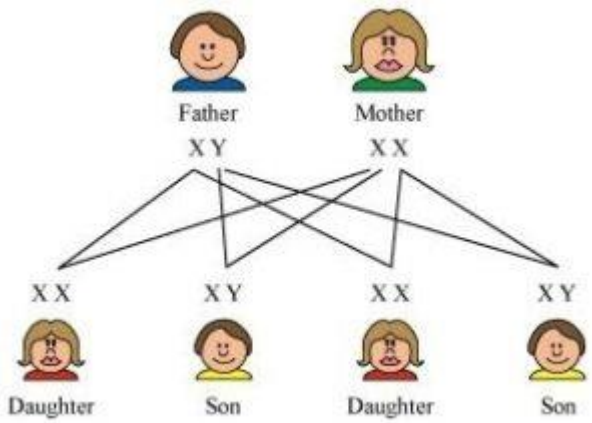
Sex Determination

Sex determination, is a system which decides the sexual characteristics of an organism or offspring. It helps to determine whether the organism will be male or a female, which are the two most common sexes. The widely used technique is chromosomal sex determination, in which sex chromosome of male i.e. X or Y chromosome, decides the sex or gender of the offspring. Female carries XX chromosome and male carries XY chromosome. It is also possible to perform genetic tests to eliminate any chromosomal or genetic disorders. Sex determination is the genetic process of determining the sex of the organism. Let us study in more detail about this.

Before we understand how sex determination is done, we need to understand the genetic makeup of a human being. As we all know, humans have 23 pairs or 46 chromosomes. Of these 23 pairs, 22 pairs are known as autosomes whereas 1 pair is known as the sex chromosome. It is this one pair that helps in determining the sex of an individual.

Sex Determination in Humans



Females in humans have 2X chromosomes- 1 each is inherited from either parent and denoted as XX. Males in humans have 1X and 1 Y chromosome, where the X is inherited from the mother and the Y from the father. In a way, we can say that it is the father who determines the sex of the unborn child. This can put to shame a lot of history in which people believed it is the woman who was responsible for not giving birth to a male heir. We can also say that the absence of the Y chromosome makes the individual a female.

At the time of spermatogenesis in males, both types of gametes are produced- one carrying the X chromosome and one carrying the Y. At the time of fertilization, the sex of the resulting zygote will depend on which gamete of the father will fuse with the X of the mother. We can, therefore, say that there is a 50% chance that the child will be a male and 50% that it can be a female.

Types of sex determination

- The XX-XY system as seen in human beings, where, XX is the female and XY is the male. This is also seen in a few insects.

Sex Linkage:

We know that the rules of Mendelian inheritance are not valid for all human traits. Besides the frequency of affected individuals is very different in the two sexes and the appearance of a trait in the offsprings is determined by the fact whether it is passed on by the father or the mother. However, for some of these traits it is now known that though the genes are located on the autosomes yet they have different phenotypic expressions in the two sexes. The traits determined by genes located on the sex chromosomes are called **sex –linked** and their mode of transmission as sex linked inheritance. Thus there are two types of **sex- linked inheritance** depending upon whether the genes are located on X or Y chromosome. It is claimed that in addition to completely X or Y linked genes, there are probably other genes which, by crossing over, are able to change their localization from X to Y chromosome and vice versa in the course of generations. Such genes are called partially or incompletely sex linked.

While considering the details of X and Y linked inheritance it is necessary to explain that a female with XX chromosomes naturally has two alleles at each locus. She may be homozygous or heterozygous for them. But in case of a male, there is only one X chromosome, he is called hemizygous for the single allele of X- linked genes. It may be pointed out that amongst the X and Y chromosomes which are not equal in size, there may exist a small section of Y chromosome which is homologous with the section of the X- chromosome. Thus there could be three possible sites for genes on the sex chromosome.

1. The non- homologous section of the Y- chromosome; such genes are totally Y-linked.

2. The non-homologous section of the X- chromosomes; such genes are totally X-linked.

3. The homologous section of the X and Y-chromosomes; such genes are said to be partially sex-linked as they can pass on from X to Y chromosomes or from Y to X- chromosome or from Y to X-chromosome during crossing over.

- **Sex linkage** describes the sex-specific patterns of inheritance and presentation when a gene mutation (allele) is present on a sex chromosome (allosome) rather than a non-sex chromosome (autosome). In humans, these are termed X-linked recessive, X-linked

dominant and Y-linked. The inheritance and presentation of all three differ depending on the sex of both the parent and the child. This makes them characteristically different from autosomal dominance and recessiveness.

- 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. As such, X-linked recessive conditions affect males much more commonly than females.
- 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 (and may occasionally present with symptoms due to aforementioned skewed X-inactivation).

Examples

- Aarskog–Scott syndrome
- Adrenoleukodystrophy (ALD)
- Bruton's agammaglobulinemia
- Color blindness
- Complete androgen insensitivity syndrome
- Congenital aqueductal stenosis (hydrocephalus)
- Duchenne muscular dystrophy
- Fabry disease
- Glucose-6-phosphate dehydrogenase deficiency
- Haemophilia A and B
- Hunter syndrome
- Inherited nephrogenic diabetes insipidus
- Menkes disease (kinky hair syndrome)
- Ornithine carbamoyltransferase deficiency
- Wiskott–Aldrich syndrome

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.

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 (CLS)
- Fragile X syndrome
- Idiopathic hypoparathyroidism
- Incontinentia pigmenti
- Rett syndrome (RS)
- Vitamin D resistant rickets (X-linked hypophosphatemia)

In classical genetics, a mating experiment called a reciprocal cross is performed to test if an animal's trait is sex-linked.

Y-linked

Y linkage, also known as **holandric inheritance** (from Ancient Greek ὅλος *hólos*, "whole" + ἀνδρός *andrós*, "male"), describes traits that are produced by genes located on the Y chromosome. It is a form of sex linkage.

Y linkage can be difficult to detect. This is partly because the Y chromosome is small and contains fewer genes than the autosomal chromosomes or the X chromosome. It is estimated to contain about 200 genes. Earlier, the human Y chromosome was thought to have little importance;. The Y-chromosome is sex-determining in humans and some other species: not all genes that play a role in sex determination are Y-linked. The Y-chromosome, generally does not undergo genetic recombination and only small regions called pseudoautosomal

regions exhibit recombination. The majority of the Y-chromosome genes that do not recombine are located in the "non-recombining region". A Y-linked condition will only be inherited from father to son and will always affect every generation.

For a trait to be considered Y linkage, it must exhibit these characteristics:

- occurs only in males
- appears in all sons of males who exhibit that trait
- is absent from daughters of trait carriers; instead the daughters that are phenotypically normal and do not have affected offspring.

These requirements were established by the pioneer of Y linkage, Curt Stern. Stern detailed in his paper genes he suspected to be y-linked. His requirements at first made Y linkage hard to prove. In the 1950s using human pedigrees, many genes were incorrectly determined to be Y-linked. Later research adopted more advanced techniques and more sophisticated statistical analysis. Hypertrichosis (Hairy ears) are an example of a gene once thought to be Y-linked in humans; however, that hypothesis was discredited. Due to advancements in DNA sequencing, Y linkage is getting easier to determine and prove. The Y-chromosome is almost entirely mapped, revealing many y-linked traits.

Y linkage is similar to, but different than X linkage; although, both are forms of sex linkage. X linkage can be genetically linked and sex-linked, while Y linkage can only be genetically linked. This is because males' cells have only one copy of the Y-chromosome. X-chromosomes have two copies, one from each parent permitting recombination. The X chromosome contains more genes and is substantially larger.

Y-chromosome deletions are a frequent genetic cause of male infertility.

Sex-influenced traits

Sex-influenced traits are those characters in which the dominance of an autosomal gene depends on the sex of the individual. In case of sex influenced traits the heterozygous express differently in male and females because of the differences in the male and female hormones. Thus a trait may be dominant in one sex and recessive in the other. Sex influenced traits is also known as sex controlled traits. This phenomenon is also known as Sex-influenced dominance. Example: baldness in humans. Even in a homozygous dominant or recessive female the condition may not be expressed fully.

Sex-limited traits

Genes are expressed only in one of the sexes, while they may be present in both the sexes. Sex limited characters are those whose expression is determined by the presence or absence of one of the sex hormones. The phenotypic effect is thus limited to one sex or the other. Thus one sex is uniform in the expression of a particular trait and yet the same genes produce a different phenotype or do not express

at all in the other sex in its prog*e*ny. Genes for the sex limited traits are autosomal and are present in both the sexes but their expression is limited to only one sex.

Example: Milk production in mammals, age of onset of menstruation, width of pelvis, distribution of body hairs are the examples of sex-limited traits.

Multifactorial or Polygenic Inheritance

Traits that are determined by the **aggregate effects of alleles at many loci** are called polygenic or multifactorial traits. For such traits the segregation of individual loci is extremely difficult and almost impossible to detect because effect of individual allele is quite small as compared to background effect of all other loci and the environment. Many traits which we observe are multifactorial. The measurement of body height shows continuous gradation in a population. similar grading can be observed for weight or skin pigmentation. Thus these are the traits which do not fall in clear cut classes. Haemophilia or blood types A, B, O or AB etc. traits are called discontinuous traits as they can be sharply distinguished between one group and the other. On the other hand traits like height, weight, skin pigmentation which cannot be clearly distinguished into one or the other group but vary bit by bit from one extreme to another in a quantitative manner are called continuous traits. Such traits cannot be explained on the basis of one or two alleles at a certain chromosome locus. They are in fact controlled by multiple genes which may be located on different loci of the same chromosome or different chromosomes. Such quantitative characters are found among traits that can be classified according to a numerical scale.

Genetic interpretation of inheritance of quantitatively graded characters was suggested by Mendel. He tentatively suggested that perhaps more than one pair of genes was responsible for the observed variation of colour of the flowers when he crossed white and purple red flowering beans. The hypothesis of multifactor or polygenic inheritance was later on proved to be correct by Nilsson- Ehle in an analysis in a graded series of seed pigmentation in wheat crosses (stern 1960:361). Genes with cumulative effect are important in understanding the determination of characters such as stature, body weight, body built, intelligence etc.

Examples of Polygenic Inheritance

Skin Colour

The pigment melanin is responsible for dark coloration in the skin and there are at least three genes, which control for human skin color. Using a hypothetical example where the production of melanin is controlled by *contributing alleles* (denoted here as A, B and C), resulting in dark skin color, and therefore light skin color is produced by *non contributing alleles* (denoted here as a, b and c), it is possible to see how the spectrum of different skin colors can result in the offspring.

It is important to remember here that in polygenic inheritance, alleles do not display dominance over others, rather, each contributing allele gives an additive effect rather than a masking effect, and so the way that the alleles interact is different to those in Mendelian genetics. The additive effect means that each contributing allele produces one unit of colour.

Human Height

Human height is an extremely complex inheritance pattern as there are over 400 genes controlling for it, it is therefore extremely difficult to predict the height that an offspring will be; two short parents may produce a tall child, whereas two tall parents can produce a short child and parents with completely different heights may produce a tall, short or intermediary child.

In addition, height is known as a *multifactorial trait*, which means that the trait is influenced by multiple genes as well as being affected by the environment. For example factors relating to general health of a growing child such as access to food and exposure to disease, could significantly affect the final height of a person. A large majority of our traits are multifactorial so it is often difficult to assess the effect that single genes have on a resulting phenotype

Dermatoglyphics

Dermatoglyphics is a study of configurations of epidermal ridges on certain body parts, namely, palms, fingers, soles, and toes. The term is derived from ancient Greek: derma = skin, glyph = carving. Dermatoglyphic patterns **begin to develop in the 10th week of gestation and are complete by the 24th week.** Fingerprints of both hands are not the same and persist lifelong unless dermis is damaged. They are mainly under genetic control and can be used in the diagnosis of congenital malformations. Their uniqueness has led to the analyses of one's potential and preferences. Dermatoglyphics, a study of configurations of epidermal ridges on the volar aspect of hands and feet, is an example of this. Dermatoglyphics (from ancient Greek derma = skin, glyph = carving) is the **scientific study of ridge patterns of the skin of the finger, palms, toes and soles.** The human body is covered with hairs and sebaceous (oil) glands except the palmar and plantar regions which are continuously corrugated with narrow ridges. The ridges make certain patterns. Dermatoglyphic patterns can deviate from normal in a wide array of disorders. Uses of dermatoglyphic traits are manifold, but the anthropologists are more concerned in establishing **variations in respect of traits among different human populations**. However the anthropologists are interested also in the study of dermatoglyphics in the context of **twin diagnosis, paternity diagnosis, primatology** etc.

Dermatoglyphics fulfil many of the conditions laid down by boyd for a good racial criterian.
Dermatoglyphics traits are **not modified by environmental factors**. Dermatoglyphics traits **are non adaptive.**

These are **not subjected to a high rate of mutation**. These traits are identifiable without any subjective bias. However the genetic process of dermatoglyphics traits is complex and is not perfectly known.

Henry has classified the various finger patterns into four main types. These are: arches, loops, true whorls and composites. The composites form a heterogenous assemblage of patterns. Again three types have been identified by Galton. His three types are arches, loops and whorls. A loop may be open to the ulnar side or to the radial side and accordingly it is termed as ulnar or radial loop. The classic and widely used notation is A = arches; Lr = radial loops; Lu = ulnar loops and W= whorls. The whorls possess two triradii, while only one triradius is present in loops. On the other hand triradius is absent in the arches. Thus generally speaking the patterns may be identified from the occurrence of the triradius.

It is evident that whorls are most frequent among the mongoloid population and least among the Caucasoid population. On the other hand loops appear most frequently among the Caucasoid groups, while among the Mongoloid and Negroid groups loops are found in equal frequencies. Again arches appear in very small number in the Mongoloid. It is most frequent in the Negroid. The position of Caucasoid is intermediate.

Usually three indices are calculated on the basis of the frequency distribution of the different finger patterns. These are as follows:

$$\text{Furuhat's Index} = \frac{\text{Whorl} \times 100}{\text{Loops}}$$

$$\text{Dankmeijer's Index} = \frac{\text{Arches} \times 100}{\text{Whorls}}$$

$$\text{Pattern Intensity Index} = \frac{2 \times \text{Whorl} + \text{Loops}}{n}$$

n being the total number of subjects.

arch pattern

loop pattern

whorl pattern

Dermatoglyphics analysis is an integration of brain science, medicine, genetics, psychology and behavioral science. Through nearly five centuries of observation and study of genetic medicine, amount and distribution of neurons is reflected in regular patterns on our fingerprints. According to European and American experts, they found that fingerprints show different kinds of characteristics, even with monozygotic (identical) twins, their fingerprints are different. At the same time, the same fingerprints will appear again after healing of wound, as long as the injury has not affect the cells.

The surface of the skin and its deeper structures show various linear markings. Over 35 different names, many of them synonyms, have been applied to such lines, relating to various systems of grooves, raised areas, preferred directions of stretching, lines of nervous occurrence, and spread of infection. Some of these are clearly evident in intact skin, others only appear after some sort of intervention, for example, pinching, while the actual existence of others is debatable.

Cummins and Midlo classified various pattern types on fingertips as:

a. Arch

b. Loop

c. Whorl

d. Composites.

They also advocated ridge counting in biological studies and its application to various pattern types as a measure of pattern size. The palmar surface is divided into dermatoglyphic areas, which are hypothenar, thenar, and the four interdigital areas numbered I to IV. There are four digital triradii and one or two axial triradii (t).

Uchida *et al.*, classified fingerprints into arch, loop, and whorl. Ridge count was used as a dermatoglyphic indicator. Digital triradii a, b, c, d, and axial triradius “t” were described. The position of “t” was described by measuring “atd” angle. Three major palmar flexion creases were mentioned alongwith a full or partial Simian crease present in some. They also worked on and described dermatoglyphic patterns in chromosomal abnormalities.

Triradius

A triradius is located at the meeting point of three opposing ridge system.

Patterns

There are three main types of patterns:-

Arch

The ridges pass from one margin of the digit to the other with a gentle, distally bowed sweep. There is no triradius

Loop

It possesses only one triradius. The ridges curve around only one extremity of the pattern, forming the head of the loop c. From the opposite extremity of the pattern, ridges flow to the margin of the digit, this extremity of the pattern may thus be described as 'open'. According to this, loops may further be of two types:

1-Ulnar loop – When the loop opens to the ulnar margin

2-Radial loop – When the loop opens to the radial margin.

Whorl

It is distinguished by concentric design. The majority of the ridges make circuits around the core. True whorls typically possess two triradii a. There are also composite patterns in which two or more designs are combined in one pattern area. They have two or more triradii. They are included under whorls.

Open fields (O)

These are configurations in which the ridges are essentially straight, and therefore, form no patterns.

Vestiges (V)

They lack the sharp recurvature of ridges which distinguish true patterns. It is merely a local

disarrangement of ridges.

Pattern intensity

This is the number of triradii on all the ten fingers of an individual. The value ranges from 0 to 20.

Ridge count

These are made from triradii point to point of the core. After locating the triradii point and point of the core, as outer and inner termini of the count, the line is set in position to connect them. Triradial point and print of core are not included in the count. As there are two counts in whorls, only the higher one is used. In single arches, the score is "zero." The count on the ten fingers of each individual is then summed up to give a single value, the total ridge count.

Palmar Dermatoglyphics

Interdigital areas

Interdigital intervals, the clefts between digits, are numbered in sequence beginning with the interval between the thumb and index finger. The palmer surface is divisible into dermatoglyphic areas or configurational fields, which are hypothenar, thenar and the four interdigital areas numbered I to IV. Each area is a topographic unit, and there is in some palms a discrete pattern and partial boundaries formed by triradii and their radiants for each area. Characteristically, there are four "digital triradii" located in proximal relation to the bases of digits II, III, IV, and V. In radioulnar sequence, they are named a, b, c, and d Axial triradius (t) is located at or near the proximal margin of the palms, in the interval between thenar and hypothenar eminences. The configurational area lying between digital triradii "a" and "b" is interdigital II, that between triradii "b" and "c" is interdigital III, and the area between triradii "c" and "d" is interdigital IV. When a digital triradius fails or is much displaced the midpoint of the base of the corresponding digit affords a landmark separating the interdigital areas on either side. The configuration may be a true pattern (whorl or loop), a vestige or an open field. Whenever there are two patterns in an area, the one on the radial side is written first.

Whorls are designated as "W"; loops as "L" or "l" if the pattern is small (ridge count is less than six), vestiges are 'V' and open fields "O."

The following three main line formulae are frequently observed in man**; 11.9.7, 9.7.5 and 7.5.5.** According to the Wilder the 11.9.7. is the European formula and 7.5.5. is the Negro formula.

Palmar flexion creases

The main flexion creases-distal transverse, proximal transverse and radial longitudinal.

Hypothenar patterns

There are three primary true patterns in the hypothenar area: whorls, loops, and tented arches.

“atd” angle

It is formed between lines drawn from the triradii at the bases of the index and little fingers to the axial triradius . The more distal the axial triradius, the larger is the angle. Positions of the axial triradii forming angles greater than 56° are designated “distal.” If more than one axial triradius is present, the most distal one is used in the analysis.

Simian line

Usually, three flexion creases are present on the palm. In some cases, however, the two distal horizontal creases are fused to form a single horizontal crease. This line is designated as a simian crease