L.2 Polygenic inheritance (Continuous variation):

A trait produced by multiple factors whether these factors are genetics, environmental or a mixture of both .If a character is determined by just one pair of alleles A and a ,of equal frequency, the population falls into 3 different classes AA, Aa, aa in proportions 1:2:1.

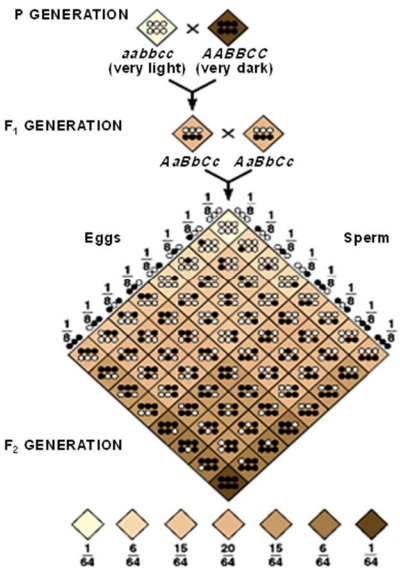
If 2 pairs of alleles, A and a, B and b are concerned, there are five phenotypic classes in the proportions 1:4:6:4:1.Because all of the genes that play role in determining phenotypes such as height or weight, segregate independently of each other, one sees a gradation in the degree of difference when many individuals are examined . Polygenic traits tend to have a bell-shaped distribution in a population. Most individuals inherit various combinations of dominant and recessive alleles. These individuals fall in the middle range of the curve, which represents the average range for a particular trait. Individuals at the ends of the curve represent those who either inherit all dominant alleles (on one end) or those who inherit all recessive alleles (on the opposite end). Traits such as height, weight, skin color, finger prints and intelligence are under polygenic control. In addition, congenital malformations such as neural tube defects, cleft palate, and club foot as well as genetic disorders such as diabetes, hypertension, and behavioral disorders are polygenic or multifactorial traits.

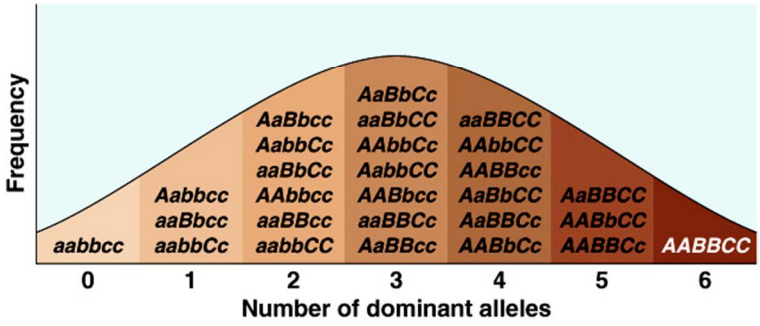
Height in human:

It is influenced by at least 10 different genes, each of which has two or more allelic forms as well as effect of diet. Most people in a population fall in middle of the curve and are average height. Those on one end of the curve are tall individuals and those on the opposite end are short individuals.

Human skin color:

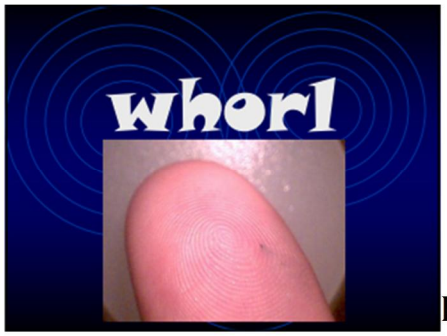
There are three pair of alleles A, a and B, b and C, c to form seven phenotypes: black ,darkest brown, dark brown ,medium brown, light ,lighter brown and white in ratio 1:6:15:20:15:6:1. The intermediate colors would result from the effects of environmental factors such as sun –tanning.





Finger prints:

Fingerprint patterns are classified by shapes as loops, whorls and arches and by ridge counts. Ridge counts are the most useful feature of finger prints to the study of phenotypic variance.



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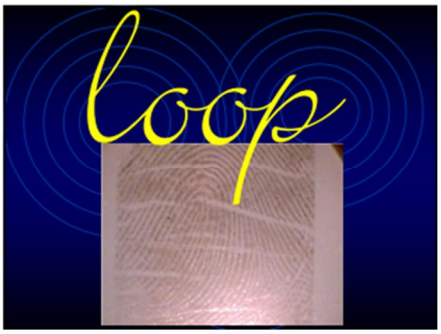
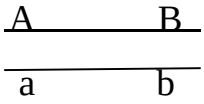


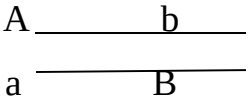
Linkage and crossover

Each chromosome has thousands of genes, and these genes are located close together on the same chromosome called linked genes, tend to be inherited together, they generally do not follow Mendel's principle of independent assortment. When genes are not linked, they assort independently; the gametes then represent all possible allele combinations; the expected phenotypic ratio of a dihybrid cross is 9:3:3:1.If genes are linked on the same chromosome, only two allele combinations are expected in the gametes; the phenotypic ratio is 3:1 the same as for a monohybrid cross.

Linkage may be in coupling (cis), in which the dominant genes are located at the same chromosome or repulsion (transphase), the dominant alleles are located on the different chromosome.



Coupling



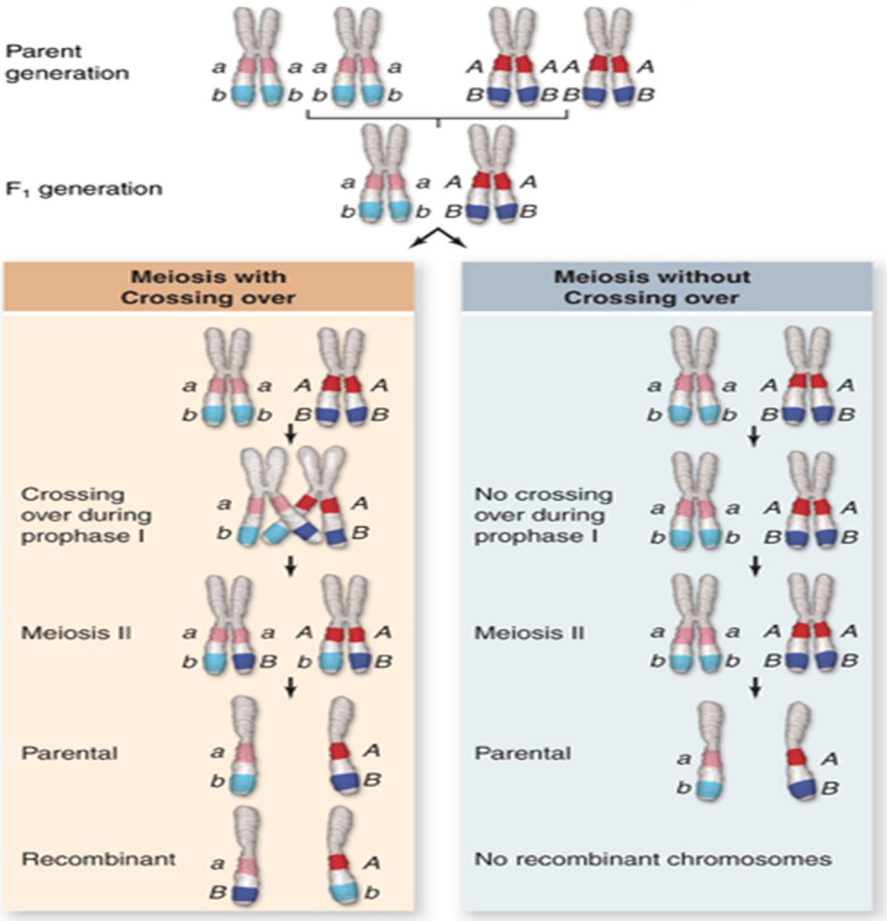
Repulsion

Human being have 24 linkage groups (2n=46,n=22+x or y),**An example of linked genes are blonde hair and blue eyes.** This combination tends to be inherited together at autosomal chromosome, other example, **linkage between G6PD deficient and color blind** on the X-chromosome.

Crossover:

The genes on the same chromosome does not remain identical from generation to generation because of this phenomenon called crossover, during the prophase 1 (Diplotene) of meiosis the homologous maternal and paternal chromosomes come together to form bivalents .The point of attachment between homologous chromatids called chiasmata or called crossover points because it is here that the joined chromatids may break and then reunite with wrong chromatid segments of two chromatid are exchanged from cross over forms four haploid cells two of them paternal types called non-recombinants and the other called new recombinants.

The genes that are far apart have a greater chance of crossing over and the genes that are closer have a less likely chance of crossing over. The frequency of crossing over between genes can be used for construct genetic maps.



Gene mapping

Refer to the physical locations of genes on chromosomes resulted from frequency of crossing over between particular pairs of genes. The tracking crossing over helps determine where genes are located on the chromosome. The gene map is useful for to identify one gene as a marker that can infer the presence of the other gene and to identifying disease predisposition.

There are two other ways in which chromosomes are mapped as well as the previous way. One way is to map a **cytogenetic map** in which chromosome bands, each representing 1 million to 5 million bases, are stained and the investigator finds a correlation between people who show a particular trait and exhibit a similar staining pattern.

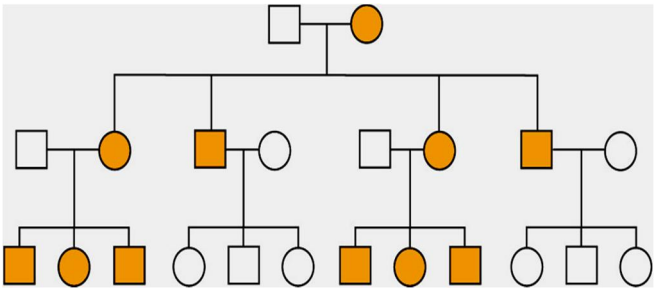
Another way to is producing a **physical map** using enzymes to cut pieces of DNA into fragments containing markers along with genes whose location is to be

determined. By using computers to "walk" or overlay these fragments into their proper sequence we can produce a map of a long strand of DNA.

Maternal inheritance and mitochondrial gene:

The bases of law of segregation are that both parents contribute genes equally to offspring. This is not the case for genes in mitochondria, the organelles that house the biochemical reactions that provide energy. Mitochondria in human cells contain several copies of mini –chromosome that carries just 37 genes.

The inheritance pattern and mutation rates for mitochondrial genes differ from those for genes in the nucleus .Mitochondrial genes are maternal inherited .They are passed only from an individual's mother **because sperm almost never contribute mitochondria when they fertilize an oocyte.** Pedigrees that follow mitochondrial genes show a woman passing the trait to all her children, while a male cannot pass the trait to any of his children.

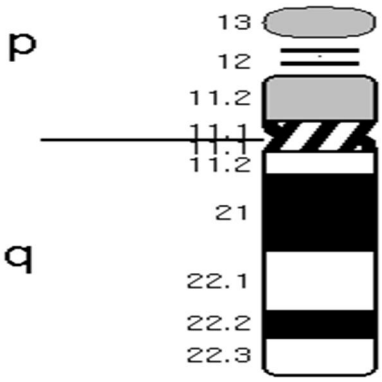


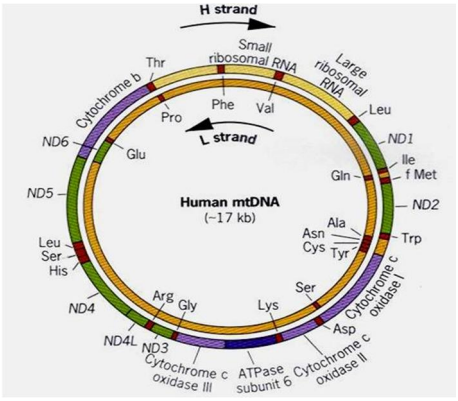
Inheritance of mitochondrial genes: mothers pass mitochondrial genes to all offspring .Fathers do not transmit mitochondrial genes

The mDNA is differs from nuclear DNA by the following:

1- Mitochondrial DNA (mDNA) does not crossover
2- Mitochondrial DNA also mutated faster than nuclear DNA for 2 reasons: It lacks DNA repair enzymes and the mitochondrion is the site of the energy reactions that produce oxygen free radical that damage DNA.
3- mDNA is not having histones, and intron (sequences that do not encode protein).

4- Finally, inheritance of mitochondrial genes differs from inheritance of nuclear genes simply because a human cell has one nucleus but many mitochondria and each mitochondrion harbors several copies of its chromosome.





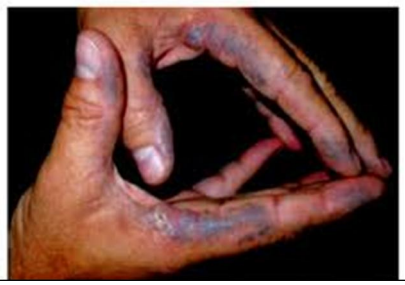
a. Nuclear chromosome **b. Mitochondrial chromosome**

Phenocopies:

A trait that appears inherited but is caused by the environment. Such a trait can either produce symptoms that resemble those of Mendelian disorder or mimic inheritance patterns by occurring in certain relatives. For example, the limb birth defect caused by the drug thalidomide, is a phenocopy of the inherited illness phocomelia . A birth defect caused by exposure to a teratogen was more likely than a sudden increase in incidence of rare inherited disease.

Other example: a phenocopy of alkaptonurea occurred in some women with dark brown skin who used a bleaching cream that contained a chemical called hydroquinone .It caused darkening of the fingers and ears, just like alkabtanuria.

Phenocopy phocomelia



Phenocopy of alkaptonurea