Genetics

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Genes are the basic structural and functional unit of genetics that are made from a string of codons that specify a protein. About 3000 genes make up one chromosome, and all the chromosomes in a signal organism in a nucleus is the genome.

Genetics is the science of hereditary transmission that explains how traits and diseases are passed down through generations.

Humans have 44 autosomes and 2 sex chromosomes which make up the 46 chromosomes. The chromosomes are paired together forming 22 pairs. Haploid cells contain only one copy of each chromosome, and the haploid number is 23 (n). Diploid cells contain homologous (same) pairs of chromosomes which is 46, (2n).

Chromosome abnormalities reflect on the number or structure of chromosomes. Euploidy is a normal number of 46 chromosomes (2n), polyploidy refers to a chromosome number that is a multiple of the normal haploid chromosome set, such as triploidy which is 69 chromosomes (3n), and aneuploidy refers to the loss of gain of 1 chromosome 45 or 47 chromosomes (2n-1 or 2n+1).

A genotype is a specific constitution of an organism in which the allele combination of an individual causes a particular trait or disorder. Genotypes can be homozygous or heterozygous. A phenotype is the form of a gene which is expressed. Phenotypes disregard the allele pairing for a gene and only focuses on the trait. We can figure out an organism's phenotype from looking at it, but it's genotype can only be determined with genetic analysis.

When genes are passed down from parent to offspring, they are passed down in alleles. An allele is one of the possible forms of genes. Most genes have two alleles, a dominant and recessive allele. If the organism is a heterozygous for a trait it possesses one of each allele, then the dominant trait is expressed. A recessive allele is only expressed if an organism is homozygous for that trait or possesses two recessive alleles. Alleles were first defined by Gregor Mendel in the law of segregation in four parts. First, it defines an allele, second It states that organisms inherit one allele from each parent. Third, it states that gametes only carry one allele for each trait and fourth, it defines the difference between dominant and recessive genes. Mendel's law of independent assortment states that when genes are inherited, they are

inherited independent of each other. Linked genes are exceptions to the law of independent assortment because two genes are located on the same chromosome.

Non-mendelian genetics describe conditions that do not follow Mendel's laws of inheritance. An example of non-mendelian genetics is mitochondrial inheritance in which is only inherited from the mother. Mitochondrial inheritance is common and accounts for genetically pre-disposed hearing loss.

When a disorder is an autosomal dominant disorder, it only needs one copy of the gene to produce the phenotype. The chance of reoccurrence of autosomal dominant inheritance is 50% and the pedigrees will show the disease in a vertical family pattern.

When the disorder is an autosomal recessive disorder, a double dose of the gene is required to produce the phenotype. When assuming the parents are heterozygous, the chance of reoccurrence is 25% and the pedigrees will show a horizontal family pattern.

When the disorder is sex-linked, the disorder is carried on the sex chromosome (most common on the X chromosome). An X-linked recessive inheritance typically only shows up in the male offspring, in order for a female to express an X-linked recessive disorder phenotype the mother must be a carrier and the father must be affected. Carrier females of an X-linked disorder have a 50% chance to have a son with the trait and a 50% chance to have a daughter who is a carrier. Sex-linked inheritance accounts for 1-2% of genetically pre-disposed hearing loss.