



Easy reader

## **22q11 - deletion syndrome**

For adults



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## 22q11 - deletion 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** can be called a **syndrome**, like 22q11 - deletion syndrome.

It is good to have a name for the symptoms. That helps the medical services know how to treat them.

You can get help and support from your community.

That is what Doctor Sólveig Óskarsdóttir says, and she knows lots about 22q11.

### **What is 22q11 - deletion syndrome?**

22q11 - deletion syndrome has many names.

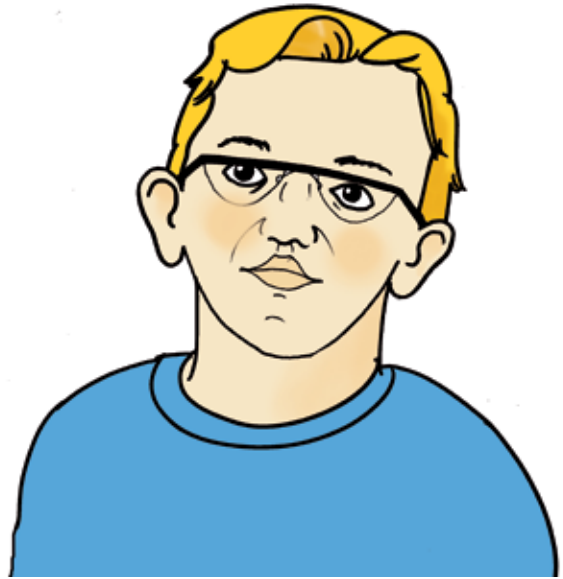
But often people just say 22q11. It used to be called CATCH22.

People with 22q11 may have a number of different symptoms.

The symptoms can be more severe in some people than in others.

Some people may feel almost completely healthy. Others may have lots of serious symptoms.

The symptoms can also vary depending on your age.



It is very unusual for one person to have all the symptoms.

Here are some symptoms typical of people with 22q11:

*Physical symptoms:*

- a heart defect
- calcium deficiency
- a cleft palate
- tooth problems
- impaired vision  
such as astigmatism or squinting
- impaired hearing
- short stature
- a curved spine (scoliosis)
- immune deficiencies
- late gross motor development  
such as balance and strength problems  
or difficulties running and jumping
- slight fine motor weaknesses

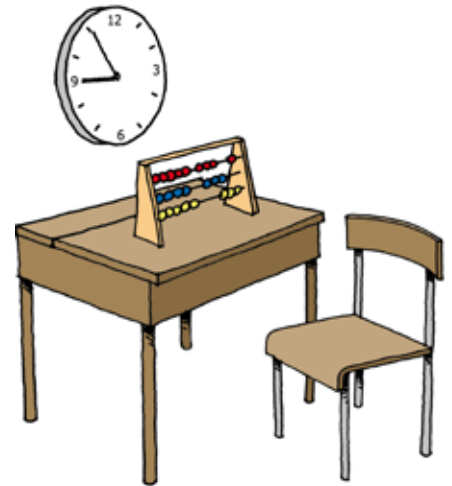


*Learning difficulties:*

About half of everyone with 22q11 have minor intellectual disabilities.

They often have difficulty

- learning new things
- learning to read and write
- learning speech and language
- concentrating



And even people with 22q11 who do not have an intellectual disability may have difficulty

- concentrating
- paying attention
- being patient and persistent

Some people with 22q11 have low energy levels and are underactive. Others may be overactive.

Some are diagnosed with ADHD. These people are overactive and have trouble being patient and paying attention.

*Other symptoms:*

Some people may also have trouble interpreting what they see and hear. Sometimes they need to get information both verbally and in writing.

Many people have trouble understanding how other people think



and what they feel.

This may make it difficult for them  
to co-operate with others.

### **How common is 22q11?**

22q11 is not common.

In Sweden, about 25 children a year  
are born with 22q11.

### **What causes 22q11?**

The human body is made up of  
millions 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.

And yet each cell  
is fully programmed.

This is called our genetic make-up.

The program determines what  
each cell will become and do.

Sometimes the programming goes wrong.

When that happens,  
the person may have a sickness  
or a syndrome,  
such as 22q11.



22q11 are the numbers and a letter  
that show where  
in our genetic make-up  
the problem is located.

### **Can children inherit 22q11?**

Usually, a child born with 22q11 has two  
parents without the syndrome.

Then it is caused by a problem that  
developed in the child's genetic make-up  
at conception.

If one of the parents has 22q11,  
the children can inherit it.

### **When can you see that a child has 22q11?**

Children with 22q11  
have lots of different symptoms.

It is possible to suspect early  
that a child has 22q11 if

- the child has many of the typical  
symptoms
- a heart defect is one of them
- cleft palate is one of them
- the child has delayed and blurry  
speech

But some children do not have  
so many symptoms.  
When those children start school  
it may become clear  
that they have trouble learning.





Then doctors and psychologists can examine the child, who may be diagnosed with 22q11.

Many people never find out that they have 22q11. That happens if they have very few symptoms.

### **How is the diagnosis made?**

If the doctors suspect that a child has 22q11, they may take a special blood sample.



After it is analyzed, they can usually tell if the child has 22q11.

### **Children with 22q11 often have a heart defect**

About half of all children with 22q11 are born with a heart defect.

This defect is usually discovered when the child is just a few days or weeks old.



Heart defects may be serious or slight.

Serious heart defect include interrupted aortic arch, blockage or stoppage in the pulmonary artery, or a hole in the wall separating the chambers of the heart.



Heart defects need to be corrected  
by surgery  
when the child is small.  
Sometimes another operation is needed  
when the child is older.  
After that there are not usually  
any severe heart problems.

### **Cleft palate is common**

Cleft palate and poor palate function  
are common.

Babies with a cleft palate  
may have sucking difficulties.  
And their food may come up  
through their noses.

These children may also have  
unclear speech.



### **Calcium in the blood**

We need calcium to build up  
our skeleton and teeth.

We also need calcium so that our  
muscles and nerves  
will work well.

We have four parathyroid glands  
in our bodies.

They regulate the amount of calcium  
in our blood.

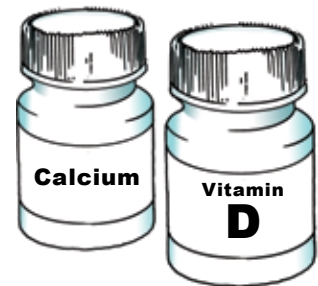
Sometimes these glands do not work as they should.

Sometimes they may be missing.

Then we may have too little calcium in our blood.

Too little calcium in the blood may cause

- tiredness
- weakness, cramps, numbness and prickly feelings in our muscles
- bone pain
- tooth enamel problems



Too little calcium may be treated with active vitamin D and calcium pills.

Infants and adults with 22q11 often have too little calcium in their blood.

Older children and young people seldom have too little calcium.

### **Immune defense problems**

Problems with the immune defense system are common in people with 22q11.

This is often because they have an underdeveloped thymus.





The thymus is an important organ for our immune defense system. Reduced immune defense may result in respiratory infections and ear infections.

When a child with 22q11 starts school, his or her immune defense system usually improves. But some people with 22q11 continue to have immune defense problems as adults.

### **What other symptoms may adults with 22q11 have?**

Adults with 22q11 may have these symptoms:

- headaches
- muscle pain
- joint pain
- thyroid problems
- gluten intolerance
- overweight
- tooth problems
- tiredness
- sensitivity to stress
- anxiety
- depression
- other psychological problems

### **Sometimes adults with 22q11 need treatment**

Many symptoms need to be treated.

It is important that adults with 22q11 go for health checkups. You and your doctor can decide together how often you should have an appointment.

If you feel anxious, sad,  
and have no energy,  
you should make a doctor's appointment.

There are good medicines to treat  
anxiety and depression.

It is also a good idea to be in contact with  
the adult habilitation services.



### **Physical exercise**

Exercise strengthens

- your immune defense system
- your heart
- your lungs

Exercise also helps reduce stress  
and overweight.

It is good for you  
to get some exercise  
for at least half an hour every day.



A gym class, pole walking or swimming  
are good ways of getting exercise.

It is also a good idea to be in contact  
with a physiotherapist.

If you have a heart defect,  
you should talk to your doctor  
about a good level of exercise.

## **Tooth problems**

Adults with 22q11 may have problems with their dental and oral hygiene.

Common problems include

- thin, weak enamel
- missing teeth
- small, narrow and pointed teeth
- too little saliva and dry mouth
- increased risk of cavities



## **Dental care is important**

It is important to brush your teeth both morning and evening.

Always use fluoridated toothpaste.

Adults with 22q11 get cavities more easily than others.

They may need to use chewing gum with added fluoride or fluoride tablets.

If your mouth is dry the risk of cavities increases. That makes it extra important to use fluoride and to drink water both with and after meals.



It is good to go to the dentist regularly. The dentist can give you lots of good advice about how to take good 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.

### **The National Insurance Office (Försäkringskassan)**

works with  
benefits and support for  
people with illnesses or a disability.

Below are a few examples of benefits  
you can apply for  
from the National Insurance Office.

### **Activity compensation**

is for people  
between the ages of 19 and 29  
who have an illness or a disability.

You are eligible for activity compensation  
if you have been unable to work  
for at least a year  
because of a disability.

The size of your activity compensation  
will depend on  
how much you are able to work.

You are also eligible  
for activity compensation  
if you have to extend your schooling  
for extra years  
because of a disability.

This applies both to  
secondary and upper secondary school.





## **Sickness compensation**

is for people  
between 30 and 64  
who have an illness or a disability.

If you will probably never  
be able to work full time  
you may be eligible for  
sickness compensation.

The size of your sickness compensation  
will depend on  
how much you are able to work.

## **Automobile compensation**

People with a disability  
may be eligible for automobile compensation.  
For example,  
if you must have a car  
in order to  
be able to work or study.

You can get automobile compensation,  
for instance to

- buy a car
- have a car adapted to your needs
- take driving lessons

You might need to have your car adapted  
if you are particularly short,  
for example.

## **Occupational aids**

Your employer is responsible for  
adapting the workplace  
to the people who work there.



An employer can receive support for aids  
or for adapting the workplace  
so it is well suited.

If you need aids  
for your life outside the workplace,  
you can get help from  
the health and medical care services.

### **Housing supplement**

If you are receiving  
activity or sickness compensation  
you will also be eligible for  
a housing supplement.  
How much supplement you receive  
depends on how much money you earn and  
how high your rent is.

### **Children with disabilities**

Parents taking care of  
a child with a disability  
can get a childcare allowance  
if the child  
needs special care or looking after  
for at least six months.

You can also get a childcare allowance  
to cover additional costs.  
Additional costs may be,  
for instance,  
if the child needs special food  
or wears out his or her clothes  
very fast.



## Interview

### **Sebastian is an adult with 22q11**

Sebastian is 24 years old.

### **What is it like to be an adult with 22q11?**

I feel like everyone else  
except that I am sometimes in pain.  
Sometimes my muscles hurt.  
Sometimes my muscles are weak.  
When that happens I need a wheelchair.

I need to rest several times a day, too.

I accept my illness.  
That makes things easier.

I also find it easy to talk to people  
and I am not shy.  
That helps me a lot.

I have an occupation I like  
and several other interests.

### **Do you take any medicines?**

I need medicine for my muscle pain.

I also take growth hormone  
to give me more energy.

Another medicine I take helps with my parathyroid function. My medicines make a big difference in how I feel and live.

### **What was your childhood like?**

I was very sick as a child. In seventh and eighth grade I was in the hospital a lot of the time or had to stay home from school.

At school there was a special room where I could rest. I liked school and did well. I was never bullied.

I did not miss out on very much. But it was a struggle to keep my grades up.

I was motivated. I also got valuable help from my parents and the school staff.

My medicines were also important and helped me to manage my schoolwork well.

### **What kind of education do you have?**

I went to an adult education center and studied voice and music.



I liked the adult education center.  
I think that school form is good  
for a lot of people.  
You learn a great deal  
and meet new people.



At the lectures I had an assistant  
who helped me by taking notes  
on what the teachers said.  
Then I could read them afterwards.  
That was very useful.

I also plan to study photography.

### **Do you have a job now?**

I have been working with music  
and photography  
for a few years now.  
I work at a day center  
and get a grant  
from the employment services.



I write poetry and short stories.  
I just entered a story into a contest.  
I also write songs,  
both the lyrics and the music.

Sometimes I perform at  
libraries and cafés.

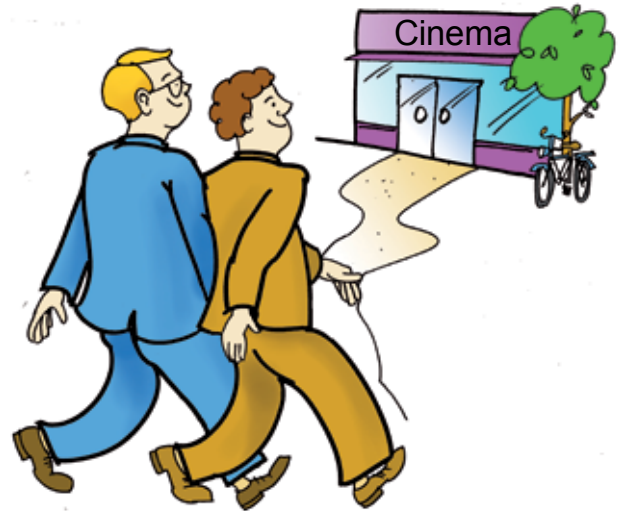
### **What is your living situation?**

I first tried living on my own  
in a practice apartment.  
That was when I was at  
upper secondary school.

Now I have an apartment of my own.  
Today I have a home helper.  
He helps me plan my groceries,  
and also with the cooking, laundry  
and cleaning.

I also have a companion  
to keep me company  
when I want to go  
to the movies  
or do some other activity.

I think I get all the help I need.



### **What problems do you have today?**

I have problems with visual perception,  
how I process what I see.

My joint problems are quite serious.  
They tire me out.  
Sometimes I just don't have any energy,  
either.

I'd like to get a driver's license.  
But I can't.  
It's because of my problems with  
interpreting what I see  
and my weak muscles.

### **What do you think about the future?**

I feel good about the future.  
I hope there will be research progress,  
so I can get even better  
medical care and support.



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