Gregor Mendel and the Principles of Genetic Inheritance

American Society of Human Genetics

Decades before the discovery of DNA and genes, Gregor Mendel's experiments on pea plants laid the foundation of human genetics research. While the field became more sophisticated in the 150 years since his experiments were conducted, his pioneering research laid the foundation for us to better understand human health and learn how to treat diseases we did not know how to treat before! It all starts with curiosity and questions.

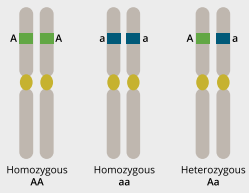
Our current understanding of how traits are inherited comes from the principles of inheritance proposed by Mendel:

1) The inheritance of each trait is determined by 'factors' (now known as genes) that are passed on to offspring.

Before Mendel's experiments, most people believed that an offspring's traits were due to the blending of each parent's traits (like how mixing red and yellow paint will produce orange). However, Mendel showed that when two varieties of purebred plants created offspring, it resembled one of the parent plants, not a blend of the two.

Mendel hypothesized that each parent contributes an underlying element to the offspring. We now know these elements as genes.

2) Individuals inherit one allele from each parent for each gene.



For any trait, an individual inherits one allele from each parent. An allele is one of two or more versions of a gene and can lead to homozygous or heterozygous offspring.

Homozygous means two alleles that form the pair for a trait are identical.

Heterozygous means each parent contributes a different allele of a given gene.

3) A trait may not show up in an individual but can still be passed onto the next generation.

Alleles can be dominant or recessive. Dominant alleles show their effect even if the individual only has one copy of the allele (is heterozygous). Recessive alleles will only show their effect if the individual has two copies of the allele (is homozygous). An individual with one dominant and one recessive allele for a gene will show the effect of the dominant allele.

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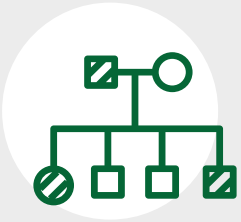
From his experiments, Mendel concluded that an individuals' genetic makeup (or genotype) cannot be determined by simply observing its external characteristics (or phenotype). This can be illustrated with family pedigree diagrams, which show patterns of how specific phenotypes are passed down in families.

Examples of Dominant Traits:



- Widow's Peaks
- Cheek Dimples
- Neurological disorder, Huntington disease (learn more with ASHG's fact sheet)

Keep exploring dominant traits with resources from Genetics Generation and Michigan Genetics Resource Center!

Examples of Recessive Traits:

Recessive traits only occur in individuals who have two recessive alleles from each parent. A recessive single-gene traits can disappear in one family generation, only to reappear in a later one.

Example of Sickle Cell Anemia Red Blood Cells

- Colorblindness
- Straight hairline (as opposed to a widow's peak)
- Sickle cell disease

Keep exploring recessive traits with resources from Scitable, Genetics Generation, and Michigan Genetics Resource Center

Why this matters today?

We now know that although some traits are due to single genes, most traits in humans are influenced by multiple genes as well as one's environment. However, the most basic occurrences of inheritance can still be explained using these principles.

If you visit a genetic genealogist to find out more about your family history, they may use DNA test results in combination with other methods to help explain the inheritance of certain traits. This can provide clues about where your ancestors may have come from, the relationships between individuals in a family, and how some traits and diseases are inherited.

Find out more about DNA genealogy on ReGeneration's website