

Individual Poster Presentation Script

Down Syndrome -

Part 1

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 commonly 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, problem-solving abilities, 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, a flattened facial profile, upward-slanting eyes and small hands and feet, although these features are not present in all individuals and vary widely (Mayo Clinic, 2024). Antonarakis *et al* (2020) maintains that in addition to developmental and physical traits, Down syndrome is associated with an increased likelihood of certain medical conditions, such as congenital heart defects, hearing and vision impairments, thyroid disorders and gastrointestinal abnormalities. Research has shown that these health outcomes are linked to altered gene expression caused by the additional chromosome, which disrupts normal biological pathways involved in neurological, cardiovascular, and immune system development (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, participate in employment, and maintain active social lives. Understanding Down syndrome therefore requires recognition of both its genetic origins and its diverse developmental and social outcomes.

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. These methods include maternal serum screening and ultrasound markers, which are often combined to improve accuracy. 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 to screen for chromosomal abnormalities. 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, which allow direct analysis of fetal chromosomes. These diagnostic tests confirm the presence of an extra copy of chromosome 21 using karyotyping or molecular cytogenetic techniques and are considered the gold standard for prenatal diagnosis.

Wilmot *et al* (2023) focus more broadly on clinical practice and postnatal identification, noting that Down syndrome may first be suspected at birth based on physical characteristics. Common features include hypotonia, distinctive facial features, and single transverse palmar creases, although these signs can vary between individuals. Due to clinical features alone not being sufficient enough for an accurate diagnosis, Wilmot *et al* (2023) also highlight the importance of postnatal genetic testing to confirm the condition. Standard karyotype analysis is used to identify full trisomy 21, mosaic Down syndrome, or translocation forms, ensuring accurate classification. This genetic confirmation is essential for guiding medical care, long-term management, and genetic counselling. Together, the evidence presented by Luo *et al* (2024) and Wilmot *et al* (2023) demonstrates that identification of Down syndrome relies on a combination of screening, diagnostic testing, and clinical evaluation to achieve reliable and timely diagnosis.

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, leaving them unprepared to make fully informed decisions. Moreover, The UK National Screening Committee supports such screening, noting that non-invasive tests can improve accuracy and reduce the need for riskier invasive procedures (GOV.UK, 2019). Although prenatal screening for Down syndrome can identify pregnancies at increased risk, its results are not diagnostic and require confirmation through invasive tests, which carry risks (Lutgendorf and Stoll, 2016). Furthermore, NIH (2023) suggests that diagnostic testing provides a definite diagnosis, nevertheless, NHS (2025) argue that even though diagnostic tests offer high accuracy, they come with a slight risk of complications, such as miscarriage, and may still occasionally be inconclusive. Finally, BJM (2025) maintain that clinical evaluations offer a comprehensive understanding of cognitive,

physical, and developmental needs, allowing healthcare providers to personalise interventions such as early speech and occupational therapy while supporting family-centered care and improving quality of life for individuals with Down syndrome. Although Hendrix (2021) believes that clinical evaluations rely on a multidisciplinary approach and continued research to close scientific gaps are needed to be able to carry this out sufficiently.

Part 2

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.

To illustrate, 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 have made it harder to manage associated health conditions. These systemic barriers reduce children's physical well-being and limit their participation in education, physical activity, 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, insufficient resources, and a lack of personalised teaching approaches. The study highlights the need for tailored and multisensory strategies to support effective learning outcomes. Furthermore, Solgrid (2023) builds on this by discussing that children with Down syndrome often face limited access to education adapted to their needs, which can create challenges in learning as individual support is vital to help them succeed.

In result, the barriers that children with down syndrome face, along with the lack of support they receive can significantly impact these individuals later in life. For example, lack of employment opportunities, difficulty expressing their emotions and social isolation all which may lead to the feeling of exclusion (Down Syndrome Support, 2025).

What Impact Does Down Syndrome Have on the Child?

As previously mentioned, 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, social inclusion, and overall daily life.

Not only this but the medical model of disability views disability as an individual condition caused by physical or biological impairment. It focuses on identifying abnormalities through diagnosis and aims to treat, manage, or correct these impairments using medical intervention. Within this model, disability is seen as a departure from typical functioning, and healthcare professionals are regarded as the key decision-makers. This approach has been criticised for reducing disability to medical issues and 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 needing medical treatment to make individuals "normal." It focuses on the individual and family, overlooking social and environmental factors that contribute to disabling experiences.

Robertson and Jaswal (2024) suggest that this perspective can negatively affect children with down syndrome by encouraging lower expectations, labelling, and exclusion in schools and social settings. By prioritising medical treatment over inclusion, it may limit opportunities for participation, hinder social interaction, and reinforce stigma. As a result, children can experience reduced self-esteem, social isolation, and fewer chances to achieve their potential.

Part 3

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, with intervention strategies built into everyday activities rather than delivered separately. The focus is on meaningful participation, social interaction, and supporting development within natural environments. EERA (2023) also describes inclusive early intervention as a coordinated and individualised process that aligns closely with inclusive education principles. Early intervention is therefore seen as a key part of inclusion, ensuring children have access to appropriate support and reasonable adjustments from an early age. Together, these approaches show that inclusive early intervention combines developmental support with a rights-based approach that promotes belonging and participation.

Inclusive early intervention is widely recognised for its social and emotional benefits for young children. Research published by Agrahari *et al* (2025) highlights that inclusive early intervention supports positive social interactions by allowing children with and without disabilities to learn and play together. This shared participation helps children develop cooperation, communication skills, and peer relationships. The article also emphasises that inclusive early environments promote a strong sense of belonging and reduce stigma by fostering acceptance and

understanding of diversity from an early age. The benefits of inclusive early intervention extend to all children, not only those with disabilities, as inclusive settings encourage empathy and positive classroom cultures. Overall, the research presents inclusive early intervention as an equity-focused approach that supports social development and inclusion for all learners.

UNICEF (2025) maintains several challenges associated with inclusive early intervention. Many children with developmental difficulties are identified too late because early detection systems are weak, fragmented, and poorly coordinated. Specialist services often focus on medical or rehabilitative approaches rather than providing holistic, family-centered support integrated into daily life. In addition, access to high-quality, inclusive early childhood education remains limited for many children due to inaccessible learning environments, insufficiently trained staff, and ongoing stigma or low expectations toward children with disabilities. These factors can reduce the effectiveness of early intervention and limit opportunities for children to fully participate and benefit from inclusive programs.

Conclusion

In conclusion, Down syndrome is a lifelong genetic condition that can affect physical growth, cognitive development, and social skills, with individuals experiencing varying levels of developmental delay and health challenges. Early identification through screening and diagnostic testing is important for providing timely support, although it can sometimes be limited by late detection or the need for invasive procedures. Children with Down syndrome often face barriers in healthcare, education, and social inclusion, which can impact their independence and quality of life. Inclusive early intervention has been shown to support development, social interaction, and participation, while fostering a sense of belonging and acceptance. Despite challenges such as limited access and insufficiently trained staff, providing early and inclusive support is essential to helping children with Down syndrome reach their full potential and actively participate in their communities.

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