



Something is Just “Not Right” – A Suspected Genetic Disorder

What to do if you think your child has a problem that is not being addressed

The following information is meant to help parents and caretakers to understand the need to seek medical advice when they feel their child is “just not right.” This should not be construed as medical advice in and of itself and should never replace medical advice. Always consult a physician if your child is experiencing problems.

Trust your instincts

The XXYY Project began as a group of parents who had one thing in common: something was just not right with our sons. As parents and as an organization, we are here to tell you to trust your instincts. Even if you have explained unusual characteristics about your child to your doctor or school, **don't stop pursuing this** until you have an answer that satisfies you.

Don't rule out anything

The information in this document covers aspects of many types of conditions but is in no way comprehensive. As you read on, you may find things that don't apply to your child. Don't rule out anything because of this. There are so many disorders that we cannot possibly tell you everything. As we have said already, trust your instincts. *Write down everything that seems unusual* to you and take the information to your doctor and get other opinions if you still feel you are not getting an answer.

Does your child have developmental problems, learning problems, or behavioral problems?

There are many genetic conditions and chromosomal problems which can now be diagnosed with a blood test. It is important to test children and adolescents with these problems so that appropriate therapy and intervention can be started. Chromosomal and genetic testing should be done on all children with developmental delays, speech delays, behavioral problems, learning disabilities, autism or autistic spectrum disorders, and intellectual disabilities. In addition to genetic testing, which would reveal these kinds of disorders, your child should have a full evaluation to determine the cause of their problems, even if genetic testing comes up negative.

Sex chromosome aneuploidy

The XXYY Project focuses on XXYY Syndrome, which is one type of sex chromosome aneuploidy, but there are many types of chromosome variations. A typical boy has one X and one Y Chromosome. A typical girl has two X chromosomes. A sex chromosome anomaly occurs when there are differences in the X and/or Y chromosomes. Some of these disorders, like Fragile X Syndrome, can run in families and it is important that families with the condition are identified.

Not all children with the behavior/developmental/physical signs described will have a genetic condition, however, anomalies in the sex chromosomes are very common and may cause a variety of problems ranging from speech delay and learning disabilities to intellectual disabilities. Children with sex chromosomal variations may also have ADHD, impulsivity, or autism.

The information contained here is not only about XXYY Syndrome. There are many characteristics of XXYY Syndrome listed here that can also apply to other conditions. This information is meant to get you started in developing your own list of things to bring to your doctor.

Even if your “child” isn’t a “child” anymore

There are many adolescents and adults with severe learning disabilities, intellectual disabilities or autism who were not tested in childhood. When so much time has passed, one tends to think that if this could have been an issue, somebody would have tested him or her. But this may not be the case. These tests were not always available or widely used and may have changed over time. If your child was tested many years ago, some things could still have been missed because medical research and development have evolved over time.

What if you already have some other diagnosis?

Many of the symptoms associated with sex chromosome anomalies (or X&Y chromosome variations as we prefer to call them) *look like other disorders* but could instead be a manifestation of the genetic condition. What we have seen, time and again, is that children are diagnosed with other conditions without ever having genetic testing. This means that just because your child already has some type of diagnosis, does not mean they have their full diagnosis, if there are also certain other symptoms. Some of these conditions include:

Bi-Polar Disorder
ADD/ADHD
Sensory Integration Dysfunction
Autism or Autism Spectrum Disorder
Pervasive Developmental Disorder
Learning Disabilities
Intellectual Disability (formerly called Mental Retardation)
Conduct disorder
Psychosis
Obsessive Compulsive Disorder

Important:

Not all children who have sex chromosome anomalies would have the above conditions but if your child has been diagnosed with any or many of these conditions and has other symptoms, there could be more going on.

Warning signs

If your child, adolescent or adult child has any of the following symptoms, it is very important to talk to your doctor about genetic testing. We are presenting these symptoms in terms that parents can understand and recognize, from your perspective. Remember that your child may have grown out of some of these things, but they may still be signs:

Developmental delay, issues or signs:

- Late to walk or difficulty just putting one foot in front of the other when walking
- May have crawled late or perhaps never crawled on his or her hands and knees – scooted instead
- Problems sitting up without support or “floppiness”
- Baby felt heavier or harder to hold in your arms than other babies
- May have had trouble learning to climb stairs or playground equipment
- Losing balance
- Runs or walks in an unusual way
- Slow or unusual sexual development
- Trouble picking things up with fingers or hands
- High or low on growth charts, especially considering your family history
- Displays difficulty in gym class, unable to keep up with peers
- Has had physical or occupational therapy

Speech delay or issues related to facial/throat muscles

- Late to talk or doesn't talk at all
- Speech that is unrecognizable to pretty much anyone but you
- Child uses sign language of some type, but you can't find hearing problems
- Older children, adolescents and adults may have problems with word retrieval and may talk more slowly as a result (people often interrupt them when they are trying to say something, too)
- Often drooling
- Trouble sucking
- Gags easily or some trouble swallowing

Behavioral problems or unique "behaviors"

- Outbursts that seem different than other children, happen at unusual times, are more frequent than they should be, are difficult to stop, or are not age-appropriate. Such as: cries easily, angers easily, lashes out at people, kicks things, throws things, cusses, mutters under breath, doesn't "filter" his or her thoughts before saying what he or she thinks, emotional outbursts.
- Arm flapping
- Hiding in small places (under bed, under desk at school, in closets – any small space).
While this is actually NOT a behavioral problem but instead a sensory problem which is common in kids with these conditions, many people view it as behavioral (as if the child is anti-social, extremely shy or uncooperative).
- Repetitive, unusual behaviors (having to do things in a certain order, turning switches on and off constantly, gets upset if his or her day is not going in the order they thought it would).
- Unusual eating problems such as eating too much sugar or salt, hoarding food, eating any food too much or not enough food of any kind.
- Stealing
- Lying or telling "tall tales"
- Doesn't seem to respond correctly/appropriately when interacting (seems like he or she doesn't hear you, doesn't understand the question or what to do, low or no response or over reactive.)
- Jumpy
- Gets upset in crowds or when there is a lot of noise
- Impulsivity
- Seems to over-exaggerate aches and pains

Cognitive, learning and attention problems

- Intellectual impairment (formerly called mental retardation)
- Learning disabilities or slow to learn in school
- Difficulty following directions
- Difficulty staying on-task (or tasks take much longer than they should)

Social issues

- Too shy, prefers to be alone or is left out
- Can't make or keep friends
- Fights with other children often
- Is victimized by other children/people
- Too trusting, easily duped or persuaded
- Unusual interactions with others
- Doesn't interact well with people his or her own age

Other physical issues

Oftentimes, syndromes of various kinds can have very specific physical characteristics. There are so many that we can't possibly list them all here. Think about your child's unique physical features and write them down. Some of these may include:

- Joint problems, unusual elbows, feet, knees, hands, hips, arms, mouth
- Unusual facial features (such as eyes set far apart)
- Unusual body shape/type
- Unusual breast development (boys and girls)

Other medical issues

Again, many syndromes have related medical issues. Write down as many medical issues your child has had as possible. Some of these may include:

- Allergies, asthma, constant coughs
- Ear infections
- Heart issues
- Digestive problems of any type
- Seizures, staring spells or just seems to blank out sometimes
- Sleep disturbances of any type, night terrors

Now What?

We recommend that you first consult your primary care physician. We also recommend seeking advice from a **developmental pediatrician, geneticist and genetic counselor**.

When dealing with your school system, consult with the special education director to find out how to obtain an evaluation for your child. If your child has not yet started school, look for the early intervention programs in your area. You can find them through the Department of Education.

Genetic Testing

As a first step, a standard karyotype or chromosomal microarray (cma) can be performed on peripheral blood, amniotic fluid or buccal swab. Fluorescence in Situ Hybridization (FISH) can investigate a larger number of cells to rule out mosaicism.

Once testing is complete, if your son is diagnosed with XYY Syndrome, please contact us for support, programs and services for your son. Email: xyyprojectsupport@genetic.org

For more information on other known Chromosome Variations, please visit the AXYS Website: <https://genetic.org/variations/>

We wish you the best and hope you find an answer to your child's issues.

The XYY Project