

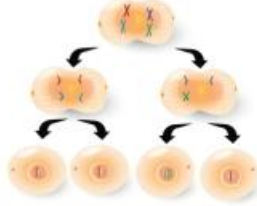
Module 5: Mendelian Genetics and Genetic Disorders

Topic 3 Content: Chromosome Disorders Notes

Introduction

Chromosome Disorders

Introduction



Chromosome Disorders

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.

Down Syndrome

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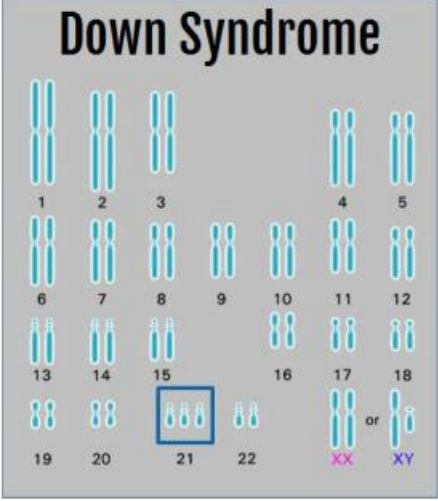
Down Syndrome

Chromosome Disorders

Introduction

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

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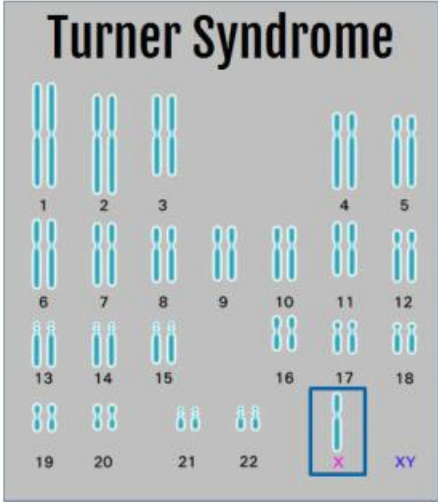
Turner Syndrome

Chromosome Disorders

Down Syndrome

Turner Syndrome

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.

The diagram shows a karyotype with 22 pairs of autosomes and one X chromosome. The X chromosome is highlighted with a blue box.

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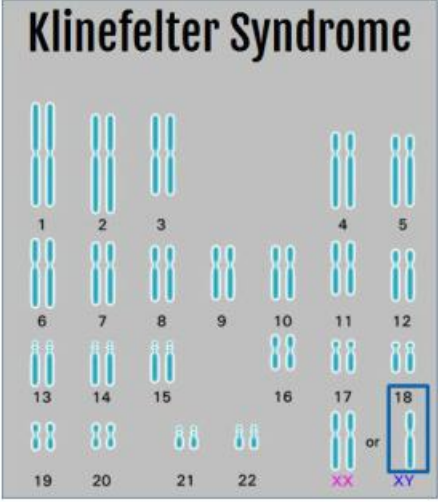
Klinefelter Syndrome

Chromosome Disorders

Turner Syndrome

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.



The diagram, titled "Klinefelter Syndrome", shows a human karyotype with 47 chromosomes. The chromosomes are arranged in pairs from 1 to 22, followed by sex chromosomes. The 18th pair (X chromosomes) is highlighted with a blue box and labeled "18". Below the box, it says "or XY", indicating the presence of two X chromosomes and one Y chromosome (XXY).

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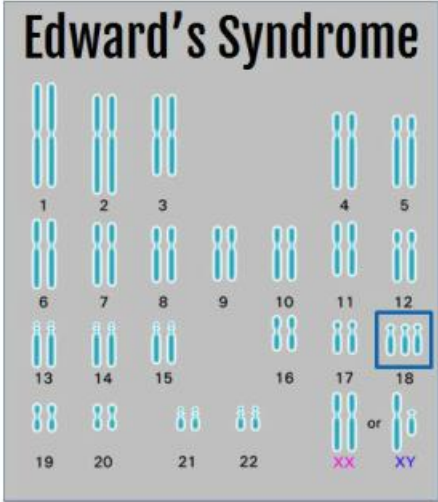
Edward's Syndrome

Chromosome Disorders

Klinefelter Syndrome

Edward's Syndrome

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.

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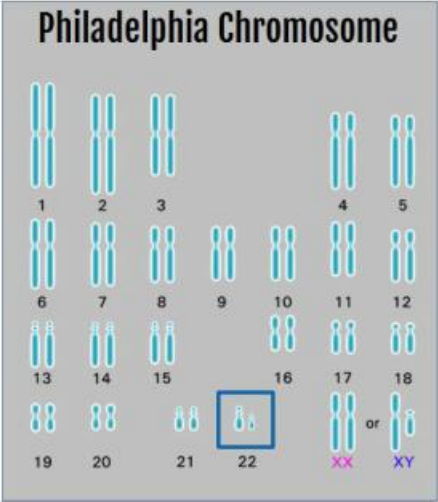
Philadelphia Chromosome

Chromosome Disorders

Edward's Syndrome

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.



The diagram illustrates a human karyotype with 22 pairs of autosomes and sex chromosomes. The 22nd pair is highlighted with a blue box, showing a shorter chromosome and a longer one, representing the Philadelphia chromosome. The chromosomes are arranged in four rows: 1-5, 6-12, 13-18, and 19-22. Sex chromosomes are labeled XX or XY.

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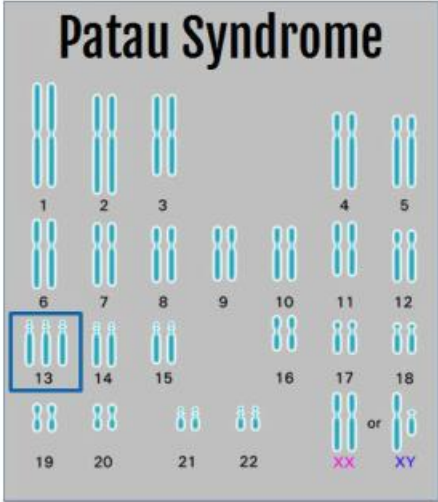
Patau Syndrome

Chromosome Disorders

Philadelphia Chromosome

Patau Syndrome

Patau Syndrome



The diagram shows a human karyotype with 22 pairs of autosomes and sex chromosomes. The 13th pair is highlighted with a blue box and contains three chromosomes instead of a pair, representing trisomy 13. The chromosomes are labeled 1 through 22, with XX for females and XY for males.

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.

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