heart defects
- Delayed menses
- Weight deficiency
- Vision disturbances

Management Changes

- Refer to pediatric neurologist for seizure monitoring and management
- Refer to early intervention and individualized education program
- Refer for cardiology evaluation
- Monitor and manage diet
- Vision testing and ophthalmology referral
- Test parents and discuss family planning and recurrence risk

Case Study Summary

- Complex genetic mechanism identified by FirstStep[Dx] would have been missed by standard chromosome analysis or microarrays that lack SNP probes
- Genetic counseling: The family was facing relocation for job purposes; however, the company reconsidered given this diagnosis and determined it was best to keep the child with her current healthcare providers
- Genetic counseling sessions also focused on understanding the risk of recurrence

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Detailed information about FirstStep[Dx] PLUS is available at www.firststepdx.com or call us at 888-888-6736 (OPEN).