DOWN SYNDROME NEW SOUTH WALES

THE Inclusion INSTITUTE

A SHORT HISTORY OF DOWN SYNDROME

FOR PARENTS | FAMILIES | MEDICAL PROFESSIONALS | COMMUNITY

Down syndrome is not a disease, it is a genetic condition. People with Down syndrome are not sufferers, they are people first with diverse talents, strengths, desires and contributions. Join us as we explore the history of Down syndrome, touching on a little biology along the way.

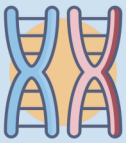
It is important to note that in the representation of the history of Down syndrome, many now outdated and inappropriate names have been associated with Down syndrome in the past. This fact sheet serves merely as a historical overview and in no way condones the use of this language or approach in the present day. We wish to reflect this history to show how far we have rightly come.

A Quick Lesson In Biology

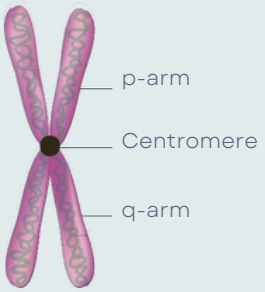
An exciting development of the 20th century was the discover of DNA – or deoxyribonucleic acid – composed of nucleic acids linked with sugars capable of forming long chains and reproducing itself.

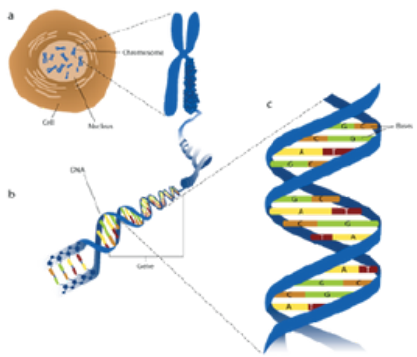
Appearing in human cells as a double stranded helix or spiral, DNA comprises ladder like formations connected by struts of chemical called purines and pyrimidines – namely, adenine and thymidine (purines) and cytosine and guanine (pyrimidines) – the sequencing of which is critical to cell activity and the production of protein. Any disruption in this can lead to issues pertaining to cell structure or cell function, or both. Most DNA within a human cell is concentrated in the cell nucleus, within which chromosomes form a key part.

Humans typically have 23 pairs of chromosomes – 22 of which are identical pairs though varying in size to other pairs, while the 23rd pair of chromosomes determines the sex of an individual, often referred to as X and Y chromosome.



Each chromosome is made up of two strands of DNA – known as a chromatid – joined together by chiamata at a specific site known as the centromere Despite it's name indicating otherwise, this often is not in the centre, resulting in each chromatid having a long arm (referred to as q) and a short arm (referred to as p).





Down syndrome is a result of extra chromosomal material found on chromosome 21, between segments 21q22.1 and 22.3, meaning it is between the first and third parts of the 22nd segment on the long arm of chromosome 21. The whole of the human genome carries over 30,000 genes. The section affected by Down's syndrome accounts for around 50 to 100 genes, a strikingly small amount in comparison to the whole. Of the 50 to 100 genes affected, only a few have been identified to date. The structure and function of cells becomes very specialised as the foetus grows – some cells make up the brain, others the muscles, some the skin and so on. This process is known as differentiation, which is governed by the genetic codes within the cells which science is only beginning to understand. This forms the basis of the commonalities we see across people with Down syndrome, derived from this cell differentiation.

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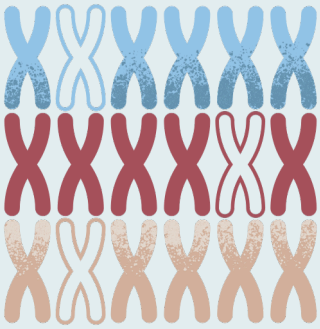
The Identification of Down Syndrome

In 1866, Langdon Down first described the characteristics of the most common cause of learning and intellectual disability. He set the condition apart from mental disability by coining the first term used to describe those with Down syndrome: "mongoloid". In Down's opinion, the children with the genetic condition shared similar characteristics to the people of the Mongolian race. The term "Down syndrome" was suggested later by a group of geneticists who wrote to the British medical journal, The Lancet, with four alternatives to "mongoloid." Down syndrome was later endorsed by the World Health Organization and officially accepted as standard terminology in 1965.

Theories on the causation of Down's syndrome and its impairment mechanisms have varied greatly. Through to 1978, it was asserted that Down syndrome was caused by the foetus adopting radical characteristics during the developmental stages. This theory persisted right up to 1978, as evidenced in "The Psychology of Mongolism". Also during this period, it was also thought that Down syndrome was caused by stress during pregnancy.



Some aspects of the condition were described earlier by French psychiatrist Jean-Étienne Dominique Esquirol in 1838 and by French physician Édouard Séguin in 1844. The genetic cause of Down syndrome was discovered in 1959. In this year, French pediatrician and geneticist Jérôme Leguenge demonstrated that Down's syndrome was in fact due to additional genetic material carried on chromosome 21, identifying it as Trisomy 21. Since this time, the characteristics of Down's syndrome correlate to a relatively small part of the long arm of chromosome 21.

It remains unknown what may predispose cells to carry additional material during the splitting process, how this in turn affects development and function. There is, however, a current state of knowledge that provides some insights in what is an ever growing field of research. To learn more of this, see our fact sheet series.

In 1910, the expected age of survival for children with Down syndrome was nine years. After the discovery of antibiotics, the average age of survival had increased to roughly 19 or 20 years. In 1980, the life expectancy was 25 years old. With recent advancements in clinical treatment such as heart surgery, about 80% of adults with Down syndrome are living to the age of 60 and at times even longer.

A SHORT HISTORY OF DOWN SYNDROME

DOWN SYNDROME REPRESENTED IN RENAISSANCE ART

History depicts many people with Down syndrome, including the Madonna and Child painting by Andrea Mantega data around 1460 depicting a child with characteristics of Down syndrome.

Another painting by Mantegna entitled The Virgin and Child from 1485 depicts similar.

As well as Virgin and Child with Saints Jerome and Louis of Toulouse, dated 1455.

In 1982, Dr. Brian Stratford, a specialist in developmental disabilities at the University of Nottingham, suggested in the journal Maternal and Child Health that the Italian Renaissance painter Andrea Mantegna used a little boy with Down syndrome as the model for his Christ child. Stratford made a “clear characteristic diagnosis” of the baby based on his distinctive facial features and the shape of his hands and toes.

This 16th century Flemish Nativity painting, The Adoration of the Christ Child, may be one of the earliest European depictions of Down syndrome. The angel on Mary's left and possibly the shepherd in the center of the background are identified as having characteristics of Down syndrome including: a flattened mid-face, epicanthal folds, upslanted palpebral fissures, a small and upturned tip of the nose, and downward curving of the corners of the mouth. The hands, crossed over the breast, have short fingers, especially on the left.

According to Levitas and Reid, authors of An angel with Down syndrome in a sixteenth century Flemish Nativity painting, the angelic depiction of an individual with Down syndrome was symbolic and may denote that the artist had an affinity for individuals with disabilities. It also raises questions of the status and value of individuals with special needs in Medieval society. Or perhaps the physical indicators of Down syndrome were not recognised by society as an abnormality.

Dr. Brian Stratford infers that Mantegna and Gonzaga wanted to highlight their shared appreciation of the humanity of children with Down syndrome “Perhaps Mantegna saw in this child something beyond the deficiencies which now so occupy our attention and perhaps then, the qualities of love, forgiveness, gentleness, and innocence were more readily recognized. Maybe Mantegna saw these qualities as more representative of Christ than others we now regard so highly.”

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History shows many depictions of people with Down syndrome from years past:

Down Syndrome NSW is the peak organisation in NSW, proudly representing people with Down syndrome, their families and carers. We offer advocacy, services and supports across the lifespan. We work with passion to ensure that all people with Down syndrome achieve their full potential in all life stages. We champion the rights of people with Down syndrome to be valued and to take their rightful place in the community.