

11-29-2020

The Science Behind Down Syndrome

Nicole Haase

Follow this and additional works at: <https://dsc.duq.edu/duquark>

Recommended Citation

Haase, N. (2020). The Science Behind Down Syndrome. *D.U.Quark*, 5 (1). Retrieved from <https://dsc.duq.edu/duquark/vol5/iss1/1>

This Staff Piece is brought to you for free and open access by Duquesne Scholarship Collection. It has been accepted for inclusion in D.U.Quark by an authorized editor of Duquesne Scholarship Collection.

The Science Behind Down Syndrome

By Nicole Haase

D.U. Quark 2020. Volume 5(Issue 1) pgs. 2-5

Published November 29, 2020

Staff Article

Imagine being at your ultrasound appointment (or with someone at theirs'), and the doctor tells you the fetus has Down syndrome. What would be your initial reaction? If this diagnosis scares you, allow me to inform you on the scientific facts, so you fully understand the diagnosis. Important topics to be discussed are the causes of Down syndrome, its three different types, and challenges that may arise from this diagnosis.

Let's first define what Down syndrome is. Down syndrome, also called Trisomy 21, is when an individual has an extra copy of chromosome 21. A mistake in cell division may lead to an individual having 47 instead of 46 chromosomes, and can result in Down syndrome (1).



Intellectual disabilities that a child diagnosed with Down syndrome may experience are as follows: mild to moderate cognitive impairment, delayed language, and an affected short and long-term memory (1). Physical characteristics of a child with Down syndrome include: flattened face, upward slanted almond-shaped eyes, a short neck, small ears, protruding tongue, tiny white spots on iris, small hands and feet, a palmar crease (a single line across the palm of the hand), small pinky fingers that may curve towards the thumb, poor muscle tone or loose joints, and shortened height (1, 2). Some of these symptoms can be seen in the image to the left (3).

The three types of Down syndrome are: Trisomy 21, Translocation Down syndrome, and Mosaic Down syndrome. The most common type is Trisomy 21, where 3 separate copies of chromosome 21 exist in each cell (2). A smaller percentage experience Translocation Down

syndrome, which is when a whole or part of an extra chromosome 21 is relocated and attached to a different chromosome (2). Mosaic Down syndrome is even less common, and is when some cells in an individual have 3 copies of chromosome 21 and some do not (1). With Mosaic Down syndrome, symptoms can be the same as with other types of Down syndrome, but may be less severe due to the individual having more normal cells than trisomy 21 cells (2).

Down syndrome is a random genetic disease, but there are risk factors that can increase the probability of Down syndrome in a fetus. A mother's age can affect her likelihood of having a child with Down syndrome. If 35 years or older, women who become pregnant are more likely to have a pregnancy affected by Down syndrome; however, Down syndrome is more common in pregnancies of younger women because being pregnant at a younger age is more common (4). Overall, approximately 1 in every 700 babies born in the United States each year is born with Down syndrome (5).

During pregnancy, there are two tests that can be performed for Down syndrome detection in the fetus. These are screening and diagnostics tests. A screening test is used to tell a woman and her healthcare provider her risk (having a lower or higher chance) of her baby having Down syndrome (6). Diagnostics tests can detect whether the fetus will have Down syndrome or not (6). Between these two tests, the diagnostics test is more definitive, but riskier for the mother and developing baby than the screening test (6). Oftentimes, a diagnostic test is used after a screening test to confirm a Down syndrome diagnosis (6).

The abnormal cell division involving chromosome 21 is random, and therefore there is nothing you can do prevent having a child with Down syndrome (1). It is expected that someone with Down syndrome can live more than 60 years, depending on severity of health issues (1). Such health complications that are common in people with Down syndrome are: heart defects, gastrointestinal defects, immune disorders, sleep apnea, obesity, spinal problems, leukemia, and dementia (1). Although complications may accompany a Down

syndrome diagnosis, raising a child with Down syndrome is very doable. If you are concerned or at high risk for having a child with Down syndrome, consulting a genetic counselor before getting pregnant would be beneficial (1).

There is a variety of organizations to check out if you have a child with Down syndrome. These include the National Down Syndrome Congress (NDSC), the National Down Syndrome Society (NDSS), and the Global Down Syndrome Foundation (7). The NDSC provides support for those who are raising a child with Down syndrome, and provides information for whoever wants to learn (8). The NDSS and Global Down Syndrome Foundation attempt to improve the lives of people with Down syndrome by also providing education, research, and advocacy programs to those who need it (7). Every state also has their own Early Intervention (EI) program, which consists of services that help and provide support to families with a child who experiences developmental delays (9). The EI program helps children in need, from birth to age 6, grow and develop (9). Thus, there are multiple programs and organizations to help children with Down syndrome and their families.

Works Cited

1. Mayo Clinic Staff. Down syndrome. *Mayo Clinic*.
<https://www.mayoclinic.org/diseases-conditions/down-syndrome/symptoms-causes/syc-20355977>
2. (2019). Birth Defects: Down Syndrome.
<https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>
3. Down syndrome. https://en.wikipedia.org/wiki/Down_syndrome
4. Allen EG, Freeman SB, Druschel C, et al. (2009). Maternal age and risk for trisomy 21 assessed by the origin of chromosome nondisjunction: a report from the Atlanta and National Down Syndrome Projects. *Hum Genet. Human Genetics*; 125(1): 41-52. <https://doi.org/10.1007/s00439-008-0603-8>
5. Mai, C. T., Isenburg, J. L., et al. (2019). National population-based estimates for major birth defects, 2010-2014. *Birth Defects Research*; 111(8): 1420-1435.
<https://doi.org/10.1002/bdr2.1589>
6. (2020). Facts About Down's Syndrome. *Unique Times*.
<https://www.magzter.com/article/Business/UNIQUE-TIMES/FACTS-ABOUT-DOWNS-SYNDROME>
7. Down Syndrome Organizations.
https://www.dsaatl.org/down_syndrome_organizations
8. (2020). National Down Syndrome Congress. <https://www.ndsccenter.org/>
9. Infant and Toddler Early Intervention.
[https://www.co.washington.pa.us/296/Early-Intervention#:~:text=The%20Early%20Intervention%20\(EI\)%20Program,children%20who%20have%20developmental%20delays.](https://www.co.washington.pa.us/296/Early-Intervention#:~:text=The%20Early%20Intervention%20(EI)%20Program,children%20who%20have%20developmental%20delays.)

Haase, N. (2020). The Science Behind Down Syndrome. *D.U.Quark*, 5 (1). Retrieved from <https://dsc.duq.edu/duquark/vol5/iss1/article1>