



The XXYY Project connects parents, medical professionals, and service providers to guide males living with XXYY Syndrome through social, physical and emotional challenges.

Do You Know this Guy?



How to identify a boy or man who may have
XXYY Syndrome

A layman's guidebook for people who may be in a position to help identify undiagnosed boys and men

A letter to men diagnosed with XYY Syndrome or spouses, parents/family members of males with XYY who do not identify with the signs and symptoms in this booklet:

The information in this booklet is based on what we know about infants, toddlers, teens and men with XYY from families who have come forward. If you or a family member has been diagnosed with XYY Syndrome and you have not contacted the XYY Project, **please do so—even if you do not need us.**

The reasons we need to hear from you:

- Medical research on XYY has been largely based on those identified with XYY who have sought support. We need your participation in order to have the most accurate information about the needs, medical and other issues related to XYY.
- We are in the process of developing a new patient registry. We need you to be part of that.
- Families of boys who are significantly impacted by the signs and symptoms of XYY outlined in this booklet need to know that you exist. It gives them hope.
- We may have identified (or will continue to identify) medical issues related to XYY that you need to be aware of. Research so far has already identified some.
- We would like to know about your success so that we can help families to replicate them.
- Because we want to know if this booklet reached you and how it reached you.

. It has been our mission to find you since 1998!

To families of deceased individuals who had XYY Syndrome

We are so sorry for your loss and we would like to honor your loved one. We would truly appreciate it if you would contact us as well. We are in the process of developing a patient registry that will document every medical aspect of XYY possible. Your input into our registry on your loved one's medical issues and cause of death would provide extremely valuable information for others. We hope you will consider contacting us.

To Contact the XYY Project:

Through the AXYS Helpline:

Toll Free: (888) 999-9428

Outside U.S. and Canada: (303) 400-9040

E-mail: support@xyysyndrome.org

www.xyysyndrome.org

Facebook: [facebook.com/xyyproject](https://www.facebook.com/xyyproject)

Twitter: [@xyyproject](https://twitter.com/xyyproject)

(It is best to contact by email for the purpose of this booklet)

© XYY Project 2015

This booklet may not be used for any other purpose than to provide information for potential XYY families.

The photos in this booklet may not be reproduced by anyone for any reason.

Purpose of this booklet

Since 1998, parents of boys and men with XYY Syndrome have been on a mission to find as many people diagnosed with XYY Syndrome as possible throughout the world. Initially an informal group, the founding parents of what is now the XYY Project have maintained a Web site and strong Internet presence and have connected with organizations serving people with disabilities around the world to find every diagnosed person we can find. While we have located hundreds, we know there should be thousands living today.

There are several reasons we believe we are not locating all of these families:

- Some people do not have Internet access.
- There are countries we are still not able to reach, but even in countries we do reach we are not finding the thousands that should exist.
- Language barriers limit us. We are a small organization using English as our primary language but we are still not reaching all English-speaking families.
- There may be individuals with XYY Syndrome, parents and spouses who are not looking for a support organization either because they don't need it or because they don't think we exist.

But we believe these are the main reasons we are not locating them:

- These people are either **not diagnosed or they are mis- or under-diagnosed** with other conditions (especially Autism and ADHD), because there are many symptoms of XYY Syndrome that either look like other conditions or often co-exist with other conditions (technically called comorbid conditions).
- There may be people living with XYY who think they have Klinefelter syndrome (XXY).
- There may be individuals with XYY Syndrome who do not have significant signs and symptoms outlined in this booklet. **We especially need to find these individuals.** If you have XYY or if you are a parent, sibling or friend of someone who has XYY who DOES NOT have these symptoms, please contact the XYY Project!
- There may be individuals with XYY Syndrome who are institutionalized whose families are no longer a part of their lives.

This booklet was created to help parents, spouses, other organizations, teachers, speech and language pathologists, occupational therapists, disability service providers, psychologists and mental health providers, babysitters, neighbors, family members, Autism organizations, learning disability organizations, ADHD organizations, and basically anyone to identify a boy or man who may have XYY Syndrome. In so doing, we are hoping to dramatically increase the rate of diagnosis.

Receiving a full and accurate diagnosis is extremely important for boys and men with XYY Syndrome to receive the medical attention and service supports they may need to live full lives. The more individuals we can identify, the more they can be included in medical research that can help develop therapies and understand the medical issues they have.

We ask you to review this booklet and if you think you know someone with XYY Syndrome, suggest that they obtain genetic testing. Share this booklet with any person or organization that you think might be in a position to identify an undiagnosed boy or man. **You could change someone's life by doing so.**

This booklet is not a medical paper although it does utilize information we know so far from medical research about XYY Syndrome. It is meant to help those who may be in contact with a boy or man with XYY to recognize the condition. This booklet should not be construed as medical advice nor predictive of the lives of any boy or man with XYY. Until more males are diagnosed with XYY and/or we find those without these symptoms, we do not know the full spectrum of XYY.

To Autism, ADHD, Learning Disability, Intellectual and Developmental Disability, Mental Health Communities:

The XYY Project is specifically reaching out to you because **we have strong evidence** that people with XYY Syndrome are being served in your communities and many of them are undiagnosed. Some are diagnosed but have not found the XYY Project. These individuals may not be receiving the benefit of interventions that could help them.

So many of the people the XYY Project knows who have been diagnosed with XYY Syndrome were not diagnosed until they were teens or even adults. It is extremely important that it is not assumed that someone would have recognized that they needed genetic testing. Their parents may have been satisfied with the diagnoses they do have, even though there are other symptoms that are not explained by those. Some parents may have been told that genetic testing should be done, but they decided not to pursue it. The first case of XYY was not reported until 1960. The testing for XYY was not available before that time and not widely used for a long time. It is safe to assume that there may be many people with XYY who were diagnosed with intellectual and developmental disabilities in the years after 1960 who were never tested. Most of the adult cases we see are individuals who were not tested until the 80's and 90's and beyond. As a result, undiagnosed people with XYY may be facing medical problems they do not know about, including some life-threatening conditions such as blood clots, heart problems, severe food allergies and more.

We need your help to reach them.

Please review this booklet thoroughly and use it as a training tool for your staff. Feel free to distribute it to families that might benefit.

The Hallmarks of XYY:

There are many symptoms of XYY outlined in this booklet by age group. Not all people with XYY have all of the symptoms and sometimes they are only mildly affected. However, there are a few symptoms that do apply to a majority of those who have XYY:

- Low muscle tone
- Speech delay
- Developmental delay
- Small testicles, delayed or stalled sexual development, low testosterone (hypogonadism)
- Taller than his family and in puberty and adulthood if he was not treated, he may be extremely tall

When added to the other symptoms such as learning disabilities or ADHD, Autism Spectrum Disorder, mental health diagnoses or intellectual disability, elbow deformities and other medical problems, a male with the above symptoms who has the body types and facial features shown in this booklet should have genetic testing. Some of his medical problems may be unexplained. For example, he may have unexplained white matter brain lesions. He may have tics, tremors and seizures. Again, when added to the above list, he needs to be tested.

**This booklet contains photos of over 50 different men and boys with XYY
Although we have used photos of the same person more than once in some cases,
it is important to realize how many different people are represented.**

What is XYY Syndrome?

48, XYY is a sex chromosome variation that affects one in every 17,000 males who are born.¹ XYY Syndrome is the name attached to the cluster of symptoms that arise as a result of the 48, XYY chromosome pattern, or an extra set of X & Y chromosomes. People of all races and ethnicities can have 48,XYY.

48, XYY is one of several types of sex chromosome variations, including those that are considered as variants of Klinefelter Syndrome, 47,XYY and 48,XXYY, 49,XXXXY, XY/XYY and another syndrome called Jacob Syndrome or 47,XYY, all of which affect males. Similar female syndromes include Turner Syndrome (XO) and several variations with added X's, such as Trisomy X (47,XXX). There are important differences, between boys and men with 47,XYY and those with 48,XYY and some authors have questioned whether 48, XYY males should be included under the umbrella of Klinefelter Syndrome.²

How many people in the world should have XYY Syndrome?

Based on the rate of incidence, this is a random sampling of countries:

Country	Number of males with XYY Syndrome that should be living
Worldwide	212,335
United States	9,262
Canada	1,026
China	42,219
United Kingdom	1,855
India	38,079
Germany	2,389
Indonesia	7,393

Although the XYY Project does have members in the United States, many European countries, Canada and a few members in Asia and some other countries, our numbers are only in the hundreds.

How is a person diagnosed with XYY Syndrome?

Diagnosing 48, XYY requires a genetic test called a [karyotype](#). The test is done by drawing blood and an analysis is done on the cells of the blood to determine the boy or man's chromosomal make-up. In some instances, a boy can have some normal XY cells and some XYY cells. This is called mosaicism.

Where should a person go if they think they may have XYY Syndrome?

The first step is to visit a pediatrician, a developmental pediatrician or your primary care doctor and ask for a referral for genetic testing with a geneticist.

References

- [1] The incidence of 48,XYY syndrome was originally estimated at 1/50,000 (Sorensen et al., 1978), but a recent report found the 48,XYY karyotype in 1/17,000 males in a newborn screening (Nielsen et al., 1991).
- [2] Grannatico et al., 1990 Males with XYY have two X and two Y sex chromosomes, instead of one each. XYY is sometimes considered a variant of another syndrome called Klinefelter Syndrome, or 47 XYY. There are other types of sex chromosome anomalies such as XYY, XXXY, XXXXY, and variations in girls such as XO (Turner Syndrome), XXX, XXXX. and XXXXX.

What does XXYY look like?

People ask us all the time if you can tell by looking at someone if they have XXYY Syndrome like you can with Down Syndrome. The answer is yes and no. If you have ever seen a group of boys and men with XXYY together, you do realize that they have many similarities and you can actually identify them. Parents were the first to realize that this was true. However, average people would not immediately notice their features unless they had seen a group of them before. Therefore, we present to you what XXYY looks like in boys and in teens and men, because there are differences as they get older. XXYY affects people of all races and ethnicities.

Physical features

Height

The first, most obvious physical feature of a person with XXYY Syndrome is that they are typically **taller than their family**. This is not as obvious when they are small but they do tend to be in the upper percentile for growth or taller and bigger than classmates. When they are teens and adults and have not been diagnosed or treated, they may be significantly taller than their family—and by that we mean they can reach 7 feet tall.



Parents, sibling and son with XXYY at 15 years



Mother & son with XXYY at 31 years and his sibling at 20



Parents, siblings and son with XXYY at 19 years



Mother & son with XXYY at 19 years



Parents & son with XXYY at 19 years



Mother & son with XXYY at 16 years



Father & son with XXYY at 22 years



Mother & son with XXYY at 19 years



Father & son with XXYY at 16 years

What does XYY look like?

Physical Features (continued)

Facial Features

Boys and men with XYY Syndrome have several facial features that can be distinct. They may not all have all of these features: **wide-set eyes, large bridge of the nose, full lips, folds in the eyelids, large foreheads, prominent brow and sometimes pronounced indentations in the temples.**



What does XXYY look like?

Facial features (continued)

As we have said previously, boys with XXYY often look very much alike. These photos of eight different boys and teens with XXYY show the most classic facial features and how they look similar.



What does XYY look like?

Physical features (continued)

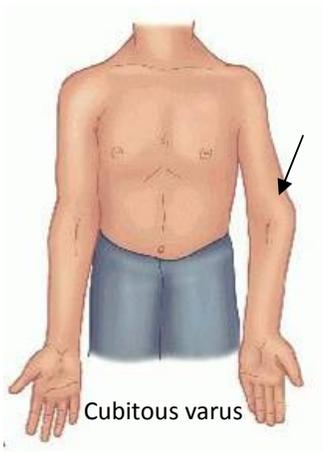
Other

There are several other physical features associated with XYY. Not all males with XYY may have all of them: **Flat feet (pes planus), curved pinky fingers (clinodactyly), breast development (gynecomastia), chest indentation (pectus excavatum, not as prominent as shown in the graphic below), deformities of the elbow (cubitus varus and radioulnar synostosis), long arms, deformity in the knee joint where it bends backwards (genu valgum).**

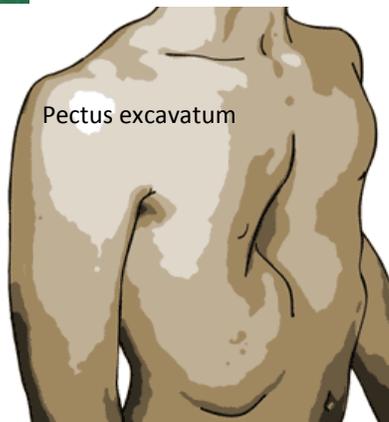
As a result of the features of the legs and arms, you may notice that a boy or man with XYY walks or runs in an unusual way or that his arms do not lay flat against his body. His elbows may protrude unusually or his arms may appear to be turned in an unusual way.



clinodactyly



Cubitus varus



Pectus excavatum



What does XXYY look like?

Physical features (continued)

Body types

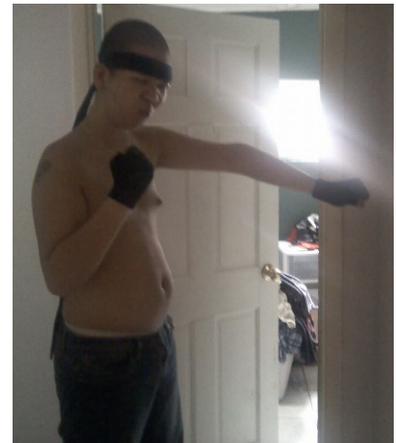
Boys and men have distinguishable body types. They may be very thin or heavy and have little body hair until they receive testosterone replacement therapy. Again, his elbows may be very prominent.



17 year old teen with XXYY (left) with his 22 year old brother



30 year old man with XXYY with his aunt



What should we look for?

Since many, if not most boys and men are never diagnosed with XYY, it is important that you never take for granted that he should have already been diagnosed, especially if he is a teen or adult. XYY impacts each person differently, on a spectrum similar to Autism. A person may not have all of these issues. Here, we have broken down the signs of XYY by age groups.

Newborns & infants

A few of the newborns whose parents have contacted us have received a diagnosis at birth because their baby had a heart condition, club foot or doctors thought the child might have a genetic condition because of various signs they saw. Heart conditions are associated with some but not all males with XYY. Instead, the signs of XYY typically begin with developmental issues and feeding problems beginning at birth into toddler age.



Newborns and infants with XYY may:

- have trouble latching on and sucking.
- sleep more than other babies and fall asleep often such as while eating or playing.
- appear to be “floppy” or can’t hold themselves or their necks up. They may fall over while sitting more than other babies their age.
- begin having mild to severe delays in developmental milestones like rolling over.
- have difficulty with fine or gross motor skills (for example, may not be able to pick up a Cheerio from their high chair tray).
- not crawl like other babies. They may scoot on their behinds instead.
- have small and/or undescended testicles.
- have hernias.
- be allergic to milk products/formula and may have projectile vomiting.
- gag a lot.
- have trouble holding a baby bottle.

What should we look for?

Toddlers

Delays in development become more obvious in toddlers with XXY. In addition to some of the signs outlined in the newborn and infant phase, there are other signs of XXY in toddlers.



Toddlers with XXY may:

- talk late or not at all. They may get very frustrated during attempts to communicate. They may have a “language of their own” that only those close to them understand.
- not be able to pull themselves up.
- walk late (beyond 2 years old).
- have trouble with balance. May fall often.
- potty train late and may not even potty train fully until much later.
- begin having temper tantrums which may continue beyond the “terrible twos”.
- have trouble learning to ride a tricycle.
- begin showing lack of judgment and impulsivity.
- hit, pinch and bite more than other toddlers.
- have croup-like coughs often, asthma, allergies or hospitalizations for respiratory problems.
- have trouble learning to drink from a straw or sippy cup.
- have a far-away look in his eyes as if in a fog.
- when they do walk, they may have an unusual gait and may have difficulty climbing stairs in an alternating step fashion. They may not be able to run.
- refuse to sleep in his bed. Instead, he might be found sleeping under his bed, beside his bed on the floor or in other places like closets after he has been safely tucked into bed.
- have staring spells or seizures.
- have night terrors.
- have heart problems/abnormalities

What should we look for?

School-aged

School-aged boys with XYY begin to show deficits in learning, socialization and self-regulation along with other signs outlined. They will also continue to have many of the issues they had when they were younger.



School-aged boys with XYY may:

- still have speech problems that require speech therapy.
- have learning disabilities that require special education supports. They may have low IQ and/or adaptive functioning scores on tests.
- have trouble waiting for his turn to talk in school.
- have trouble with voice regulation.
- be behind his peers in reading and math.
- continue to have temper tantrums or frustration-based outbursts.
- be extremely shy, withdrawn or introverted. (It is important to know that boys with XYY are not always shy.)
- have difficulty making friends.
- behave in socially inappropriate ways.
- fall asleep in school often even when he has an appropriate bed-time.
- tell many tall tales.
- have trouble in physical education classes or difficulty with some sports.
- have difficulty respecting other's space.
- have extreme reactions to noise and light.
- have unusual dental problems such as missing adult teeth, large tooth pulp, more decay than other children.
- may act out in unusual ways to get the attention of other children who are not socializing with him.
- talk back to parents, teachers, authority figures.
- talk to himself out loud.
- have difficulty following multi-step directions.
- have difficulty writing and taking notes.
- have a tremor in his hand or thumb.
- have tics.
- have trouble taking tests.
- be diagnosed with ADHD.
- be diagnosed on the Autism spectrum (PDD-NOS, Asperger's) or executive function issues.
- have sensory issues.
- have auditory processing disorder or sensory processing disorder.
- be diagnosed with asthma, food and environmental allergies.
- have difficulty with personal hygiene.
- have extreme sugar-cravings.
- bite his nails a lot.
- have scoliosis

What should we look for?

Teen-aged

Teen-aged boys with XYY begin showing signs of delayed puberty and their social immaturity becomes more of a barrier in their interactions. Most boys with XYY are behind their peers in social age, meaning that they may be 16 years old but have the maturity of a 10 or 12 year old.



Teen-agers with XYY may:

- still have speech problems but the verbal signs of speech delay have diminished or disappeared.
- have great struggles with social interactions and peer relationships. They may have few or no friends.
- have difficulty in social interactions with girls due to low social age.
- be bullied—a lot.
- still have outbursts.
- say anything that they are thinking about out loud without filtering it for appropriateness and/or cuss.
- still have difficulty with respecting other people's space.
- still have difficulty in physical education classes.
- have delayed or stalled puberty or no signs of puberty such as no body hair.
- have difficulty staying on task.
- dominate conversations without giving others a chance to speak.
- have accidents such as falling, tripping, trouble on bicycles.
- lack judgment.
- be impulsive.
- steal from others.
- not be able to stay home alone.
- continue to be sensitive to noise, light.
- continue to have food and environmental allergies and asthma.
- have difficulty with personal hygiene.
- need to be constantly prompted to do things.
- have great difficulty cleaning their rooms or conversely be overly neat.
- have increased tics and/or tremor.
- continue to have dental problems and may need braces.

What should we look for?

Adults

Adult males with XYY continue to have the issues they had in the past. However, they may also begin to have new features and health problems.



Adults with XYY may:

- have sleep apnea.
- be unusually thin OR unusually heavy.
- have low testosterone levels (hypogonadism).
- be exceptionally tall for their family, especially if they have not received testosterone replacement therapy.
- begin to have more pronounced facial features.
- have pronounced hand tremors.
- have diabetes.
- have severe food allergies.
- continue to have significant dental problems.
- continue to be behind in social age.
- have trouble keeping jobs.
- have trouble with interactions with coworkers.
- not have been able to attend or succeed in college.
- complain of a lot of pain in joints and back.
- have headaches.
- have thyroid problems.
- have blood clots.
- be in community programs for people with disabilities.
- still have frustration-based outbursts.
- have vision problems.
- have difficulties in relationships.
- still be interested in activities that teens are interested in doing.
- be easily influenced by bad people and make friends with the wrong crowd.
- have diabetes.
- have acid reflux.
- have digestive problems.
- have other health problems.
- have leg ulcers.

As they grow up

Below are photos of individuals as they have grown from boys to teens or men. Each line is a single individual. The photos show how boys with XYY change over time. Each line is one person as they grew.



Other diagnoses

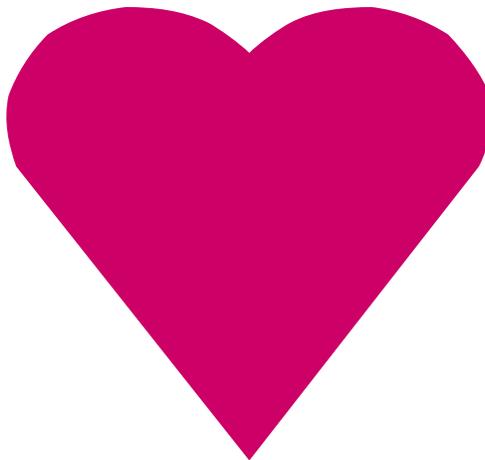
It is important to note that in addition to the symptoms we have listed in this booklet that have been commonly seen in XXYY, there is a list of other diagnoses that have been noted in various cases. Some of these are not in medical literature (yet):

- Polydactyly
- Cleft palate/Pierre-Robin
- Palatoplasty for VPI (no cleft)
- Tourette Syndrome
- Unilateral kidney
- Various brain abnormalities
- Thrombotic Thrombocytopenic Purpura (TTP)
- Colon cancer
- Pulmonary embolism
- Pituitary adenoma
- Eosinophilic esophagitis (EoE)
- Schleroderma
- Non-Hodgkin lymphoma

We have also noticed anecdotally that our families have twins running in their families. We have many boys and men with XXYY in our group who are fraternal or identical twins.

Please refer to: *A New Look at XXYY Syndrome*, Tartaglia et al. 2008
American Journal of Medical Genetics Part A 146A:1509–1522 (2008)

In memory of:
Willie, Ryan, Joey, Buzz and Jimmy



Deepest Thanks

To individuals with XYY and their families

The XYY Project is deeply thankful to the individuals with XYY and their families for being so incredibly generous in sharing your photos for this booklet. We know that if we reach even one undiagnosed boy or man with XYY, we have truly accomplished something. The families that are part of the XYY Project have always been very concerned with finding individuals and families and are ready to provide support to whomever finds this booklet and receives the XYY diagnosis.

If this booklet made you realize...

...that you may “know that guy” (or you may be that guy)

You are not alone. In spite of the fact that XYY is rare, we are here and we have been here for many years, searching for you. We are like your long-lost family.

Bring this booklet to your doctor. Tell them to also look up this medical literature:

A New Look at XYY Syndrome, Tartaglia et al. 2008

American Journal of Medical Genetics Part A 146A:1509–1522 (2008)

Ask for genetic testing.

Once you have positive XYY genetic testing results:

Join us in our quest to get answers for every person with XYY Syndrome.

If you got genetic testing And the result is XXY, XYY or other...

Because XYY looks somewhat similar to Klinefelter Syndrome (XXY) or Jacob Syndrome (XYY), some people who identify with this booklet may end up diagnosed with one of these conditions instead of XYY. There are also other variations such as XXXY, XXXXY, XYYY and so on. If you are diagnosed with any of these, please contact AXYS. The XYY Project is a project of AXYS:

AXYS (formerly known as KS&A)

www.genetic.org

Toll Free: (888) 999-9428

Outside U.S. and Canada: (303) 400-9040