

Down's Syndrome Awareness

Factsheet

Chromosome 21

Down syndrome is a chromosomal condition that occurs when an error in cell division results in an extra 'copy' of chromosome 21, this is called Trisomy.

Down syndrome can affect a person's cognitive ability and physical growth, cause mild to moderate developmental issues, and present a higher risk of some health problems.

Diagnostic tests

Diagnostic tests can be used to detect Down's syndrome while in the womb. However, they increase the risk of miscarriage, foetal injury, and preterm labour.

Screening tests

Nuchal translucency testing	At 11–14 weeks
Triple screen or quadruple screen	At 15–18 weeks
Integrated screen	A combination of the first two
Cell-free DNA	This is a blood test that analyses fetal DNA
Genetic ultrasound	At 18–20 weeks, combination of all of the above

Characteristics of Down's

- Hypotonia, reduced muscle tone leading to floppiness
- Eyes that slant upwards and outwards
- Palmer crease, may only have one palm crease
- A below average weight and length at birth
- Flat nasal bridge and protruding tongue
- Short stature
- Large space between large and second toe
- Single flexion furrow of the fifth finger

Early development

- Mild to moderate learning disability but very variable
- Developmental milestones are achieved later
- Delay in speech and clarity are common
- Motor skills delayed

Down's syndrome

Down's syndrome or Down syndrome, is a genetic condition that typically causes some level of learning disability and a characteristic range of physical features.



Diagnosis of Down's syndrome predominately occurs after birth.

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Outlook

Children may take longer to acquire skills such as walking and talking, but with stimulation, they can acquire key life skills and attend school and, in some cases, college.

Depending on how the condition affects a person, it is often possible for someone to work and to live semi-independently with Down syndrome.

Later in life....

- Life expectancy is significantly shorter in Downs but it is increasing all of the time
- Higher risk of Alzheimer's and epilepsy

Treatment for Downs

Treatment for Down syndrome varies by individual.

- Speech therapy to improve communication
- Physical therapy to help strengthen muscles and improve motor skills
- Occupational therapy to help refine motor skills and make daily tasks easier
- Behavioural therapy to help manage the emotional challenges that may accompany Downs

Day to day care

With help and support, lots of adults with Down's syndrome can lead an active and fairly independent life including work.

Although it may not be possible to live completely independently, some adults with Down's syndrome leave home and live in their own accommodation with support.

Staff can provide different levels of support depending on the person's particular needs

- An occupational therapist can offer practical advice to help make independent living easier
- Lots of people with Down's syndrome have loving relationships, although they may need some support

Relationships

- People with Down's syndrome tend to have lower fertility, this makes it difficult to have children, but not impossible
- If one partner in a couple has Down's syndrome, there's around a 1 in 2 chance of each of their children having Down's syndrome, too
- The risk of miscarriage and premature birth is also higher in women with Down's syndrome

Birth statistics

1 in 700 pregnancies on average

Higher if mother is over 35

1:1000 if mother is under 30

12:1000 if mother is over 40

Downs development

On average a child with Down's syndrome will.....

- Sit at 11 months
- Crawl at 17 months
- Walk at 26 months

There may also be problems with attention, a tendency to make poor judgments, and impulsive behaviour.