The Focus Foundation

research · awareness · treatment

The Focus Foundation presents...

A PARENT'S GUIDE TO A 47,XXY DIAGNOSIS

You've just found out your son has an extra X chromosome. **What's next?**











Critical info and hope for today...

and every day forward, based on 20 years of experience!

Table of Contents

01 What is 47,XXY?

02 Treatment

04 Benefits of early diagnosis

05 Developmental milestones

06 47,XXY: Fact vs. Fiction

08 Looking Ahead

09 About Us

10 Glossary

What is 47, XXY?



If you've just heard the words "47, XXY (Klinefelter Syndrome)," it's normal to feel worried, sad, or overwhelmed. 47, XXY means a boy has one extra X chromosome. It's more common than many realize, and many people go through life without ever being diagnosed.

Each child is unique. Some boys may experience speech or learning challenges, motor delays, or increased anxiety and for some, very little is challenging. With caring specialists, targeted care at home and school, and treatment when advised, children with 47, XXY keep growing, learning, and reaching their goals. They are independent and successful in life!

Physical traits can include lower muscle tone, more easily fatigued, taller height, and, later on, lower fertility. Many families find comfort in knowing that therapies, medical care and fertility options continue to improve. Most importantly, your child is loved, capable, full of potential and able to thrive.

Treatment

Many children do not require any special services but benefit from working with a multidisciplinary team that are experts in 47,XXY and their varied abilities and challenges.

Physical therapy, speech therapy, and/or neurodevelopmental therapy (NDT) are helpful tools that can improve your son's developmental outcome and help him build a strong foundation for the future. These therapies address the possible speech, motor, cognitive, and developmental delays that occur in boys with 47,XXY.

Another avenue to explore is **hormonal replacement therapy (HRT)**. Boys with 47,XXY have decreased levels of testosterone, a male sex hormone, in their bodies. Lower levels of testosterone may contribute to developmental delays and can be treated by supplementing testosterone during key periods of their lives.

"Your child will be happy, healthy, and a normal kiddo! You have the gift of knowledge, a road map of where your child will most likely need support- and all kids need support!"
-Angie, mother of 6 year old with 47, XXY

Treatment Continued

Infancy (EHT)

- The first period for intervention is during the mini-puberty of infancy, with treatment at **4, 5 and 6 months of age**. During this time, infants experience increased hormonal levels that are crucial for their neurodevelopment. Early hormonal treatment (**EHT**) involves giving three testosterone injections over three months. Scientific research indicates that **EHT** enhances cognitive, social, and behavioral capabilities in boys.

Childhood (HBT)

- The second intervention period occurs between the ages of **5 and 8**. During this time, a hormonal booster treatment (**HBT**) can be administered. This involves three testosterone injections over three months to help stimulate the onset of puberty.

Puberty (TRT)

- The third intervention period is during and after puberty. Testosterone replacement therapy (**TRT**) can be provided through daily Androgel applications or weekly injections. **TRT** is designed to maintain adequate testosterone levels during and following puberty.

Benefits of early diagnosis

- Prenatal and early diagnosis of 47,XXY is the most important step in ensuring a positive outcome for your son. When diagnosed early, these boys are able to begin important interventions while they are still developing, improving their outcome.

- Additionally, research has shown that boys with 47,XXY who begin HRT as early as possible show improved behavioral, cognitive, speech, and language skills. Their health and well-being are significantly improved.

- Research has shown that early intervention and treatment is the key in helping certain developmental difficulties such as motor delays, Childhood Apraxia of Speech (CAS), language-based learning disorders, reading deficits, and increased anxiety.

Developmental milestones



Sitting	6 months
4 point Crawling	9 months
Walking	12 months
First Words	12 months
Two-word phrases	18 months



With help along the way, boys with 47, XXY develop within normal limits. With targeted care and EHT, many boys meet the milestones above and some even earlier, which is great to see!

47,XXY: Fact vs. Fiction

Many people haven't heard of 47,XXY, so it can be misunderstood and myths can spread—*especially* online. Here are a few common myths and what the research actually shows:

"Testosterone does not influence brain development in boys with 47, XXY."

Research shows early care matters. Studies find benefits for learning and behavior when specific strategies are implemented at strategic times.

"Boys with 47, XXY are clumsy and not athletic."

Not true. Many boys with 47,XXY thrive in sports and are skilled in active play. Our boys play soccer, basketball, water polo, run cross country and track, and much more!

"There is an increased incidence of autism in 47, XXY."

In our population, the incidence of ASD is quite low. We address social interactions beginning in infancy and onward.

"Boys with 47, XXY are sterile."

Many boys with 47, XXY often have reduced sperm production, but many are not infertile. With newer procedures, sperm can often be retrieved and preserved for future family planning. There are more options now than ever!

"Boys with 47,XXY are extremely delayed and don't reach their milestones."

With the right early support from specialists familiar with 47, XXY, boys reach their developmental milestones right on time. Early interventions such as speech, physical therapy, and hormone therapies can make a big difference in helping them thrive.

"Boys with 47,XXY can't go to college or make friends."

Many boys thrive academically, attend college, and even pursue graduate degrees! They build close friendships, have meaningful relationships, and lead full, happy lives. With understanding and encouragement, your child can reach his goals academically, socially and personally.



Looking Ahead

When you first hear that your child has 47, XXY, it's normal to feel unsure of what to expect. Your son can absolutely live a full, happy, and meaningful life! With love, support, and understanding, boys with 47,XXY grow into confident young men who achieve their goals, build friendships, and thrive in their own unique ways. They are tall, handsome, and lovely young men.

"XXY kids are often indiscernible from other kids in school and teachers say he's one of the best in the class. He may have some struggles, but tends not to stick out from his peers."
-Mother of 2-year-old with XXY

You're not alone in this journey. Dr. Samango-Sprouse and her staff are here to support you, care for your child and make sure he becomes the best he can be! We are here to help and have physicians, therapists and interventionists throughout the world. Together, we can help your son shine—not despite his diagnosis, but because of the strength, empathy, and perspective it helps him develop. Every boy's path is different, but with the right tools and belief in his abilities, he can flourish.

About Us

The Focus Foundation is a non-profit organization dedicated to helping children with X & Y chromosomal variations reach their full potential. For two decades, we have been at the forefront of research, early identification, and neurodevelopmental care for boys with 47, XXY, 47,XYY, 47,XXX, and related conditions.

Our Expertise

The foundation was co-founded by **Jackie Bucksbaum** and **Dr. Carole Samango-Sprouse,** a world-recognized leader in X and Y chromosomal variation research.

- She is the author of the book *X & Y Chromosomal Variations: Hormones, Brain Development, and Neurodevelopmental Performance*.
- She has published over **120 peer-reviewed scientific papers**.
- She is a Clinical Professor of Pediatrics at George Washington University, as well as an Adjunct Associate Professor at Florida International University.
- Her work has shaped how clinicians worldwide understand early development in children with 47, XXY.

What We Do

We bridge the gap between cutting-edge science & everyday life by offering:

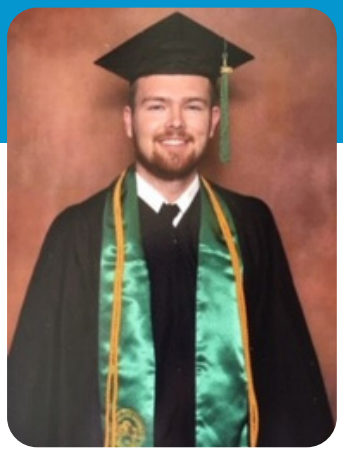
-Syndrome-specific evaluations by experts who understand the unique developmental needs of our patients

-Guidance on early intervention + individualized support

-Access to ongoing research + the newest findings

-A family-centered approach that emphasizes strengths, progress, and hope

Glossary of Terms



Helpful terms for parents to understand their son's 47,XXY diagnosis.

Amniocentesis- a prenatal test involving extracting amniotic fluid from the uterus to screen for certain health conditions.

Androgen deficiency- a condition when the body does not produce enough male hormones, such as testosterone.

Childhood Apraxia of Speech (CAS)- a motor disorder in which a child struggles to motor plan and produce words, sentences, and speech. It is highly associated with later reading dysfunction.

Chromosomal microarray analysis (CMA)- a genetic test used to screen for chromosomal abnormalities, specifically duplications (extra chromosomal material) or deletions (missing chromosomal material).

Hormonal replacement therapy (HRT)- the suggested treatment for boys with 47,XXY to combat their androgen/testosterone deficiency. It involves supplementing testosterone at 3 key periods:

- *Early hormonal treatment (EHT)*- three intramuscular injections administered at 4, 5, and 6 months of age.
- *Hormonal booster treatment (HBT)*- three intramuscular injections administered between 5- and 8-years-old.
- *Testosterone replacement therapy (TRT)*- daily administration of testosterone via Androgel or weekly injections of testosterone.

Hypotonia- a condition characterized by low muscle tone in the face, trunk, lower extremities (legs), and/or upper extremities (arms) causing weakness or floppiness. It is often present in infants with 47,XXY but may be overlooked because it is mild.

Karyotype testing- a genetic test that looks at the size, shape, and number of chromosomes to further screen for abnormalities and mosaicism.

Klinefelter Syndrome (KS)- a sex chromosome condition that occurs when a male has at least one extra X chromosome. 47,XXY is the most common chromosome constellation of this disorder.

Mini-puberty- a period of increased hormonal activity during infancy, between 2-6 months, with a peak around 3-4 months. Mini-puberty has a significant impact on cognitive and physical development of all males.

Mosaicism- a condition where an individual has two ore more genetically different sets of cells in their body. Mosaicism in boys with KS can mean they have some cells that are 47,XXY and some that are 46,XY (normal male karyotype) and can cause milder symptoms. The typical level of mosaicism is between 15-20%

Muscular Torticollis- a condition in which the infant's neck muscles are tight or shortened, resulting in the head being tilted or turned to one side. This can restrict motion, affect muscles on that side of the body, or cause plagiocephaly (flattening on one side of the head). It is addressed with early pediatric physical therapy.

- *Congenital muscular torticollis*- when the condition is present at birth
- *Positional muscular torticollis*- when the condition presents later in infancy, often associated with hypotonia

Neurodevelopmental therapy (NDT)- a treatment approach focused on enhancing motor, movement, and executive function. It can also be referred to as the Bobath method.

Tongue tie- a condition present at birth where the tongue's range of motion is restricted due to a short, thick, or tight band of tissue known as the lingual frenulum. The tongue tie can be placed anteriorly (front), posteriorly (back), or buccally (side).

CONTACT US

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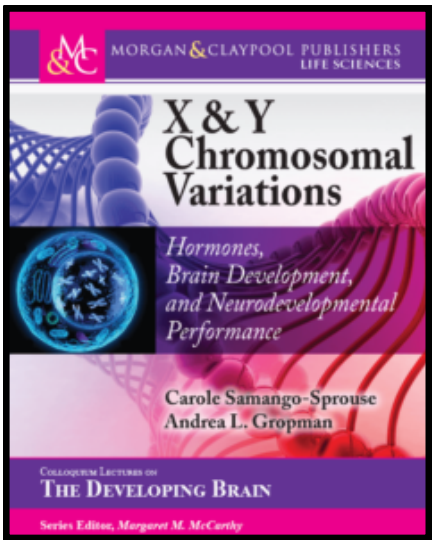
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Check out Dr. Samango- Sprouse's book! (Linked on our website)



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