

Characteristics and Causes of Down Syndrome

Jaclyn Brennan

Trinity Christian College

Characteristics and Causes of Down Syndrome

Down Syndrome is a genetic condition in which a child is born with an extra chromosome. According to The National Association of Down Syndrome, Down Syndrome occurs in about 1 in every 792 live births, making it the most common chromosomal disorder. While Trisomy 21 is the most common form of Down Syndrome, Translocation and Mosaicism are also two less common forms. While the factors that cause Down Syndrome are still unknown, there are common characteristics among individuals with Down Syndrome including physical, cognitive, and behavioral features.

There are three main kinds of Down Syndrome: Trisomy 21, Translocation, and Mosaic. While Trisomy 21 is the most common type, they all are caused by the Chromosome 21. According to Anna Kessling and Mary Sawtell of the University of Hertfordshire (2016), Trisomy 21 occurs when there is an unusual cell division in which the mother's egg or father's sperm contains an extra chromosome, chromosome 21. Instead of the normal 46 chromosomes (23 from each parent), a child then has 47 chromosomes. While extensive research has been conducted, there has been no known cause for this cell division and therefore no known factors that result in Down Syndrome. However, there has not been sufficient evidence to support that nationality, diet, medication, or illnesses cause Trisomy 21. The Centers for Disease Control and Prevention (2019) stated that there can be a correlation between the age of the mother and increased risk for Trisomy 21. Mother's 35 and older are more likely to have a child with Trisomy 21, but still majority of mother's to children born with Trisomy 21 are under the age of 35 due to more births among the age group of 35 and younger. This type of Down Syndrome is found in around 94% of people with Down Syndrome.

The next main type of Down Syndrome is Translocation, which is found in about 4% of people with Down Syndrome (Kessling & Sawtell, 2016). Differing from Trisomy 21, people with Translocation do

not have an entire and separate chromosome 21. Instead, they contain an additional part of chromosome 21 attached to another chromosome. This occurs when chromosomes break off and rejoin to a different chromosome. According to the National Association for Down Syndrome (2018), translocation may be inherited, about 1%, due to one of the parent carrying chromosomal material that is arranged in unusual manner. While the process of Translocation and Trisomy 21 are different, they both have no known reason for the occurrence and the features of Down Syndrome are no different (Kessling & Sawtell, 2016).

While this type is the least common, occurring in just under 2% of people with Down Syndrome, the next and final main type of Down Syndrome is Mosaicism. According to the Centers for Disease Control and Prevention (2019), children with mosaic Down Syndrome may have three copies of chromosome 21 but other may also have the normal two of chromosome 21. While mosaic means combination, “children with mosaic Down Syndrome may have the same features as other children with Down Syndrome” (2019) but may have fewer features. Again, there is no known cause for mosaic Down Syndrome, meaning, that there is no real known root cause for any type of Down Syndrome.

While there is no known cause for Down Syndrome, there are, however, many common characteristics among individuals with Down Syndrome. It is important to make note that each person is different. There are physical, developmental and behavioral characteristics that are often prevalent in those with Down Syndrome. The Centers for Disease Control and Prevention (2016) listed many physical characteristics, the most common being a. Again, there are many other physical features and also not all seen within each individual. In terms of development, the National Institute of Child Health and Human Development (2017) found that delayed speech development and ability to walk are common struggles for those with Down Syndrome. They also tend to have challenges with attention span, temper and stubbornness, as well

as impulsivity. Most individuals with Down Syndrome are visual learners and benefit from manipulatives (physical objects), images, and observation.

Along with physical, developmental, and behavioral characteristics, there are also health conditions that are associated with Down Syndrome. Again, it is important to note that each individual is different and not every person with Down Syndrome will have a serious health problem. There are some conditions that occur more commonly among children, with the most prominent being heart defects.

According to the National Institute of Child Health and Human Development (2017), “one-half of babies with Down Syndrome have congenital heart disease (CHD)”. While some are minor and treated through medication, others require surgery. Two other extremely dominant are vision problems, such as cataracts and near-sightedness, and hearing loss. There are many other conditions associated with Down Syndrome, but these were among the most common.

Despite the fact that every 1 in 792 people have Down Syndrome, there is still information that is unknown. Trisomy 21 is the most common type of Down Syndrome, accounting for about 94% of those with Down Syndrome, and may be associated with the age of the mother, but the primary cause is somewhat of a mystery. Common characteristics linked with Down Syndrome are, but not limited to, specific physical features, behaviors such as impulsivity and stubbornness, and associated health conditions. Overall, individuals with Down Syndrome can learn and develop new skills throughout their lives and reach goals and a different pace.

References

- Kessling, A., & Sawtell, M. (2016, May 03). The Genetics of Down's Syndrome. Retrieved July 03, 2020, from <http://www.intellectualdisability.info/diagnosis/articles/the-genetics-of-downs-syndrome>
- Facts about Down Syndrome. (2019, December 05). Retrieved July 03, 2020, from <https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>
- Facts About Down Syndrome. (2018). Retrieved July 03, 2020, from <https://www.nads.org/resources/facts-about-down-syndrome/>
- What are common symptoms of Down syndrome? (2017). Retrieved July 03, 2020, from <https://www.nichd.nih.gov/health/topics/down/conditioninfo/symptoms>
- What conditions or disorders are commonly associated with Down syndrome? (2017). Retrieved July 03, 2020, from <https://www.nichd.nih.gov/health/topics/down/conditioninfo/associated>