Easy reader
Klinefelter syndrome
For adults
contents

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Klinefelter syndrome

People are alike and different.

Everybody has some things that work well and some things that do not. In this information material, things that do not work so well are called symptoms.

A group of symptoms taken together is known as a syndrome. Klinefelter syndrome is one.

It is good to have a name for the symptoms. Then the health services can give you better treatment. You can also get help and support from your community.

That is what Doctor Stefan Arver says. He knows lots about Klinefelter syndrome.

What is Klinefelter syndrome?
Klinefelter syndrome only affects boys and men. It is something you are born with.

People with Klinefelter may have many different symptoms. But no one has all the symptoms and they may be more or less severe.
Here are some of the symptoms of Klinefelter in children and teenagers:

Boys with Klinefelter grow taller than other boys of the same age.

About half of all boys with Klinefelter learn to speak later than others.

Some boys with Klinefelter need extra time to learn, for instance,
- how to do new things
- how to read and write.

Many boys with Klinefelter have problems
- with concentration
- doing one thing for a long time
- sitting still.

Just over half of all boys with Klinefelter are also diagnosed with ADHD.

Some have trouble understanding how others think and what they feel. For this reason they may find it difficult to cooperate with others.

Many learn to walk, run or jump later than other children.
People with Klinefelter often have low energy levels. They may find it hard to get started doing something.

**How do you know if a boy has Klinefelter syndrome?**

Boys with Klinefelter syndrome may have many different symptoms. You cannot see from any sign on his body that a boy has Klinefelter syndrome.

If a boy is having any of these problems at school

- language problems
- learning problems
- concentration problems
- attention problems.

then psychologists and doctors can investigate him. He can possibly be diagnosed with Klinefelter syndrome.

Klinefelter syndrome is almost always diagnosed in the teen or adult years.

There are boys and men who have Klinefelter syndrome but never know it because they have so few symptoms.
How is the diagnosis made?
If a doctor suspects that a boy has Klinefelter syndrome, a blood sample may be taken and the boy’s hormones investigated.

Sometimes other tests are also needed to be certain of the diagnosis. Such as investigating the boy’s chromosomes.

The surest sign of Klinefelter syndrome in teenagers and adults is if the testicles are as little as plum pits. Normal testicles are the size of whole plums.

How common is Klinefelter syndrome?
It is thought that at most 90 boys are born with the syndrome every year in Sweden.

Symptoms in adults with Klinefelter syndrome
Men with Klinefelter syndrome have smaller testicles than others.

98 out of 100 men with Klinefelter syndrome have no sperm in their seminal fluid. This makes it difficult for them to become biological fathers.
Men with Klinefelter syndrome often have less body hair and thinner whiskers than others. They may have weak muscles.

Many feel tired a lot of the time and sleep a great deal.

Men with Klinefelter syndrome may also develop other diseases and disorders. These include
- diabetes
- cardiovascular disease
- high blood pressure
- brittle bones
- sleep problems
- joint pain.

Why is it called Klinefelter syndrome?
Harry Klinefelter was a doctor in the United States. As early as 1942, he discovered the syndrome in 8 of his patients.

These patients had
- small testicles
- very little body hair and whiskers
- poorly-developed muscles
- enlarged breasts
- no sperm.
**What causes Klinefelter syndrome?**
The human body is made up of billions of cells. Different cells have different functions.

Some cells develop into the heart, for example, and make it work well. Other cells develop, for instance, into the kidneys, the skin or the brain.

Each cell is so tiny it can barely be seen. Yet each cell is fully programmed. This is called our genetic make-up. Our genetic make-up determines what each cell will become and do.

In every cell in the human body there are 23 pairs of chromosomes. One pair is the sex chromosomes, which are also known as the X and Y chromosomes.

Men have one X and one Y chromosome in every cell. Women have two X chromosomes in every cell.
Sometimes the programming of our genetic make-up goes wrong. When that happens, the person may have a sickness or a syndrome.

Boys with Klinefelter are born with an extra X chromosome or sometimes even with more than one extra X chromosome.

That affects how the testicles develop. In Klinefelter syndrome the testicles stop growing in puberty. Instead of continuing to grow they become smaller.

The testicles produce a male hormone called testosterone. Our hormones govern how our bodies develop.

Testosterone, the male hormone, does a number of important things. Men with Klinefelter often have too little testosterone.

If you have too little testosterone you may
- be tired and depressed
- have low energy levels and very little sex drive
- develop less muscle strength than others.
Men with Klinefelter may need additional testosterone. The doctor can prescribe a testosterone medication.

Testosterone can be given as an injection or applied as a gel.

This treatment is often needed from puberty and on.

**Can men with Klinefelter become fathers?**
The testicles are also where the sperm develop. Men with Klinefelter have fewer sperm than other men. Some have no sperm at all.

Men with Klinefelter can get help from the medical care services when they want to be fathers.

**Can children inherit Klinefelter syndrome?**
It does not appear that children can inherit Klinefelter.

**Do people often have more than one child with Klinefelter syndrome?**
If you have one child with Klinefelter, the risk of having another child with Klinefelter is not increased.
Adults with Klinefelter syndrome need contact with the medical care services

Adults with Klinefelter should have the following checked regularly:

- Blood pressure
- Cardiovascular system
- Lipids
- Bone density
- Testosterone
- Diabetes.

It is good to plan in advance and know what questions you want the doctor to answer.

**Blood pressure**

Many men with Klinefelter syndrome develop high blood pressure when older. This is because the walls of the blood vessels become more rigid with age.

High blood pressure is hard work for the heart so it is important to have your blood pressure checked regularly.

If you have high blood pressure you need to take medication to bring your blood pressure down to normal.
**Cardiovascular system**
Klinefelter syndrome may raise your lipid levels. When this happens, fat sticks to your blood vessels. The blood vessels become very narrow. Then it is difficult for the blood to pass through them.

**Brittle bones**
If your skeleton becomes very weak your bones may break too easily.

Exercise strengthens the skeleton. Sometimes medication is also needed.

**Diabetes**
Men with Klinefelter may develop diabetes. Diabetes means having too much glucose in your blood.

That can lead to other illnesses. It is important to know what lifestyle can help you prevent more illnesses.
Exercise
Exercise strengthens your muscles, your heart and your joints.

Exercise also lowers your stress and helps you sleep better. It is good for you to exercise at least half an hour a day.

You can improve your fitness by taking walks or by taking the stairs instead of the elevator.

Exercise strengthens your muscles. A physiotherapist can help you plan an exercise program.

Choose exercise you enjoy. Then you will be sure to do it.

Dental care is important
It is important to brush your teeth both morning and evening. Always use fluoridated toothpaste.

Go to the dentist often. Dentists give you good advice about taking care of your teeth.
Support and help in the community

People with illnesses and syndromes may need help and support from community services.

You may need help in finding a job or some other occupation.

If you are living on your own, you may also need help to manage things like shopping and cleaning.

There are laws regulating the rights of people with an illness or a syndrome to support and help.

For instance, some help is available under the **Social Services Act (SoL)**.

What determines your right to support is the extent of your difficulties and needs.

**The Health and Medical Care Act (HSL)** may entitle you to
- health care
- habilitation
- personal aids
- transportation to and from the hospital or health centre
You can get help from the habilitation centre in applying for medical and transportation services.

**Interview**
Stefan is 47 years old. He describes what it is like to live with Klinefelter syndrome.

“Things were pretty good at school. Maybe I was ‘lucky’. My mother wanted me to go to a private school. The classes were smaller. Everyone got help.

But sometimes I didn’t understand what the teachers said. Swedish and maths were difficult. English was easier.

Sports were a problem. I knew what I was supposed to do but my body wouldn’t do it.

I moved to the United States at the age of 15. Things were better there. As I said, English was easy.
In the US, the teachers explained until I understood.

I did not think about being different. But I did sleep more than others, day and night.

Also, my breasts grew a little. My muscles did not grow.

I didn’t really have any problems until I grew up. I was working at a restaurant where I had to move between hot and cold spots. My testicles ‘went up into my groin.’

I saw a doctor. He explained that I have Klinefelter syndrome. He also told me I wouldn’t be able to become a biological father.

After seeing that doctor I started getting testosterone shots. They changed my life.

When you have too little testosterone you have no energy.
You don't reach out to others and you don't work on your problems. That gives you poor self-esteem.

Now that I have the right testosterone level I feel much better physically. It was like I had been missing a piece of a puzzle.

Nowadays I understand what I read. I can pay attention. The biggest change was my sex drive. It just exploded!

There is nothing bad about the testosterone except I have some pimples.

A few years after I got my diagnosis I met my present girlfriend. She likes me just the way I am.

After three years she and I wanted to try to start a family. But I knew that I did not have any sperm. So we went to the doctor for help.

We had to use a donor. Another man's sperm
fertilized my girlfriend’s eggs. At first this felt strange. But a counsellor told me it was like seeds. You buy seeds at the store. Then you go home and plant them. You take care of them and water them.

When they sprout and grow you think of them as your plants. I think that is a good comparison with donated sperm.

Using artificial insemination my girlfriend and I first had a son and then a daughter.

I am a trained occupational therapist. My job is to help others to find their way forward in life and at work.”