CASE STUDY #6

17p12-p11.1 DUPLICATION

firststep Dx PLUS BUCCAL

For individuals with autism and other disorders of childhood development

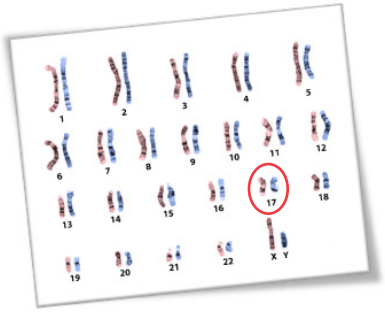
Patient and Clinical Presentation

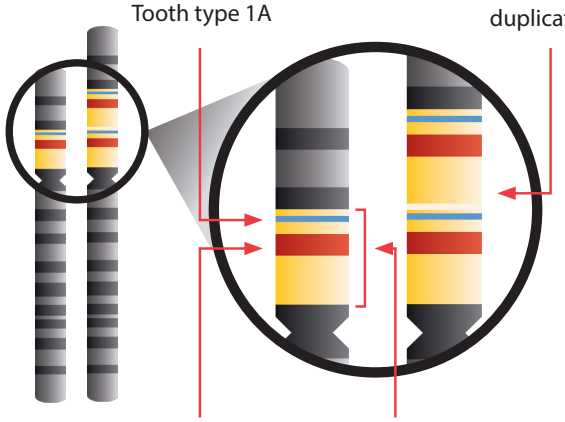
Neurologist ordered FirstStep Dx for an 18-year-old male with intellectual disability

- Genetic testing done at age 2 identified a duplication on chromosome 17
- At that time, the duplication had unknown clinical significance

FirstStep Dx Results

- 7.8 Mb duplication of chromosome 17p12-p11.1
- 51 genes, including the critical region for both Charcot Marie Tooth Type 1A and Potocki-Lupski syndrome





Syndrome Characteristics*

CHARCOT MARIE TOOTH TYPE 1A

- Distal muscle weakness
- Sensory loss
- Slow nerve conduction velocity
- Slowly progressive

POTOCKI-LUPSKI SYNDROME

- Hypotonia
- Failure to thrive
- Heart defects
- Facial features
- Intellectual disability
- Autism spectrum disorder

* This person's diagnosis is expected to encompass features of both these syndromes; he may have additional features not listed here due to the large number of duplicated genes.

Management Changes

- Cardiology evaluation
- Genetics evaluation
- Orthopedic evaluation
- Psysiatry evaluation
- Continued neurology evaluations
- Physical and occupational therapy
- Daily heel cord stretching
- Avoid certain medications

Case Study Summary

- The clinical significance of this person's duplication was reclassified
  - This is a possibility for every result of unknown significance
  - It is important to periodically check-in with a genetics professional for new information
- Genetic counseling session focused on understanding this new information

LINEAGEN
2677 E Parleys Way
Salt Lake City, Utah 84109
www.lineagen.com

affymetrix Powered by Affymetrix™

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