Rare-Ed

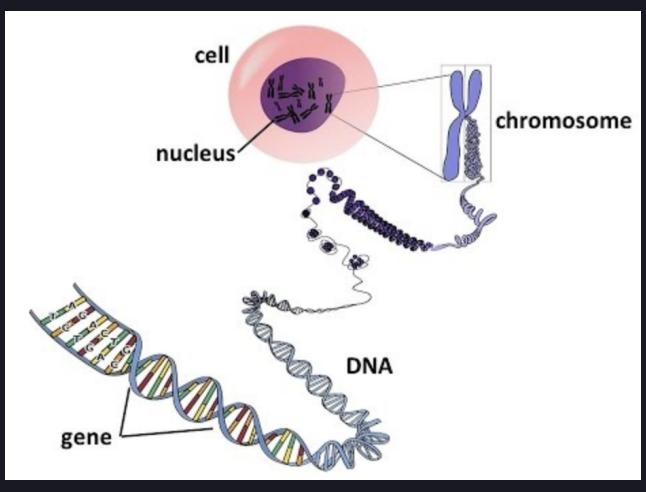
Rare Genomic's New Education Campaign

Genes & How they make us

As the basic functional and physical unit of heredity, genes are specific segments of DNA that provide the necessary instructions for proteins that build, regulate and maintain your body.

In other words, a gene is a sequence of nucleotides in DNA or RNA that codes for a molecule that has a function. The order of the nucleotides sends a message much like a sentence.

Genes are organized and packaged in units called “chromosomes.” A human being has 20,000 to 25,000 genes located on 46 chromosomes (23 pairs). These genes are known, collectively, as the human genome.



One set of chromosomes comes from a person’s mother, and the other set of chromosomes comes from the father. Thus, every person has two copies of each gene, one inherited from each parent.

Alleles are forms of the same gene with small differences in their sequence of DNA bases. However, the order of DNA base pairs does not differ very much from one person to the next. In fact, the order is the same 99.9% of the time. That .1% difference is why people are different. These differences are what makes us have or be prone to certain diseases.

There may be as many as 7,000 rare diseases. The total number of Americans living with a rare disease is estimated at between 25-30 million. This estimate has been used by the rare disease community for several decades to highlight that while individual diseases may be rare, the total number of people with a rare disease.

References

Genetics Home Reference, US Department of Health & Human Services National Institutes of Health, University of Leicester, GeneEd from the National Institutes of Health

Rare Disease Inheritance

The majority of rare diseases are caused by changes in genes. In some cases these genetic changes are passed down from one generation to the next. In other cases, they occur randomly in the person.

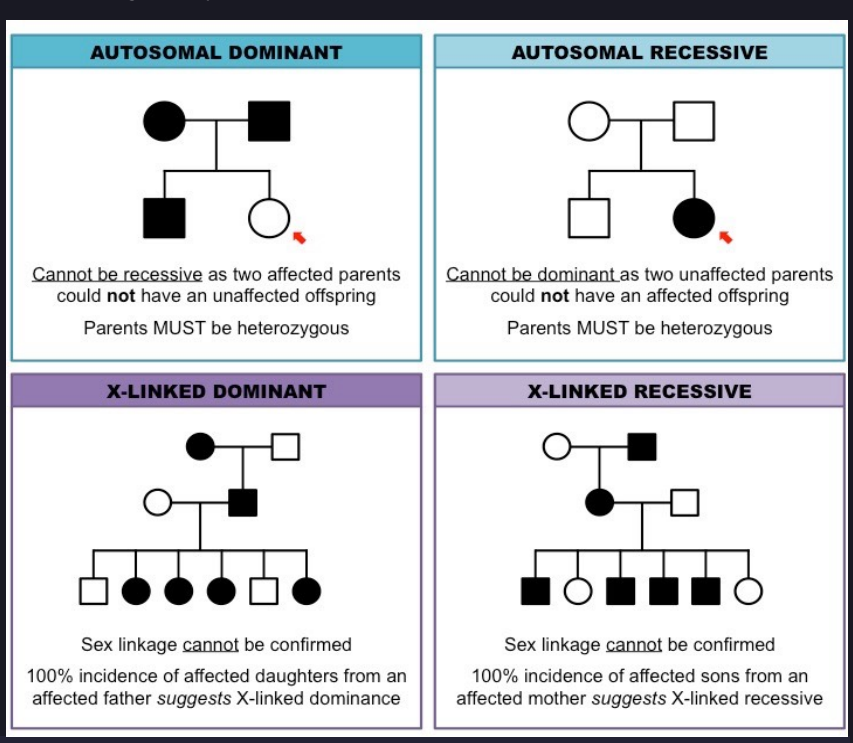
Inheritance is the process by which genetic information is passed on from parent to child.

Genes are specific segments of DNA that provide the necessary instructions for proteins that build, regulate and maintain your body. They are organized and packaged in units called “chromosomes.” They are also the basic functional and physical unit of heredity.

One set of chromosomes comes from a person’s mother, and the other set of chromosomes comes from the father. Thus, every person has two copies of each gene, one inherited from each parent.

Genetic conditions can be inherited in two main ways:

- **Simple or Mendelian: a defective gene can cause disease**
  - One copy from one parent= dominant
  - One copy from both parents= recessive
- **Complex or Multifactorial: interaction of multiple genes and environmental factors result in a complex pattern of inheritance**
- Mitochondrial: defective gene is present in the Mitochondria and only passed from the mother.
- Sex-linked: defective gene is present only on one sex chromosome.



References

Genetic and Rare Diseases Information Center, NHS, University of Leicester, GeneEd from the National Institutes of Health