

Down Syndrome: What we know!

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Introduction.

Down syndrome is the term used to describe a new-born who has an extra copy of chromosome 21. The additional chromosome increases the likelihood of health problems and is associated with delays in the child's physical and mental development. (Powell-Hamilton, 2022). The developmental patterns of children with Down syndrome are just as diverse as those of children with conventional development. Every person is an individual with a distinct personality and set of interests. In addition to their unique qualities, they also have Down syndrome, which will influence them in varying degrees (Downs Syndrome Association, no date). Since Down syndrome affects numerous systems, it is linked to a variety of clinical disorders. Many signs and symptoms, such as neurological traits, congenital heart defects, distinctive facial features and intellectual and developmental difficulties, are present in these patients (Akhtar, Rizwan and Bokhari, 2023).

Learning Difficulties.

Walking and talking are developmental milestones that are often reached later in life by most children with Down syndrome than by other children. In addition to potentially having issues with verbal memory, expressive communication, and attention span, they commonly have mild to severe intellectual deficiencies (Boston Children's Hospital, no date). Children with Down syndrome exhibit unique patterns of relative strengths and weaknesses and reach a cognitive functioning level comparable to children between the ages of five and eight. Even while new-borns with Down syndrome frequently have early tendencies towards social vocalising, young children with Down syndrome typically exhibit delays in the commencement of spoken first words and more significant difficulties in the development of expressive language than in the acquisition of cognitive skills (Schworer, Fidler and Daunhauer, 2021). Any notable language delay will inevitably result in an increasing cognitive delay because language is such a powerful tool for comprehension, thinking, reasoning, and memory (Down Syndrome Education International, no date).

Hearing Impairment

Children with Down syndrome have a significant prevalence of hearing impairments, according to conventional behavioural testing for hearing. Due mostly to physical abnormalities in the head and neck region, Down syndrome has been linked to a variety of otorhinolaryngologic symptoms (Chin, Khami and Husein, 2014). Individuals with hearing loss may be more likely to experience delayed language and speech acquisition, which could hinder them from realising their full potential (Laws and Hall, 2014). The successful detection of hearing loss existing at birth has been made possible by the establishment of universal neonatal hearing screening. After that, behavioural audiometry is used to examine the auditory systems of young children with Down syndrome (Bull, 2011).

Diagnosis

Up to 90% of pregnancies afflicted by foetal Down syndrome can be detected by conventional prenatal screening procedures (Cuckle and Benn, 2010). Pregnant women have long been offered screening for foetal trisomy 21 (T21) in various countries across the world.

The combination of a blood test and an ultrasound scan is the most accepted screening technique for the first trimester of pregnancy. A blood test enables a medical practitioner to search for potential "markers" of a higher chance of Down syndrome in the mother's blood, such as certain proteins. The doctor then conducts an ultrasound test, which uses high-frequency sound waves to create images. Ultrasonography can sometimes identify Down syndrome in a foetus if there is fluid at the back of its neck. The ultrasound test is called nuchal translucency measurement. (National Institute for Health, 2023).).

Diagnosis tests can provide a clear answer. They examine the fluid or placenta's cells surrounding your child. A diagnostic test can result in miscarriage in 1 out of 200 women (0.5%). Chorionic villus sampling and amniocentesis are the two forms of diagnostic tests (Office for Health Improvement and Disparities, 2022).

The majority of Down syndrome cases are discovered after the baby is delivered because many pregnant parents elect not to have prenatal diagnostics performed. Physicians will typically suspect Down syndrome in cases when specific physical traits are seen, like a flat facial profile, a small nose, an upward tilt to the eyes, and low muscle tone. The physician will do a karyotype test. This test examines the quantity, form, and size of chromosomes in a sample of your body's cells.

Speech and Language Therapy.

It could be suggested that if we want to support the social and cognitive development of children with Down syndrome, speech and language therapy is the most crucial component of intervention services. It is recommended that they receive regular evaluations from a Speech and Language Therapist, who may suggest tailored exercises aimed at fostering their verbal and communicative abilities and enhancing their articulation and flow (Down Syndrome Education International, no date). Downs Syndrome Association (no date) believe encouraging children with Down syndrome to utilise gestures and signs, such as Makaton or Signalong, can greatly improve their comprehension and communication skills. Signs and speech are used concurrently; often by the time a child is five years old, they have stopped using the signs as they develop better at saying the words. Every day, all day, language is learned by children with Down syndrome. In order to fully use all of the language acquisition opportunities that arise during play and daily activities, parents and educators must make the most of the conversations and interactions their children have at home, preschool, and school (Down Syndrome Education International, no date). Using visual supports pictures, written words, encouraging imitation, and repeating words are some strategies that have been found to help children with Down syndrome acquire words and sentences.

Source: Therapy and Wellness Connection (no date)



Conclusion.

The resulting abnormal cell division that results in the genetic disorder known as Down syndrome is an extra complete or partial copy of chromosome 21. This extra genetic material causes the developmental defects and physical traits of Down syndrome (Antonarakis, *et al.*, 2020). There is no one-size-fits-all cure for down syndrome; it is a lifetime illness. Early access to medical care can assist children and infants with Down syndrome become more cognitively and physically capable, enabling them to integrate more easily into society (MedlinePlus, 2020)

Script - What is Down Syndrome?

Down syndrome is the term used to describe a new-born who has an extra copy of chromosome 21. The additional chromosome increases the likelihood of health problems and is associated with delays in the child's physical and mental development. (Powell-Hamilton, 2022). Abukhaled, *et al.*, (2023) agree and state the most prevalent genetic cause of intellectual disability is Down syndrome. It results from the homo sapiens chromosome 21 having three copies as opposed to two. Every child with Down syndrome will struggle with learning to some extent, from moderate to severe. People with Down syndrome will function at the lower end of the average ability range in a tiny percentage of cases. A small number of students will also experience severe and multiple learning challenges. The majority will hover around these two extremes. The developmental patterns of children with Down syndrome are just as diverse as those of children with conventional development. Every person is an individual with a distinct personality and set of interests. In addition to their unique qualities, they also have Down syndrome, which will influence them in varying degrees (Downs Syndrome Association, no date). The most frequent genetic cause of developmental delay is down syndrome, which affects 1 in 700 live new-borns. People with Down syndrome might be of any race or socioeconomic status. If a mother's risk increases with age. 1 in 1250 at age 25, 1 in 1000 at age 30, 1 in 400 at age 35, and around 1 in 100 at age 40. (Minnesota Department for Health, 2024). An additional, vital segment of chromosome 21 is present in all or some of the cells in every person with Down syndrome. It is currently unknown what is causing the additional whole or half chromosome. Age is the only known factor associated with a higher likelihood of having a child with Down syndrome. However, women under the age of 35 give birth to 51% of infants with Down syndrome since these women have greater birth rates. (Graaf, *Buckley and Skotko*, 2022). Since Down syndrome affects numerous systems, it is linked to a variety of clinical disorders. Many signs and symptoms, such as neurological traits, congenital heart defects, distinctive facial features and intellectual and developmental difficulties, are present in these patients (Akhtar, Rizwan and Bokhari, 2023). The life expectancy and infant survival rates of patients with Down syndrome have significantly increased due to recent developments in general care, surgical treatments for the correction of congenital impairments, and medical practice. Akhtar, Rizwan and Bokhari (2023) said 60 years ago, research conducted in Birmingham, revealed that only 40% of infants would survive to be five years old, and only 45% would survive their first year of life. Fifty years later, follow-up research revealed that 78% of patients with Down syndrome and a congenital heart abnormality survived for a year, compared to 96% of patients without the abnormalities. The advancements in medical research should result in a significant increase in these patients' life expectancies. Healthcare facilities want to give these patients appropriate and timely care so they can lead happy and productive lives (Skotko, Davidson and Weintraub, 2013)

Diagnosis.

Up to 90% of pregnancies afflicted by foetal Down syndrome can be detected by conventional prenatal screening procedures (Cuckle and Benn, 2010). Pregnant women have long been offered screening for foetal trisomy 21 (T21) in various countries across the world. The approved programme entails educating women about the condition and their unique risk of T21, followed by the provision of a sensitive screening test to identify a high-risk population (Verwell, de Boer and Oepkes, 2012). This combination of a blood test and an ultrasound scan is the most accepted screening technique for the first trimester of pregnancy. A blood test enables a medical practitioner to search for potential "markers" of a higher chance of Down syndrome in the mother's blood, such as certain proteins. The doctor then conducts an ultrasound test, which uses high-frequency sound waves to create images. Ultrasonography can sometimes identify Down syndrome in a fetus if there is fluid at the back of its neck. The ultrasound test is called nuchal translucency measurement. (National Institute for Health, 2023). However, although there is no risk to the mum or the unborn child from screening tests, it is nevertheless necessary to carefully evaluate whether to get them. Certain screening tests during pregnancy may force parents into making tough choices. For instance, screening Down syndrome can result in challenging judgements regarding the risk of miscarriage associated with diagnostic procedures like amniocentesis (National Health Service, 2021). Diagnosis tests can provide a clear answer. They examine the fluid or placenta's cells surrounding your child. A diagnostic test can result in miscarriage in 1 out of 200 women (0.5%). Chorionic villus sampling and amniocentesis are the two forms of diagnostic tests (Office for Health Improvement and Disparities, 2022). Ghi, *et al.*, (2016) discuss these tests and say amniocentesis and chorionic villus sampling are examples of prenatal diagnostic invasive procedures that are typically carried out to rule out fetal chromosomal or biochemical abnormalities. The danger of procedure-related fetal loss is the primary worry for prenatal invasive testing.

The National Down Syndrome Society (no date) say expectant parents have to take the option that is best for them when it comes to whether to have a prenatal screening or diagnostic test. Some parents would prefer to find out as soon as possible whether their fetus has Down syndrome while others may choose not to have the screening tests. The majority of Down syndrome cases are discovered after the baby is delivered because many pregnant parents elect not to have prenatal diagnostics performed. Physicians will typically suspect Down syndrome in cases when specific physical traits are seen, like a flat facial profile, a small nose, an upward tilt to the eyes, and low muscle tone are some characteristics shared by babies with down syndrome. But not every baby will have these traits, so the physician will do a karyotype test. This test examines the quantity, form, and size of chromosomes in a sample of your body's cells. This test will identify any extra chromosomes or extra fragments of chromosomes, as well as any missing complete or partial chromosomes (National Library of Medicine, 2022),

Barriers to Children with Down Syndrome.

Learning difficulties

Walking and talking are developmental milestones that are often reached later in life by most children with Down syndrome than by other children. In addition to potentially having issues with verbal memory, expressive communication, and attention span, they commonly have mild to severe intellectual deficiencies (Boston Children's Hospital, no date). According to Powell Hamilton (2022), Down syndrome can affect learning in a variety of ways, but it frequently results in mild to moderate intellectual limitation. Children with Down syndrome may need help with self-care activities such as dressing and grooming because of their linguistic and physical impairments. Children with Down syndrome exhibit unique patterns of relative strengths and weaknesses and reach a cognitive functioning level comparable to children between the ages of five and eight. For a very long time, people with Down syndrome have been characterised as having relatively strong visuospatial abilities but rather inferior verbal capabilities (Griego, *et al.*, 2015).

Individuals with Down syndrome frequently experience communication and language challenges, which can affect their ability to participate in social, academic, and community settings. Even while new-borns with Down syndrome frequently have early tendencies towards social vocalising. young children with Down syndrome typically exhibit delays in the commencement of spoken first words and more significant difficulties in the development of expressive language than in the acquisition of cognitive skills (Schworer, Fidler and Daunhauer, 2021). Additionally distinct in their presentation are the social-communicative underpinnings that support language acquisition in Down syndrome. When it comes to social sharing, children with Down syndrome typically use gestures, eye contact, and vocalisation. It is anticipated that children with Down syndrome will show signs of cognitive delay, acquire environmental awareness and information more slowly, and struggle with memory and reasoning. This cognitive delay could be partially the result of the challenges with language acquisition. Any notable language delay will inevitably result in an increasing cognitive delay because language is such a powerful tool for comprehension, thinking, reasoning, and memory. However, the more we can do to support children who struggle with speech and language acquisition, the more equipped they will be to acquire new information and grow cognitively (Down Syndrome Education International, no date).

Hearing Loss

Children with Down syndrome have a significant prevalence of hearing impairments, according to conventional behavioral testing for hearing. Due mostly to physical abnormalities in the head and neck region, Down syndrome has been linked to a variety of otorhinolaryngologic symptoms (Chin, Khami and Husein, 2014). Therefore, it is advised to see an Ears, Nose and Throat (ENT) professional on a regular basis (Barr, *et al.*, 2011). The most frequent ENT symptom associated with this disease is hearing loss (Park, *et al.*, 2014).

Individuals with hearing loss may be more likely to experience delayed language and speech acquisition, which could hinder them from realising their full potential (Laws and Hall, 2014). The successful detection of hearing loss existing at birth has been made possible by the establishment of universal neonatal hearing screening. After that, behavioural audiometry is used to examine the auditory systems of young children with Down syndrome (Bull, 2011). Children with hearing loss often have poorer outcomes in speech, language, schooling, social skills, cognitive capacities, and overall quality of life (Chin, Khami and Husein, 2014). Prognosis and family counselling can be improved by early detection of hearing loss and a knowledge of its aetiology. Furthermore, understanding treatment approaches, such as the many cochlear implant alternatives, assistive devices, and hearing aids, can help guide patient care to maximise outcomes.

Strategy- Speech and Language Therapy.

Treatment, care, and support are provided for children and adults with communication disorders through speech and language therapy (Royal College of Speech and Language Therapists, no date). It could be suggested that if we want to support the social and cognitive development of children with Down syndrome, speech and language therapy is the most crucial component of intervention services. Every chance to promote communication and comprehension must be taken to support and enhance a child's speaking. Furthermore, it is recommended that they receive regular evaluations from a Speech and Language Therapist, who may suggest tailored exercises aimed at fostering their verbal and communicative abilities and enhancing their articulation and flow (Down Syndrome Education International, no date). Downs Syndrome Association (no date) believe encouraging children with Down syndrome to utilise gestures and signs, such Makaton or Signalong, can greatly improve their comprehension and communication skills. Signs and speech are used concurrently; often by the time a child is five years old, they have stopped using the signs as they develop better at saying the words. Zampini, Salvi and D'Odorico (2015) say children diagnosed with Down syndrome have lexical development that is predicted by gestures, and the progression of their preverbal and verbal development can be monitored by gestural production. This can provide as a "bridge" to the start of these utterances and lead to an increase in vocabulary, which can benefit the formation of spoken language. Every day, all day, language is learned by children with Down syndrome. To fully use all of the language acquisition opportunities that arise during play and daily activities, parents and educators must make the most of the conversations and interactions their children have at home, preschool, and school (Down Syndrome Education International, no date). Intensity matters. American research has demonstrated that children who receive intervention sessions five times a week for nine months will make more progress in uttering their first words than those who receive sessions just once a week (Yoder, *et al.*, 2015). As part of a play-based intervention, a therapist encouraged imitation and directly taught words to the children while they played with objects. Effective communication and child-rearing techniques were also taught to parents.

For interventions to have an impact, they need to be of longer duration and increased intensity (Warren, *et al.*, 2020). Repetition and prompting: A recent study examined the methods parents employed to teach their toddlers with Down syndrome new words. The most successful tactics, according to the study, were getting the child to repeat words and encouraging them to pronounce words (Adamson, Bakeman and Brandon, 2015). Using visual supports pictures, written words, encouraging imitation, and repeating words are some strategies that have been found to help children with Down syndrome acquire words and sentences.

Conclusion.

The resulting abnormal cell division that results in the genetic disorder known as Down syndrome is an extra complete or partial copy of chromosome 21. This extra genetic material causes the developmental defects and physical traits of Down syndrome (Antonarakis, *et al.*, 2020). There is no one-size-fits-all cure for down syndrome; it is a lifetime illness. Early access to medical care can assist children and infants with Down syndrome become more cognitively and physically capable, enabling them to integrate more easily into society (MedlinePlus, 2020). Children with Down syndrome and other developmental disabilities may be more vulnerable to social exclusion, and speech and language impairments may make this even worse. Engaging in social activities is crucial for children with various disabilities as it can yield favourable consequences like friendship, emotional growth, support, functional skill development, and belonging. But according to data, those with intellectual disabilities are among the most marginalised social groups (Ali *et al.*, 2012). A child with a handicap has the right, as stated in Article 23 of the UNCRC, to lead a complete and respectable life, free from discrimination, and to participate fully in society (Unicef, no date). The new curriculum, according to the Welsh government (no date), is made for all children. Every child will benefit from its imaginative teachings that have practical applications. They will be able to fulfil their potential and have it meet their own specific requirements.

You have discussed the diagnosis process for down syndrome. What decisions do parents have to make after the diagnosis has been made? Hannah

Mirtabar, *et al.*, (2023) say that in a short amount of time, women must decide whether to take part in the screening in the first place, whether to undergo a diagnostic test if there is an elevated chance of fetal abnormality, and what to do if an abnormality is found. Prenatal screening for fetal anomalies is a sensitive practice because to its association with (selective) abortion. Jones (2020) says abortion is permitted up to birth, and 90% of mothers whose unborn children are identified with Down syndrome opt to have one. A more distressing conversation is hard to conceive for someone who has Down syndrome. In a culture that is actively working to eradicate individuals such as themselves, how can they find their place in society (Dimensions, no date). Abortion is currently illegal in England, Wales, and Scotland after 24 weeks; however, it is permissible until birth if the unborn child has a handicap such as Down syndrome, cleft lip, or club foot. Countries that allow abortions based on a person's impairment have been frequently condemned by the UN Committee on the Rights of Persons with Disabilities. This portion of the Abortion Act, according to the Disability Rights Commission "is insulting to many individuals; it promotes negative perceptions of disability...It is also incompatible with the equal worth of people with and without disabilities (Don't Screen Us Out, 2023).

You have used Down syndrome as a name for your condition. Are there any variations? Chantelle

Trisomy 21 - The most prevalent form of Down syndrome is trisomy 21. A chromosome that has an extra copy is referred to as trisomy. When a fetus develops trisomy 21, it means that every cell contains three copies of chromosome 21 rather than the normal two copies. 96 % percent of cases with Down syndrome are of this type (Cleveland Clinic, no date).

Translocation - A tiny proportion of individuals with Down syndrome (about 3% of cases) have this type of condition. This happens when an additional chromosome 21—either a portion or the entire chromosome—is present but is joined, or “trans-located,” to another

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