

### Mendel's laws and probability history:

**Probability:** what is the chance that a given event will occur? For us, what is the chance that a child, or a family of children, will have a given phenotype?

**Probability is expressed in numbers between 0 and 1.** **Probability = 0** means the event never happens; **probability = 1** means it always happens. The total probability of all possible events always sums to 1. The probability of an event equals **the number of times it happens divided by the number of opportunities**. These numbers can be determined by experiment or by knowledge of the system.

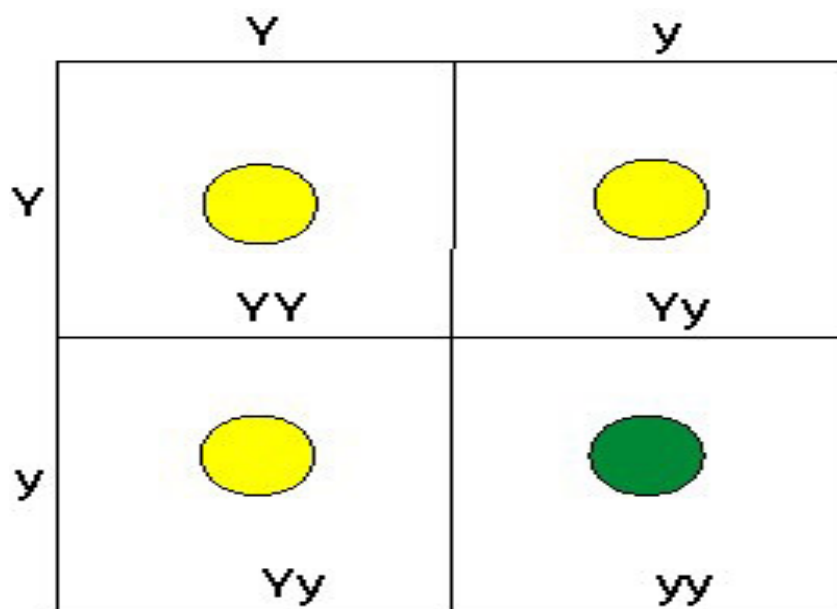
### Mendel's LAWS and probability

Mendel realized that the results of his genetic crosses followed rules of probability.

- ❖ The rule of multiplication (product rule) says that the probability of two independent events occurring together can be calculated by multiplying the individual probabilities of each event occurring alone. For example, the chance of getting two tails when you flip two coins is:  $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$
- ❖ Rule of addition (Sum rule): the probability of an event that can occur in two or more independent ways is the sum of the separate probabilities of the different ways.

Random events are independent of one another-not affected by the outcome of previous events

**Punnett Square:** A diagram uses to show the probability or chances of a certain trait being passed from one generation to another. Its discovered by **Reginald Punnett**.







- ❖ Mendel's study of inheritance dealt with simple, independent segregating traits.
- ❖ There are other patterns of inheritance more than the dominance/recessive relationship Mendel observed.
- ❖ The environment can also influence the phenotype of an organism.

**Dominance:**

- ❖ **Complete dominance:** dominant allele is expressed; recessive allele is not expressed.
- ❖ **Incomplete dominance:** Mendel discovered that when a hybrid is created in the F1 generation, one phenotype is present (dominant) while the other is absent (recessive). Dominant and recessive alleles both **expressed by blending**, the result is new phenotype which is intermediate between homozygous phenotypes, for example: Japanese four-o'clock flowers, Red flower plant genotype = RR, White flower plant genotype = WW, and Pink flower plant genotype = RW.
- ❖ **Codominance:** occurs when both alleles produce a phenotype independent of one another. Dominant and recessive alleles are both **expressed, but don't blend**, the result is new phenotype which has both homozygous phenotypes present, for example: colour of hair coat in cattle.  $c^r c^r$  = red hairs,  $c^w c^w$  = white hairs and  $c^r c^w$  = roan coat (mixture of both colors).

**Multiple Alleles:**

- ❖ Traits are the result of more than 3 or more types of alleles
- ❖ Human blood type is an excellent example of multiple alleles
- ❖ ABO blood type is controlled by the ABO gene.
- ❖ There are 3 different alleles for blood type (A, B, & O).
- ❖ The A and B alleles express A and B antigens that are expressed on the surface of erythrocytes (red blood cells), and the O type does not express any antigens.
- ❖ A is dominant to O
- ❖ B is also dominant to O
- ❖ A and B are both codominant.

Blood Type (genotype)	Type A (AA, AO)	Type B (BB, BO)	Type AB (AB)	Type O (OO)
Red Blood Cell Surface Proteins (phenotype)				

**Polygenic Inheritance:**

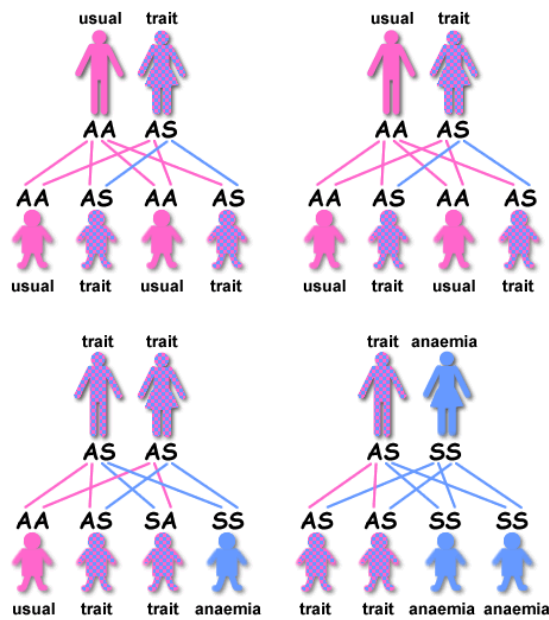
- ❖ Some traits are determined by the combined effect of two or more genes. These are referred to as polygenic traits. Often the genes are large in quantity, but small in effect.
- ❖ In polygenic inheritance, more than one pair of alleles determines the phenotype.
- ❖ Each dominant allele is additive to the overall phenotype.
- ❖ Human skin, hair, and eye color are polygenic traits because they are influenced by more than one allele at different loci.
- ❖ Human skin color is a polygenic trait determined by three genes.
- ❖ The more dominant alleles a person has, the darker their skin color.
- ❖ There are several examples of multifactorial traits.
- ❖ Skin color, Palate and lip disorders, allergies and some cancers.

### **Environment and the Phenotype:**

- ❖ In some cases, the environment can affect phenotype more than genetics.
- ❖ For example, temperature can affect the color of primroses and Himalayan rabbits.
- ❖ The control of phenotype by genetics and/or environment leads to the nature.

### **Pleiotropy:**

Pleiotropy refers to the phenomenon of a single gene affecting several traits. Sickle-cell anemia, an example of pleiotropy, is a disorder of the blood caused by an inherited abnormal hemoglobin (one defect in the hemoglobin gene - a single amino acid change in the protein). A single gene results in irregularly shaped red blood cells that painfully block blood vessels, cause poor overall physical development, as well as related heart, lung, kidney, muscle pain, eye problems (jaundice or eye damage) and early death. All of this is caused by a single mutation in one of the hemoglobin genes. Another pleiotropic example is albinism. The gene for this trait not only results in a deficiency of skin, hair, and eye pigmentation but also causes defects in vision.



### Sex-Linked Inheritance:

- ❖ The sex chromosomes, X and Y, not only carry the genes that determine male and female traits but also those for some other characteristics as well. Genes that are carried by either sex chromosome are said to be sex linked.
- ❖ Men normally have an X and a Y combination of sex chromosomes, while women have two X's. Since only men inherit Y chromosomes, they are the only ones to inherit Y-linked traits. Men and women can get the X-linked ones since both inherit X chromosomes.
- ❖ The Y allele carries 26 genes, mostly related to gender. The X allele carries genes for gender as well as genes unrelated to gender, the X-linked genes.
- ❖ There are many more X-linked conditions than Y-linked conditions, since humans have several times as many genes on the X chromosome than the Y chromosome. Only females are able to be carriers for X-linked conditions; males will always be affected by any X-linked condition, since they have no second X chromosome with a healthy copy of the gene.
- ❖ In X-linked recessive inheritance, a son born to a carrier mother and an unaffected father has a 50% chance of being affected, while a daughter has a 50% chance of being a carrier, however a fraction of carriers may display a milder (or even full) form of the condition due to a phenomenon known as skewed X-inactivation, in which the normal process of inactivating half of the female body's X chromosomes preferably targets a certain parent's X chromosome (the father's in this case). If the father is affected, the son will not

be affected, as he does not inherit the father's X chromosome, but the daughter will always be a carrier.

- ❖ In X-linked dominant inheritance, a son or daughter born to an affected mother and an unaffected father both have a 50% chance of being affected (though a few X-linked dominant conditions are embryonic lethal for the son, making them appear to only occur in females). If the father is affected, the son will always be unaffected, but the daughter will always be affected. A Y-linked condition will only be inherited from father to son and will always affect every generation.
- ❖ Linked genes are inherited together because they cannot segregate during meiosis.
- ❖ The linked alleles on the same chromosome form a linkage group