

# Down Syndrome can be Identified during Pregnancy by Prenatal Screening

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## Description

Down syndrome or down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is usually associated with physical growth delays, mild to moderate intellectual disability, and characteristic facial features. The average IQ of a young adult with down syndrome is 50, equivalent to the mental ability of an eight- or nine-year-old child, but this can vary widely. The parents of the affected individual are usually genetically normal. The probability increases from less than 0.1% in 20 year-old mothers to 3% in those of age 45. The extra chromosome is believed to occur by chance, with no known behavioral activity or environmental factor that changes the probability. Down syndrome can be identified during pregnancy by prenatal screening followed by diagnostic testing or after birth by direct observation and genetic testing. Since the introduction of screening, Down syndrome pregnancies are often aborted.

## Chromosome Abnormalities

There is no cure for Down syndrome. Education and proper care have been shown to improve quality of life. Some children with down syndrome are educated in typical school classes, while others require more specialized education. Some individuals with down syndrome graduate from high school, and a few attend post-secondary education. In adulthood, about 20% in the United States do paid work in some capacity, with many requiring a sheltered work environment. Support in financial and legal matters is often needed. Life expectancy is around 50 to 60 years in the developed world with proper health care. Regular screening for health problems common in down syndrome is recommended throughout the person's life. Down syndrome is one of the most common chromosome abnormalities in humans. It occurs in about 1 in 1,000 babies born each year. In 2015, Down syndrome was present in 5.4 million individuals globally and resulted in 27,000 deaths, down from 43,000 deaths in 1990. It is named after British doctor John Langdon Down, who fully described the syndrome in 1866. Some aspects of the condition were described earlier by French psychiatrist Jean-Etienne 1838 and French physician Edouard Seguin in 1844. The genetic cause of Down syndrome was discovered in 1959. People with Down syndrome may have some or all of these physical characteristics: a small chin, slanted

eyes, poor muscle tone and a flat nasal bridge, a single crease of the palm, and a protruding tongue due to a small mouth and relatively large tongue. These airway changes lead to obstructive sleep apnea in around half of those with Down syndrome. Other common features include: a flat and wide face a short neck, excessive joint flexibility, extra space between big toe and second toe, abnormal patterns on the fingertips and short fingers. Instability of the atlantoaxial joint occurs in about 20% and may lead to spinal cord injury. Hip dislocations may occur without trauma in up to a third of people with Down syndrome. Growth in height is slower, resulting in adults who tend to have short stature the average height for men is 154 cm and for women is 142 cm.

Individuals with Down syndrome are at increased risk for obesity as they age. Growth charts have been developed specifically for children with Down syndrome. This syndrome causes about a third of cases of intellectual disability. Many developmental milestones are delayed with the ability to crawl typically occurring around 8 months rather than 5 months and the ability to walk independently typically occurring around 21 months rather than 14 months. Most individuals with Down syndrome have mild or moderate intellectual disability with some cases having severe difficulties. Those with mosaic Down syndrome typically have IQ scores 10-30 points higher than that. As they age, people with Down syndrome typically perform worse than their same-age peers. Commonly, individuals with Down syndrome have better language understanding than ability to speak. Between 10 and 45% has either a stutter or rapid and irregular speech, making it difficult to understand them after reaching 30 years of age, some may lose their ability to speak. They typically do fairly well with social skills. Behaviour problems are not generally as great an issue as in other syndromes associated with intellectual disability. In children with Down syndrome, mental illness occurs in nearly 30% with autism occurring in 5–10%. People with Down syndrome experience a wide range of emotions. While people with Down syndrome are generally happy, symptoms of depression and anxiety may develop in early adulthood. Children and adults with Down syndrome are at increased risk of epileptic seizures, which occur in 5-10% of children and up to 50% of adults. This includes an increased risk of a specific type of seizure called infantile spasms. Many (15%) who live 40 years or longer develop Alzheimer's disease.

## Intellectual Disability

In those who reach 60 years of age, 50-70% has the disease. Hearing and vision disorders occur in more than half of people with Down syndrome. Vision problems occur in 38 to 80%. Between 20 and 50% have strabismus, in which the two eyes do not move together? Cataracts cloudiness of the lens of the eye occurs in 15%, and may be present at birth. Keratoconus a thin, cone-shaped cornea and glaucoma increased eye pressure are also more common, as are refractive errors requiring glasses or contacts. Brush field spots (small white or greyish brown spots on the outer part of the iris are present in 38 to 85% of

individuals. Hearing problems are found in 50-90% of children with Down syndrome. This is often the result of otitis media with effusion which occurs in 50-70% and chronic ear infections which occur in 40 to 60%. Ear infections often begin in the first year of life and are partly due to poor eustachian tube function. Excessive ear wax can also cause hearing loss due to obstruction of the outer ear canal. Even a mild degree of hearing loss can have negative consequences for speech, language understanding, and academics. It is important to rule out hearing loss as a factor in social and cognitive deterioration. Age related hearing loss of the sensor neural type occurs at a much earlier age and affects 10-70% of people with down syndrome.