

Patau syndrome essay essay sample



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Patau syndrome is a disorder that occurs due to the appearance of a person's thirteen chromosome three times (trisomy 13) instead of only twice in the cell. In some cases of the disorder, only a select percentage of the cells display the third thirteen chromosome while others contain the average amount of pairs, this is known as mosaicism. The extra chromosomal material in the cell can lead to many problems in the developmental stages of the humans life, these developmental issues cause severe mental retardation and physical defects or abnormalities in other body parts of that person. Patau syndrome occurs in approximately one out of every ten-thousand to sixteen thousand infants born. The disorder is primarily not an inherited trait, being caused by random events in the formation of the sperm and egg of healthy parents.

Because of the many severe defects entailed to babies born with the disease, many do not survive past the first days or weeks of their lives; only five to ten percent of babies born with the disease live through the first year of their life. Many babies born with Patau syndrome have small or poorly developed eyes, heart defects, brain or spinal cord abnormalities, a cleft lip or palate, close-set eyes, and many other defects. Infants born with the disorder also have a impeded rate of growth due to problems feeding.

Treatment for Patau syndrome is relevant due to the severity of the defects in the infant. Surgical procedures can be performed, but are usually done after the baby's first few months of life due to the high mortality rate of effected infants. Patau syndrome can be spotted post-birth through X-rays showing the placement of the heart further to the right of their chest than normal, or a CT or MRI can be performed displaying the structure of the

infant's brain. In most cases of Patau syndrome, either side of the baby's brain will be fused together (holoprosencephaly).