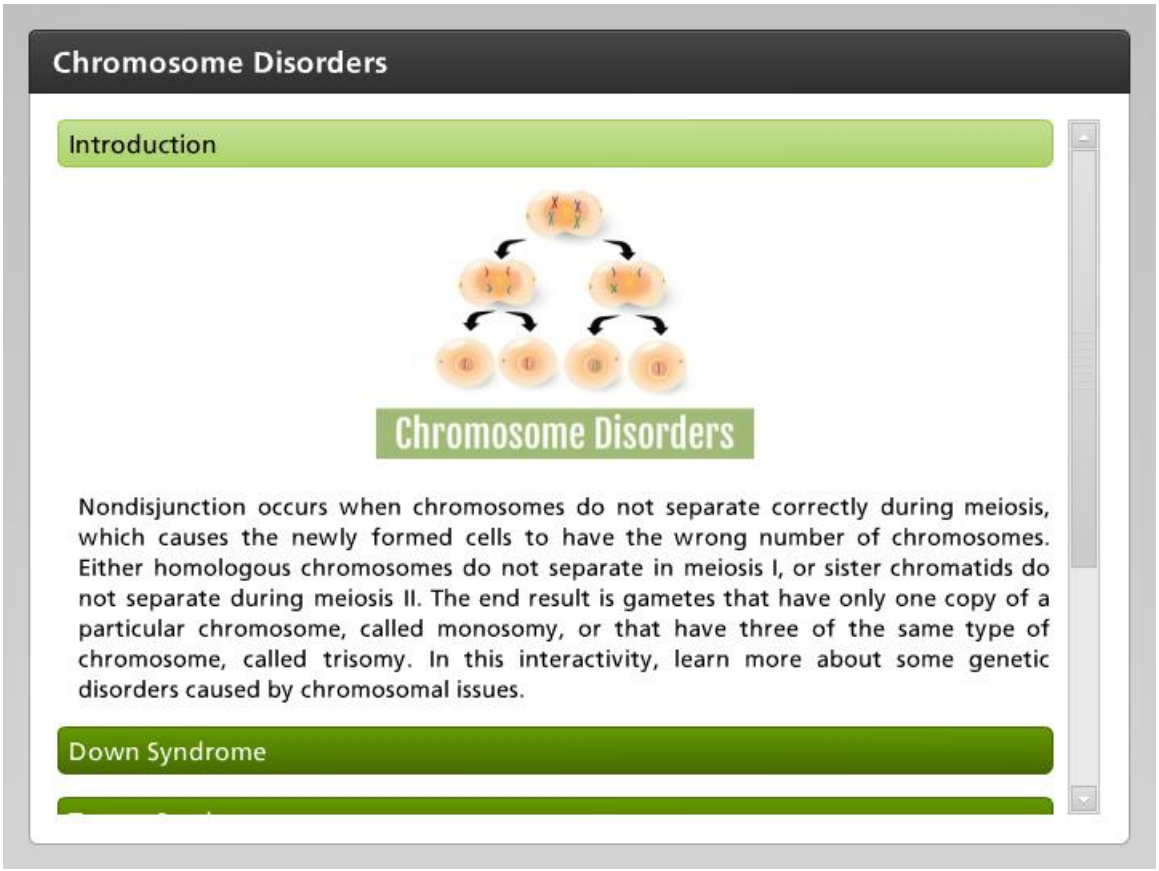
Module 5: Mendelian Genetics and Genetic Disorders
Topic 3 Content: Chromosome Disorders Notes

Introduction



Nondisjunction occurs when chromosomes do not separate correctly during meiosis, which causes the newly formed cells to have the wrong number of chromosomes. Either homologous chromosomes do not separate in meiosis I, or sister chromatids do not separate during meiosis II. The end result is gametes that have only one copy of a particular chromosome, called monosomy, or that have three of the same type of chromosome, called trisomy. In this interactivity, learn more about some genetic disorders caused by chromosomal issues.

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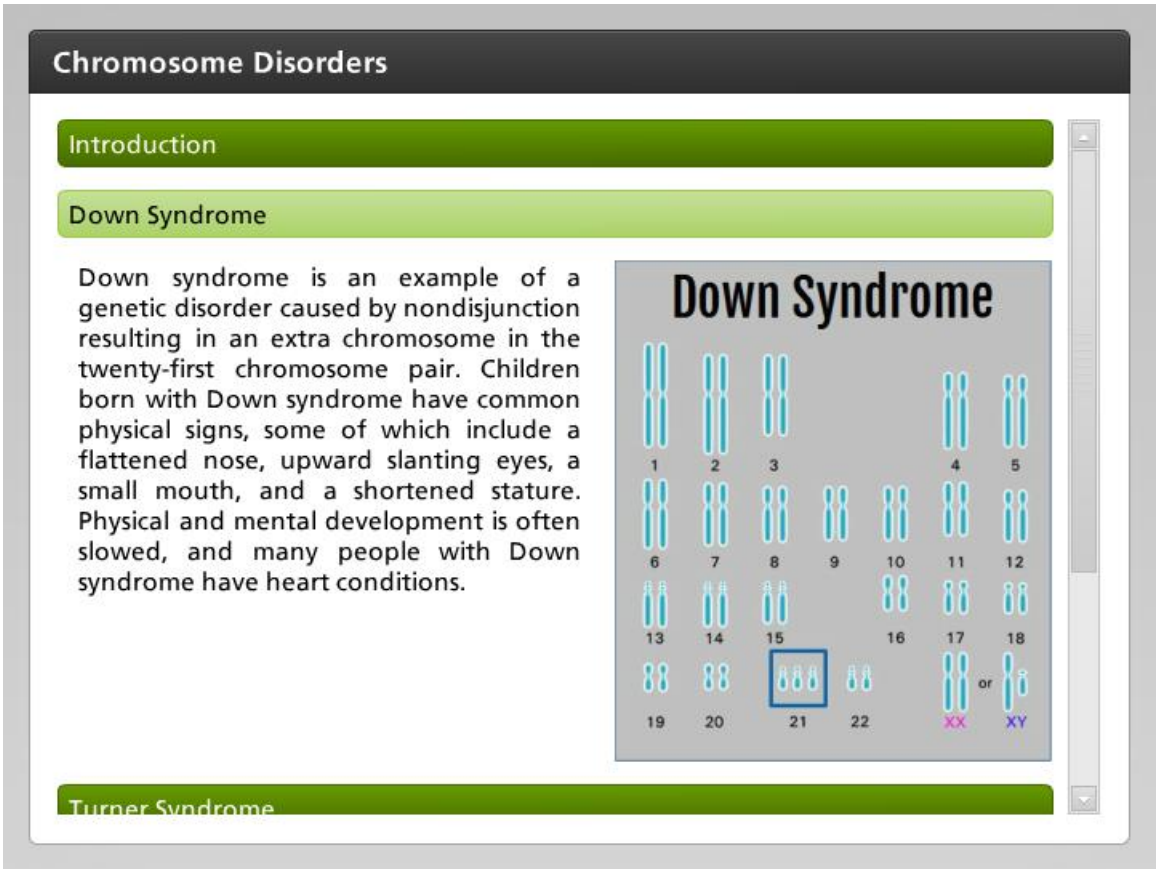
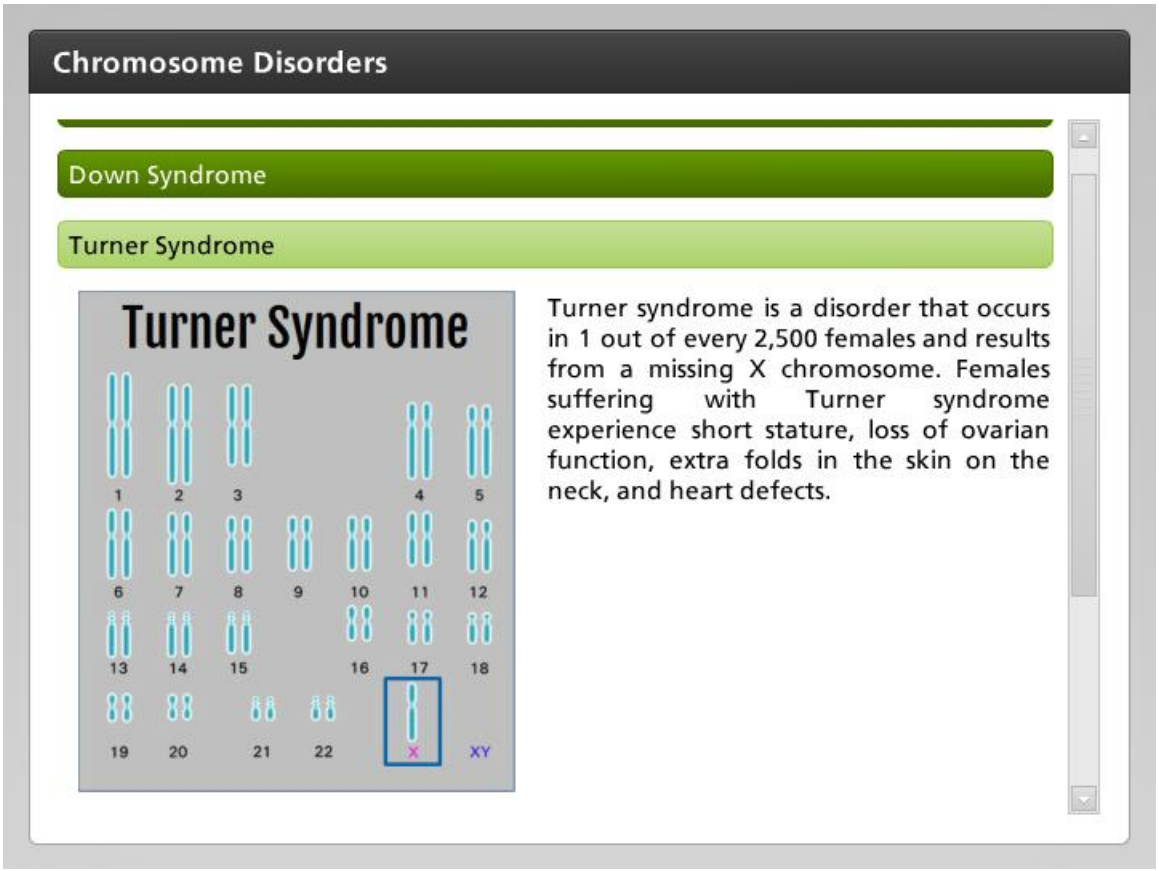
Down Syndrome



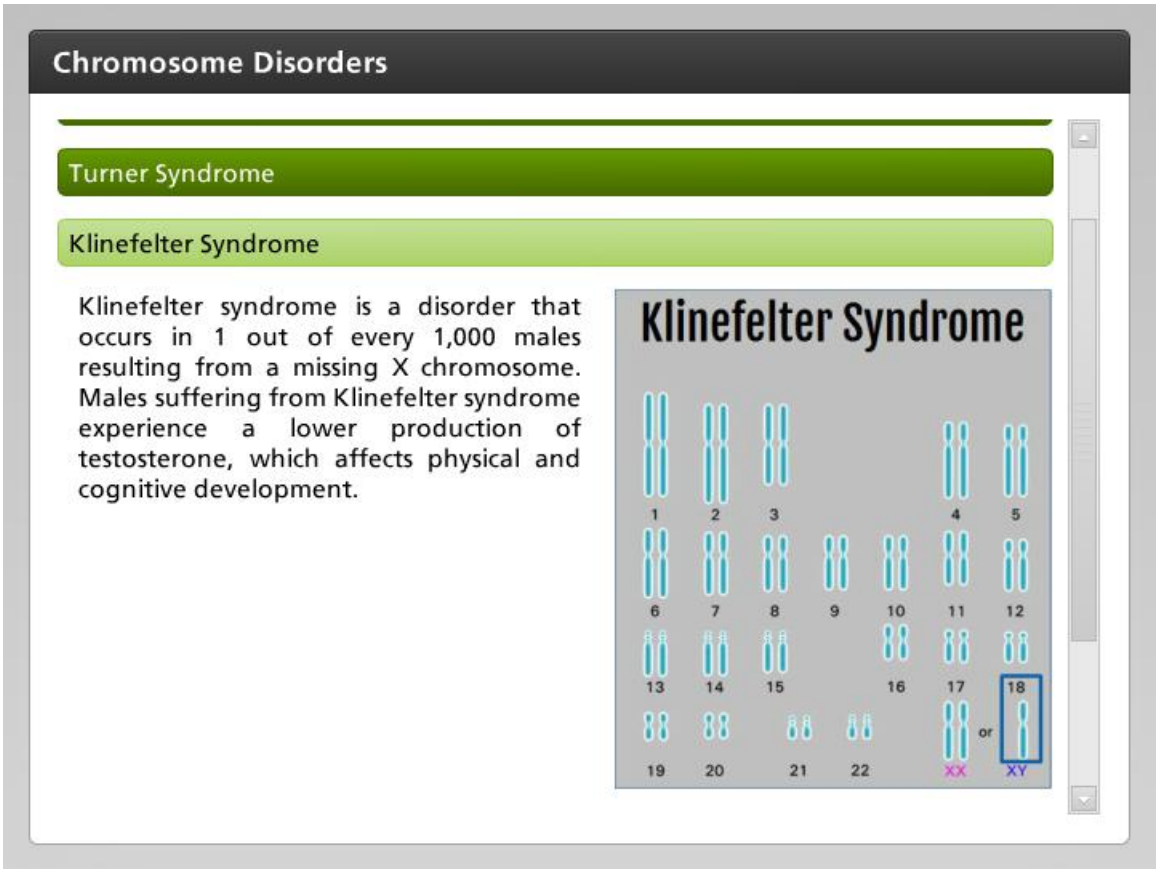
Down syndrome is an example of a genetic disorder caused by nondisjunction resulting in an extra chromosome in the twenty-first chromosome pair. Children born with Down syndrome have common physical signs, some of which include a flattened nose, upward slanting eyes, a small mouth, and a shortened stature. Physical and mental development is often slowed, and many people with Down syndrome have heart conditions.

Turner Syndrome



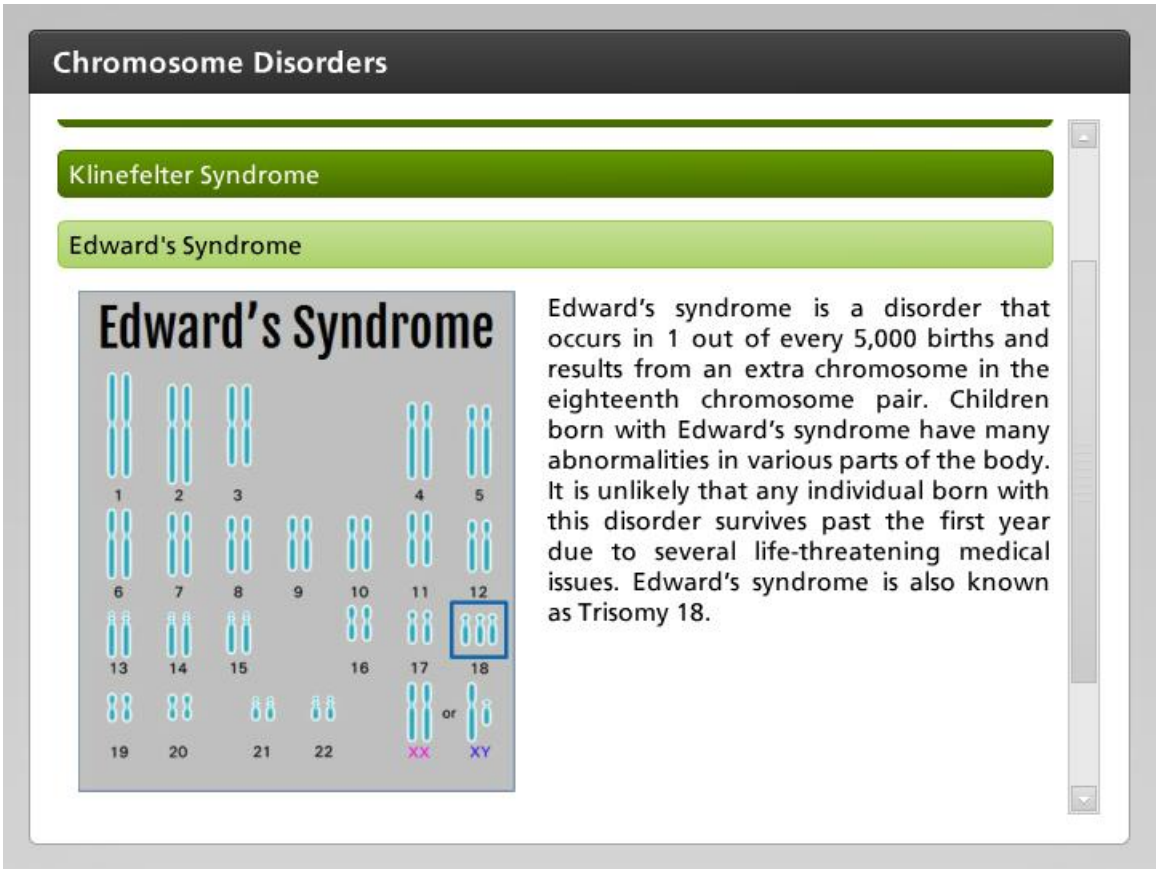
Turner syndrome is a disorder that occurs in 1 out of every 2,500 females and results from a missing X chromosome. Females suffering with Turner syndrome experience short stature, loss of ovarian function, extra folds in the skin on the neck, and heart defects.

Klinefelter Syndrome



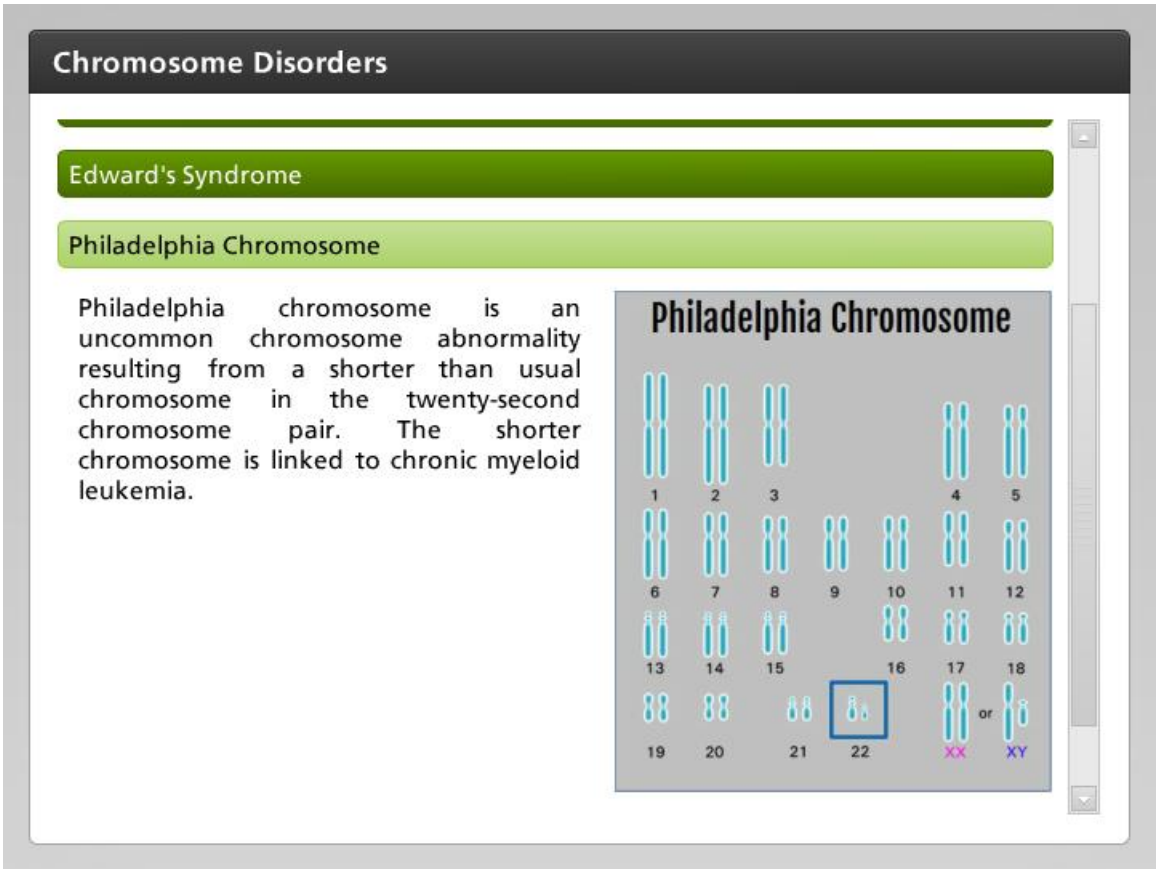
Klinefelter syndrome is a disorder that occurs in 1 out of every 1,000 males resulting from a missing X chromosome. Males suffering from Klinefelter syndrome experience a lower production of testosterone, which affects physical and cognitive development.

Edward's Syndrome



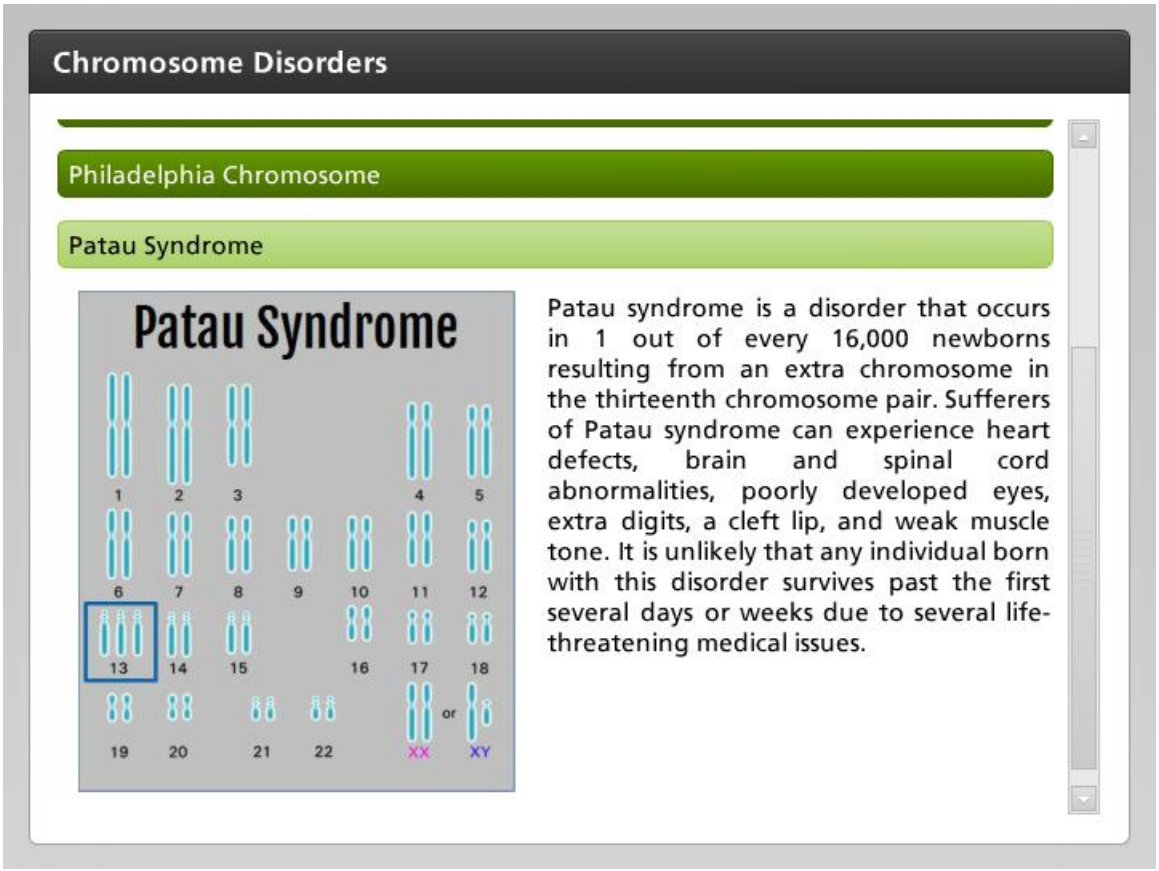
Edward's syndrome is a disorder that occurs in 1 out of every 5,000 births and results from an extra chromosome in the eighteenth chromosome pair. Children born with Edward's syndrome have many abnormalities in various parts of the body. It is unlikely that any individual born with this disorder survives past the first year due to several life-threatening medical issues. Edward's syndrome is also known as Trisomy 18.

Philadelphia Chromosome



Philadelphia chromosome is an uncommon chromosome abnormality resulting from a shorter than usual chromosome in the twenty-second chromosome pair. The shorter chromosome is linked to chronic myeloid leukemia.

Patau Syndrome



Patau syndrome is a disorder that occurs in 1 out of every 16,000 newborns resulting from an extra chromosome in the thirteenth chromosome pair. Sufferers of Patau syndrome can experience heart defects, brain and spinal cord abnormalities, poorly developed eyes, extra digits, a cleft lip, and weak muscle tone. It is unlikely that any individual born with this disorder survives past the first several days or weeks due to several life-threatening medical issues.