UNIT 1 HUMAN GENETICS

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Learning Objectives

Once you have studied this unit, you should be able to understand

- how a single gene or genes form the physical hereditary link between generations, from parent to offspring;
- the determination of sex and the relation of sex to inheritance, and sex related human traits; and
- the important factors of human diversity and ethnic variation.

1.1 INTRODUCTION

The field of anthropology is basically concerned with both biological and social/ cultural evolution as well as diversity of human population throughout the ages. For understanding human genetics one needs to understand about physical anthropology or biological anthropology which deals with the study of human biology, evolution of the human organism, the relation between environment and human organism, and genetic variations between individuals and groups. The field of human genetics (of anthropological interest may be referred to as anthropological genetics which is concerned with human population variation study) uses pattern of genetic similarity and differences among different human populations throughout the human ancestry to infer the history of human evolution, migration, admixture and diversity. This would enable the scientists in explaining how the modern *Homo sapiens* evolved through the stages of *Homo habilis* and *Homo erectus* through the millennia to the modern man and the reasons behind many of the biological differences that we observe in different ethnic groups of the world.

Genetics is a branch of biology that deals with heredity or inherited variation of genetic traits. The science of genetics studies the phenotypic (visible) differences between individuals and attempts to relate them to underlying genic or chromosomal differences. The hereditary units that are transmitted through parent to offspring are called genes.

The word 'gene' is used frequently in genetics as a designation for each of the small units of heredity within a cell. Genetics has proved to have numerous practical applicatons because man has learned to use the discoveries in many diferent fields. It is being used in such diverse areas as plant and animal breeding, medical diagnosis, and genetic counseling, and even in cases of law. Genes are biochemical instructions that are supposed to determine those inherited traits that reside in the long molecules of deoxyribonucleic acid or DNA. Long polynucleotide molecules of *deoxyribonucleic acid*, called DNA, are intimately assocaited with chromosmes and are found exclusively in chromosomes. The chemical composition of chromosomes includes histones, proteins and deoxyribonecleic acid. The DNA is found only in chromosomes and is double stranded. The genes are then, sections of the DNA ladder-like molecules; different genes are different because they contain different sequence of the "letters' A, T, C, and G. DNA in conjunction with protein matrix may form nucleoprotein and becomes organised as chromosomes that are found in the nucleus of the cell. DNA is a stable molecule, however, on rare occasions a change or heritable alteration may occur spontaneously, is called mutation which is the lead sources of biological variation. In the study of heredity, we must clearly distinguish between 'genes'and 'traits'. Genes are at the bottom of developemnt. On the other hand, traits, such as hair colour, eye colour, size, shape, etc. are end products of development. They require both the proper genes and proper environment for their development.

1.2 HISTORY, DEFINITION AND SCOPE

Human Genetics, as the name indicates, describes the study of inherited variation as it occurs in human beings. The inheritance of many traits, including human traits is at present fairly well known. The biochemical studies on the constituents of the chromosomes have given essentially the correct picture how heredity really works at the molecular level. Genes can be the common factor of the most inherited traits. Genes have become prominent in the nature versus nurture debate. Study of human genetics can be useful as it can answer questions about human nature, behaviour as well as understand the diseases and disease treatment, and genetics of human life.

The science of genetics emerged from the famous work of Father Gregor Mendel (1822-1884) while working at the Augustinian monastery of St Thomas at Bruno in Moravia with the common garden pea. He published the results of his studies in 1866 and thereby laid the foundation of modern genetics. In his paper, Mendel proposed some basic genetic principles — the law of segregation, and the law of independent assortment. The first one states that each parent contains two copies of a unit of inheritance (later called gene), however, any one of two genes (called allele) can be transmitted to the offspring through the gamete. Which allele in a parent's pair of alleles is inherited is a matter of chance. The second principle states that the segregation of such gene pair (allele) occurs independently in respect of other gene pair, i.e. the paired genes (allelic pairs) separate from one

another and are distributed to different sex cells. The result is that new combinations of genes present in neither parent are possible. However, during his (Mendel) lifetime very few people realised the importance of his path breaking research. In 1901, Hugo de Vries, Carl Correns and Erich von Tschermak realised that Mendel's observations, conclusions and hypothesis have great importance in the field of genetics. During early 1900s, researchers noted that chromosomes behave like Mendel's traits and also inherited in random combinations. In 1909 Wilhelm Johansen renamed Mendel's characters as 'gene' and William Bateson coined the term 'genetics' to study genes. Thereafter researchers repeated and confirmed Mendel's hypothesis and his (Mendel) ideas on the inheritance of traits became more widely accepted and is now termed as Mendel's laws of inheritance.

Another milestone in the field of genetics is the discovery of the model for the structure of DNA as a genetic material by J.D. Watson and F.H.C. Crick in 1953. This was probably the key stone that unlocked an explosion in the field of human genetics as a form of molecular revolution.

Following are some of the fields where human genetics may contribute its knowledge for the betterment of the human society.

- To understand basic principles of inherited variation in man and to understand application of genetics in human life, and
- To answer questions about human nature, understand the diseases and development of effective disease treatment and health care.

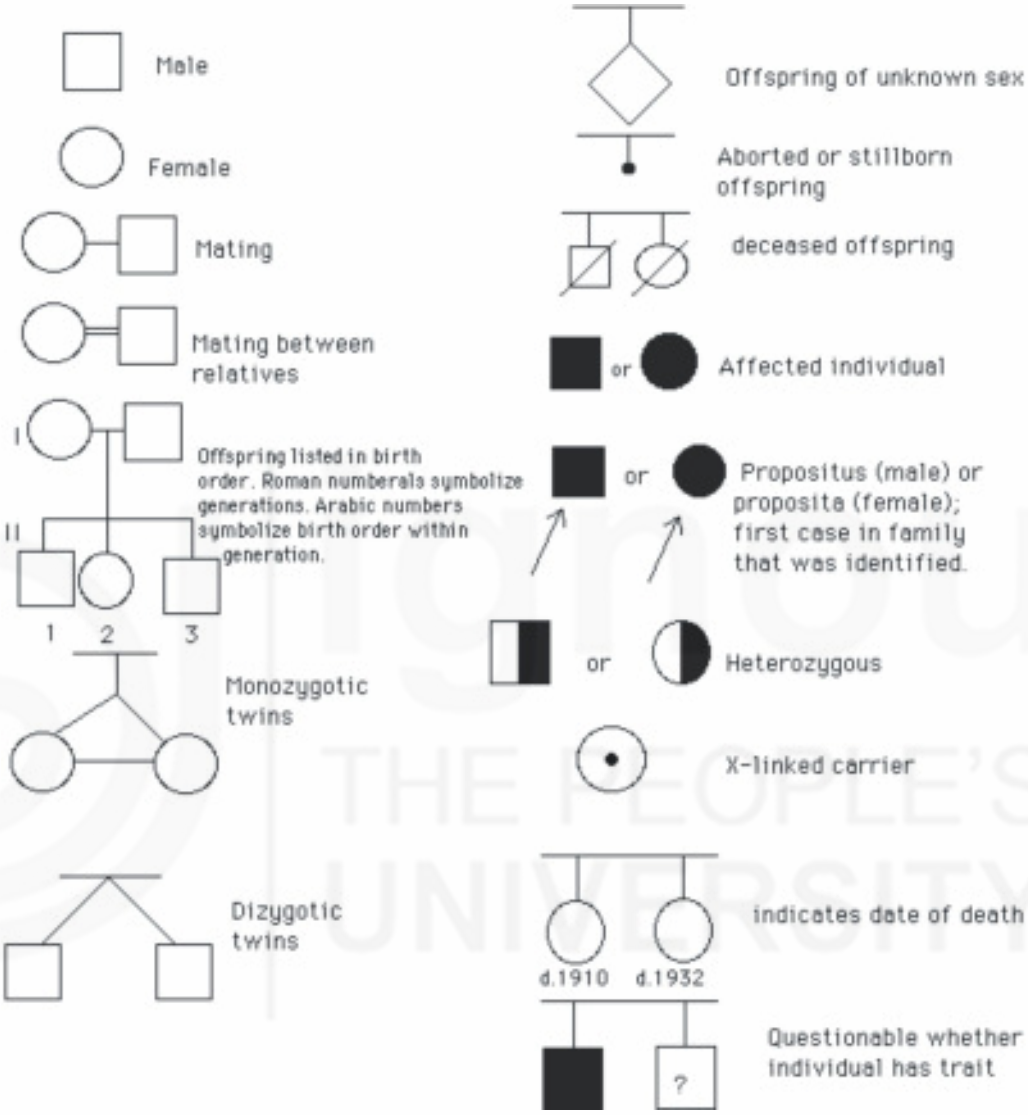
1.3 MENDELIAN INHERITANCE IN MAN

Mendelian traits or traits of simple inheritance are mainly discrete in nature and are controlled by alleles at single genetic locus. Therefore, in humans, traits or disorders that a single gene specifies are said to be Mendelian traits. Currently more than 4500 human traits are said to be inherited as per Mendelian principles; and another large conditions are suspected to be Mendelian traits. Many of the known Mendelian traits may be classified as disorders as per physical or mental disability. However, the most prevalent Mendelian disorders are very rare, usually affecting 1 in 10000 births or even less than that.

Human geneticist unlike others, who carried out experiment on plant or animal, can't have an access over experimental or controlled breeding. Hence they have to confine their study by observing the mode of inheritance in a pedigree. A Pedigree is a systematic drawing of the ancestral line of a given individual (both father and mother side) or family tree of a large number of individuals that depict blood relationship and transmission of inherited traits. A Pedigree can help to determine the genetic basis of a particular trait, especially in human, where experimental mating is not possible.

The term 'pedigree' (line of ancestors) is derived from French word '*pie de grue*' means crane's foot. The diagram of pedigree of large families with parents linked by curved lines to their offspring often resembled a bird's foot. You can tell a mode of inheritance just by looking at a pedigree. Pedigree is built of shapes connected by lines, vertical lines represent generations, horizontal lines that connect two or more shapes at their centers represent parents and vertical lines joined horizontally above them represent siblings. Matings are shown as

horizontal lines between two or more individuals. In case of shapes, *square* indicate male, *circles* indicate female and *diamonds* for unknown sex. Different shades or colours can be added to the symbols to identify different phenotype — full coloured shapes for individuals who express the trait under study and half-filled for carriers. Each generation is listed on a separate row labeled with Roman numerals, whereas, individuals within a generation labeled by Arabic numerals.



Source: www.bio.classes.UCSC.edu

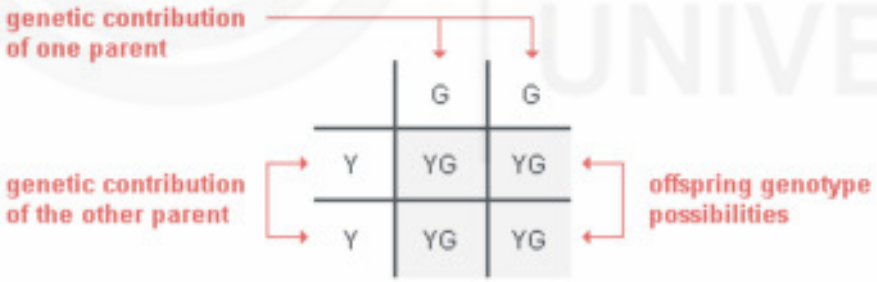
1.4 TYPES OF INHERITANCE

The patterns, in which Mendelian traits appear or transmitted in families, are called modes of inheritance. On the basis of chromosome where genes are located, you can find two types of inheritance - autosomal i.e. located on autosomes; and sex-chromosomal i.e. located on sex chromosomes, X or Y. Both autosomal and sex chromosomal inheritance may be subdivided as dominant or recessive inheritance on the basis of expression of alleles. However in respect of Y chromosome, there is no such subdivision like that described earlier. Hence, we have five modes of inheritance — autosomal recessive inheritance, autosomal dominant inheritance, X-linked recessive inheritance, X-linked dominant inheritance and Y-linked inheritance.

Mendel's observation of two different expressions of an inherited trait in a single locus (e.g. short or tall in respect of pea plant) narrates the facts that a gene can exist in alternate forms, usually called allele. An individual having two identical alleles is called homozygous, whereas the one with two different alleles is called heterozygous. Hence an individual may be homozygous either by two dominant alleles or two recessive alleles.

The allele that masks the effect of the other allele is called dominant (specifically completely dominant) and the masked one is called recessive. Whether the trait is dominant or recessive mostly depends upon the particular nature of the phenotype. Sometimes the heterozygous behave like an intermediate or a mix between homozygous dominant and homozygous recessive. Recessive disorders, in many cases, tend to be more severe or lethal and produce symptoms at an earlier age than dominant disorders.

If the genetic basis of a trait is known one can predict the outcomes of crosses. These are Punnett square method, forked line method and probability method. The ratios predicted from Mendel's law, apply to a new allele combination to each newly conceived offspring i.e. 50% chance of inheriting the allele, no matter what was the previous combination. You can compare the situation with tossing of coins; for first one the possibility of its being the head (or tail) is 50%. The same is true for second or any subsequent tossing. Therefore, if there is a 25% chance for a recessive disorder and first child is affected, there is no guaranty that next three will not be affected. The best way to calculate the probability of inherited traits was invented by Reginald Punnett and is called Punnett square. This is a simple graphical way to calculate all potential combinations of genotype for each time. You can start the same by drawing a grid of perpendicular lines. Now put the genotype of one parent across the top and other one down the left side. At last you can fill all the boxes by copying row and column letters (alleles).

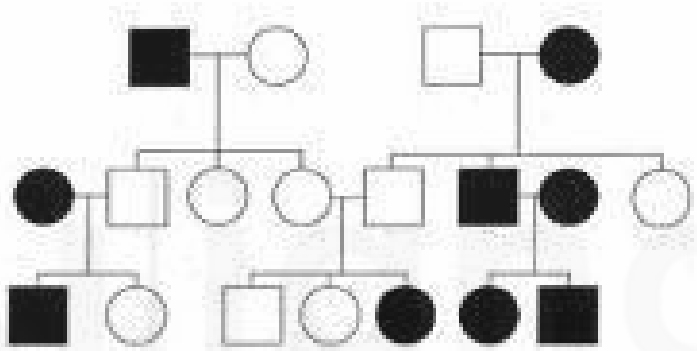


1.4.1 Autosomal Recessive Inheritance

Autosomal recessive trait can affect both sexes in equal proportions and can (but not necessarily) skip generation. The gene is carried on autosomes. For expression of recessive trait to be displayed, two copies of trait or allele needs to be present, which indicate that both the parents must be at least carrier for the specific traits. Therefore, a recessive trait can remain hidden for several generations without displaying the phenotype or diseases. The trait characteristically appears only in sibs, not in their parents, offspring or other relatives.

Sometimes a rare autosomal recessive trait may occur in families where the parents are close (blood) relatives, who are supposed to inherit the allele from a common ancestor. The situation is called consanguinity. Marriages between relatives - "consanguineous marriages", as they are often called, are important genetically.

Because closely related individuals have a higher chance of carrying the same alleles than less closely related individuals. The children from consanguineous marriages are more frequently homozygous for various alleles than are children from other marriages. In some ancient societies like the Pharaohs of ancient Egypt and the Incas of Peru favoured marriages of brothers and sisters of the ruling dynasties, to keep the 'royal blood' pure. These are extreme cases of consanguineous marriages. In some societies, more common types of close consanguinity are observed in cousin marriages. Examples of other consanguineous relations are those between uncle or aunt and nephew or niece (third degree), between cousins (fourth degree) and between second cousins (sixth degree). Consanguinity relations are identified by the number of steps from a common ancestor to only one of the related individuals, namely, the one more remote from him.



Source: www.migeneticsconnection.org

Some important characteristic features are:

- Occurrence and transmission is not influenced by sex;.
- Traits can express only in homozygous condition;
- In a pedigree you can find the trait only in siblings, not in their parents;
- On average ¼ th of the sibs of the proband are affected;
- In the instance of a rare disease, affected individuals have normal parents;
- Ratio of affected, carrier and non-affected is 1:2:1 (in sibs); and
- Parents of an affected child, in many cases, are close blood relatives.

Results from each of the six possible crosses are summarized in Table 1.1

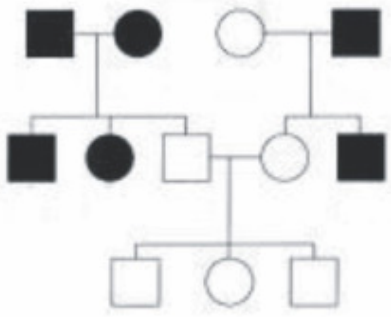
Table 1.1: Summary of Autosomal Recessive inheritance

Parents	Offspring
One parent homozygous Normal Other parent homozygous Normal	All the offspring will be homozygous normal
One parent homozygous Normal Other parent heterozygous Normal (Carrier)	50% probability that offspring will be homozygous normal 50% probability that offspring will be heterozygous normal (Carrier)

One parent heterozygous Normal (Carrier) Other parent heterozygous Normal (Carrier)	25% probability that offspring will be homozygous normal 50% probability that offspring will be heterozygous normal (Carrier) 25% probability that offspring will be affected
One parent homozygous Normal Other parent affected	All the offspring will be heterozygous normal (Carrier)
One parent heterozygous Normal (Carrier) Other parent affected	50% probability that offspring will be heterozygous normal (Carrier) 50% probability that offspring will be affected
One parent affected Other parent affected	All the offspring will be affected

1.4.2 Autosomal Dominant Inheritance

Autosomal dominant trait, like autosomal recessive traits, can affect both sexes in equal proportions; the gene is carried on autosomes but unlike previous one does not skip generations. If no offspring inherits the trait in any generation its transmission stops. The trait is called "dominant" because a single copy of the trait, inherited from either parent, is enough to cause this trait to appear; the dominant allele masks the recessive one. Hence both homozygous dominant and heterozygous individual can express the trait. This often means that at least one parent must have the trait to transmit; otherwise it may appear because of mutation. Unaffected family members do not transmit the trait to their children. Dominance and recessiveness are obviously developmental phenomena resulting from genic action. They refer to the effect of a combination of differing alleles as compared to the effect of a homozygous combination.



Source: www.migeneticsconnection.org

Some important characteristic features are:

- Occurrence and transmission is not influenced by sex;
- Traits can express in both homozygous and heterozygous condition;
- You can find the trait in every generation of a pedigree;
- Affected individuals are usually born of normal parents;
- Affected individuals are always the product of a parent carrier of the same character;

- Trait always transmitted by an affected person (if heterozygous he/she is supposed to transmit the trait to half of the children and if homozygous to all the children); and
- All children of a normal individual will be normal i.e. unaffected family members do not transmit the trait to their children.

Results from each of the six possible crosses are summarized in Table 1.2

Table 1.2: Summary of Autosomal Dominant inheritance

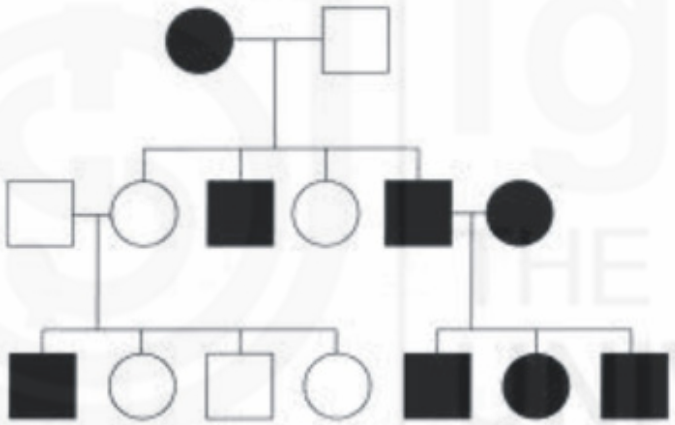
Parents	Offspring
One parent homozygous affected Other parent homozygous affected	All the offspring will be homozygous affected 50% probability that offspring will be homozygous affected 50% probability that offspring will be heterozygous affected
One parent homozygous affected Other parent heterozygous affected	25% probability that offspring will be homozygous affected 50% probability that offspring will be heterozygous affected 25% probability that offspring will be normal
One parent heterozygous affected Other parent heterozygous affected	All the offspring will be heterozygous affected
One parent homozygous affected Other parent normal One parent heterozygous affected Other parent normal	50% probability that offspring will be heterozygous affected 50% probability that offspring will be normal
One parent normal Other parent normal	All the offspring will be normal

Sex-Linkage: In the human species, the sex-chromosomes contain many more genes than those concerned with sex-determination. These affect the widest range of characters and bear no relation to sex. 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 X-chromosome, 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.

1.4.3 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.



Source: www.migeneticsconnection.org

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.

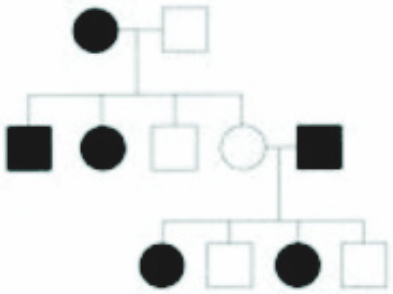
Results from each of the six possible crosses are summarized in Table 1.3

Table 1.3: Summary of 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

1.4.4 X-linked Dominant Inheritance

X-linked dominant inheritance shows the same phenotype as a heterozygote and homozygote. Incase 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.



Source: www.migeneticsconnection.org

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; and
- X linked dominant cannot distinguish from Autosomal Dominant by progeny of affected females, but only from the progeny of affected males.

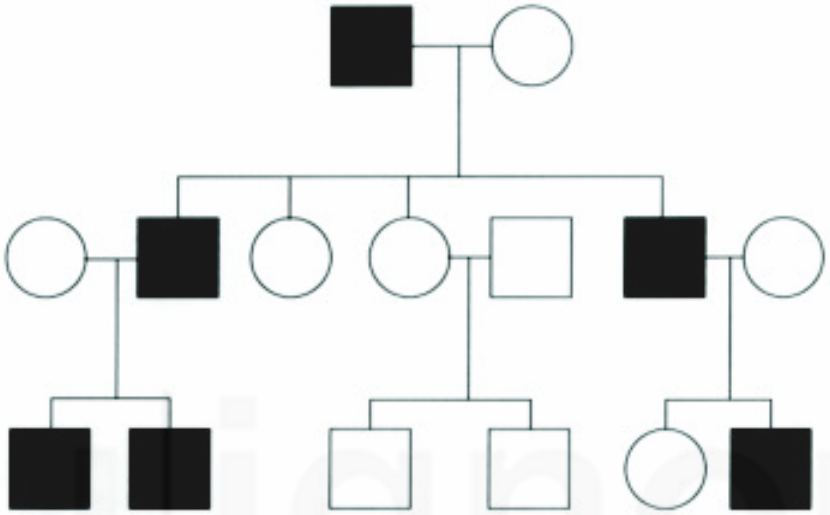
Results from each of the six possible crosses are summarized in Table 1.4

Table 1.4: Summary of 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

1.4.5 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 (www.sakshieducation.com).



Source: www.migeneticsconnection.org

Some important characteristic features are-

- In pedigree, only males are affected;
- Affected male transmitted the trait to all his sons but never to his daughter; and
- No skipping of generations.

Results from each of the two possible crosses are summarized in table 1.5

Table 1.5: Summary of Y-linked inheritance

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

1.5 EXAMPLES

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 and young adults, and may result in early death.	Cystic Fibrosis	Normal
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 imperfecta
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, $C_6H_5NHCSNH_2$, 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

Y-linked traits
Hypertrichosis of ear: growth of hair on the rim of pinna
Testis determining factor (TDF)

Note: You can find out more examples from NCBI databases OMIM: Online Mendelian Inheritance in Man

Sex-limited and Sex-controlled Traits

So far we have been discussing about sex-linked traits, but there are some such genes which are sex-limited in their effect, that is, they are expressed phenotypically in one sex only. In man sex-limited expression of genes occurs in uterine (in female) and prostate (in male) cancer. Anatomical and physiological properties of the female sex, such as width of pelvis or age of onset of menstruation is a sex-limited expression. Similarly, sex-limited male characters such as type of beard growth or amount and distribution of body hair, probably depend on genes common to both sexes, but the penetrance and expressivity of the genes are more limited to males. Sex-limitation is only the extreme example of control of the expression of certain genotypes by sex.

When a genotype is expressed in both sexes but in a different manner in each, we speak of sex-controlled, or sex-modified, genic expression. Sex-controlled dominance has been suggested as an explanation of the pattern of inheritance of baldness in man. Both sexes may be affected, but the high relative frequency of affected males is notable. Some of these traits are controlled by the sexual constitution of the individual and thus are under the influence of sex hormones.

1.6 SUMMARY

Mendel considered a single gene to be responsible for a single trait, but after the discovery of other types of non-Mendelian inheritance it is now clear that many genes may be involved for the production of single or many traits. Mendel's

laws also incorporate many of the modern discoveries which enriched these laws. For example, chromosome or meiosis was discovered after Mendel's work. Now we can correlate that Mendel's first law i.e. law of segregation is about anaphase-I where homologous chromosomes segregate from each other. Similarly as per second law, segregation of alleles for one character follow independently of the segregation of allele of other character because each pair of homologous behaves like an independent unit during meiosis. Again, especially after modern discovery, we can understand that it is the gene and not the trait (as per Mendel) that are inherited.

Since Mendel's time, understanding of the mechanisms of genetic inheritance has grown immeasurably. The simple rules of Mendelian inheritance do not apply in elucidating many of the inheritance patterns, and are understood to be non-Mendelian inheritance patterns.

1.7 GLOSSARY

Allele : an alternate form of gene that determine alternate traits or characteristics.

Autosomal dominant : the inheritance pattern of a dominant allele on autosomes.

Autosomal recessive : the inheritance pattern of a recessive allele on autosomes.

Autosomes : a non-sex determining chromosome. Human has 22 pairs of autosomes.

Carriers : a heterozygous individual who possess a deleterious recessive allele which is suppressed by dominant normal allele.

Chromosome : a structure within a cell's nucleus that carries gene and consists of a continuous molecule of DNA and proteins.

Consanguineous : relating to or denoting people descended from the same ancestor.

Dominant trait : the trait that is expressed in the F_1 generation.

DNA : a long linear polymer found in the nucleus of a cell, formed from nucleotides and shaped like a double helix; generally associated with the transmission of genetic information.

Gene : a sequence of DNA that instructs a cell to produce a particular protein.

Genetics : branch of biology that concerned with heredity and variation.

Heterozygous : having two different alleles of a gene at a single locus and produces different kinds of gametes.

Homozygous : having two identical alleles of a gene single locus and produces only one kind of gamete.

Mutant : an allele that differs from wild type allele, altering the phenotype.

Mutation : any event that changes genetic structure; any alteration in the inherited nucleic acid sequence of the genotype of an organism.

Pedigree : a chart consisting of symbols for individuals connected by lines that depict blood relationships and transmission of inherited traits.

Probability : probability is a way of expressing mathematical knowledge that an event will occur or has occurred.

Proband : proband, or propositus, is a term used most often in genetics to denote a particular subject (person in human genetics) being studied or reported on.

Protein : a type of macromolecule that is the direct product of genetic information.

Recessive trait : the trait that is masked in the F_1 hybrids.

Sex cells : sex cells are the cells that give rise to the gametes of organisms that reproduce sexually.

Sex Chromosome : a chromosome containing genes that specify sex.

Sex linked : genes that are part of a sex chromosome.

Variable expression : a genotype producing phenotype that varies among individuals.

X linked dominant : the inheritance pattern of a dominant allele on X chromosome.

X linked recessive : the inheritance pattern of a recessive allele on X chromosome.

Y linked : the inheritance pattern of a gene on Y chromosome.

Suggested Reading

Cummings, M. R. 1997. *Human Heredity: Principles and Issues*. Belmont, Wadsworth.

Gardner, E. J., Simmons, M.J. and Snustad, D.P. 1991. *Principles of Genetics*. New York, John Willey & Sons.

Lewis, R. 2003. *Human Genetics: Concepts and Applications 5th edition*. Boston, WCB McGraw Hill.

Stern, C. 1960. *Principles of Human Genetics* (2nd Edition). San Francisco and London, W. H. Freeman and Company.

Strickberger, M. W. 2003. *Genetics 3rd edition*. New Delhi, Prentice Hall.

Website

Mendel Web: www.mendelweb.org

National Center for Biotechnology Information: Online Mendelian Inheritance in Man: www.ncbi.nih.gov

Sample Questions

1) What is autosomal recessive trait? From marriages between normally pigmented carrier people and albinos what proportion of children would be expected to be albino and normal? What is the chance in a family of three children that one would be normal and two albinos?

2) What do you mean by X linked recessive trait? From marriage between carrier female and affected male what proportion of children would be expected to be Haemophilic?

3) What do you mean by Human Genetics? Write a brief note on Mendelian genetics in Man.

4) What are X linked traits? How does X linked dominant trait is differentiated from Autosomal dominant trait?

5) What is Pedigree? Draw a pedigree of X linked recessive traits in Man.

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