What is an aneuploidy?

Aneuploidy is when someone has 1 or more extra or missing chromosomes. Normally we have 46 chromosomes. They come in pairs of 23. We get 23 chromosomes from each parent. When there are extra or missing chromosomes, it can affect a person's health and development. Below are some of the common conditions that can happen from an euploidy:

Most common conditions:

Trisomy 21 (Down Syndrome)

Trisomy 18 (Edward Syndrome): This causes very serious health problems at birth. Most babies who are born with this do not live past one year.

Trisomy 13 (Patau Syndrome): This causes very serious health problems at birth. Most babies who are born with this do not live past one year.

What is Down syndrome?

- Down syndrome is the most common chromosome condition in humans.
- It is caused by an extra chromosome. People with Down syndrome have an extra copy of chromosome 21. When this happens, it causes the brain and body to develop differently.

What health problems come with Down syndrome?

People with Down syndrome are at a higher risk for having:

- Problems with the heart
- Stomach problems
- Epilepsy
- Trouble with speech, eyesight, or hearing
- Trouble with learning, problem-solving, and social skills
- Some cancers





How can I find out if my child has Down syndrome?

You can find out if your child has Down syndrome before they are born. This can be done with tests like:

- First-trimester screening (early screening that can be done 10-13 weeks into the pregnancy)
- Quad screen (best if screening is done 15-23 weeks into the pregnancy)
- Noninvasive Prenatal Test (NIPT) (test can be done as early as 10 weeks into pregnancy)

Doctors can also tell you if your child has Down syndrome at birth. This is based on what the child looks like (such as having certain facial features).

Is there a cure for Down syndrome?

There is no cure for Down syndrome. Doctors will treat the health problems that occur with Down syndrome as they come up, such as heart problems.

How long does a person with Down syndrome live?

People with Down syndrome are expected to live to be about 60 years old.

Many people with Down syndrome go on to college. Many are able to live on their own and have jobs. Many go on to live fulfilling lives with the help of:

- A good home life
- Support from family and friends
- Good educational programs
- Quality healthcare



Talk to your CHN provider if you have more questions!



