

WHAT IS DOWN SYNDROME?

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Down syndrome is a genetic condition that happens when someone has an extra copy (full or partial) of chromosome 21. Chromosomes are “packages” of genes that determine how the body develops and functions.

As a result of the extra genetic material, Down Syndrome results in:

- Intellectual delays
- Developmental delays
- Distinctive physical features
- The intensification of health issues

What causes Down syndrome?

When a baby is typically developing, these packages of genes (chromosomes) separate as part of cell division, leaving two copies of each chromosome. Down syndrome is the result of a third chromosome remaining instead of the usual two copies. All types of Down syndrome have an extra full or partial chromosome 21.

Why this occurs is still unknown. However, the age of the mother has been linked to an increased chance of having a child with Down syndrome with 49% of children born with Down syndrome born to mothers over the age of 35. Parents who have one child with Down syndrome are also more at risk of having another child with Down syndrome, regardless of their age.

What are some possible related disorders?

A small percentage of people with Down syndrome also have a diagnosis for autism spectrum disorder, anxiety, depression, aggression and social withdrawal.

What are some of the common signs/symptoms of Down syndrome? How is Down syndrome expressed?

People with Down syndrome often present specific physical characteristics, experience developmental delays, and different intellectual ability. However, these signs and symptoms are not universally present and when they do appear, can vary widely among individuals.

Every person with Down syndrome is a unique individual that will most likely look like a family member and may possess one, many or none of these characteristics. Common physical features people with Down syndrome may share:

- Low muscle tone
- Small stature (short)
- An upward slant to the eyes
- A single deep crease across the center of the palm
- A slightly flattened facial profile

In addition, physical development is often slower in children with Down syndrome compared to children without Down syndrome. Due to lower muscle tone, things like turning over, sitting, standing, and walking may take longer to learn. Despite these delays, children with Down syndrome can learn to participate in activities with other children.

Cognitive delays are common in people with Down syndrome and usually range from mild to moderate. Only rarely is Down syndrome associated with severe cognitive impairment.

Common cognitive and behavioural difficulties may include:

- Short attention span
- Poor judgement
- Impulsive behaviour
- Slow learning
- Delayed language and speech development

How common is Down syndrome?

One in every 781 babies born in Canada has Down syndrome. Down syndrome appears equally across racial, gender, and socio-economic groups. There is an estimated 45 000 Canadians with Down syndrome.

Trisomy 21 is the most common type of Down syndrome and presents itself by having 3 full copies of chromosome 21. It accounts for 95% of cases of Down syndrome.

What are some of the risks of having Down syndrome?

Children with Down syndrome reach key developmental milestones later than other children. They also have a higher risk for certain medical conditions. These include:

- Congenital heart defects (when heart, or blood vessels near the heart don't develop typically before birth)
- Gastrointestinal issues (constipation, irritable bowel syndrome (IBS), colon polyps, hemorrhoids etc.)
- Sleep issues (difficulties with onset of sleep, sleep apnea- frequent awakenings during sleep)
- Respiratory infections (pneumonia, COVID-19 etc.)
- Vision and hearing problems
- Increased risk of obtaining childhood leukemia
- Diabetes

Although this list might be intimidating, it is important to be aware of the possible risks and to remember that every person with Down syndrome will have a different experience including health risks.

Do symptoms change over time?

People with Down syndrome will experience symptoms throughout their lives. However, with adequate medical care and support, individuals with Down syndrome can live happy and fulfilling lives in their community.

How is Down syndrome diagnosed?

PRENATALLY

There are 2 kinds of test that may be performed before the baby is born: a screening tests, and a diagnostic tests.

A **screening test** estimates the probability of the baby being born with Down syndrome, but it cannot tell you for certain if your baby will have Down syndrome or not.

A **diagnostic test** provides a diagnosis with almost 100% accuracy. There are 2 prenatal diagnostic tests procedures: **Chronic villus sampling (CVS)** which takes place between week 10 and 14 of gestation, and **amniocentesis** which takes place between weeks 15 and 20 of gestation.

AT BIRTH

When a child is born, certain physical traits can signal the possibility of a Down syndrome diagnosis. A Karyotype (genetic testing) is performed to confirm or deny the diagnosis. A Karyotype is when doctors draw the baby's blood then organize the chromosomes by size, number, and shape. This test is necessary to confirm Down syndrome because these physical traits can be present in babies even without a diagnosis.

How can I support someone with Down syndrome?

There are many different ways to support someone with Down syndrome.

When it comes to case management, education, and strategies, local health professionals may refer the client to social services, community resources, and available rehabilitation services such as:

- Special needs educators (SNE)
- Speech and language pathologists (SLP) – for help with communication
- Occupational therapists (OT) – for help with their development
- Psycho-educators
- Behavior Analysts
- Physiotherapist – for help with motor skills if they have low muscle tone
- Optometrist or hearing specialist – for help with vision and hearing

Speak to a General Practitioner (GP/doctor), health care provider or your local clinic to find out how your child can get these services.

You can also support someone with Down syndrome in his or her day-to-day life. Completing daily tasks/ routines and learning new skills can be more challenging for people with Down syndrome. Each person is different, and the support they require can change with time as children get older.

Daily things that can be done at home include but are not limited to:

- Praising them when they learn something new (e.g. "I love how you washed your dishes, thank you!").
- Speaking clearly and calmly so they can learn from you.
- Playing and singing songs, and reading books together to help with sounds and words.
- Modeling/demonstrating how to do things instead of just giving instructions using words.
- **Asking your health care provider about communication technics such as Singalong, Makaton or PECS.**
- Performing routines the same way every day to keep things predictable.
- Encouraging regular physical activity.
- Looking out for changes in mood or behaviour – someone with Down syndrome might not be able to communicate with their words that something is wrong or that they are unwell.
- Taking them for regular hearing, eyesight, and health checks.

In school, special educational needs support might include things like:

- Extra support or adaptations from a teacher or assistant
- Help taking part in lessons
- Support with things like eating and getting around school

Where can clients and caregivers get more information?

Click on the links:

- [Global Down Syndrome Foundation](#)
- [Canadian Down Syndrome Society: General Information](#)
- [Canadian Down Syndrome Society: Parent support](#)
- [Canadian Down Syndrome Society: Housing](#)
- [Preferred language guide](#)

• This fact sheet is based on the medical model. It is for general information only and does not replace professional medical advice. If provided to clients and their family, it must be reviewed with them to ensure understanding and address any questions or concerns.

• Please keep in mind that a disability is only one part of a person. Identifying and fostering their strengths will be critical for supporting the person to reach their goals.

• For more information and/or support, please visit your local clinic/Community Miyupimaatisiun Centre (CMC) and ask to be connected with a case manager.