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Scope of Study: Today, more and more people are becoming interested in genetics. Most of the work done in this field does not cover human heredity for several reasons. The most important reason is that man has such a long life cycle. Geneticists get their knowledge about human inheritance by studying family pedigrees. From this information they can predict how a certain trait or characteristic is inherited. This study is an accumulation of material that will help to introduce to capable high school students some of the work in human heredity that has been done. The history of genetics really begins with Gregor Mendel who gave us the basic information for the laws of segregation and independent assortment. Most human traits are inherited by simple dominance, recessiveness, and sex linkage. There are examples of pedigree studies for most all traits or characteristics of the parts and systems of the body such as eye color and hemophilia. These studies lead to practical applications such as the prognosis of whether two people should marry and to the diagnosis of certain diseases.

ADVISER'S APPROVAL James H. Zant

A STUDY GUIDE FOR A UNIT ON HUMAN HEREDITY FOR THE ADVANCED HIGH SCHOOL BIOLOGY STUDENT

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A STUDY GUIDE FOR A UNIT ON HUMAN HEREDITY

FOR THE ADVANCED HIGH SCHOOL

BIOLOGY STUDENT

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PREFACE

Many high schools of today are offering advanced biology classes for the very capable and interested students. This paper is written to be used as a guide for teaching human genetics in such a course.

There has not been too much work done in the field of human heredity because of the long life cycle of man and other involving problems. The work that has been done, however, should be brought to the attention of more people. This paper will show some of the advantages one can gain by having an adequate knowledge of heredity. A paper, such as this one, is mostly written for those students preparing themselves for a career in the medical and zoological fields.

Indebtedness is acknowledged to Drs. James Zant and Herbert Bruneau, Oklahoma State University, and Dr. Mary Warters, Centenary College of Louisiana, for their assistance and guidance in writing this paper; and to my sister, Velma Bernice Sanders, for her assistance in typing this paper.

TABLE OF CONTENTS

CHAPTER I

HISTORY OF GENETICS

Genetics, the science of heredity, is becoming one of our most interesting and important subjects. Ever since the hydrogen bombs, there has been much more experimentation done in this field to determine the effects of radiation. In the classroom, there are questions about why are eyes blue and not brown. Therefore, it is important to try to bring to students more and more information in order to answer their questions. This chapter is a review of some of the history of genetics. Later chapters will develop human genetics more.

Ancient records show us that even the Greek philosopher, Aristotle, gave heredity some thought and made an attempt to understand its causes. However, the actual history dates back to 1857 when Sir Francis Galton began a series of studies on inheritance which resulted in significant conclusions concerning variation within a species.

It was actually, however, Gregor Mendel, an obscure Austrian monk who was the founder of this field. In 1864, he read a paper about his work with plants before a scientific society of his day. However, the significance of his great work was not appreciated for 36 years, 16 years after Mendel had died.

About 1900, three scientists working in different parts of the world came upon practically the same thing that Mendel had discovered. They were: DeVries in Holland, Correns in Germany, and Tschermak in Austria. Then Mendel's paper was republished and he received credit due

him. Thus, the science of genetics was born. His work developed into the famous Mendel Laws of segregation and independent assortment.

Mendel did not know what went on within the cell that was responsible for his 3:1 and 9:3:3:1 ratios. In 1902, W. S. Sutton and C. E. McClung in this country, and Theodore Boveri in Germany, decided that hereditary factors within the chromosomes were responsible for heredity. In 1911, another American scientist, Thomas Hunt Morgan conceived the idea of genes, lying in a linear fashion on the chromosomes.[1] He was convinced of this because whenever he saw irregular behavior in the inheritance of certain traits in his Drosophila (fruit fly) cultures he also noted irregularities in their chromosomes and, conversely, any deviation in the chromosome pattern was reflected in abnormal flies. From this information Morgan and his associates were able to construct chromosome maps showing rather definitely where the genes lay with respect to one another on the four chromosomes in Drosophila. Today, even the sex chromosomes of man have been mapped.

Many efforts were made to see the genes within the chromosomes. As early as 1881, Balbiani in Italy pointed out the giant chromosomes in the salivary glands of certain flies. In recent years these cells have been studied. The chromosomes appear under the light microscope as ribbons of alternately dark and light discs or bands. There is considerable proof now to show that each disc corresponds to a group of genes with respect to location on the chromosome.

Geneticists today know that genes are nucleoproteins and that cells contain desoxyribose nucleic acid (DNA) in chromosomes and ribonucleic acid (RNA) in both the chromosomes and cytoplasm. This is the present

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[1]Alfred Elliott, Zoology (New York, 1952), p. 585

stage in the history of genetics. Geneticists are spending most of their time now studing the chemistry phase of heredity.

CHAPTER II

HUMAN HEREDITY

At this particular time when the world is becoming so full of numerous undesirable abnormalities, it is time for man to start thinking about the principles of human heredity. Most of the basic ones have already been discussed in an elementary biolory course. Most everything mentioned in this paper will be to tell how a certain trait is inherited as students should become aware of this.

Practical Applications of Human Heredity

Man is not a particularly favorable object for genetic study. He has a long life cycle and comparatively small individual progenies. The understanding of the genetics of any organism depends first of all upon a knowledge of what characteristics are inherited. A great deal of what we know about the inheritance of different traits in man has come from the analysis of pedigrees and of twin studies.

Just what are some of the important reasons for studying human heredity? First of all it is important for genetic prognosis. This covers the field where doctors and counselors give advice in prospective marriages and prospective families. A doctor should be able to tell a prospective father affected with a dominant abnormality that half his offspring can be expected to suffer from the same thing. He should be able to give encouraging advice as well as discouraging. In making the prognosis, the doctor should observe the trait, describe, and identify

it. Then a check of the literature about the inheritance of the characteristic should be made. Next, the family history should be checked to see if the inheritance is similar to ones in the literature. Last of all, study the potential environment. Then make the prognosis.

A second application is in the diagnosis of a certain disease. On the basis of the family history, a diagnosis may be made when the conditions are too difficult to identify accurately by other means. For example in the case where a child was suffering from dry skin, sparse hair, undeveloped teeth, a doctor without checking into the background said it was low metabolism and gave thyroid treatment which proved harmful.[1] Another doctor traced history and found it was the dominant hereditary form of ectodermal dysplasia which is a trait that involves the total absence of sweat glands as well as hair and tooth deficiencies.

The third application is the instituting of preventive measures against certain diseases on the basis of the known genetic background.

The fourth application is in the testing of paternity and other medico-legal problems on the basis of various blood groups.

The fifth application is in the setting up of eugenic programs for the protection and improvement of society.

The Vocabulary of Human Genetics

DOMINANT: An inheritable condition or trait which shows up in the offspring over its opposite or recessive trait.

RECESSIVE: An inheritable condition or characteristic which does not

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[1]Adrian M. Srb and Ray D. Owen, General Genetics (San Francisco, 1952), pp. 530-531.

show in the presence of its opposite or dominant condition.

SEX LINKAGE: A condition where genes are tied in some fashion to a particular sex.

GYNEPHORIC: A condition of a sex-linked recessive where the female is the carrier of a trait.

HOLANDRIC: Inheritance where the character is transmitted only from father to son.

HEMIZYGOUS: An unbalanced condition of a gene in which it has no mate.

SYNDROME: A medical term for a condition which expresses itself in apparently unrelated effects on several different organs of the body.

PLEIOTROPIC: A term used where a gene has different effects on different organs of the body.

ANTICIPATION: A progressively earlier appearance of an inherited condition in successive generations.

HETEROPHERY: The sudden appearance of a new abnormality in several members of a family.

HOLOGYNIC INHERITANCE: Condition where the character is transmitted only from mother to daughter in the female line. It is probably caused by attached Xs.

SEX-LIMITED INHERITANCE: Difference in penetrance in the sexes.

POLYMERIC or POLYGENIC FACTORS: Those factors applying to racial features as skin color and head shape; and where two or more genes are causing a character such as goiter.

ISOPHENIC: A term for genes having similar phenotypic effects.

EXPRESSIVITY: A degree to which a gene can express itself.

POSITION EFFECT: The expression of a gene may be altered in different locations.

TRIAD INHERITANCE: This is where a trait is inherited by sex linkage; recessive, and dominant genes.

SEMI-SEX LINKED: A condition where a trait or characteristic is dominant in one sex and recessive in the other.

HOMOCHRONOUS: The appearance of a certain character at a certain age of life.

SUB-LETHAL: The case where certain characters cause death in childhood and not at birth.

AUTOSOMAL GENES: These are genes found on chromosomes other than the sex chromosomes.

Examples of Human Inheritance

As a person grows older and thinks of having a family some day, the thought of how certain traits or characters are inherited comes to one's mind. This section will be divided into some of the main areas of the human body and will describe the more common characteristics of each and how it is inherited.

EYES: There are many conditions that lead to blindness. One of the most common conditions is the cataract. This is actually a clouding up of the lens. It is a dominant characteristic with low penetrance. Another condition of the eyes is glaucoma where liquid of the iris presents pressure on nerve of cornea. It is caused by a dominant gene. Retinitis pigmentosa, a development of a pigment alongside the blood vessels of the retina, can also lead to blindness. This usually appears before one is twenty years old and is caused by triad dominant inheritance. Aniridia, absence of an iris is another cause of blindness. This is an example of a dominant gene with skips. Anophthalmus, the absence of the eyeball, is a recessive condition. Microphthalmus, a reduced eyeball condition, is generally a

recessive sex-linked trait; but sometimes is triad. Optic atrophy, a degeneration of the optic nerve is triad with sex linkage leading.

There are several defects of the eyes that cause trouble but don't lead to blindness. There are two kinds of color blindness--complete and incomplete. In complete color blindness everything is black and gray. In incomplete color blindness red is green, blue is yellow. Both kinds are caused by sex-linked genes. Nyctolpia or night blindness is a sex-linked recessive or triad trait. This condition is attributed to an insufficiency of rods and retinal purple. Hyperopia, far-sightedness, is dominant while myopia, near-sightedness, is generally recessive. Cornea astigmatism which is caused by unequal curvature of the cornea that causes objects in one place to be in sharper focus than objects in another place, is a dominant characteristic. Strabismus or cross eyes is a triad trait. Nystagmus,which is an uncontrolled movement of the eyes is a sex-linked recessive and autosomal dominant trait.

Some abnormalities of the physical appearance of the eyes are mongolian eye fold, ptosis, and cryptophthalmas. The first of these is a condition where a fold of skin from the upper eyelid extends down over the inner corner of the eye to produce this distinctive appearance. It is a dominant trait. Ptosis, another dominant trait, is a condition where a person is unable to use the muscles which raise the upper eyelids giving a drooping appearance. Cryptophthalmas is a condition where the lids fail to separate and the baby is born unable to open its eyes. This condition is a recessive autosomal trait.

Eye color is an interesting color. Brown eyes are dominant over blue eyes. Other colors are heterozygotes of brown. The blue color of the iris is due to a pigment in the back (or retinal layer) of the iris as seen through a semi-opaque, colorless layer in front of the iris. In

those with darker eyes, it will be masked by the development of melanin in the front part of the iris.

EARS: The size, shape, and structure of ears are inherited. There are two different kinds of ear lobes, if one will observe. These two kinds are attached and free lobes. The free lobes are dominant over the attached ones. A dense hair growth on the ears called hypertrichosis is a holandric trait. The complete absence of an ear is a dominant characteristic. Some people are born with natural earring holes which are depressions located in exactly the same spot where young ladies of times passed pierced their ears for earrings. This trait caused by an autosomal dominant gene shows incomplete penetrance and variable expressivity.

There are many reasons for deafness. Congenital deafness is caused by two sets of recessive genes. Deafness after maturity or otosclerosis is accompanied by ringing or buzzing in the ears. This condition is induced by abnormal growth of bone around the bones of the middle ear which transmit sound vibrations and make hearing possible. There are more women than men affected with this, and it is believed to be caused by the interaction of autosomal dominant and sex-linked dominant genes. Deafness that appears around the age of forty is caused by the atrophy of auditory nerves which transmit impulses from the hearing organs of the ears to the brain. This is caused by an autosomal dominant gene. Otitis, a tendency for inflammation of the middle ear is caused by recessive genes.

NOSE: Multiple genes affect the length, shape and width of the nose. A high convex bridge of nose is dominant over a straight bridge or concave bridge. A dominant gene caused the root of the nose at the forehead to be higher in some individuals than in others. A straight tip of the nose is dominant over an upturned tip. Wide spread of wings of the nose is dominant over a narrow spread of wings. High wings which show openings of the nose are recessive to lower types.

MOUTH: One characteristic about the mouth is the lips. Full lips are dominant over thin lips. A deformity of the lips is harelip which is usually accompanied by a cleft palate. This is caused by both recessive and dominant genes with greater penetrance in the male than in the female.

Perhaps one never thinks of different shape tongues, but there are several. Backfold and U-shaped tongues are dominant over clover-leaf shape. The inheritance of aglossia, absence of the tongue, is not known. The adherent tongue which is an example of holandric inheritance is a condition where the tongue cannot be raised to the palate to pronounce K or G.

The absence of teeth may be hereditary. If the missing teeth are the upper incisors and molars, central incisors and lateral incisors, a dominant gene is responsible. If the missing teeth are canines, a sex-linked gene is responsible. The lack of wisdom teeth is a dominant characteristic. If the enamel of teeth is missing, this is caused by a dominant sex-linked gene. Very small teeth, microdontia, is a recessive characteristic. Malocclusion is a condition of irregular teeth that results from the crossing of races with large and small jaws. The growth of the gums over the teeth, called hyperplasia, is caused by a dominant gene. Early decay of the teeth is hologynic inheritance.

A receding chin is recessive to a straight chin. A protruding chin and overhanging lower lip is caused by a dominant gene. A dimple in the chin and cheeks is a dominant characteristic.

SKIN: The pigmentation of the skin is interesting. If there is a pigment, it is dominant. There are two sets of genes responsible for color between marriages of Negro and non-Negro. Certain dominant genes cause characteristics as freckles and piebalding. Albinism is a recessive trait. Freckles is the condition where the pigment tends to accumulate

in isolated little islands which become very prominent when darkened by exposure to the sunlight. Piebalding is where there are large white spots on the skin of Negroes that tend to remain constant.

There are many skin abnormalities. A few will be mentioned here. Xeroderma pigmentosum is a condition where there is extreme sensitivity of eyes and skin to light. This is a recessive case where freckles may show in the heterozygote. This may also be an incompletely sex-linked case. As a child grows older, certain regions of the rash frequently become malignant and death nearly always results before maturity is reached. Anidrotic ectodermal dysplasis is where there is a deficiency in the sweat glands, a sparse growth of hair on the head and body, and a deficiency of teeth. This is caused by both dominant and sex-linked recessive genes. Keratosis pilaris is a condition where the eyelashes and eyebrows are absent, the scalp is more or less completely bald, and the skin is dry, hairy and more or less scaly. This is recessive inheritance. Verrucosis is a condition of warts occurring on the skin of face, scalp, trunk, and limbs. These warts are oval, grayish white or yellor in color. These are due to recessive genes and to predisposition of the disease.

HAIR: Hair color is due to pigmentation. Two pigments, granula and mellarin, are the ones responsible. Dark pigments are dominant, red is recessive. Gray hair is the lack of pigmentation with air spaces being formed. As a person gets older, the whiter their hair gets. In certain pedigrees, premature grayness is inherited as an autosomal dominant characteristic. Sometimes a person has a white forelock or blaze. This is a simple dominant trait.

It is hard to believe, but curly hair is dominant over straight hair. The abnormality of ringed hair is inherited as a simple recessive trait.

This is a condition of air spaces being between pigment. When a whorl of hair goes counterclockwise, we have a recessive trait called widow's peak.

Baldness is a sex-limited characteristic being dominant in males and recessive in females. Much hair, hypertrichosis, is dominant. Hypotrichosis, thin hair is recessive.

HANDS and FEET: When geneticists describe traits of the hands and feet, they more or less describe the abnormal traits. A few of these will be discussed here. Syndactyly, the fusion of bones of the adjacent toes and fingers, is a holandric trait. Zygodactyly, commonly known as webbed toes, is a fusion of the flesh. This also is a holandric trait. Crooked little fingers, called streblomicrodactyly, is caused by an irregularly dominant gene. Polydactyly, the presence of extra fingers and toes, is dominant with variable expressivity. Hammer toe, a condition where the big toe overlaps others, is dominant. The inheritance of clubbed feet is still uncertain. The absence of hands and feet known as acheiropodia is recessive. Flat foot, a common ailment of many people is a recessive trait. Right-handedness is dominant over left-handedness. Monodactyly, a dominant trait, is where only the little finger is present. The absence of digits, ectrodactly, is recessive. Anonychia pollicun, the absence of the thumb nails, is dominant.

SKELETON: Achondroplastic dwarfism is a dominant condition where the head and trunk of body are normal in size, but there is a great reduction in the length of the limbs which results in a short stature. This condition is accompanied by a deformity of the long bones. The legs are usually set wide apart at the hips and curve inward as they extend down toward the feet giving a bowed appearance. The head is somewhat deformed. An ateliotic dwarf is well proportioned, but small in all parts of the skeleton. This condition is due to a deficiency of the growth

hormone of the pituitary gland and may be due to two recessive genes of at least two independent loci. Osteo-chondrodystrophy is a condition resulting from an overall irregularity in the bone development which produces deformities of the trunk and the limbs. The limbs may be of normal length, but the trunk may be abnormally shortened. This is a sex-linked recessive trait. Osteopsathyrosis is a condition of fragile bones associated with a blue color of the sclera of the eye and with otosclerosis which results in deafness. This is a dominant trait. Dysostosis cleidocranialis is the absence of the clavicles or collar bones giving a person the ability to fold their shoulders inward until they touch or almost touch under the chin. Associated with this trait is a soft spot on the head. This is caused by a dominant gene. The looseness of the ligaments which hold bones in place may result in various defects of the posture and also account for the ability of affected persons to perform unusual contortions. This condition is associated with orthostatic albuminuria (presence of albumin in the urine only when the person has been sitting or standing for some time). This is a dominant trait with a high penetrance. Rickets is a bone deformity caused by a deficiency of Vitamin D. The susceptibility to this is dominant. Arthritis is a condition of soreness and stiffness of the joints between the bones. The susceptibility to this is dominant.

MUSCLES: Genes for muscular development as a whole are sex limited. Muscles of women usually contain heavier deposits of fat than those of men. Pseudohypertrophic muscular dystrophy is the gradual wasting away of muscles beginning in childhood and usually resulting in death in the early teens. This is caused by a sex-linked recessive gene occurring in boys only. Peroneal atrophy is the progressive wasting away of calf muscles usually beginning between the ages of ten and thirty. The muscles of the

forearm and hand are affected in some cases. This is triad inheritance with the sex linked form being the most severe. The complete absence of the long palmar muscle of the forearm can be easily detected since the muscle is connected with the hand by a prominent tendon that runs down the central region of the wrist. This missing muscle occurs because of a dominant gene with a slight reduction of penetrance. This absence causes no handicap in the use of the hands and arms.

DIGESTIVE SYSTEM: There are several kinds of ulcers =gastric, pepic, and duodenal. Gastric ulcer is caused by a dominant gene with no skips. Pepic ulcer is an irregular dominant trait which occurs more frequent in males than in females. Duodenal ulcer is a recessive trait. The tendency toward appendicitis is caused by a dominant gene with skips sometimes. Disturbances of the gall bladder are due to an irregular dominant gene. Hypertrophic pyloric stenosis is an infantile and sometimes a congenital condition in which there is projectile vomiting and constipation. The symptoms develop around the third or fourth week of life. There is also gastric peristalsis and a furrowed brow. This condition is caused by the thickening of the pyloric stomach wall, hypertrophy of the enormous circular muscle of the pylorus, the circular fibers of the pyloric sphincter and by a recessive gene.

RESPIRATORY SYSTEM: The susceptibility to tuberculosis is of a recessive type of gene.

CIRCULATORY SYSTEM: Pernicious anemia is where there is an insufficient number of red blood cells to transport oxygen in quantities needed for the best performance of the cells of the body. The red blood cells are not being produced by red bone marrow rapidly enough to supply the body needs. There is a deficiency of an antianemic factor formed in the cells of the stomach wall which is stored in the liver and, hence, an extract of liver

of other animals can relieve a victim. This is a dominant characteristic with considerable variation in expressivity. Hemolytic icterus results from a hyperfragility of red cells which causes their excessive destruction by the spleen. This causes the spleen to hypertrophy because of the excessive labor it is called on to perform which, in turn, leads to increased destruction of red cells. This is a dominant trait with variation in expressivity and can be corrected by removing the spleen. Thalassemia is a special type of anemia which appears in infancy or childhood and is accompanied by an enlarged spleen and liver, abnormally shaped red blood cells and an excess of leucocytes in the blood. This condition is intermediate in dominance and is confined to Syrians, Greeks and Italians. Sicklemia is a condition where red blood cells assume a sickle shape whenever they are held in a medium which is deficient in oxygen. This condition is common in Negroes and rare in whites. The cause of the sickle shape is due to a special kind of hemoglobin which is present in these cells. An intermediate gene is responsible. When the gene is homozygous, a person has sickle-cell anemia. When the gene is heterozygous, a person has sicklemia. Leukemia is a dominant condition where the white cells seem to destroy the red cells. Hemorrhagic nephritis is a disease where the capillary walls of the kidneys rupture easily and allow blood to filter into the urine. This is caused by a dominant gene. Hemophilia, a disease in which blood fails to clot properly, is caused by a sex-linked recessive gene. Phlebectasis or varicose veins is a condition where the veins of the legs bulge out under the skin because of the decline in elasticity of the walls of the vein. This is a dominant characteristic with incomplete penetrance. Hemorrhoids is a dominant characteristic caused by varicose veins in the walls of the rectum and may occur independently of the condition in the veins of the legs. High blood pressure, or hypertension,

may be accompanied by kidney trouble, heart trouble and apoplectic stroke. There may be a number of genes which contribute to the nature and severity of the symptoms but it is usually a dominant gene. Eosinophilia, caused by dominant and recessive genes, is an increase of eosinophilic leukocytes. This condition may be caused by allergy, by dermatoses or by presence of parasites in the body. Polycythemia vera (vaquez's disease) is a condition of a high count of erythrocytes and an enlarged spleen. This condition, caused by dominant and recessive genes produces a red face and tongue. Target cell anemia or leptocytosis is a disturbance of liver function; non-hemolytic jaundice, yellow skin and eyes. This is dominant inheritance. A recessive trait is methemoglobinemia which is the presence of methemoglobin in the blood in large amounts. This substance has a chocolate-brown color and cannot carry oxygen.

One of the best known characteristics in man is inheited by the multiple alleles. This characteristic is the blood groups A, B, AB and O. Three alleles are responsible for the various groups: genes A, A^B and a, of which the first two are dominant to the last. Gene A controls the formation of anti-A (agglutinogen); gene A^B controls the formation of anti-B; and gene a is without effect in that it causes no antibody formation. Neither A nor A^B is dominant to the other, so when both are present in homologous chromosomes the blood group AB results. Since these genes are inherited in a definite fashion, the knowledge of blood types has some value in addition to that needed in transfusions. In cases of questionable paternity the knowledge of blood types can be used and may rule out certain males as possible fathers of a child.

EXCRETORY SYSTEM: Nephritis, an inflammation of the kidneys is a dominant disease with skips. Polycystic disease of the kidneys may sometimes be congenital with the kidneys being enlarged before birth. It is

a dominant characteristic with skips. Addison's disease of the suprarenals is a fatal condition displaying a deep brown or bronze skin. This is a simple dominant characteristic.

ENDOCRINE SYSTEM: Diabetes mellitus is a condition where the victims produce an insufficient amount of insulin in the pancreas and sugar metabolism is deffective as a consequence. The excess sugar accumulates in the system and is excreted into the urine. The predisposition to this is dominant but can be avoided if the intake of carbohydrate foods is moderate. Diabetes insipidus is a condition due to an insufficient secretion of a hormone from the posterior lobe of the pituitary gland. This hormone (pitressin) regulates the reabsorption of water in the renal tubules. When it is deficient, an inadequate amount of water is reabsorbed and consequently an excessive amount of water is eliminated into the urine. This condition is accompanied by an extreme thirst because of the loss of water through the kidneys. This type of diabetes shows dominant inheritance with reduced penetrance. Gout is a condition resulting from a perversion of purine metabolism that results from excessive production of uric acid. Persons with this condition have an abnormally high uric acid content in the blood, have attacks of acute arthritis and sometimes have chalky deposits form in the cartilage of the joints. Many people have hyperuricemia (high uric content of the blood) who do not have gout. Hyperuricemia is a dominant characteristic with gout being a higher stage of it and is found mostly in males.

NERVOUS SYSTEM: There are two forms of amaurotic idiocy--infantile and juvenile. The infantile form appears after several months after birth. This recessive condition causes a gradual decline in mental ability, impairment of vision leading to blindness, convulsions, progressive muscular weakness and emaciation. The juvenile form appears at the age of six or seven

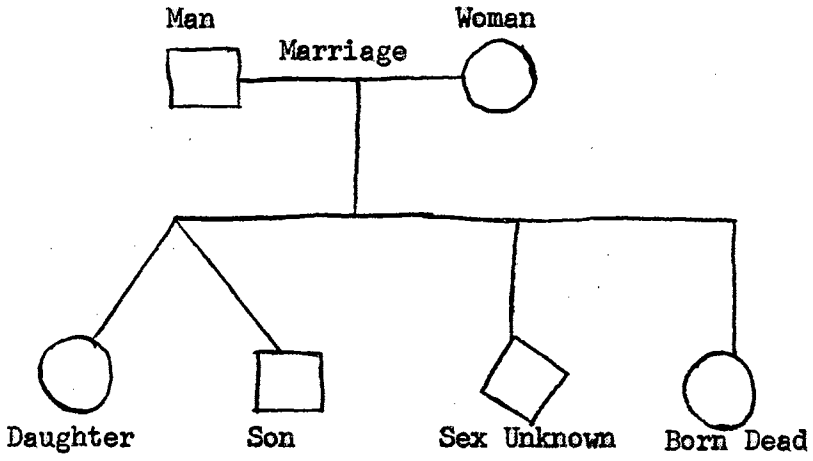
showing progressive lost of vision followed by mental deterioration, then muscular incoordination and finally the wasting away of muscles and mental obliteration with death occurring before the age of twenty-one. This is caused also by a recessive gene but a different one from the one that causes the infantile form. Huntington's chorea is a form of St. Vitus dance showing uncontrolled twitching of the voluntary muscles of the body. This dominant trait usually appears in the thirties and is accompanied by mental deterioration. Schizophrenia is a condition caused by multiple recessive genes in which the victims have almost incomplete insensibility to surroundings. Manic-depressive psychosis is a condition where the victims have periods of happiness and then sadness. This condition is caused by a dominant gene with the penetrance dependable upon some environmental agents. Senile dementia is a condition where a person has a brilliant mind in his early years but in his old age begins to show a progressive deterioration of mental faculties which may lead to complete loss of normal mentality. This condition is influenced by heredity. Epilepsy is a condition where the victim has fits which may run to unconsciousness and muscular spasms. It may be inherited by a dominant gene or brought on by a brain injury. Spinal ataxia results from a degeneration of sensory neurons in the spinal cord. With loss of sensation from the muscles comes a defective control of the muscles making the afflicted person have difficulty in maintaining his equilibrium. A person may lose all power of purposeful action through inability to control the voluntary muscles and may become a helpless invalid. This condition may be caused by both recessive and dominant genes. However, when the dominant gene is responsible, a person has brain degeneration rather than spinal cord degeneration. Hypertrophic neuritis is an enlargement of the spinal nerves followed by ataxia of the arms and legs and eventually muscular

atrophy develops between birth and maturity. This is caused by a recessive gene. Shaking palsy, gradual loss of control of muscles, appears at fifty or sixty years of age. Tremulous motions replace purposeful movements. Other characteristics are rigidity of movement, a mask-like face and a propensity to bend the trunk forward and pass from a walking to a running pace. This condition is due to the degeneration of large ganglion cells in the corpus striatum and also by dominant and recessive genes. Psychasthenia is a functional neurosis in which there is a pathological fear and anxiety with obsessions, fixed ideas, tics, unreality, and self-accusation. This is usually caused by a dominant gene. Dipsomania, a morbid craving for alcohol, is caused by a sex-linked gene expressed in the males and recessive in the females.

CHAPTER III

PROJECTS AND ACTIVITIES

1. Make a chart of your own family. A good way to start is to make the diagram of squares and circles to include as many relatives as possible. The more persons included, the more your chart will mean. Get all of the information that you can from parents, aunts, uncles and grandparents. Make a list of a number of traits and number them. Place the number of any trait in the square or circle which represents a person that had that trait.



See if you can tell from the completed chart whether certain traits have been dominant or recessive in your family. How have you been affected by any of these traits found in your relative? As help in starting the chart, you might use the suggestions from the list of traits below.[1]

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[1] B. B. Vance and D. F. Miller, Biology For You (3rd ed., Chicago, 1954), p. 509.

Hair type

1. straight
2. curly
3. black
4. brown
5. red
6. blond

Eye color

7. brown
8. gray
9. blue

Eyelashes

10. long
11. short
12. straight
13. curved

Fingers

14. long
15. short
16. tapering
17. square
18. joint missing
19. joint fused
20. extra finger
21. fingers joined
22. curved
23. short wide nails
24. long narrow nails

Special ability

25. art
26. music
27. dramatics

Health

28. susceptibility to disease
29. age at death
30. robust

2. Make a chart with movable chromosomes. On a sheet of plain paper draw circles 1 1/2 inches in diameter. Place the circles so as to represent the P_1 generation and their germ cells, the F_1 generation and their germ cells, and the F_2 generation.

Get two packets of paper matches that are made from different colors of paper or color one set differently. Cut off the match heads. Cut off pieces of the sticks about 1/2 or 3/4 of an inch long. Let these represent the chromosomes and place them in the first circles.

Choose a problem in heredity and let the match sticks represent the chromosomes carrying the genes used in the problem. Move the chromosomes so as to show what germ cells can be formed. Then move the chromosomes to show how they would combine at fertilization to form the F_1 generation. Continue through the problem.

This kind of movable chart can be adapted to different kinds of heredity problems and is a splendid aid in visualizing the chromosome behavior and the mechanics of inheritance. Try a problem with a dihybrid cross.[2]

3. Genetics of Human Blood Groups. The human blood groups are better understood genetically than any other human characteristic. A knowledge of the blood groups has important medico-legal applications. It is also important to physicians in giving transfusions.

The red blood cells carry the antigens and the serum contains the antibodies. There are four principal blood groups (phenotypes): O, A, B, and AB.

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[2]B. B. Vance and D. F. Miller, Biology for You (3rd ed., Chicago, 1954), p. 510.

The genotypes of the different groups are as follows:

Group	Genotypes
O	ii
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B

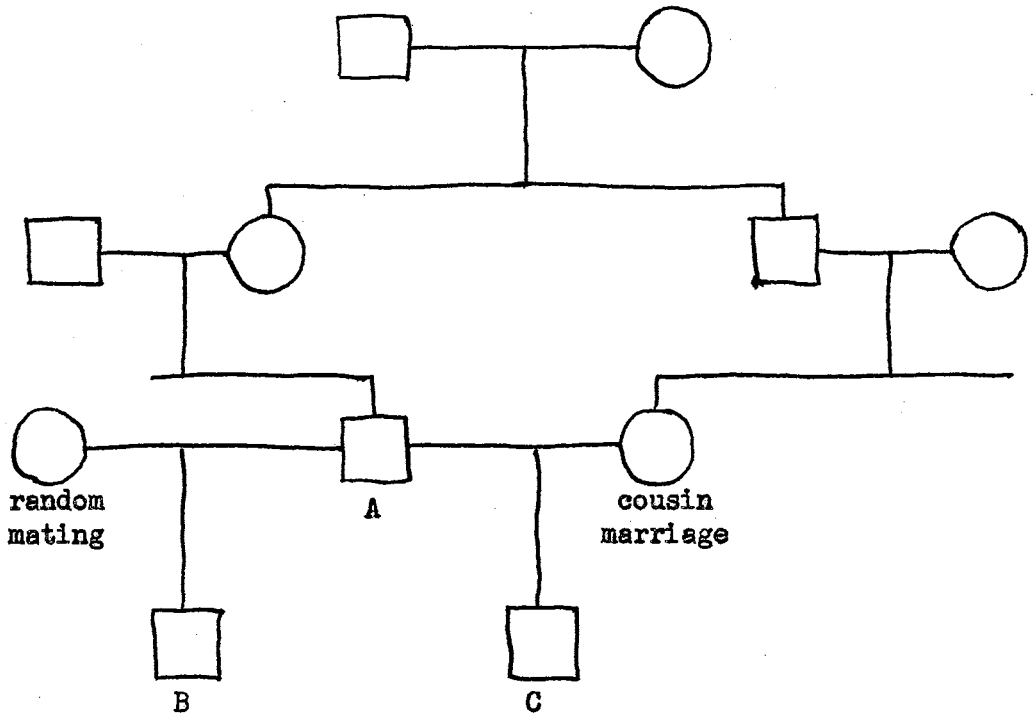
Prepare a microscope slide by marking two rings with a wax pencil, each about one-half inch in diameter; label one A and the other B. The instructor will give you a drop each of A and B test sera. Add a drop of blood to each of the test sera and rock the slide gently for a few minutes. Observe under the microscope. If neither of the blood samples clump, the blood is type O. If both samples clump, the blood is AB. If only the Anti-B serum mixture clumps, the blood is group B. If only the Anti-A serum mixture clumps, the blood is group A. To what blood group do you belong? _____. What types of cells should your own serum agglutinate? _____.

Give the phenotypic ratios expected from the following crosses:[3]

Cross	Blood Group A	Blood Group B	Blood Group AB	Blood Group O
$I^Ai \times I^Bi$				
$I^AI^B \times I^Bi$				

[3] Eldon J. Gardner, Genetics Laboratory Manual (Minneapolis, 1952), p. 15.

4. Consanguineous marriages play an important part in the expression of recessive traits. If albinism (aa) occurs in the general population at the rate of one in 40,000 people, what would be the chance of it occurring in "A's" child in the following pedigree if he married at random and if the trait was not known to occur in his family? _____.
What would be the chance if he married his cousin? _____.



If A's grandmother was known to have been an albino, what would be the chance of it occurring in B? _____.
What would be the chance of it occurring in C? _____.[4]

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[4] Eldon J. Gardner, Genetics Laboratory Manual (Minneapolis, 1952), p. 53.

5. A rather frequent inherited difference among people is in the ability to taste a compound called phenylthiourea. To about 70 per cent of North American Whites this compound tastes very bitter; the remaining 30 per cent find it virtually tasteless. The ability to taste this compound is dominant, so that we can let TT and Tt = "tasters"; tt = "non-tasters."[5]

Make a chart of your family pedigree concerning the ability to taste or not taste phenylthiourea.

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[5]Adrian M. Srb and Ray D. Owen, General Genetics (San Francisco, 1952), p. 52.

SUMMARY

Today, more and more people are becoming interested in genetics. Most of the work done in this field does not cover human heredity for several reasons. The most important reason is that man has such a long life cycle. Geneticists get their knowledge about human inheritance by studying family pedigrees. From this information they can predict how a certain trait or characteristic is inherited.

This study is an accumulation of material that will help to introduce to capable high school students some of the work in human heredity that has been done. The history of genetics really begins with Gregor Mendel who gave us the basic information for the laws of segregation and independent assortment. Most human traits are inherited by simple dominance, recessiveness, and sex linkage. There are examples of pedigree studies for most all traits or characteristics of the parts and systems of the body such as eye color and hemophilia. These studies lead to practical applications such as the prognosis of whether two people should marry and to the diagnosis of certain diseases.

A SELECTED BIBLIOGRAPHY

Elliott, Alfred M. Zoology. New York: Appleton Century Crofts, Inc., 1952.

Gardner, Eldon J. Genetics Laboratory Manual. Minneapolis: Burgess Publishing Co., 1952.

Gates, Reginald Ruggles. Human Genetics. New York: MacMillan Co., 1946.

Horowitz, Norman. "The Gene." Scientific American, (October, 1956), 17-27.

Mirsky, A. E. "The Chemistry of Heredity." Scientific American, (February, 1953), 28-38.

Snyder, Laurence H. and Paul R. David. The Principles of Heredity. Boston: D. C. Heath and Co., 1946.

Sonneborn, T. M. "Partner of the Genes." Scientific American, (November, 1950), 39-49.

Srb, Adrian M., and Ray D. Owen. General Genetics. San Francisco: W. H. Freeman and Co., 1952.

Stein, Curt. Principles of Human Genetics. San Francisco: W. H. Freeman and Co., 1960.

Sturtevant, A. H. "Social Implications of the Genetics of Man." Classic Papers in Genetics. Ed. James Peters. Englewood Cliffs, N. J.: Prentice - Hall, Inc., 1959, pp. 259-263.

Sutton, Walter S. "Chromosomes in Heredity." Classic Papers in Genetics. Ed. James Peters. Englewood Cliffs, N. J.: Prentice - Hall, Inc., 1959, pp. 27-41.

Vance, B. B., and D. F. Miller. Biology For You. Chicago: J. B. Lippincott Co., 1954.

_____, C. A. Barker, and D. F. Miller. Biology Activities. Chicago: J. B. Lippincott Co., 1954.

Winchester, A. M. Genetics. Cambridge, Mass.: Houghton Mifflin Co., 1958.

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