

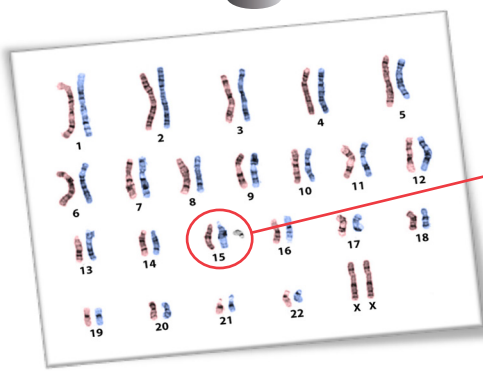
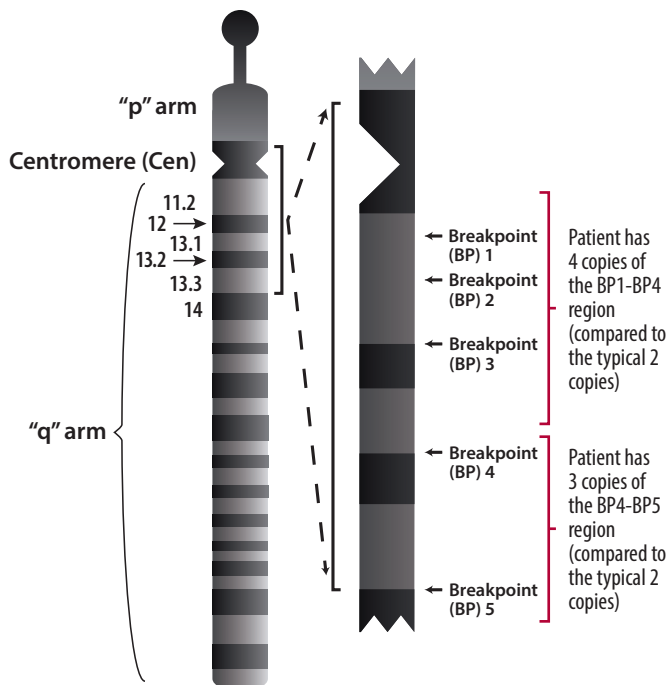
## INVERSION DUPLICATION 15 SYNDROME

### Patient and Clinical Presentation

Neurologist ordered FirstStep<sup>Dx</sup> for a 7-month-old female with congenital hypotonia and developmental delay

### FirstStep<sup>Dx</sup> Results

- Asymmetric gain of chromosome 15 from bands 15q11.2 to 15q13.3
- Consistent with the inv dup(15) syndrome (also called isodicentric chromosome 15 syndrome)



### Chromosome 15

A karyotype is a genetic test that is often used to identify isodicentric chromosome 15 syndrome. FirstStep<sup>Dx</sup> has the ability to identify the exact gene content of the additional chromosome material (in gray)

### Syndrome Characteristics

- Developmental delay, intellectual disability, autism spectrum disorder (ASD)
- Congenital hypotonia
- Seizures between ages 6 months to 9 years
- Increased risk for gastroesophageal reflux disease (GERD)
- Hypogonadism and genital abnormalities
- Increased risk for scoliosis
- Joint hypermobility

### Management Changes

- Evaluation by neurologist for seizure risk (can help prevent unforeseen medical crisis)
- Contraindicated medications include GABA agonists
- Screening and evaluation for ASD
- Initiation of early intervention therapies with focus on strengths (memory) and challenges (verbal language)
- Endocrinology evaluation
- Medication for GERD
- Monitoring of scoliosis and joint problems
- Evaluation with medical geneticist

### Case Study Summary

- The exact genomic content of this asymmetric gain was determined (due to increased probe coverage)
- Identification of genetic pathology led to significant changes in clinical management
- Family was able to connect with the Dup 15q Alliance Support Group
- Low recurrence risk for the family because inv dup(15) syndrome is nearly always *de novo*