B.A/B.Sc Sem IV Paper 7 Unit 1&2

Dr Aishwarya Awasthi

Non Mendlean Inheritance

Sex Limited & Sex Influenced traits

Some traits are carried on the sex chromosomes, X and Y. Most traits carried are present on only the X-chromosome. The Y-chromosome is smaller, and so, very few genes are located on this chromosome.Sex traits can be categorized into three types of inheritance: sex-limited, sex-linked, and sex-influenced. Sex-limited traits are traits that are visible only within one sex. For instance, barred coloring in chickens normally is visible only in the roosters.

Sex-linked traits would be considered traits like sickle cell anemia and color blindness. They are said to be linked because more males (XY) develop these triats than females (XX). This is because the females have a second X gene to counteract the recessive trait. Thus, the trait is more likely to be visible in the male.

Sex-influenced traits are autosomal traits that are influenced by sex. If a male has one recessive allele, he will show that trait, but it will take two recessive for the female to show that same trait. One such gene is baldness.

A lot of sex-limtied traits can determine parental carriers by using a pedigree. The colored blocks indicate a recessive trait phenotype, and from there, carriers can be traced back. This is an effective method when determining the probability of an offspring receiving that trait.

The traits limited to only one sex due to anatomical differences are called a sex limited treat. Such trait affects a structure or function of the body of males or females only. These traits are controlled by sex-linked or autosomal genes.

Chest Development in women

Beard growth in humans is limited to men.

A woman does not grow a beard hersell But she can pass the genes of heavy beard growth to her sons.Sex hormones and other ph siologic differences between males and females effect on expression of certain gene. For example, premature baldness is an autosomal dominant trait. But this condition is rarely expressed in the female due to female sex hormones. It appears in them only after menopause. Thus. sex limited inheritance, perhaps more correctly called sex-influenced inheritance.

Autosomal Inheritance

Sex linked InheritanceThe inheritance of a trait (phenotype) that is determined by a gene located on one of the sex chromosomes is called sex linked inheritance. The expectations of sex-linked inheritance in any species depend on how the chromosomes determine sex. For example, in humans, males are heterogametic. It has one X chromosome and one Y chromosome. But females are homogametic. They two X chromosomes. In human males, the entire X chromosome is active. But one of a female's X chromosomes is largely inactive. Random inactivation of one X chromosome occurs during the early stages of female embryogenesis. Therefore, every cell that forms from a particular embryonic cell has the same X chromosome inactivated. This pattern of sex determination occurs in most vertebrates, but in birds and many insects and fish the male is the homogametic sex.

In general terms, traits determined by genes on sex chromosomes are not different from traits determined by autosomal genes. Sex-linked traits are distinguishable by their mode of transmission through successive generations of a family. In humans it is called X-linked or Y-linked inheritance.

X-linked recessive Traits:

These are expressed in allheterogametic and homogametic which are homozygous for the recessive allele. An example is the sex-linked recessive is horns in sheep that appear only in males. The recessive phenotypes of such genes are more common in males than in females. The examples of X-linked recessive trait in human are Color blindness, Duchenne muscular dystrophy, Hemophilia. Kennedy disease.

Males are affected almost exclusively

Transmission occurs through carrier females to their sons

Male-to-male transmission does not occur

Affected males are at risk of transmitting the disorder to their grandsons through their carrier daughters

X linked dominant inheritance:

Pattern of X- linked dominant inheritance is different from X- linked recessive. It is more common in females than males. All daughters are affected by their father. But none of his sons are affected. A heterozygous affected mother passes the trait equally to half of her sons and half of her daughters. Hypophosphatemic rickets is an X — linked dominant trait. It is a rare hereditary disease. It is different from common dietary rickets. Other examples of X-linked dominant traits are Retinitis pigmentosa and Rett syndrome.

Y linked trait:

The genes present on the Y chromosomes are called Y-linked genes and their traits are called Y-linked traits. Y chromosome is not completely inert it carries a few genes. These genes have no counterpart on X chromosome.

Example: gene is present on Y chromosome of man. It determines maleness. Y — linked traits are found only in males. These traits directly pass through Y chromosome from father to son only. Females do not inherit Y chromosome. So such traits can not pass to them.

X and y linked Traits: The genes present on both X and Y chromosomes are called X and Y linked genes. For example, the bobbed gene in Drosophila are present on X and Y both. The pattern of inheritance of X and Y linked gene is like autosomal genes. So these are also called pseudoautosomal genes.

Genes are inherited from our biological parents in specific ways. One of the basic patterns of inheritance of our genes is called X-linked recessive inheritance.

X-linked inheritance

X-linked inheritance means that the gene causing the trait or the disorder is located on the X chromosome. Females have two X chromosomes; males have one X and one Y. Genes on the X chromosome can be recessive or dominant. Their expression in females and males is not the same. Genes on the Y chromosome do not exactly pair up with the genes on the X chromosome. X-linked recessive genes are expressed in females only if there are two copies of the gene (one on each X chromosome). However, for males, there needs to be only one copy of an X-linked recessive gene in order for the trait or disorder to be expressed. For example, a woman can carry a recessive gene on one of the X chromosomes unknowingly, and pass it on to a son, who will express the trait:

There is a 50 percent chance that daughters carry the gene and can pass it to the next generation. There is a 50 percent chance that a daughter will not carry the gene and, therefore, cannot pass it on. There is a 50 percent chance that sons do not have the gene and will be healthy. However, there is a 50 percent chance that a son will have inherited the gene and will express the trait or disorder.

Examples of X-linked recessive conditions include red-green color blindness and hemophilia A:

Red-green color blindness.

Red-green color blindness simply means that a person cannot distinguish shades of red and green (usually blue-green). Their visual acuity (ability to see) is normal. There are no serious complications; however, affected individuals may not be considered for certain

occupations involving transportation or the Armed Forces where color recognition is required. Males are affected more often than females, because the gene is located on the X chromosome.

Hemophilia A. Hemophilia A is a disorder where the blood cannot clot properly due to a deficiency of a clotting factor called Factor VIII. This results in abnormally heavy bleeding that will not stop, even from a small cut. People with hemophilia A bruise easily and can have internal bleeding into their joints and muscles. The occurrence of hemophilia A (Factor VIII deficiency) is around 1 in 4500 live male births. The occurrence of hemophilia B (Factor IX deficiency) is one in 20,000 live male births. Hemophilia A accounts for most cases. Treatment is available by infusion of Factor VIII (blood transfusion). Female carriers of the gene may show some mild signs of Factor VIII deficiency, such as bruising easily or taking longer than usual to stop bleeding when cut. However, not all female carriers present these symptoms. One-third of all cases are thought to be new mutations in the family (not inherited from the mother).

Each child of a mother affected with an X-linked dominant trait has a 50% chance of inheriting the mutation and thus being affected with the disorder. If only the father is affected, 100% of the daughters will be affected, since they inherit their father's X chromosome, and 0% of the sons will be affected, since they inherit their father's Y chromosome.

There are less X-linked dominant conditions than X-linked recessive, because dominance in X-linkage requires the condition to present in females with only a fraction of the reduction in gene expression of autosomal dominance, since roughly half (or as many as 90% in some cases) of a particular parent's X chromosomes are inactivated in females.

Examples

Alport syndrome

Coffin–Lowry syndrome

Fragile X syndrome

Idiopathic hypoparathyroidism

Incontinentia pigmenti

Rett syndrome (RS)

Vitamin D resistant rickets (X-linked hypophosphatemia

Y Linked traits

Y-linked traits never occur in females, and occur in all male descendants of an affected male. The concepts of dominant and recessive do not apply to Y-linked traits, as only one allele (on

the Y) is ever present in any one (male) individual. Males with a single Y- or X-linked allele are described as hemizygotes, because only one allele is present.

Inheritance for genes on the Y chromosome **Supplement**Sex chromosomes are not only relevant for sex determination. They carry genes that are associated with other traits. Thus, it is not uncommon to see certain traits manifesting as sex-linked. In humans and other animals, there are two types of sex-linked inheritance:
X linked inheritance
Y linked In heritance

Y-linked inheritance is a form of inheritance for the genes located on the Y chromosome. In human and other mammalian males, the sex chromosomes are the X and Y chromosome. Y chromosome is smaller than the X chromosome and thus, carries relatively fewer genes. Apart from the genes for sperm development, appropriate hormonal output, and other traits characterizing males, the Y chromosome also carries genes other than for sex determination. Hypertrichosis of the ears, webbed toes, and porcupine man are examples of Y-linked inheritance in humans. Hypertrichosis of the ears (or hairy ears) is a condition wherein there is a conspicuous growth of hair on the outside rim of the ear. Webbed toes condition is characterized by having web-like connection between second and third toes. Porcupine man is a condition when the skin thickens and gradually becomes darker, scaly, rough, and with bristle-like outgrowths. Since Y-linked inheritance involves the Y chromosome, Y-linked inheritance is passed on from father to son.

Multifactorial traits or Polygenic triats

Polyallelic Traits "multiple gene inheritance" is a member of a group of non-epistatic genes that interact additively to influence a phenotypic trait. The term "monozygous" is usually used to refer to a hypothetical gene as it is often difficult to characterise the effect of an individual gene from the effects of other genes and the environment on a particular phenotype. Advances in statistical methodology and high throughput sequencing are, however, allowing researchers to locate candidate genes for the trait. In the case that such a gene is identified, it is referred to as a quantitative trait locus (QTL). These genes are generally pleiotropic as well. The genes that contribute to type 2 diabetes are thought to be mostly polygenes.[1] In July 2016, scientists reported identifying a set of 355 genes from the last universal common ancestor (LUCA) of all organisms living on Earth.[2]

Traits with polygenic determinism correspond to the classical quantitative characters, as opposed to the qualitative characters with monogenic or oligogenic determinism. In essence instead of two options, such as freckles or no freckles, there are many variations, like the color of skin, hair, or even eyes.

Polygenic locus is any individual locus which is included in the system of genes responsible for the genetic component of variation in a quantitative (polygenic) character. Allelic substitutions contribute to the variance in a specified quantitative character. Polygenic locus may be either a single or complex genetic locus in the conventional sense, i.e., either a single gene or closely linked block of functionally related genes

In modern sense, the inheritance mode of polygenic patterns is called polygenic inheritance, whose main properties may be summarized as follows:

Most metric and meristic traits are controlled by a number of genetic loci.

Main mode of nonallelic genes interaction in corresponding gene series is addition of mainly small particular allele contributions.

The effects of allelic substitution at each of the segregating genes are usually relatively small and interchangeable which results that identical phenotype may be displayed by a great variety of genotypes.

The phenotypic expression of the polygenic characters is undergoing considerable modification by environmental influence.

Polygenic characters show a continuous rather than discontinuous distribution.

Balanced systems of polygenic inheritance in a population contain a great deal of potential genetic variability in the heterozygous condition and released by small increments through genetic recombination between linked polygenes.

Multiallelic traits

it is one of two or more versions of a known mutation at the same place on a chromosome. It can also refer to different sequence variations for a several-hundred base-pair or more region of the genome that codes for a protein. Alleles can come in different extremes of size. At the lowest possible end one can be the single base choice of a single nucleotide polymorphism (SNP). At the higher end, it can be the sequence variations for the regions of the genome that code for the same protein which can be up to several thousand base-pairs long.

Sometimes, different alleles can result in different observable phenotypic traits, such as different pigmentation. A notable example of this trait of color variation is Gregor Mendel's discovery that the white and purple flower colors in pea plants were the result of "pure line" traits which could be used as a control for future experiments. However, most alleles result in little or no observable phenotypic variation.

Most multicellular organisms have two sets of chromosomes; that is, they are diploid. In this case, the chromosomes can be paired: each pair is made up of two homologous chromosomes. If both alleles of a gene at the locus on the homologous chromosomes are the same, they and the organism are homozygous with respect to that gene. If the alleles are different, they and the organism are heterozygous with respect to that gene

A B O Blood group

A population or species of organisms typically includes multiple alleles at each locus among various individuals. Allelic variation at a locus is measurable as the number of alleles (polymorphism) present, or the proportion of heterozygotes in the population. A null allele is a gene variant that lacks the gene's normal function because it either is not expressed, or the expressed protein is inactive.

For example, at the gene locus for the ABO blood type carbohydrate antigens in humans, classical genetics recognizes three alleles, I^A, I^B, and i, which determine compatibility of blood transfusions. Any individual has one of six possible genotypes (I^AI^A, I^Ai, I^BI^B, I^Bi, I^AI^B, and ii) which produce one of four possible phenotypes: "Type A" (produced by I^AI^A homozygous and I^Ai heterozygous genotypes), "Type B" (produced by I^BI^B homozygous and heterozygous genotypes), "Type AB" produced by heterozygous genotype, and "Type O" produced by ii homozygous genotype. (It is now known that each of the A, B, and O alleles is actually a class of multiple alleles with different DNA sequences that produce proteins with identical properties: more than 70 alleles are known at the ABO locus. Hence an individual with "Type A" blood may be an AO heterozygote, an AA homozygote, or an AA heterozygote with two different "A" alleles.)

Allelic dominanace

A number of genetic disorders are caused when an individual inherits two recessive alleles for a single-gene trait. Recessive genetic disorders include albinism, cystic fibrosis, galactosemia, phenylketonuria (PKU), and Tay–Sachs disease. Other disorders are also due to recessive alleles, but because the gene locus is located on the X chromosome, so that males have only one copy (that is, they are hemizygous), they are more frequent in males than in females. Examples include red-green color blindness and fragile X syndrome.Other disorders, such as Huntington's disease, occur when an individual inherits only one dominant allele.

Sex determination

A sex-determination system is a biological system that determines the development of sexual characteristics in an organism. Most organisms that create their offspring using sexual reproduction have two sexes. Occasionally, there are hermaphrodites in place of one or both sexes. A sex-determination system is a biological system that determines the development of sexual characteristics in an organism. Most organisms that create

their offspring using sexual reproduction have two sexes. Occasionally, there are hermaphrodites in place of one or both sexes. There are also some species that are only one sex due to parthenogenesis, the act of a female reproducing without fertilizationThe XX/XY sex-determination system is the most familiar, as it is found in humans. The XX/XY system is found in most other mammals, as well as some insects. In this system, most females have two of the same kind of sex chromosome (XX), while most males have two distinct sex chromosomes (XY). The X and Y sex chromosomes are different in shape and size from each other, unlike the rest of the chromosomes (autosomes), and are sometimes called allosomes. In some species, such as humans, organisms remain sex indifferent for a time after they're created; in others, however, such as fruit flies, sexual differentiation occurs as soon as the egg is fertilized

Spermatogenesis

Spermatogenesis is the process by which haploid spermatozoa develop from germ cells in the seminiferous tubules of the testis. This process starts with the mitotic division of the stem cells located close to the basement membrane of the tubulesThese cells are called spermatogonial stem cells. The mitotic division of these produces two types of cells. Type A cells replenish the stem cells, and type B cells differentiate into primary spermatocytes. The primary spermatocyte divides meiotically (Meiosis I) into two secondary spermatocytes; each secondary spermatocyte divides into two equal haploid spermatids by Meiosis II. The spermatids are transformed into spermatozoa (sperm) by the process of spermiogenesis. These develop into mature spermatozoa, also known as sperm cells. Thus, the primary spermatocyte gives rise to two cells, the secondary spermatocytes, and the two secondary spermatocytes by their subdivision produce four spermatozoa and four haploid cells

Spermatozoa are the mature male gametes in many sexually reproducing organisms. Thus, spermatogenesis is the male version of gametogenesis, of which the female equivalent is oogenesis. In mammals it occurs in the seminiferous tubules of the male testes in a stepwise fashion. Spermatogenesis is highly dependent upon optimal conditions for the process to occur correctly, and is essential for sexual reproduction. DNA methylation and histone modification have been implicated in the regulation of this process.[4] It starts at puberty and usually continues uninterrupted until death, although a slight decrease can be discerned in the quantity of produced sperm with increase in age .

Spermatogenesis starts in the bottom part of seminiferous tubes and, progressively, cells go deeper into tubes and moving along it until mature spermatozoa reaches the lumen, where mature spermatozoa are deposited. The division happens asynchronically; if the tube is cut transversally one could observe different maturation states. A group of cells with different maturation states that are being generated at the same time is called a spermatogenic wave

Spermatocytogenesis is the male form of gametocytogenesis and results in the formation of spermatocytes possessing half the normal complement of genetic material. In spermatocytogenesis, a diploid spermatogonium, which resides in the basal compartment of the seminiferous tubules, divides mitotically, producing two diploid intermediate cells called primary spermatocytes. Each primary spermatocyte then moves into the adluminal compartment of the seminiferous tubules and duplicates its DNA and subsequently undergoes *meiosis I* to produce two haploid secondary spermatocytes, which will later divide once more into haploid spermatids. This division implicates sources of genetic variation, such as random inclusion of either parental chromosomes, and chromosomal crossover that increases the genetic variability of the gamete. The DNA damage response (DDR) machinery plays an important role in spermatogenesis. The protein FMRP binds to meiotic chromosomes and regulates the dynamics of the DDR machinery during spermatogenesis.[13] FMRP appears to be necessary for the repair of DNA damage.

Each cell division from a spermatogonium to a spermatid is incomplete; the cells remain connected to one another by bridges of cytoplasm to allow synchronous development. Not all spermatogonia divide to produce spermatocytes; otherwise, the supply of spermatogonia would run out. Instead, spermatogonial stem cells divide mitotically to produce copies of themselves, ensuring a constant supply of spermatogonia to fuel spermatogenesis

Oogenesis

It starts with the process of developing primary oocytes, which occurs via the transformation of oogonia into primary oocytes, a process called oocytogenesis Oocytogenesis is complete either before or shortly after birth.

Number of primary oocytes

It is commonly believed that, when oocytogenesis is complete, no additional primary oocytes are created, in contrast to the male process of spermatogenesis, where gametocytes are continuously created. In other words, primary oocytes reach their maximum development at ~20weeks of gestational age, when approximately seven million primary oocytes have been created; however, at birth, this number has already been reduced to approximately 1-2 million.

Two publications have challenged the belief that a finite number of oocytes are set around the time of birth. The renewal of ovarian follicles from germline stem cells (originating from bone marrow and peripheral blood) has been reported in the postnatal mouse ovary. In contrast, DNA clock measurements do not indicate ongoing oogenesis during human females' lifetimes.Thus, further experiments are required to determine the true dynamics of small follicle formation.