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TOPIC- GENETICS OF SEX

Sex determination, the establishment of the sex of an organism, usually by the inheritance at the time of fertilization of certain genes commonly localized on a particular chromosome. This pattern affects the development of the organism by controlling cellular metabolism and stimulating the production of hormones that trigger the development of sexual glands or organs. An excess or lack of hormones during embryological development may cause an individual to develop the superficial appearance of one sex while retaining the genetic constitution of the other sex.

Sex-Linkage: the 23rd pair of chromosomes present in humans is called allosomes, which differ in size and function when compared to autosomes. They determine the sex of an individual. They are labeled using letters (XY). Allosomes comprise either two X chromosomes (women), or one X chromosome and one Y chromosome (male). These are present in reproductive cells (egg cell and sperm cell) of the body. When both fuse together during sexual reproduction, a new offspring having 46 chromosomes with XX (female) and XY (male) is formed.

Genes carried in the same chromosome are said to be 'linked' because they are assorted together. Haemophilia is due to the operation of a recessive sex-linked gene. A woman, heterozygous for it is therefore unaffected, since she carries the haemophilia gene (*h*) in one Xchromosome, and its normal allelomorph (*H*) in the other. Normal women can transmit haemophilia while a normal man cannot do so.

Sex-linked Inheritance: Colour blindness is an example of sex-linked inheritance in man. Women are much less often colour blind than men. But if a woman does happen to be colour blind, and if she marries a normal man, all of her sons are colour blind but none of her daughters are.

In humans, there are two forms of sex linkage: (1) X linkage and (2) Y linkage. The difference lies on the type of sex chromosome involved.

X linkage is the state of a genetic factor (gene) being borne on the X chromosome. The inheritance of these genes from the X chromosome is referred to as X-linked inheritance, and may either be recessive or dominant.

X-linked Recessive Inheritance

Sex-linkage was first discovered by Thomas H. Morgan (father of modern genetics) in 1910. Sex-linked traits affect male and female differently. As human male is hemizygous for X-linked traits, any gene on a male's X chromosome is expressed in his phenotype because there is no such second allele to mask its expression. Therefore, the condition of dominant and recessive trait is limited to female only. Females express X-linked traits or disorders when they are homozygous for the disorder and become carriers when they are heterozygous.

Therefore female can transmit the trait as affected if her father is affected and mother at least carrier. However male can transmit the trait if any of the parents is affected or carrier (for mother). Therefore, the incidence is much higher in males than females. These patterns of inheritance are also called crisscross inheritance or skip generation inheritance, in which a character is inherited to the second generation through the carrier of first generation. X-linked (both recessive and dominant) traits are always passed on by the X chromosome from mother to son or from either parent to daughter. The trait never passed from father to son.

The human male is hemizygous in respect of X-linked inheritance as they have single copy of X chromosome.

Some important characteristic features are:

- Occurrence and transmission is influenced by sex; males are more affected than females;
- Affected male does not transmit the trait to his sons but always transmits to all his daughters;
- Carrier female can transmit the trait to half of her children of either sex;
- The trait is transmitted from affected male through all his daughters to half of his grandsons; and
- The trait may be transmitted through a series of carrier females; carrier shows variable expression of the trait.

X linked Recessive inheritance

Parents	**Offspring**
Mother homozygous Normal Father Normal	All the offspring will be homozygous normal
Mother homozygous Normal Father affected	All the daughters will be heterozygous normal (carrier) All the sons will be normal
Mother heterozygous Normal (carrier) Father Normal	50% probability that daughter will be homozygous normal 50% probability that daughter will be heterozygous normal (carrier) 50% probability that son will be normal

	50% probability that son will be affected 50% probability that daughter will be heterozygous normal (carrier) 50% probability that daughter will be affected
Mother heterozygous Normal (carrier) Father affected	50% probability that son will be normal 50% probability that son will be Affected
Mother affected Father normal	All the daughters will be heterozygous normal (carrier) All the sons will be affected
Mother affected Father affected	All the offspring will be affected

X-linked Dominant Inheritance

X-linked dominant inheritance shows the same phenotype as a heterozygote and homozygote. In case of an X-linked dominant inheritance, male to male transmission is not there. This also makes it distinct from autosomal traits. X linked dominant cannot be distinguished from Autosomal Dominant by progeny of affected females, but only from the progeny of affected males. Affected females are more common than affected males (but heterozygous females have milder expression); on the other hand the traits (especially disorder) are more severe in males than their female counterparts.

Some important characteristic features are-

- Occurrence and transmission is influenced by sex; females are more affected than males but may be with variable expressions;

- Homozygous female transmitted the trait to all the children;
- Male transmitted the trait to all the daughters but never to a son;
- Affected males have no normal daughter;
- Affected heterozygous females transmit the trait to half of their children of either sex. Affected homozygous females transmit the trait to all their children;
- X linked dominant cannot distinguish from Autosomal Dominant by progeny of affected females, but only from the progeny of affected males.

X linked Dominant inheritance

Parents	**Offspring**
Mother homozygous affected Father affected	All the offspring will be homozygous Affected
Mother homozygous affected Father normal	All the daughters will be heterozygous affected All the sons will be affected
Mother heterozygous affected Father affected	50% probability that daughter will be homozygous affected 50% probability that daughter will be heterozygous affected 50% probability that son will be affected 50% probability that son will be

	normal
Mother heterozygous affected Father normal	50% probability that daughter will be heterozygous affected 50% probability that daughter will be normal 50% probability that son will be affected 50% probability that son will be normal
Mother normal Father affected	All the daughters will be heterozygous affected All the sons will be normal
Mother normal Father normal	All the offspring will be normal

Y-linked Inheritance

The genes located on the Y chromosome, whose alleles are absent on the X chromosome are Y-linked genes or holandric genes (also hemizygous). Y-linked inheritance occurs when a gene is transmitted through the Y chromosome. Since Y chromosomes can only be found in males, hence Y linked genes are only passed on from father to son and never appear in females. Therefore, there is no skipping of generation and affected males have all affected sons, no females are said to be affected for the trait

Some important characteristic features are-

- Only males are affected;

- Affected male transmitted the trait to all his sons but never to his daughter;
- No skipping of generations.

Y-linked inheritance

Father	**Offspring**
Father affected	All the sons will be affected
Father normal	All the sons will be normal

Y-linked traits examples

Hypertrichosis of ear: growth of hair on the rim of pinna

Testis determining factor (TDF)

Traits/ Description	**Autosomal Recessive**	**Autosomal Dominant**
Albinism: is a form of hypopigmentary congenital disorder, characterised by a partial or total lack of melanin pigment in the eyes, skin and hair (or more rarely the eyes alone).	Albinism	Normal pigmentation
Thalassemia: Human haemoglobin molecule consists of two alpha and two beta globin chains conjugated with heme. In alpha Thalassemia alpha chain is missing or defective and in beta Thalassemia beta chain is absent which leads to iron overload and anemia.	Thalassemia	Normal
Cystic Fibrosis: Cystic fibrosis is an inherited disease that causes thick, sticky mucus to be built up in the lungs and digestive tract. It is one of the most common chronic lung diseases in children	Cystic fibrosis	Normal

and young adults, and may result in early death.		
Tay-Sachs disease: Tay-Sachs disease is caused by a mutation on chromosome 15. Tay-Sachs disease occurs when the body lacks hexosaminidase A, a protein that helps break down a chemical found in nerve tissue called gangliosides.	Tay-Sachs disease	Normal
Xeroderma pigmentosum: A disruption of affected person's DNA's ability to repair damage caused by ultraviolet radiation of sunlight.	Xeroderma pigmentosum	Normal
Hitchhiker's thumb: More formally known as "distal hyper extensibility of the thumb" can extend the top of the thumb backwards nearly 90° when the thumb is extended in a "thumbs-up" position.	Hitchhiker's thumb	Straight thumb
Dentinogenesis imperfecta: Pulp chambers and root canals of teeth are obliterated with abnormal dentin. There is also an increased constriction and junction between the crowns and the roots of the molar.	Normal teeth	Dentinogenesis Imperfect
Cleft Chin: It is a Y-shaped fissure on the chin with an underlying bony peculiarity.	No Cleft Chin	Cleft Chin
Brachydactyly: Better known as clubbed thumb, is a condition where the thumbs are shorter and stubbier than normal. Brachydactyly is a genetically inherited trait that is often dominant.	Normal thumb	Brachydactyly

PTC taste sensitivity: Phenylthiocarbamide also known as PTC is anorganosulfur thiourea containing a phenyl ring. A crystalline compound, C6H5NHCSNH2, that tastes intensely bitter to people with a specific dominant gene and tasteless to others.	Non-taster	Taster
Achondroplasia: Characterized by prominent forehead, low nasal root, redundant folds in arms and legs accompanied by short-limbed dwarfism.	Normal	Achondroplasia:
Familial Hypercholesterolemia: Characterized by high LDL in blood resulting to deposition of cholesterol in arteries, tendons, skin, etc., which may leads to coronary artery diseases.	Normal	Familial Hypercholesterolemia

Traits/ Description	**X-linked Recessive**	**X-linked Dominant**
Duchene muscular dystrophy: It is an inherited disorder that involves rapidly worsening muscle weakness.	Affected	Normal
Haemophilia A: Occurs due to the deficiency of factor VIII in blood. Affected persons are unable to produce a factor needed for blood clotting, therefore the cuts, wounds, etc., of haemophilic persons continue to bleed and sometimes (if not stopped by clotting factors) leads to death.	Haemophilia A	Normal

Red green color blindness: Colour perception is mediated by light absorbing protein in the cone cells of the retina in the eye. Colour blindness is caused by an abnormality in any of the receptor protein. Red green colour blindness is the ability to perceive the colour green and red.	Red green color blindness:	Normal vision
G6PD deficiency: It is an inherited disorder in which the body doesn't have enough enzyme glucose-6-phosphate dehydrogenase, or G6PD, which helps red blood cells (RBCs) function normally, and deficiency may cause hemolytic anemia.	G6PD deficiency:	Normal
Incontinentia pigmenti: Incontinentia pigmenti is a genetic defect on X chromosome which leads to unusual blistering and changes in skin color.	Normal	Incontinentia pigmenti
Fragile X syndrome: Fragile X syndrome is a genetic condition involving changes in part of the X chromosome resulting in mental retardation.	Normal	Fragile X syndrome
Congenital generalized hypertrichosis: Person has more hair follicles which lead to dense and more abundant terminal hair. Generally, it causes excess facial and upper body hair that covers extensive areas of skin.	Normal	Congenital generalized hypertrichosis

Sex-limited traits

Some genes in the autosomal chromosomes are expressed in one of the sexes. This happens either because of the differences in the internal environment of the hormones or due to the dissimilarities in the anatomy. (They are traits constricted limited to one sex only due to differences in sex hormones of each sex).

Examples

1. Milk production is limited only to the females, because they have certain hormones help the gene to express its effect.
2. The appearance of beard trait is limited to males only and it is one of the secondary sex characteristics in human males.

Sex-influenced traits

They are traits that their genes are carried on autosomes and the sex of individual acts to modify their dominance where the act of these genes is influenced by the hormones secreted from gonads of adult males and females , Sometimes, the sex of living organism acts to modify the dominance of some traits that are called sex-influenced traits, such as the presence of horns in cattle trait and genetic baldness trait in humans .

Baldness it spreads among males of some families more than the females, its appearance is controlled by a dominant gene that carried on autosomes and influenced by the masculinity hormones only.

Sex-influenced traits are autosomal traits that are expressed based on the influence of the sex, in particular the sex hormones: e.g. testosterone in males and estrogen and progesterone in females. Although these traits can be seen in both the sexes, the degree or frequency of the phenotypic expression varies according to the sex. The genes are dominant in males but recessive in females.