

What Is Down Syndrome?

According to Mayo Clinic (2024) Down syndrome is a chromosomal condition that occurs when an individual has an additional copy of chromosome 21, a situation most referred to as full trisomy 21. This genetic difference originates from an error in cell division during the formation of reproductive cells and is not caused by environmental or lifestyle factors (NHS, 2023). Mayo Clinic (2024) also states that the presence of extra genetic material influences physical growth, brain development, and overall health across the lifespan. One of the defining features of Down syndrome is intellectual disability, which typically ranges from mild to moderate and may affect learning speed and communication skills. Children with Down syndrome often experience developmental delays, meaning that milestones such as walking and speech are reached later than in typically developing children (NHS, 2023). Individuals with

Down syndrome may also share certain physical characteristics, including reduced muscle tone, although these features are not present in all individuals (Mayo Clinic, 2024). Antonarakis et al (2020) maintains that Down syndrome is associated with an increased likelihood of certain medical conditions, such as congenital heart defects, hearing and vision impairments, and thyroid disorders.

Research has shown that these health outcomes are linked to altered gene expression caused by the additional chromosome (Chapman et al 2024). NHS (2023) suggests that while Down syndrome is a lifelong condition, it is not an illness and does not inevitably determine quality of life. With access to early intervention services, appropriate healthcare and inclusive educational opportunities, many individuals with Down syndrome are able to develop independence and maintain active social lives.

How is Down Syndrome Identified and Diagnosed?

Down syndrome is identified through a structured process that includes prenatal screening and diagnosis as well as postnatal clinical and genetic confirmation.

According to Luo et al (2024), prenatal identification begins with screening methods that estimate the likelihood of trisomy 21 rather than providing a definitive diagnosis. More recently, non-invasive prenatal testing (NIPT) has become widely used due to its high detection rate; this test analyses cell-free fetal DNA in maternal blood. Despite its accuracy, Luo et al (2024) emphasises that NIPT remains a screening tool and cannot confirm Down syndrome on its own. A definitive prenatal diagnosis therefore requires invasive diagnostic procedures such as chorionic villus sampling or amniocentesis. These diagnostic tests confirm the presence of an extra copy of chromosome 21 and are considered the gold standard for prenatal diagnosis.

Wilmot et al (2023) note that Down syndrome may first be suspected at birth based on physical characteristics. Due to clinical features not being sufficient enough for an accurate diagnosis, postnatal genetic testing is required. Standard karyotype analysis is used to identify full trisomy 21, mosaic Down syndrome, or translocation forms, ensuring accurate classification and guiding medical care and genetic counselling.



(Source: Vecteesy, 2022)

Strengths and Limitations of Identification and Diagnosis

Prenatal screening for Down syndrome provides parents with information about the likelihood of their baby having the condition, allowing them to make informed choices while remaining safe for both mother and child (NHS, 2025). However, Down Syndrome UK (2025) point out that many women report receiving insufficient information and emotional support.

Although prenatal screening can identify pregnancies at increased risk, its results are not diagnostic and require confirmation through invasive tests, which carry risks (Lutgendorf and Stoll, 2016). NHS (2025) argue that even though diagnostic tests offer high accuracy, they come with a slight risk of complications, such as miscarriage.

BJM (2025) maintains that clinical evaluations offer a comprehensive understanding of cognitive, physical, and developmental needs, allowing healthcare providers to personalise interventions while supporting family-centered care.

What are the Barriers to Down Syndrome?

According to Down Syndrome UK (2024) and NHS (2023), children with down syndrome will face many barriers including health issues, learning difficulties, educational access and social and communication challenges.

Vasquez et al (2025) highlight that health issues present major barriers for children with Down syndrome due to restricted access to healthcare. Limited specialist services and delayed early intervention reduce children's physical well-being and limit their participation in education and everyday life.

Buckley et al (2024) emphasises that the learning difficulties that children with down syndrome may face are delayed speech, language, and literacy skills. Sakti et al (2025) expresses that these difficulties are compounded by limited teacher training and insufficient resources. Solgrid (2023) discusses that children with Down syndrome often face limited access to education adapted to their needs.

As a result, the barriers that children with down syndrome face can significantly impact these individuals later in life, including lack of employment opportunities, social isolation and feelings of exclusion (Down Syndrome Support, 2025).

What Impact Does Down Syndrome Have on the Child?

Down syndrome impacts a child's development, learning, health, and social experiences. Children may experience delayed milestones, learning and communication difficulties, and increased health needs, which can affect education and social inclusion.

The medical model of disability views disability as an individual condition caused by physical or biological impairment and aims to treat or manage these impairments. This approach has been criticised for failing to consider how social and environmental factors contribute to people's experiences (Burghs et al, 2016). Francis et al (2024) says that the medical model sees disability as a negative condition.

Robertson and Jaswal (2024) suggest that this perspective can negatively affect children with down syndrome by encouraging lower expectations, labelling, and exclusion, which may limit opportunities for participation and reinforce stigma.

Conclusion

In conclusion, Down syndrome is a lifelong genetic condition that can affect physical growth, cognitive development, and social skills. Early identification through screening and diagnostic testing is important for providing timely support.



(Source: Alamy, 2021)

An Appropriate Support Strategy for Children with Down Syndrome

According to Ueda et al (2021) inclusive early intervention refers to the support provided to young children with disabilities within mainstream early childhood settings. This approach involves children learning and participating alongside typically developing peers. EERA (2023) describes inclusive early intervention as a coordinated and individualised process that aligns closely with inclusive education principles. Agrahari et al (2025) highlights that inclusive early intervention supports positive social interactions and promotes a strong sense of belonging while reducing stigma. UNICEF (2025) maintains several challenges associated with inclusive early intervention, including weak early detection systems, limited access to inclusive education, insufficiently trained staff, and ongoing stigma toward children with disabilities.