CASE STUDY #3

ANGELMAN SYNDROME

firststep Dx PLUS BUCCAL

For individuals with autism and other disorders of childhood development

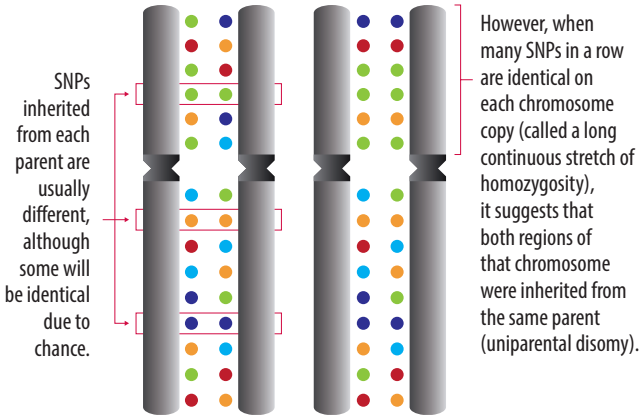
Patient and Clinical Presentation

Pediatrician ordered FirstStep^{Dx} for 5-year-old male with intellectual disability, ADHD, and excessive salivary secretions

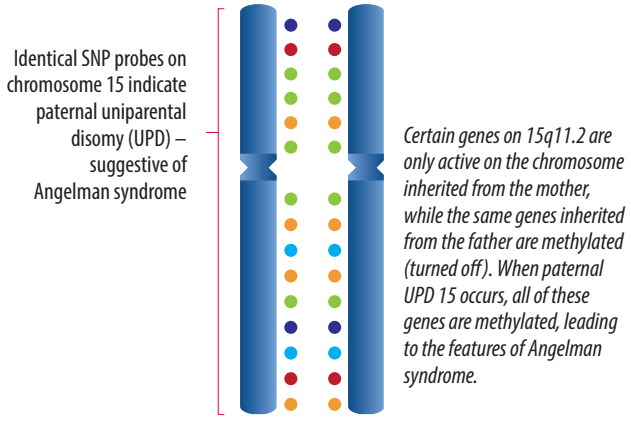
FirstStep^{Dx} Results

- Suggestive of uniparental disomy (UPD) of chromosome 15
- This indicates either Angelman or Prader-Willi syndrome depending on parent-of-origin of the UPD 15
- Lineagen confirmatory testing is consistent with Angelman syndrome

Long Continuous Stretch of Homozygosity



Long Continuous Stretch of Homozygosity Indicating UPD 15



Angelman Syndrome Characteristics

- Developmental delay
- Lack of speech
- Seizures
- Walking and balance disorders/gait ataxia
- Apparent happy demeanor
- Abnormal EEG
- Incidence of 1 in 15,000 births

Management Changes

- Switched from intensive speech and language therapy to sign and picture communication
- Neurological evaluation ordered due to commonly occurring seizures in children with AS
- Informed individualized education program
- Management of sleep disturbances
- Evaluation and management of scoliosis

Case Study Summary

- UPD identified by FirstStep^{Dx} would have been missed by standard chromosome analysis or microarrays that lack SNP probes
- Speech therapy is contraindicated for AS – child is excelling in alternate communication methods
- Genetic counseling: The diagnosis accurately informed family planning decisions and helped the child qualify for a special education program in his school district

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).

FSDXP080912 Ver2.1