# Down Syndrome Descriptive Essay

## Abstract

Down Syndrome is a genetical disorder. Before 1862, children born with all or several symptoms such as lower IQ, stunted growth, tongue larger than usual, low muscle tone, slanted eyes, among others fell in the category of mentally disabled. The syndrome often appears in babies born to older parents especially the mother. Screening fetus from ten weeks can reveal whether the pregnancy would lead to a baby with the syndrome or not. Research shows that the condition is not hereditary. Apart from Washington, Down syndrome occurs in all other states as seen in Appendix A. According to Global Disease Burden, the annual death rate of the world's population having Down syndrome was 0.5 per 100,000 people as of 2013. Down syndrome patients need close attention because the condition increases the risk other illnesses to the patient. Although Down syndrome is as old as humankind, the 1866 description of John Langdon shed light into the knowledge of the condition. Down syndrome does not have any form of treatment. A baby diagnosed with the condition will have to live with it until death.

Keywords: Trisomy 21, Chromosomes, Karyotype, Prevalence.

## Table of Contents

[Abstract 2](#_heading=h.gjdgxs)

[Table of Contents 3](#_heading=h.30j0zll)

[List of Tables 4](#_heading=h.1fob9te)

[List of Figures 4](#_heading=h.3znysh7)

[Introduction 5](#_heading=h.2et92p0)

[Analysis of the Epidemiology 6](#_heading=h.3dy6vkm)

[Persons Affected 6](#_heading=h.1t3h5sf)

[Place of Occurrence 9](#_heading=h.lnxbz9)

[Time of the Outbreak 12](#_heading=h.2jxsxqh)

[Conclusion 13](#_heading=h.3j2qqm3)

[References 14](#_heading=h.1y810tw)

[Appendices 15](#_heading=h.4i7ojhp)

[Appendix A 15](#_heading=h.2xcytpi)

[Appendix B 16](#_heading=h.3whwml4)

## List of Tables

[Table 1: Summary of Births with Down Syndrome cases 7](#_heading=h.4d34og8)

[Table 2: Summary of Prevalence by Maternal Age 8](#_heading=h.17dp8vu)

[Table 3: Likelihood of Down Syndrome and Maternal Age 8](#_heading=h.26in1rg)

## List of Figures

[Figure 1: Trisomy Prevalence Rate 7](#_heading=h.2s8eyo1)

[Figure 2: Prevalence by Maternal Age 8](#_heading=h.3rdcrjn)

[Figure 3: Prevalence Rate of Babies per State 9](#_heading=h.35nkun2)

[Figure 4: Prevalence Rate of Maternal Age per State 10](#_heading=h.1ksv4uv)

[Figure 5: Mortality Rate of Down Syndrome per 100,000 People 11](#_heading=h.44sinio)

[Figure 6: Arrangement of Chromosomes in Trisomy 21 (National Association for Down Syndrome) 12](#_heading=h.z337ya)

#

## Down Syndrome

## Introduction

Down Syndrome is a genetical disorder. Before 1862, children born with all or several symptoms such as lower IQ, stunted growth, tongue larger than usual, low muscle tone, slanted eyes, among others fell in the category of mentally disabled. John Down was the English doctor who gave a full description of the condition. It earned the name courtesy of the physician (Wright, 2011). Ordinarily, a human being should have 46 chromosomes. However, during sperm or egg development, the twenty-first chromosome does not separate making it have twenty-four. Consequently, during reproduction, the combination results in 47 chromosomes. Karyotype studies revealed that there are three chromosomes at the 21st, hence the name Trisomy 21. The added genetic material causes an overexpression of genes associated with the 21st chromosome (Zbucka-Kretowska et al., 2017).

The syndrome often appears in babies born to older parents especially the mother. In some cultures, parents killed babies who had such symptoms. With time screening of fetus could reveal some signs of Down Syndrome. That gave mothers the will to decide on whether to keep the pregnancy or terminate it. Ethics guide some parents. Whereas a section believes that abortion is wrong, others take it as a way of stopping a problem before it occurs. The patients do not have a normal life expectancy, in fact, it is between 50 and 60 years in developed countries. That implies that countries that do not have advanced health schemes and facilities would notice them die much earlier. A probable reason is that they are vulnerable to other infections that require close monitoring, which might not be affordable in developing countries. The patients go to school but rarely advance their studies in the U.S.; most of them are partially-independent.

This research project will analyze Down Syndrome by looking at the persons affected, the location of the occurrence, and when the condition broke. It will incorporate statistical analysis of obtained data from several sources as a way of expounding the above issues. The statistical focus will dwell on U.S. states.

## Analysis of the Epidemiology

### Persons Affected

Screening fetus from ten weeks can reveal whether the pregnancy would lead to a baby with the syndrome or not. However, depending on the type of test, the screening might give a false positive. For instance, using ultrasound alongside other tests results in 5% false positive cases while Cell-free fetal DNA contributes to just 0.3% (Sehnert et al., 2011). Trisomy 21 affects people of all race and ethnicity in the U.S. and all over the world (Zbucka-Kretowska et al., 2017). Its detection is in infancy or sometime after birth by examining the physical traits. There is a higher risk of giving birth to a down syndrome baby if one of the parents has it. Men have a lower chance of fathering such babies because most are unable to sire. The most common way of ending up with babies having the condition is from the mother. The likelihood increases with age.

Conversely, research shows that the condition is not hereditary. It occurs due to random cell division even when the fetus develops. That explains why the maternal age of less than 35 is at an elevated risk of giving birth to the babies. Considering that it is of three types apart from Trisomy 21 there is Mosaicism; it happens due to nondisjunction of the twenty-first chromosome, Translocation; it occurs during cell division where a section of the twenty-first chromosome disjoins and merges with another chromosome. Maternal parents having the gene translocation tend to transfer the condition to babies more frequent than paternal parents. Table 1 shows the prevalence rate of Trisomy 21 in 10,000 births for all the States and the Department of Defense between 2010 and 2014 (National Birth Defects Prevention Network). Appendix A shows a detail of the statistics per State.

*Table 1: Summary of Births with Down Syndrome cases*

| **State** | **White, Non-Hispanic** | **Black, Non-Hispanic** | **Hispanic** | **Asian or Pacific, Islander, Non-Hispanic** | **American Indian, Non-Hispanic** |
| --- | --- | --- | --- | --- | --- |
| Department of Defense | 588 | 103 | 84 | 29 | 12 |
| Total of States | 9428 | 2394 | 5375 | 787 | 206 |

Figure 1 shows the prevalence rate as per Table 1.

*Figure 1: Trisomy Prevalence Rate*

Table 2 shows the prevalence by mother’s age in the U.S. between 2010 and 2014.

*Table 2: Summary of Prevalence by Maternal Age*

| State | Below 35 | Over 35 | Total  |
| --- | --- | --- | --- |
| Department of Defense | 506 | 295 | 801 |
| Total of States | 9704 | 8885 | 18589 |
| **Total of U.S.** | **10210** | **9180** | **19390** |

Figure 2 represents the above information on a pie chart showing the prevalence Total of U.S.

*Figure 2: Prevalence by Maternal Age*

The more extended bar of White, Non-Hispanic implies that the White population is higher so is the Hispanic. Therefore, the likelihood of down syndrome occurring also rises. Figure 2 disapproves the premise that the prevalence increases with age. In fact, the percentage is higher in mothers under the age of 35. National Institute of Health gives the contrary of the above. See Table 3 for the comparisons.

*Table 3: Likelihood of Down Syndrome and Maternal Age*

| Age | Likelihood of 1 occurrence in |
| --- | --- |
| 25 | 1,300 |
| 30 | 900 |
| 35 | 350 |
| 42 | 55 |
| 49 | 25 |

### Place of Occurrence

Apart from Washington, Down syndrome occurs in all other states as seen in Appendix A. There is a similarity between the population of babies and the mothers, which justifies the reliability of the data. Figure 3 and Figure 4 show the prevalence rate of babies and maternal age in 10,000 from 43 States respectively.



*Figure 3: Prevalence Rate of Babies per State*



*Figure 4: Prevalence Rate of Maternal Age per State*

From the two plots, Texas has the highest rate followed by New York while Vermont and Hawaii have the least. Once again, it appears so because of the total number of babies born is higher than the rest of states. Washington had a relatively higher live births value but recorded no Down syndrome from 2010 through 2014 the data in Appendix B (National Birth Defects Prevention Network) resulted in Figure 4.

 According to Global Disease Burden, the annual death rate of the world's population having Down syndrome was 0.5 per 100,000 people as of 2013. That said, the annual mortality rate in Africa was at 1.5. that is three times the world’s value. Asia, America, and Europe registered the lowest value of 0.3 per 100,000 people. Similarly, Africa recorded the highest number of years of healthy life lost. It was 137.9 against America’s 42.7 while Asia and Europe recorded 37.7 and 36.3 per 100,000 people. The good news is that the mortality rate has been decreasing since 1990. Figure 5 below shows a cluster bar that gives an overview of the mortality rate.

*Figure 5: Mortality Rate of Down Syndrome per 100,000 People*

The reasons for the elevated rate in Africa compared to the rest of the world are lack of education and improved healthcare. Down syndrome patients need close attention because the condition increases the risk other illnesses to the patient. Often children born with Trisomy 21 develop congenital heart defects, which threatens their lives more. There is no treatment. Therefore, developed countries can avail the necessary health care attention to prolong the patient’s life. Proper healthcare schemes also require adequate finances while some African countries have their citizens living below poverty, which makes constant medical attention to Down syndrome patients a secondary need. Further, there being special programs for patients who exhibit moderate impairments such as special schools, frequent visit to physicians, among others makes the U.S. and other parts of the world have reduced the risk of mortality.

### Time of the Outbreak

Although Down syndrome is as old as humankind, the 1866 description of John Langdon shed light into the knowledge of the condition. Jerome Lejeune gave the world more understanding of the disease in 1959. He discovered that patients with the condition had an added chromosome see Figure 6 (The arrow shows the Trisomy 21) (Wright, 2011). As a condition that appears due to genetical disorders, the exact date, time and season is unknown. Shortly after and the beginning of the 1980s some physicians suggested to parents of babies born with Down syndrome not to undergo surgeries that would correct some atresia conditions. They aimed to eliminate the babies by starving them to death.



*Figure 6: Arrangement of Chromosomes in Trisomy 21 (National Association for Down Syndrome)*

 A typical example that brought a change of perception about babies born with Down syndrome was the 1982 Indiana case. The Baby Doe was born with the syndrome, which manifested esophageal atresia that required correction through surgery. Efforts to convince the physicians, parents and the State’s supreme court could not save the baby (Wright, 2011; Sayeed, 2005). The challenge informed law experts to persuade Congress to legislate against such behavior in future. It is logical to argue that the first time of detection usually ten weeks or more of pregnancy would be the time of the outbreak of the condition. However, as discussed, the tests are not 100% accurate because there is a chance that they could give false negative or positive. As a result, the surest proof would be between birth and a few years after birth by testing the DNA of the child.

## Conclusion

 Down syndrome does not have any form of treatment. A baby diagnosed with the condition will have to live with it until death. What parents should do is to offer all the necessary physical, emotional, and medical support to the child as he or she grows. Where the child will need particular attention, they should allow then undertake the special education. On the other hand, when the child can go to normal schools because the condition is mild, the parents should assist them especially now that the federal law protects such children against exclusion. The mortality rate has significantly decreased thanks to the improved medical facilities and attention to the people with the condition. One fact that turns out is that the condition begins during fertilization or cell division. There is a need for further research to find out why maternal age below 35 has a higher prevalence rate unlike expected rate increase with an increase in age. Moreover, Washington State could act as a reference while conducting further research.

## References

Global Disease Burden. Down syndrome in Africa: Statistics on overall impact and specific effect on demographic groups. Retrieved from http://global-disease- burden.healthgrove.com/l/79921/Down-Syndrome-in-Africa

National Association for Down Syndrome. Facts about Down syndrome. Retrieved from http://www.nads.org/resources/facts-about-down-syndrome/

National Birth Defects Prevention Network. Birth defects and data directory. Retrieved from https://www.nbdpn.org/docs/bdr21145-sup-0003-suppinfo03.pdf

National Institute of Health. How many people are affected by or at risk for Down syndrome. Retrieved from https://www.nichd.nih.gov/health/topics/down/conditioninfo/Pages/risk.aspx

Sayeed, S. (2005). Baby Doe redux? The department of health and human services and the born- alive infants protection act of 2002: A cautionary note on normative neonatal practice. *Pediatrics, 116*(4). 576-585.

Sehnert, A. J., Rhees, B., Comstock, D., Feo, E., Heilek, G., & Rava, R. (2011). Optimal detection of fetal chromosomal abnormalities by massively parallel sequencing of Cell- Free fetal DNA from maternal blood. *Clinical Chemistry, 57*(7). 1024-1049.

Wright, D. (2011). *Downs: The history of a disability*. Oxford: OUP Oxford.

Zbucka-Kretowska, M., Charkiewicz, K., Goscik, J., Wolczynski, S., & Laudanski, P. (2017). Maternal plasma angiogenic and inflammatory factor profiling in foetal Down syndrome. *PLOS ONE, 12*(12): e0189762. doi: 10.1371/journal.pone.0189762

## Appendices

### Appendix A

| State | White, Non-Hispanic | Black, Non-Hispanic | Hispanic | Asian or Pacific, Islander, Non-Hispanic | American Indian, Non-Hispanic |
| --- | --- | --- | --- | --- | --- |
| Alaska | 38 | <6 | 0 | <6 | 63 |
| Arizona | 181 | 17 | 190 | 16 | 33 |
| Arkansas | 136 | 36 | 24 | 4 | 0 |
| California | 103 | 22 | 325 | 27 | 0 |
| Colorado | 273 | 31 | 181 | 15 | 4 |
| Delaware | 44 | 17 | 12 | 5 | 0 |
| Florida | 640 | 295 | 405 | 54 | <5 |
| Georgia | 128 | 119 | 76 | 24 | 1 |
| Hawaii | 5 | 0 | 2 | 14 | 0 |
| Illinois | 549 | 129 | 352 | 48 | 3 |
| Indiana | 282 | 37 | 27 | 6 | 0 |
| Iowa | 214 | 15 | 20 | 4 | 0 |
| Kansas | 125 | 9 | 46 | 11 | <5 |
| Kentucky | 261 | 28 | 19 | 6 | 1 |
| Louisiana | 130 | 61 | 27 | <5 | 0 |
| Maine | 68 | 4 | 2 | 2 | 0 |
| Maryland | 132 | 98 | 69 | 12 | 0 |
| Massachusetts | 520 | 70 | 128 | 52 | 0 |
| Michigan | 405 | 104 | 31 | 21 | 0 |
| Minnesota | 121 | 49 | 29 | 23 | 2 |
| Mississippi | 72 | 60 | 3 | 1 | 3 |
| Missouri | 348 | 76 | 39 | 10 | 2 |
| Nebraska | 179 | 5 | 13 | 6 | 1 |
| Nevada | 73 | 17 | 104 | 11 | 2 |
| New Jersey | 243 | 92 | 230 | 34 | 2 |
| New Mexico | 45 | 4 | 102 | 1 | 20 |
| New York | 698 | 262 | 363 | 144 | 2 |
| North Carolina | 447 | 134 | 150 | 24 | 15 |
| North Dakota | 42 | 0 | 2 | 3 | 6 |
| Oklahoma | 198 | 26 | 76 | 11 | 28 |
| Oregon | 266 | 11 | 110 | 17 | 7 |
| Puerto Rico | - | - | 251 | - | - |
| Rhode Island | 49 | 7 | 17 | 0 | 1 |
| South Carolina | 195 | 69 | 48 | 7 | 0 |
| Tennessee | 387 | 104 | 75 | 10 | 2 |
| Texas | 857 | 226 | 1583 | 93 | 4 |
| Utah | 307 | 4 | 78 | 18 | 2 |
| Vermont | 31 | 0 | 0 | 1 | 0 |
| Virginia | 318 | 130 | 119 | 31 | 0 |
| Washington | - | - | - | - | - |
| West Virginia | 58 | 3 | 1 | 0 | 0 |
| Wisconsin | 260 | 23 | 46 | 21 | 2 |
| Department of Defense | 588 | 103 | 84 | 29 | 12 |
| **Total of States** | **9428** | **2394** | **5375** | **787** | **206** |

### Appendix B

| State | Below 35 | Over 35 | Total  |
| --- | --- | --- | --- |
| Alaska | 37 | 26 | 63 |
| Arizona | 241 | 203 | 444 |
| Arkansas | 128 | 75 | 203 |
| California | 235 | 272 | 507 |
| Colorado | 254 | 257 | 511 |
| Delaware | 40 | 39 | 79 |
| Florida | 731 | 712 | 1443 |
| Georgia | 174 | 213 | 387 |
| Hawaii | 13 | 16 | 29 |
| Illinois | 493 | 566 | 1059 |
| Indiana | 224 | 138 | 362 |
| Iowa | 156 | 109 | 265 |
| Kansas | 117 | 87 | 204 |
| Kentucky | 194 | 122 | 316 |
| Louisiana | 143 | 83 | 226 |
| Maine | 50 | 29 | 79 |
| Maryland | 171 | 164 | 335 |
| Massachusetts | 316 | 507 | 823 |
| Michigan | 332 | 245 | 577 |
| Minnesota | 128 | 97 | 225 |
| Mississippi | 82 | 67 | 149 |
| Missouri | 286 | 205 | 491 |
| Nebraska | 136 | 102 | 238 |
| Nevada | 99 | 83 | 182 |
| New Jersey | 263 | 332 | 595 |
| New Mexico | 112 | 67 | 179 |
| New York | 727 | 768 | 1495 |
| North Carolina | 405 | 393 | 798 |
| North Dakota | 36 | 20 | 56 |
| Oklahoma | 206 | 135 | 341 |
| Oregon | 244 | 182 | 426 |
| Puerto Rico | 145 | 105 | 250 |
| Rhode Island | 35 | 43 | 78 |
| South Carolina | 187 | 150 | 337 |
| Tennessee | 346 | 228 | 574 |
| Texas | 1457 | 1337 | 2794 |
| Utah | 219 | 198 | 417 |
| Vermont | 18 | 13 | 31 |
| Virginia | 301 | 303 | 604 |
| Washington | - | - | - |
| West Virginia | 44 | 19 | 63 |
| Wisconsin | 179 | 175 | 354 |
| Department of Defense | 506 | 295 | 801 |
| **Total of States** | **9704** | **8885** | **18589** |