CONDITION

Down syndrome

Down syndrome is a lifelong chromosome difference that affects development and may cause certain health problems.

WHAT IT IS
Lifelong developmental condition caused by an extra chromosome 21

BODY SYSTEM
Whole body, especially development, heart, hearing, and thyroid

MAIN SYMPTOMS
Developmental delay, low muscle tone, learning disability, characteristic features

WHAT CAUSES IT
Extra chromosome 21, usually from random cell division

HOW IT IS DIAGNOSED
Prenatal chromosome testing or a postnatal blood karyotype

MAIN TREATMENT
Developmental support, health monitoring, and treatment of associated conditions

CAN IT BE CURED?
No - the chromosome difference remains lifelong

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or sudden weakness

LONG-TERM OUTLOOK
Lifelong condition; many people live fulfilling adult lives

Overview

Down syndrome is a lifelong genetic condition caused by extra genetic material from chromosome 21. Chromosomes are packages of DNA that contain instructions for how the body grows and works.

Most people have two copies of chromosome 21. A person with Down syndrome usually has three, which is why the condition is also called trisomy 21.

The extra genetic material affects physical development, learning, and health, but the effects vary widely. Every person with Down syndrome has their own personality, strengths, needs, and level of independence.

Down syndrome is not an illness someone catches and does not define a person's quality of

life. With suitable health care, education, communication support, and opportunities to participate in community life, many people have active and meaningful lives.

Some people prefer identity-first language, such as "a Down syndrome person," while others prefer "a person with Down syndrome." Respect the language each person uses for themselves.

Symptoms

Features may be noticed during pregnancy or after birth, but physical features alone cannot confirm the diagnosis.

Common early features include:

Children usually reach milestones such as sitting, walking, speaking, and using the toilet later than other children. Timing varies considerably.

Learning disability is expected, but its extent cannot be predicted in infancy. Speech may develop more slowly than understanding. Hearing loss, differences in mouth movement, and low muscle tone can also affect speech.

Emotional health and behaviour vary. Some people also have attention difficulties, autism, anxiety, or depression. New behaviour should not automatically be attributed to Down syndrome: pain, hearing loss, poor sleep, thyroid disease, stress, or another treatable problem may be responsible.

- Low muscle tone, or hypotonia
- A flatter facial profile and upward-slanting eyes
- A short neck, small ears, or a tongue that tends to rest forward
- A single crease across the palm
- A wider space between the first and second toes
- Slower growth and development

Causes and risk factors

Down syndrome begins before birth when a baby receives extra genetic material from chromosome 21. Nothing a parent did during pregnancy causes it.

There are three chromosome patterns:

Most cases result from a random error when an egg or sperm forms. This is called

nondisjunction, meaning the chromosomes did not separate in the usual way.

The chance of an affected pregnancy rises with maternal age, but Down syndrome can occur at any reproductive age. It is not caused by diet, stress, prescribed medicines, or anything either parent could have controlled.

Most cases are not inherited. However, a translocation can sometimes run in a family, even when the parent carrying it does not have Down syndrome. Chromosome testing and genetic counselling can explain the chance in a future pregnancy.

- Trisomy 21: Every tested cell has three separate copies of chromosome 21. This is the most common pattern.
- Translocation Down syndrome: Extra chromosome 21 material is attached to another chromosome.
- Mosaic Down syndrome: Some cells have an extra chromosome 21 and others do not.

Diagnosis

During pregnancy, ultrasound and blood tests, including cell-free DNA screening, can estimate the chance that a fetus has Down syndrome. Screening cannot provide a definite diagnosis, and an increased-chance result does not mean the fetus certainly has the condition.

Prenatal diagnosis requires chromosome testing on cells collected through:

These procedures have benefits and risks that should be discussed with an obstetric professional or genetic counsellor.

After birth, physical features and low muscle tone may suggest Down syndrome. A blood test called a karyotype confirms the diagnosis by examining chromosome number and structure. It can distinguish trisomy 21, translocation, and mosaic Down syndrome.

An initial health assessment usually checks for associated heart, feeding, hearing, vision, and thyroid problems.

- Chorionic villus sampling , which takes a small sample from the placenta
- Amniocentesis , which takes fluid from around the fetus

Treatment and management

No treatment removes the extra chromosome. Care supports development, identifies health problems early, and treats them when needed.

Early support may include physical therapy for movement and strength, occupational therapy for everyday skills, and speech and language therapy. Communication support can include signs, pictures, or electronic devices as well as speech.

School support should reflect the child's learning profile, not only the diagnosis. Some children attend mainstream classes with extra help; others benefit from specialised teaching or a combination of settings.

Health monitoring commonly covers:

Treatment depends on the problem found. A heart defect may need monitoring or surgery. Hearing loss may require hearing devices or other treatment. An underactive thyroid can be treated with thyroid hormone, while obstructive sleep apnoea may require airway treatment or breathing support during sleep.

Routine vaccinations and preventive services remain important. Adults should receive ordinary primary care, reproductive health care, and age-appropriate cancer screening rather than having every concern attributed to Down syndrome.

- Heart health
- Hearing and vision
- Thyroid function
- Growth and nutrition
- Sleep and breathing
- Dental health
- Digestion and bowel habits
- Emotional health, behaviour, and development

Complications

Down syndrome increases the chance of several health conditions, although no one develops all of them.

Possible complications include:

Alzheimer-type brain changes become more common with age, but not every adult develops dementia at the same time or in the same way. Memory loss, reduced daily skills, personality changes, or confusion should be assessed. Depression, thyroid disease, hearing loss, poor sleep, medication effects, and pain can cause similar symptoms.

Upper-spine instability usually causes no symptoms. New neck pain, weakness, walking

changes, loss of bladder or bowel control, or reduced use of the limbs needs prompt assessment, especially after an injury.

- Congenital heart defects
- Hearing loss, repeated ear infections, and vision problems
- Obstructive sleep apnoea
- An underactive thyroid
- Feeding difficulties, constipation, coeliac disease, or bowel abnormalities
- Dental and gum problems
- Seizures
- Certain blood disorders, including a higher risk of leukaemia
- Neck instability involving the upper spine
- Earlier Alzheimer-type changes in the brain

Prevention

Down syndrome cannot usually be prevented. It most often results from a random chromosome event before pregnancy, not an avoidable exposure or behaviour.

Prenatal screening and diagnostic testing provide information but do not prevent the condition. Testing is a personal decision.

If testing shows translocation Down syndrome, genetic counselling may help determine whether either parent carries a balanced translocation and explain what this means for future pregnancies. Counselling should provide information and support without pressuring a family toward a particular decision.

Living with it and what to expect

Down syndrome remains throughout life, but needs and abilities change over time. Development usually continues through childhood and adulthood, even when progress is slower or uneven.

Children may benefit from repeated teaching, clear routines, visual supports, and time to practise. Their interests, relationships, and personalities are as individual as those of other children.

Adolescent planning may cover puberty, personal safety, relationships, consent, education, work skills, and increasing independence. People with Down syndrome need clear, respectful information about bodies, sexuality, and healthy relationships.

Adults may live with family, in supported housing, or more independently. Many participate in paid or volunteer work, sport, education, friendships, and community activities. Support needs range from occasional help to daily assistance.

Women with Down syndrome may be fertile and can become pregnant. Most men have greatly reduced fertility, although rare exceptions occur. Reproductive and sexual health care should be offered directly and respectfully.

Health needs may become more complex with age. Hearing, vision, mobility, sleep, thyroid function, mental health, memory, and daily skills require continued attention. A noticeable loss of existing abilities is not simply an expected part of Down syndrome and should be assessed.

Life expectancy has improved substantially because of better treatment for heart defects, infections, and other conditions. No single lifespan can be predicted; overall health, associated conditions, access to care, and social support all influence long-term outlook.

When to get medical help

Arrange routine follow-up for recommended health monitoring. Make a non-urgent appointment for reduced hearing or vision, persistent constipation, unusual tiredness, weight changes, loud snoring, pauses in breathing during sleep, loss of learned skills, or continuing mood or behaviour changes.

Seek prompt medical advice for new neck pain, weakness, numbness, reduced coordination, or a change in walking, particularly after a fall, collision, or other neck injury.

Get emergency help for:

These are not routine features of Down syndrome and need assessment based on their cause.

- Severe trouble breathing, blue or grey lips, or inability to wake normally
- Fainting with ongoing symptoms or severe chest discomfort
- Sudden weakness, loss of movement, or loss of bladder or bowel control
- A seizure lasting several minutes or repeated seizures without recovery
- Signs of severe dehydration, such as very little urine with marked drowsiness

Common questions

Is Down syndrome inherited?

Usually not. Most cases result from a random chromosome-separation error. Translocation Down syndrome can sometimes be inherited, so chromosome testing may lead to parental testing and genetic counselling.

Can a prenatal blood test tell for sure?

No. Cell-free DNA and other prenatal blood tests are screening tests. They estimate the chance of Down syndrome but cannot confirm it. Chorionic villus sampling or amniocentesis with chromosome analysis is needed for prenatal diagnosis.

Can Down syndrome be cured?

No. The extra chromosome material remains throughout life. Therapies and medical care can support development and treat associated health problems, but they do not remove Down syndrome.

Is Down syndrome a disease?

No. It is a genetic condition. A person may have associated illnesses, such as thyroid disease or a heart defect, but Down syndrome itself is not an infection or a disease acquired later.

Can people with Down syndrome live independently?

Some live independently or with occasional support; others need regular or daily assistance. Independence can include making choices, working, travelling, managing parts of daily life, and maintaining relationships, even when practical support is needed.

Do all adults with Down syndrome develop dementia?

No. The risk of Alzheimer disease is higher and symptoms may appear earlier than in the general population, but timing and effects vary. New memory or behaviour changes need assessment because several treatable conditions can resemble dementia.

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