



The XXYY Parent Network, an informal association of parents, was founded in 1998 and became the XXYY Project in 2003. In 2015, we merged with AXYS, the Association for X & Y Chromosome Variations.



## RECOGNIZING XXYY Syndrome



### What is XXYY Syndrome?

Boys are typically born with one X and one Y in each cell. Boys and men with XXYY have an extra X and an extra Y chromosome. XXYY is one of several conditions known as X&Y Chromosome variations. Approximately 1:18,000-50,000 boys are born with XXYY. It is characterized as a developmental disability and a neurodevelopmental disorder.

### How is XXYY Diagnosed?

A diagnosis for XXYY is done through genetic testing called a karyotype or microarray, which is performed on a blood sample for chromosome analysis. In some cases, the test may show multiple findings, such as typical XY cells as well as XXYY cells, which is called mosaicism.

### How is XXYY Treated?

Boys with XXYY benefit from evaluations and therapies to address developmental, psychological, and medical problems. Developmental evaluations and therapies may include speech/language, physical, and occupational therapies. Psychological evaluations may include cognitive, ADHD, executive functioning and autism assessments. Interventions may include academic services or accommodations, social skills groups or behavior therapies. Medical evaluations may include developmental behavioral pediatrics, endocrinology, gastroenterology and neurology. Most boys will start testosterone replacement therapy during adolescence prescribed by their endocrinologist. Medical treatments, psychological interventions, and academic services can help promote well-being and activities of daily living.

#### XXYY characteristics recognized by parents and medical professionals:

- Tall (taller than expected for family)
- Curved pinky finger (Clinodactyly)
- Developmental delays/Delayed milestones
- Speech and language impairment
- Behavior outbursts & mood swings
- Learning disabilities & possible low IQ
- ADHD & Autism Spectrum Disorders
- Allergies, asthma & respiratory illnesses
- Tremors
- Flat feet
- High arches (pes cavus)
- Testosterone deficiency & delayed or lack of sexual development
- infertility
- Undescended testes
- Significant dental problems
- Seizures
- Breast development (gynecomastia)
- Low muscle tone (hypotonia)
- Heart problems
- Deep vein thrombosis
- Eosinophilic Esophagitis (EOE)

**Not all individuals with XXYY have all these traits.**

### Interesting Facts

- Cognitive abilities vary among boys with XXYY.
- Many men with XXYY can live independently with community supports, while some may require supervision in a residential setting.
- Babies with XXYY are typically born with normal male genitalia.