47, XYY syndrome

Other names:
- Jacob’s syndrome
- XYY karyotype

Causes & risk factors

47, XYY syndrome is caused by
- An extra copy of the Y
- Each cell has a total of 47 chromosomes instead of the usual 46.

Most cases of 47, XYY syndrome are not inherited.
The disorder usually occurs as a random event during the formation of sperm cells.
- 47, XYY syndrome occurs in about 1 in 1,000 newborn boys.

Symptoms

47, XYY syndrome typically causes
- Often no unusual physical features.
- Most males with this chromosomal change and are able to father children.
- Taller than average height
- Normal Testosterone levels
- Learning problems

Complications

- Increased risk of learning disabilities
- Delayed development of speech and language skills.
- Hand tremors or other involuntary movements (motor tics)
- Behavioral and emotional difficulties
- Autistic spectrum disorders

Treatment

Because 47, XYY syndrome typically has minimal symptoms associated with it, there are no specific treatments for the disorder itself.

REFLECTION QUESTION

What symptoms could make a male think he has 47, XYY syndrome?
Klinefelter syndrome
Other name: 47 X-X-Y syndrome

Causes and risk factors

Klinefelter syndrome is the presence of an extra X chromosome in a male.

• This would be written as XXY.

Klinefelter syndrome occurs in about 1 out of 500 - 1,000 newborn boys.

Women who get pregnant after age 35 are slightly more likely to have a boy with this syndrome than younger women.

Symptoms

• Abnormally long legs
• Abnormally large breasts
• Small testicles
• Tall height

Treatment

Testosterone (male hormone) therapy may be prescribed. This can help:

• Grow body hair
• Improve appearance of muscles
• Improve concentration
• Improve mood and self esteem
• Increase strength

Most men with this syndrome are not able to get a woman pregnant.

Complications

Klinefelter syndrome also increases your risk of:

• ADHD
• Breast cancer in men
• Depression
• Learning disabilities, including dyslexia.

Finding Joy in Misery

Unlike most males, I have an additional chromosome in my genetic make-up -- XXY. Unfortunately, it wasn't uncovered until I was in my early 30s. Had it been discovered earlier testosterone replacement therapy (TRP) would have started at an earlier point and I probably would have avoided many problems in my 20s.

People who develop Klinefelter Syndrome have a tendency later to develop autoimmune diseases and disorders. The autoimmune disorder I have causes pain in joints and connective tissue -- a lot of pain. For some people (like me), the disease progresses to a point where the remissions are only partial and the episodes grow in intensity.

REFLECTION QUESTION

Why can Hormone Replacement Therapy (HRT) be a useful treatment method for individuals with Klinefelter syndrome? Can you think of any downsides to this form of treatment?
XXX Syndrome (Trisomy X)

Other names: Trisomy X, Triple X

Causes and risk factors

- XXX syndrome is caused by the presence of an extra ‘X’ chromosome in every cell.
- Typically, a female has two X chromosomes in every cell of her body.
- The extra ‘X’ chromosome is typically inherited from the mother, but is a random event—not caused by anything she did or could prevent.
- Trisomy X is often not diagnosed until later in life, if ever.
- The extra ‘X’ chromosome occurs in about one in every 1,000 newborn girls.

Symptoms

Many girls and women with Triple X have no signs or symptoms. Signs and symptoms can include:

- Physical:
  - Tall stature (height)
  - Smaller head size
  - Abnormal curving or bending of the pinkies.
- Developmental:
  - Learning disabilities- Normal IQ, but may be 10-15 points below siblings
  - Speech and language delays
  - Poor coordination, awkwardness, clumsiness

Diagnosis & Treatment

- XXX syndrome is either diagnosed prenatally (before birth) or after the child is born.
- These tests look at a person’s chromosomes (karyotype.)
- Treatment depends on what needs the child has. Girls with XXX syndrome may need to be seen by physical, developmental, occupational, or speech therapists if they have developmental or speech problems.

Breaking the News

Today, I told my teachers that I have triple x syndrome. That way they don't have to worry if I'm not doing as well as the rest of the students in my class. I didn't want to tell them personally, so I had my mom write a note for me to give each of them. I also told my best friends. I think they felt bad for me, but I don't think they look at me any differently. Thank goodness. I don't know what I would do without my friends by my side.

I also found out that my tallness isn't from Triple X Syndrome. It's just from my dad's side of the family. That was a good thing to hear! Also, I don't even learn that slowly, so I don't even understand why everyone was worried in the first place. I mean, the doctor told my mom that most girls with it stay undiagnosed with it. I guess I was one of the lucky ones to know I do have it...sike! It's not something I'm proud of, but I decided just to tell my closest friends. And if anybody else finds out, it's not that big of a deal. It's not like I'm a different person or anything!! I'm still the same ole me! :)

REFLECTION QUESTIONS

If you suspected that you had Triple X syndrome, would you want to know for sure? Why?

What tests might the doctor do to confirm a diagnosis?
**Turner syndrome**

Other names: Monosomy X, 45 XO

**Causes and risk factors**

- Humans have 46 chromosomes. Two of these chromosomes, the sex chromosomes, determine if you become a boy or a girl.

- Females normally have two of the same sex chromosomes, written as XX. Males have an X and a Y chromosome (written as XY).

- Most commonly, a female with Turner Syndrome has only one X chromosome.

- Turner syndrome occurs in about 1 out of 2,000 live births.

**Symptoms**

Possible symptoms in young infants include:

- Swollen hands and feet
- Wide and webbed neck

A combination of the following symptoms may be seen in older females:

- Absent or incomplete development at puberty
- Broad, flat chest shaped like a shield
- Drooping eyelids
- Dry eyes
- Infertility (not able to have children)
- No periods (absent menstruation)
- Short height

**Treatment**

Growth hormone may help a child with Turner syndrome grow taller. Estrogen (female hormone) replacement therapy is often started when the girl is 12 or 13 years old.

Women with Turner syndrome who wish to become pregnant may consider using a donor egg.

**Complications**

- Arthritis
- Cataracts
- Diabetes
- Heart defects
- High blood pressure
- Kidney problems
- Middle ear infections

**Brianna’s Story**

I'd like to tell you a little story about my family, and living with Turner Syndrome. Our smallest child is our miracle baby girl. She is our angel sent from above, a gift from God. Brianna was diagnosed with Turner's syndrome while I was still pregnant. In the hospital we noticed our baby didn't want to nurse or eat from a bottle, they took her to the NICU and ran many tests, x-rays, blood work and after 1 wk of being in the hospital she had to go into surgery to have heart surgery, which is common in Turner babies.

My baby was a little fighter, she fought the nurses when they put in her feeding tube! One week after her surgery they found out that she would need a 2nd surgery to fix a diaphragmatic hernia that didn’t let her right lung expand enough for her to breathe on her own. So she was put on this C-pap breathing machine to help her breath. Brianna stayed in the hospital for about a month. I stayed with her the whole time and believe me doctors and nurses would tell me, go home and get some rest. I refused to leave without my baby girl. Now she is three years old and is this healthy happy child, who drives me crazy like any other three year old would. I hope you enjoyed my long story.

**REFLECTION QUESTION**

Which complications do you think are the most severe? Why do you think this?