SYLLABUS FOR A UNIT ON HUMAN HEREDITY

FOR SECONDARY BIOLOGY

By

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A SYLLABUS FOR A UNIT ON HUMAN HEREDITY

FOR SECONDARY BIOLOGY

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CHAPTER I

THE PROBLEM

The Problem Stated

It is evident that almost everyone is interested in heredity, especially the heredity that is related directly to their lives. It is a subject which appeals to all age groups, the adolescent boy or girl, the bride and groom, and the mature person. High school students have shown unusual interest in genetics and generally want to know more about human heredity. However it should be noted that the average youth leaves high school with very little knowledge about human heredity. This is unfortunate in the light of the fact that completion of high school ends the formal schooling of many students.

Because of these reasons the writer decided to prepare a syllabus for a unit on human heredity for high school biology.

Purpose of this Study

The purpose of this study is three fold: (1) to provide the writer with a satisfactory knowledge of a few selected characteristics of the field of human heredity that may be easily communicated to high school biology students; (2) to present a general guide which could serve as resource material for teachers in the teaching of human heredity on the high school level, and (3) an attempt to build up a background of information about human heredity and provide sufficient reference for the

teacher to proceed to further studies in particular areas of this topic that he may choose.

Limitations of this Study

This study is limited by several factors. First, since there is so much literature on this subject, hence this study will be a survey of the field rather than an attempt to thoroughly investigate it. Secondly, in order to limit the area of study so that adequate treatment could be given to the ideas presented, it has been necessary to select a few topics out of many related and equally important topics. Thirdly, since the mechanism of heredity is so linked with the process of reproduction, it is not possible to get a clear picture of heredity without some understanding of this body function. However information on this topic has been omitted from this study because it is likely that most individuals who read this report will have a basic knowledge of reproduction.

Plan of Procedure

Generally, this study has been based mostly upon elementary literature in this field. Information was gathered from many different sources. A variety of textbooks on genetics have been surveyed as well as many other books, articles, bulletins, and other research studies devoted to some specific area of the study of genetics.

The first part of the study has been devoted to the historical background of genetics. Mendelian Laws and their application to human heredity have been studied.

Throughout the study the use of hereditary characteristics in man has been used to illustrate such factors as multiple alleles, sex-determination,

sex-linked, and sex-influenced characteristics. The last part of this report involved the study of linkage, crossing over, and several characteristics inherited by man.

Most of the study presents data from the literature used. It is quite basic to the expert in this field. However, the material has been presented in such a way that it may be easily communicated to the high school student.

CHAPTER II

HISTORICAL BACKGROUND

Most of our knowledge in the field of genetics has been developed since the beginning of the present century. As we search back into the history of scientific thought, however we find that speculations on the nature of heredity are as ancient as the history of mankind.

The first accounts of definite attempts to formulate an explanation for conception and inheritance were largely speculation, with little, if any, support. Pythagoras, 500 BC, proposed the theory that a moist vapor descends from the brain, nerves, and other body parts of the male during coitus and that from this, similar parts of the embryo are formed in the uterus of the female. This would give us some explanation of the transmission of likeness from the male parent to the offspring, but leaves unclear how the female could transmit any qualities.

About two hundred years later Aristotle proposed that the semen of the male was produced from the blood, and that it was, in fact, highly purified blood. In such a purified state, it possessed the ability to give form to a new life. The menstrual fluid of the woman was supposed to be semen also, but the woman did not have the power to acheive so high a degree of purification of blood as the man. This less purified semen was supposed to furnish the building material while the male furnished the life-givin power, which enabled this material to be formed into an embryo.

Little of importance relating to heredity was discovered, from the

time of the Greeks until about the eighteenth century. The idea that both male and female must contribute something to the embryo, as Aristotle had done, was recognized and some amendments were made in these ideas.

In the latter part of the seventeenth century Anton van Leeuwenhock with the use of his microscope, observed living sperms in the semen of animals. He also noted the association of the sperms with the eggs of frogs and fishes and believed that the sperm furnished the life of the embryo while the egg provided a place for the nourishment and development of the embryo.

Jan Swammerdam developed the preformation theory, which held that the development of the embryo was actually only the enlargement of parts that were already present in the sperm or egg. Some scientist of this time even imagined that they could see a miniature human being inside the sperm.[1]

There was much disagreement with the idea that the sperm was the sole agent of heredity and it was pointed out that children often show physical characteristics which are found in the mother and not in the father.

A Dutch scientist, Regnier de Graaf studied the ovaries of mammals and noted the protuberances which he interpreted as eggs which were similar to the eggs of birds. He believed that the egg breaks from the ovary, is fertilized, and makes its way to the uterus for development.

During the eighteenth century a Frenchman, Pierre-Louis Moreau de Maupertuis, thoroughly disagreed with the idea of preformation, for he felt that a consideration of the plain facts of biparental inheritance ruled out the idea that one parent formed a miniature preformed embryo.

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[1]A. M. Winchester, Genetics (Cambridge, Massachusetts, 1958), p. 18.

He proposed the belief that both parents produced semen which united during coitus to form the embryo. The semen, he believed, was made up of tiny particles, each of which was destined to form some specific body part, and that two such particles, one from each parent, unite to form each body part. One of these might dominate the other, however, to make a child more like one parent in this one respect. All of this sounds very much like the conclusions drawn by Mendel over a century later.

Many other theories were advanced such as the encasement theory which stated that each preformed embryo contained the still smaller preformed embryos of the generations to come.

In the latter part of the eighteenth century Jean Baptiste Lamarck proposed the significance of "use and disuse" and the inheritance of acquired characteristics. He held, for instance, that giraffes have long necks because they have stretched them reaching for food through the ages. He believed it was the habits and manner of life and the conditions in which its ancestors lived that have in the course of time fashioned its bodily form, its organs, and its qualities.

Charles Darwin proposed the hypothesis of pangenesis. According to this every cell, tissue, and organ produces minute pangenes. These are collected throughout the body by the currents of blood or other fluids and become a part of the germ cell. These pangenes are passed on to the offspring and they recreate these cells, tissue and organs from whence they came, thus a child represents a blending of the qualities of its two parents.

August von Weismann formulated the germ plasm theory, according to which all organisms possess a special germplasm which preserves itself by repeated divisions. These germ cells were merely carried by the body and

were in no way influenced by what happened to the body during its lifetime. The hereditary factors which a person can transmit to his offspring are determined at the time his life begins as a single cell and there is nothing he can do to alter the trait which may be transmitted through his reproductive cells.

This theory does not appeal to the imagination as much as the idea of the inheritance of acquired characteristics, but it is essentially correct. Many people would prefer to believe that somehow their attainments in life can be transmitted to their children; it seems frustrating to think that each individual must build each achievement anew.[2]

In 1901, Hugo De Vries proposed his mutation theory, that living organisms occasionally but regularly produce new types of offspring through sudden changes, mutations, in the hereditary mechanism. These mutations, he believed, were a much greater factor in evolution than the small gradual changes which Darwin thought to be so important. It was evolution through a series of sudden jumps. We now know that sudden changes in the units of heredity do take place to produce new types of organisms and that these changes are very important in providing new characteristics for selection.

Gregor Mendel discovered the principles of heredity for which he is called "the father of modern genetics". Mendel's greatest discoveries were made possible by the care with which he planned his experiments, his careful records of all phases of his work, and the mathematical precision with which he analyzed his results. Also he continued his experiments long enough to accumulate data in large enough quantities to have statis-

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[2]A. M. Winchester, Heredity and Your Life (New York, 1956), p. 31.

tical significance.

Some of the details of Mendel's results are given later. Suffice to say at this point that he discovered the fundamental basis for the inheritance of characteristics, which hold true for all forms of life. Mendel presented the results of his work in 1865. In spite of the great significance of his findings, the world took little note of his paper.

It was not until the beginning of the present century that the result of Mendels work and their great significance were to become known to the scientific world. At this time several investigators began checking the literature in an effort to explain some of their own results from hybridization, and they soon recognized the importance of Mendel's work. Most of our present day knowledge of genetics dates from the time of this rediscovery.

Within the scope of this report the writer cannot begin to mention all the many outstanding men who have built our genetic knowledge during the present century. However the work of many of them will be referred to in later chapters.

CHAPTER III

MEIOSIS

In man as in other plants and other animals, the key to an understanding of the laws of heredity lies in an understanding of the behavior of the chromosomes at the time of cell division. This is because the units of heredity, the genes, are, with very few exceptions, as integral part of the chromosomes.

There are regularly two of each kind of gene in each cell. These two genes are found on two different chromosomes within the cell, and the two chromosomes will always be similar to one another. The chromosomes within any cell may be of different lengths and shapes, but for every chromosome of a given length and shape there is, as a rule, another like it. These are called homologous chromosomes, and each bears the same type of genes in the same sequence. For instance if you carry a gene near the center of a V shaped chromosome which affects the pigmentation in your eyes, there will always be another V-shaped chromosome of the same kind which will carry a gene for eye pigmentation at a corresponding locus. This is true of all chromosomes with an exception which will be covered in connection with sex-determination. Thus the human chromosomes may be grouped into twenty-four pairs. These twenty-four pairs are found in all of the body cells except the reproductive cells, but the mature human sperm and egg each contain only twenty-four single chromosomes, one of each of the different kinds. This condition is brought about through

the process of meiosis. Essentially the process consists of two successive cell divisions accompanied by only one splitting of the chromosomes, resulting in a reduction in the number of chromosomes from the diploid number to the haploid number.

This number of chromosomes (48) has had general acceptance since 1923 however since 1956 fifteen individuals have been reported to have 46 chromosomes. Some Japanese state that there is no doubt that this number (46) exists in man, but this is not the only possible number in the species; besides individuals with 46 chromosomes, some with 47 and others with 48 exist.[1]

The number of Whites and Japanese studied so far are too small to provide the basis of reliable estimates of the frequencies of the three karyotypes in the two ethnic groups.

An obvious function of meiosis is the maintenance of a constant chromosome number in the species, for without it as a consequence of fertilization, there would be a doubling of the chromosome sets in each generation. A second function of meiosis is to serve as a physical mechanism for the segregation, assortment, and recombination of the genes.

A complete discussion of the process of meiosis will not be given in this report because this information is obtainable from almost all the references used. It was the belief of the writer that simplified methods of presenting this material to high school students would be of more value.

In order for the student to better understand what happens to the chromosomes when a cell divides a model using several strings of beads

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[1]"Three Chromosome Numbers in Whites and Japanese," Science (June 6, 1958), 127:3310.

may be used to represent the chromosomes.

We will assume in this model the cell has four chromosomes: two long and two short chromosomes. Use a red string of polyethylene pop-beads to represent one of the long chromosomes and a green string of beads to represent the other long chromosome. A short blue string and a short yellow string represent the pair of short chromosomes.

00000000	00000000	000000	000000
red	green	yellow	blue

Fig. 1. The four chromosomes

The first significant thing that happens during meiosis is the forming of partnerships. The two long chromosomes come together. Also the two short chromosomes come together as shown in Fig. 2.

red	0000000000	000000	yellow
green	0000000000	000000	blue

Fig. 2. The partnership

The next thing that happens is duplication, however, this does not yet involve the centromeres, the bodies shown as small circles, in the center of the chromosome.

red	0000000000 0000000000	000000 000000	yellow
green	0000000000 0000000000	000000 000000	blue

Fig. 3. The chromosomes duplicated

In meiosis there are two divisions. First, there is the separation of the partner chromosomes. That is to say, the original red chromosome and its duplicate separate from the original green chromosome and its

duplicate. The yellow chromosome and the blue chromosomes separate in a similar manner. Now we have two pairs of chromosomes in each of the two cells, one long pair and one short pair.

Fig. 4. First division in meiosis

In the second division in meiosis there is the splitting of the centromeres. Thus in the splitting one of the red and one of the yellow chromosomes go into one cell. The other red and yellow chromosomes go into a second cell. Precisely the same thing occurs, in the cell, with the green and blue chromosome.

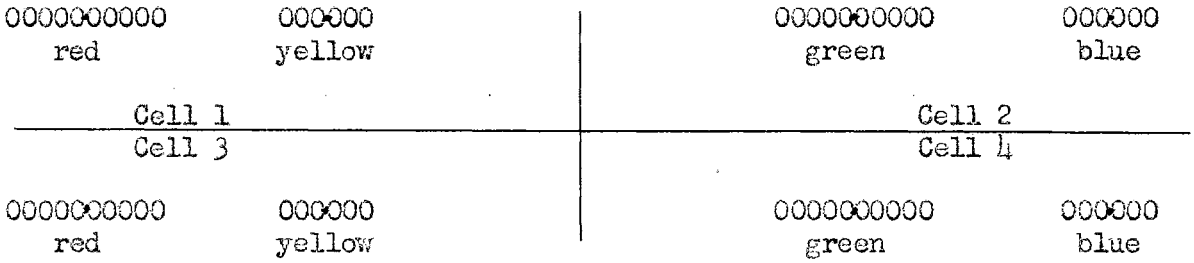

Fig. 5. Second division in meiosis

We now have four cells and each one has two chromosomes, one half the number present in the cells from which they developed. This is meiosis in its simplest form.[2]

Meiosis would always occur like this if it were not for the fact that the individual strands that make up the chromosome become intertwined and entangled. This is crossing over and will be discussed later

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[2]N. B. Abraham et al., Laboratory and Field Studies in Biology (Washington, D. C., 1957), pp. 671-673.

in this report.

Maturation of the male sex cell is known as spermatogenesis; maturation of the female sex cell is known as oogenesis. Four spermatozoa result from the maturation of one spermatogonium. The result of the two maturation division in the female is one large cell, the ovum, and three small, nonfunctional cells, the polar bodies.

The human ovum has twenty-four chromosomes, and the spermatozoon has the same number; thus at fertilization the chromosome number, which is forty-eight, is restored.

There are several points a student should understand after a study of meiosis. First, the chromosomes (genes) are passed on from parent to offspring. Second, the chromosome number is reduced by one half when the gametes are formed. Third, the chromosome number is restored at the time of fertilization. Fourth, chance operates in reduction division and fertilization. Fifth, each parent contributes one half of the chromosome present in the offspring and one of each homologous pair. Sixth, in the offspring there is a "mixing" and some chromosomes present in the parent and grandparents will be absent.

A very likely question that might be asked at this point is: what is there in living organisms that cause the development of different characteristics in humans? What we inherit is nothing more than a very tiny bit of cytoplasm and some chromosomes.

There is "something" in the chromosome that determines these characteristics called genes. Some geneticists think of genes as separate molecules--tiny invisible chemical particles. Unfortunately, we can't see these tiny genes. No one knows how many genes there are in a single human cell.

We can say that a gene is a molecule or part of a molecule located in a specific position in a chromosome and is responsible for controlling certain chemical changes in the cytoplasm. Thus determines different characteristics in different individuals.

In a living chromosome each gene is found only in a certain position on the chromosome. A gene which can produce its effects even though the other gene is present is known as a dominant gene. The other gene is known as a recessive gene.

CHAPTER IV

MENDELIAN LAWS

The first person to conduct decisive experiments in heredity and to formulate the basic laws of genetics was Gregor Mendel. Mendel planned carefully his experiments with garden peas and spent two years in selecting races with distinctive and contrasted characters and in making certain that each of his original stock was pure. During the next six years he made many crosses by artificial pollination, each of which was carried through three or more generations. Mendel kept an accurate statistical record of all plants and seeds of each kind that was produced, analyzed these results, and from them deduced the two most important fundamental laws of heredity.

The first of these laws states that the factors for a pair of characteristics are segregated. The factors responsible for a pair of alternative or contrasted characters are known as alleles. This can be illustrated by a monohybrid cross. A cross in which the parents differ in one pair of alternative characters. It will be easy to follow if we take a specific case as an example. Sufficient families have been studied in which it has been found that the gene for albinism is recessive, while the gene for normal pigmentation is dominant over it.

First lets consider a marriage between an albino and a homozygous normally-pigmented person. All the individuals of the F_1 generation are pigmented, no matter which parent is pigmented and which is an albino.

All these children will carry the gene for albinism and can pass it on to their children.

If one of the heterozygous pigmented children marries a heterozygous pigmented individual, on the average three fourths of the children are pigmented and one fourth have no pigment in their skin. Further study of the pedigree would show on the average one fourth would be homozygous pigmented, one half would be heterozygous pigmented and one fourth would be albinos.

In case of a marriage between two albinos all the children from such a marriage will be albinos.

In the case of intermediate genes, inheritance prediction is much simpler because there are no hidden genes. It is possible to tell the type of genes which a person carries when they are of this nature simply by observation of the person. For instance, if a member of the Caucasian race has wavy hair we know that he has a gene for curly and a gene for straight hair since waviness is the intermediate expression of these two genes. We do not need to depend upon ancestry in such cases.

When races differ from each other in two or more pairs of factors, the inheritance of one pair of factors is independent of that of the others.[1] This is Mendels second law and can be illustrated with a dihybrid cross, that is a cross between parents which differ in two pairs of characters. This can be illustrated by the use of two well known human heritable characters, polydactyly and attached ear lobes. Polydactyly a condition in which there is an extra finger on one hand an an extra toe on one foot. This is dominant over the gene for normal fingers and toes.

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[1]Tracy I. Storer, General Zoology (New York, 1957), p. 156.

In the case of attached ear lobes, free ear lobes are dominant over attached ear lobes.

If an individual homozygous for both of the two dominant traits marries an individual homozygous for the two recessive traits all their offspring will be heterozygous for each of the two traits and will express the dominant trait in each case.

If an individual heterozygous for each of the two traits marries an individual heterozygous for each of the traits and a large number of offspring were examined from this same type of cross, on the average the phenotypic results would be as follows: 9/16 polydactyl with free ear lobes; 3/16 polydactyl with attached ear lobes; 3/16 with normal finger and free ear lobes; 1/16 with normal fingers and attached ear lobes.

The different genotypes noted under these conditions would have been: 1/16 pure bred polydactyl and pure bred for free ear lobes, 1/8 pure bred polydactyl and hybrid for free ear lobes, 1/8 hybrid polydactyl and pure bred for free ear lobes, 1/4 hybrid polydactyl and hybrid for free ear lobes, 1/16 pure bred polydactyl and attached ear lobes, 1/8 hybrid polydactyl and attached ear lobes, 1/16 normal fingers and pure bred for free ear lobes, 1/8 normal fingers and hybrid free ear lobes, 1/16 normal fingers and attached ear lobes.

This can be very clearly illustrated by using symbols for the characteristics and 16 squares, filling in the genotype of each of all possible types of offspring.

There are some very good class demonstrations which can be carried on an illustrate random or chance combination. For instance, if we toss a penny in the air, the chance of it falling heads is one half. The chance of it falling tails is one half. Yet we do not think it impossible if we

toss a penny four times and get heads four times. If we should toss the penny four hundred times, however, we would get a ratio very close to 1:1. This will illustrate what happens to a gene pair during meiosis. It is just a 1:1 chance as to which gene will be found in a given gamete.

Two coins can be used in the toss to illustrate random or chance combination of two objects which can combine in only three ways. This is essentially what occurs during fertilization, when a pair of entities, the gene pair, recombine after being separated during meiosis.

There are three types of head-tail combinations that might appear, a head-head combination, a head-tail combination, or a tail-tail combination. If a large number of tosses were made the results would be about 1/4 head-head, 1/2 head-tail, and 1/4 tail-tail combinations.

This can also be shown by the use of red and white marbles, beans, beads or some other small object. Place equal numbers of red and white beans in a container on a table. In another container place equal numbers of red and white beans. To minimize errors due to picking of unequal numbers of red and white beans from the container, many more beans should be provided than are needed. This helps to insure that the chance selection of beans is not interfered with through an inbalance of colors present in any one box.

Select a group of students and each will take his turn in taking one bean from each container. Be sure the student does not look at the beans as he picks them from the container. Place the two beans next to each other on the table. Have each student pick several pair of beans from the containers and place them on the table. Now the group will decide into how many rows the pairs of beans can be grouped. The decision will be based on two facts: the beans selected together must remain together as

a pair, and beans of two different colors are being selected. The students will find there are three groups into which the pairs of beans can be placed. The red-red combination, red-white combination, and the white-white combination. If a large number of samples are picked the ratio will be close to 1:2:1.

These exercises can be used to allow the students to discover for themselves certain principles applicable to genetics.

CHAPTER V

SEX CHROMOSOMES AND INHERITANCE RELATED TO SEX

Attempts of all sorts have been made artifically to cause an unborn child to be a male or female at will. But all such attempts have failed. The fact is a person is a male or female at the moment he begins his existence as a fertilized egg.

The difference between the sexes is due to a difference in their chromosomes. Of the twenty-four pairs of chromosomes in each cell of human beings twenty-three pairs are similar in both sexes. The twenty-fourth pair determines the sex of the individual. In females this pair consists of two similar chromosomes called X-chromosomes. In males the pair consists of one X-chromosome and one Y-chromosome. The Y-chromosome is smaller. Thus the genotype of males is XY and the genotype of females is XX.

When reduction division takes place in the female all the eggs receive an X chromosome. In the male, half the sperm cells receive an X and half receive a Y. An egg (X) may be fertilized by either an X or Y sperm. The fertilized eggs, therefore, will be either XX or XY. All the cells of the body produced by mitosis are like the fertilized egg and are either XX or XY, and so they develop into either a girl or a boy. Half the sperm cells contain an X and half a Y. Hence, the number of fertilizations that produce girls (XX) and boys (XY) are about equal.

However, there is a slight inequality in the sex ratio. Male births

somewhat exceed the female births -- about 106 males to 100 females.

Several explanations have been suggested to account for the slight departure from equality in the sex ratio. However none of the theories are supported by any definite evidence. It has been suggested that the Y containing sperm cells can move a little faster than the ones containing the X chromosome and so reach the egg a little more often at the time of fertilization. Other suggestions are perhaps the Y containing sperm enters the egg more readily after contact than do the X containing sperms.

It has been suggested that at fertilization the ratio is 150 males to 100 females. At birth the males outnumber females by a ratio of about 106 to 100, but the death rate is greater for males at every stage of life. During childhood the rate is about 103 males to 100 females. During the young adult stage or the time for marriage the proportions of the two sexes is about equal. At fifty years of age we find that the males have shrunk from equality to a point where there will be about 85 men to every 100 women. At eighty-five the women outnumber men almost two to one and at one hundred there are about five times as many women as men.

Knowledge of the X and Y chromosome has explained certain traits associated with sex, such as color blindness in man. Women are much less often color blind than men. But if a woman does happen to be color blind, and if she marries a normal man, all of her sons are color blind but none of her daughters are.

We can readily explain this fact with a simple illustration. The X chromosome contains genes not only for sex but also for other traits, such as normal color vision, or a recessive color blind gene. We can designate the recessive color blind gene as c, and its normal allele as C. A color blind woman carries a c in each of her X chromosomes. A

normal man carries C, the normal allele in his X chromosome. We may regard his Y chromosome as a partially empty sack that carries no gene at the locus under discussion.

If a color blind woman, X^cX^c, marries a normal man, X^CY^-, the possible types of offsprings would be as in Fig. 6.

X^CY^-	X^cX^c
X^C or Y^-	X^c or X^c

X^cY^-	X^CX^c

Fig. 6. First generation

All the males of the first generation would be color blind and all the females would be carriers of the color blind gene but would have normal vision. If one of the females of these parents marries a normal male the types of children would be as in Fig. 7.

X^CY^-	X^CX^c
X^C or Y^-	X^C or X^c

X^CX^c	X^CX^C	X^cY^-	X^CY^-

Fig. 7. Second generation

All the daughters would have normal vision but might be carriers of the color blind gene and the sons might be color blind or have normal vision. The expected ratio would be 50% sons color blind and 50% with normal vision.

It is now apparent why color blindness is less often found among women than men. The male may show a trait when only one gene is present. Both recessives must be present in the female for the trait to show.

Hemophilia is also inherited as a single sex-linked recessive. The primary symptoms of the disease is an abnormal tendency to bleed because of an extremely slow rate of coagulation of the blood. In hemophilious the coagulation time is greatly prolonged, varying from one-half hour to 22 hours or more, according to the severity of the disease. Thus it is a lethal gene because it causes death before maturity of the majority of individuals expressing the gene.

The fact that it is a sex-linked recessive indicates that a female can develop the disease only if she receives the gene from her father who has the gene and therefore the disease, as well as from her mother, who may be heterozygous. In view of the rarity of the disease, and the failure of most affected males to leave offsprings, this coincidence would seldom happen. Thus explains the fact, with a few doubtful exceptions, hemophilia has not been reported in women.

The most famous pedigree of hemophilia is that of Queen Victoria of England.[1] Victoria was heterozygous for the gene, perhaps as the result of a new mutation since there is no record of hemophilia among her ancestors.

Many false conceptions related to sex-linkage should be corrected. A popular misconception is that boys tend to inherit most of their characteristics from the mother while girls inherit most from the father. It is true that a man receives all of his sex-linked genes from his mother and none from his father, but this represents only about one twenty-fourth part of his total inheritance.

Sex-limited genes are often confused with sex-linked genes but are

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[1]Edward C. Colin, Elements of Genetics (New York, 1956), p. 245.

entirely different in methods of inheritance. Sex-limited genes are those which produce characteristics that are expressed in only one of the sexes.[2] They may be located on any of the chromosomes, whereas sex-linked genes are located on the X chromosome. Sex-limited genes are responsible for some secondary sexual characteristics, such as the beard on men, as well as primary sexual characteristics.

All the evidence indicates that the expression of sex-limited characteristics depends upon the presence or absence of one of the sex hormones. A woman, normally does not have a beard, yet she carries all the genes necessary to produce a beard, and they will express themselves only when the male hormone is present. In rare cases abnormalities in hormone secretion may occur in a woman and allow these genes to express themselves.

Sex-influenced genes are genes that are dominant in man and recessive in women. Baldness is the only one which is commonly observed. To determine the heredity of daughters, we treat the trait as a recessive and to determine the heredity of sons we treat it as a dominant. Thus a woman may carry the gene for baldness without showing it, and she will pass it on to one-half of her children. The sons who receive the gene will become bald, but the daughters will have normal hair growth, provided the father gave them the normal gene. A bald man, may have only one gene for baldness and, in this case, one-half of his sons will be bald, but all the daughters will be normal if the mother carries normal genes. Other combination of genes for this trait can be worked out by

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[2] A. M. Winchester, Genetics (New York, 1951), p. 122.

reference to a chart.[3]

Sex-linkage can be shown with pipe cleaner "chromosomes" and test tube "cells".[4] If we use a sex-linked character such as color blindness, the results of the mating of parents of any genotype can be shown. To show the carrier woman and normal man mating, for example, we need four "cell" (test tubes) to represent the zygotes. In addition to the cells we need eight "chromosomes", (pipe cleaners), 2Y's, $4X^C$ and 2 X^c. X^C represents the X chromosomes with the normal gene and X^c represents the X chromosomes with the color blind gene.

Fill each test tube about 3/4 full of water and add a little bromthymol blue indicator to each test tube and add just enough acid or base to get a light blue color.

Soak the X^C chromosomes in a weak solution of sodium hydroxide, and the X^c chromosomes in about a 10% hydrochloric acid solution. Do this about half an hour before using them. Leave them in solution until the last minute. Remove them with forceps and place them on paper towels labeled X^C, X^c, and Y. Make the desired cross and the results are as indicated in Fig. 8.

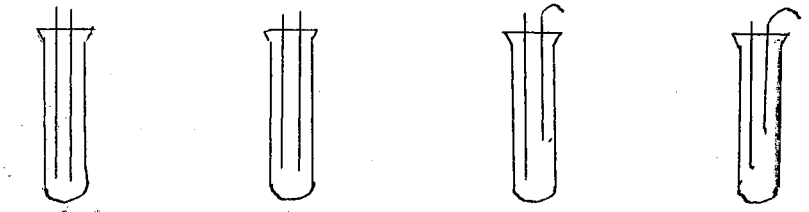

Fig. 8. Results of different combinations of chromosomes

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[3] A. M. Winchester, Heredity and your life (New York, 1956), p. 154.

[4] N. B. Abraham et al., Laboratory and Field Studies in Biology (Washington, D. C., 1957), p. 703.

CHAPTER VI

MULTIPLE ALLELES AND MULTIPLE GENE INHERITANCE

Many cases are now known of more than two alternative factors that affect the same character; these are called multiple alleles. Multiple alleles refers to all genes that occupy the same locus on a chromosome.

One of the best examples of multiple-alleles in man is the four blood types called, A. B. AB, and O. It was found that blood cells of man may contain two distinct antigens. Persons with type O blood posses neither of these antigens, but posses both anti-A and anti-B antibodies in the plasma. Type A persons have the A antigen in the red blood cell and anti-B antibodies in the plasma. Type B persons have the B antigen and anti-A antibody and people with type AB have both antigens and no antibodies in the plasma. Neither A nor B is dominant over the other, but both are dominant to O.

The genotype of individuals with type A blood could be AA or AO, of type B either BB or BO, of type AB it is AB, and of type O it is OO.

The method of inheritance of blood types is well understood. It is easy to see from Fig. 9 the possible blood types of children from parents of various blood types.

By knowing the possible genotypes a parent of a given blood type might have, one can readily follow the table and see why children of the given type are possible.

The human race is also subdivided into three MN blood groups; M,

N, and MN. These three have no relationship to the ABO blood group; that is, a person of group A might belong to any one of the three MN blood groups. However, unlike what applies in the case of ABO groups, people who belong to M, N or MN blood groups do not contain natural antibodies against those of another M or N group. Generally a transfusion from a person with a different M N group than the recipient leads to no complication.

BLOOD TYPE OF PARENTS	POSSIBLE BLOOD TYPES OF CHILDREN
O x O	O
O x A	O, A
O x B	O, B
O x AB	A, B
A x A	A, O
A x B	O, A, B, AB
A x AB	A, B, AB
B x B	B, O
B x AB	A, B, AB
AB x AB	A, B, AB

Fig. 9. Possible blood types

There is another interesting series of multiple alleles related to the blood known as the Rhesus blood groups or Rh blood groups. Actually there are about eight kinds of Rh alleles but these will not be considered in this report. For simplicity and ability to explain to high school students the writer selected to present this material as if some people have the Rh antigen, while others do not. Those with the antigen are designated Rh-positive; those without it Rh-negative. About 85% of the people in the United States are Rh-positive and 15% Rh-negative. No one has been found who contains the natural Anti-Rh antibody, but it has been found that an Rh-negative person can develop these antibodies if exposed to the Rh antigen. This is important in transfusion and in childbirth.

If a Rh-negative mother and a Rh-positive father have children the

child may be either Rh-negative or positive, because the Rh antigen is produced by a dominant gene. When this gene is absent a Rh-negative individual results. A Rh-negative person is homozygous for the recessive allele. If the child inherits the Rh antigen from the father and the mother's blood has been sensitized either from a previous Rh positive child or a blood transfusion and there is a leakage across the placenta so the antibodies of the mother may pass into the blood streams of the child it destroys the red blood cells of the child and is often fatal to the child unless special precautions are taken in advance by the physician.

There are many characteristics which are not clearly defined or divided into distinct groups, but show variations from one extreme to another. It is not possible to explain such variations on the basis of two or more alleles at a certain chromosome locus. However this can be explained on the basis of a number of different genes located at different loci on the chromosome. Characters such as this show multiple gene inheritance.

Skin color inheritance in man depends upon quite a number of genes. Also the body height in man illustrates multiple gene inheritance.

The color of the skin is dependent upon the amount of a pigment, melanin, deposited in the skin. This depends upon the genes and of course the amount of sunlight the skin receives. According to the hypothesis negroes differ in skin color from members of the Caucasian race mainly because of the nature of two gene loci which influence skin color.

A cross between a white person and a negro show segregation of genes according to the dihybrid ratio. The first generation offsprings results in a mulatto, an intermediate skin pigmentation. When two mulattos marry

their children may have any one of five different skin shades, ranging from white to the negroid pigmentation. The number of genes for the negroid pigment could range from zero to four genes for dark skin. The offsprings with zero genes were white; one gene, brunette; two genes, mulatto; three genes, chocolate; and four genes, negroid.[2]

Body height in man may be illustrated if we assume there are four genes involved. Actually there are more involved but the principle of transmission is the same.

Two medium sized parents can produce a tall child. The parents may be heterozygous for a number of recessive genes for tallness and an offspring receive these recessive genes and be tall.

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[2] Ibid.

CHAPTER VII

LINKED GENES AND CROSSING OVER

It is very obvious, that the number of genes in man is far greater than the number of chromosomes, and hence not all genes can be independent. A given chromosome must contain numerous genes. The genes located on the same chromosome are said to be "linked". Linkage is the tendency of two genes to remain together in the process of inheritance.

Two genes may be closely linked, that is, they almost always stay together in transmission, or they may be loosely linked, and frequently are not transmitted together to the offspring. These genes are subjected to frequent alterations by a segmental interchange between chromatids of the homologous chromosomes. When this segmental interchange occurs, the genes are said to have "crossed over". The frequency of crossing-over between two genes is used as a measure of the relationship of their location on the chromosome.

The number of linkage groups corresponds to the haploid number of chromosomes, also the linear order of the genes within a linkage group can be specified by a study of the frequency with which genes within this group cross over with one another. The distance between these genes are specified in terms of their recombination frequencies.

The amount of crossing over between linked genes varies with their distance apart in the chromosome. The further apart they are, the greater amount of crossing over between them.

Crossing over occurs during the first division of meiosis. At this stage each chromosome consists of four chromatids. The chromatids become twisted about one another, and often a breakage and a reattachment of portions of the chromatids from homologous chromosomes.

This process can be illustrated better than words can describe it. The teacher or students can make very adequate drawings on charts to illustrate the process of crossing over and linkage. Beads can also be used to show this. Strings of two different colored beads can represent two chromosomes. Part of one string can be removed and attached to the other string showing crossing over during meiosis.

It is very difficult to study autosomal linkage in man. Sex-linkage, however is much more readily studied, and considerable progress has been made in the mapping of the sex chromosomes of man. In man there should be 24 linkage groups, but it will probably be a long time before marker genes are known for all 24 pairs of chromosomes.

On the basis of present knowledge, a list of a number of more or less definite autosomal linkage groups or linked pairs, and a list of genes known to be in the X and Y chromosome can be given.[1]

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[1] R. R. Gates, Human Genetics (New York, 1946), p. 82.

CHAPTER VIII

OTHER INHERITED CHARACTERISTICS IN MAN

Many human characteristics have been studied besides the ones mentioned so far, and since the knowledge of some of these may be useful in high school teaching and are interesting, this chapter will be devoted to some of them.

Eye color is probably one of the most often talked about human characteristic. Ordinary eye colors are due to the presence of pigment in the cells of the iris. The difference in eye color depends largely upon the melanin pigment which develops in the front layer of the iris. It is an optical effect. In fact, all eyes are basically just as "blue" as blue eyes. The other eye colors are due to the addition of pigment in the front of the iris.[1]

In blue eyes no pigment whatsoever is produced in front of the iris. In green eyes there are diluted brown pigment cells in front of the iris superimposed on the blue background produces the effect of green. Gray eyes are due to scattered dark pigment in front of the iris screening the blue. Brown eyes are due to the concentration of dark pigment in front of the iris masking the blue. Pink "albino" eyes are due to the reflection from pink blood vessels, because they have no pigment in their eyes at all.

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[1] Amram Scheinfeld, You and Heredity (New York, 1950), p. 56.

With the exception of mutations when both parents are blue-eyed, all their children have blue eyes; when one parent is heterozygous brown-eyed and one blue-eyed the children are partly brown-eyed, partly blue-eyed. Marriage of two brown-eyed persons usually results only in brown-eyed children; yet blue-eyed ones may appear. These facts indicate that brown is dominant to blue, and blue-eyed individuals are homozygous recessive for eye color.

Hair color follows the same general principles as does that of the eyes. In hair the color is due to the pigmentation of hair cells.

There seems to be two primary pigments in the hair each of which is subjected to variation through multiple genes. One is black the other light blond. In addition to this there seems to be another pigment which ranges from sandy red to yellow. Various genes influence the intensity of these pigments in such a way as to produce the wide variations in hair color. Red hair seems to result from a single pair of recessive genes. A marriage between two light blondes almost always yields light blonde children, a fact which indicates the recessive nature of the blonde gene. Children resulting from marriages between dark haired persons are typically dark haired. This indicates a dominance of the genes for dark hair over the genes for light hair.

There is not complete agreement as to the exact cause of the graying of hair. It is known that the age at which the hair begins turning gray is influenced by heredity. It is difficult to establish the exact method of inheritance, however, because of environmental effects of diet and other factors.

Some persons are born with a white forelock of hair which is given the genetic designation of blaze. This isolated island of white hair

grows out from the central portion of the scalp where it joins the forehead and is usually combed back over the head to produce a white streak in a head of hair otherwise normal in pigmentation. This condition results from an autosomal dominant gene.

The development of the skeleton is also affected by genes. There are about two hundred bones in the adult human body, and their size, shape, and arrangement to a large extent determines the body build. Achondroplastic dwarfism seems to result from the influence of a dominant gene. The head and trunk of the person with this type of dwarfism are normal size, but there is a great reduction in the length of the limbs, and this results in short stature. The gene inhibits normal growth of the long bones. The genes for this condition are found in entire human groups such as the Congo pygmies. A similar condition has been achieved by selection in one breed of dogs. The dachshund has short bowed legs and was bred for this character so that it could get into the burrows of foxes and badgers easily.

The Atelliotic dwarf is well proportioned but small in all parts of the skeleton. This characteristic is due to a deficiency of the growth hormone of the pituitary, which influences the growth of the skeleton. Normal-sized children have been born of parents both of whom were midgets.

Many studies in human genetics dealing with the relative effect of heredity an environment have been made of twins, "Identical", monozygous, or one-egg twins, which arise from a single zygote; and the fraternal, dizygotic, or two-egg twins, which arise from two zygotes. Monozygous twins have identical genes and, as a result, make ideal subjects for studies of the effect of environment upon development, since any difference which they show must be environmental. Fraternal twins serve very nicely

to illustrate the effects of difference in heredity in a constant environment. They will have differences in many of their genes, since they are no more alike genetically than any other brother or sister in the family, yet they are most likely to have a very similar environment. The results of many of these studies makes it apparent that heredity places its imprint upon almost every aspect of human development and behavior and the environment modifies the effects of these genes. Thus the individual is a product of his environment as it acts upon his hereditary potentialities.

The inheritance of twinning is not well established, however, certain facts associated with twin birth seem to be reasonably well established: (1) twin births tend to cluster in specific families, and (2) the frequency of twinning varies with maternal age and some extent with parity. It has been estimated that a mother who has given birth to one set of twins will at her next delivery repeat with another twin birth approximately 3.6% of the time. This per cent varies on the basis of whether the first twin pair was monozygous or dizygous. If the first pair was dizygous, then the probability that the mother will repeat with another set of twins is 4.55%. However, if the first pair was monozygous, then the chance of a second set of twins is only 1.43%, not much higher than the probability of a twin birth in the general population.

One of the most important human factors influenced by heredity for the future of man is intelligence. There is ample evidence to show that the extreme variations in mental capacity among human beings are partly hereditary and partly environmental. Training plays an important part in bringing out inherent potentialities, but even among persons with similar training there are great variations in general intelligence, aptitudes,

and other characteristics. Since these variations are generally continuous and not clear-cut, it is obvious that multiple genes are involved. Thus there are many cases in which variations in a single gene may have a very obvious effect on mental ability. A person may be born with a whole gene complex for a brilliant mind, yet also inherit a single pair of recessive genes for a particular type of idiocy, which would be epistatic to the entire complex and thus make the person an idiot.

In spite of the extremely complicated problem of the inheritance of mental characteristics, a great deal of information has been given on the relative significance of hereditary and environmental factors in determining mental capacity. In general bright parents tend to have bright children, and dull parents, dull ones, however there are many exceptions to this case. Generally the children of parents at either extreme of the intelligence-rating scale will show a tendency to regression toward the average of the population.

Special aptitudes, are independently inherited and may not have any close correlation to general intelligence. This phenomenon is well illustrated by the interesting case of a woman moron with an I.Q. of about 60 who could never learn to tell time by a clock, but could play the piano very nicely by ear.[2]

Epilepsy is a disease characterized by sudden seizures known as epileptic fits. In the most extreme forms this develops into unconsciousness and muscular spasms. Some people have the disease in a milder form in which the fits are minor. Brain injury is known to be an environmental agent which can induce the onset of epilepsy, but the majority of cases

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[2]A. M. Winchester, Genetics (New York, 1951), p. 336.

arise without such injury and have a hereditary basis. Sufficient studies have been made to indicate that it is inherited as a dominant trait.

The study of hereditary mental defects is of increasing importance. It seems that mental defects are increasing in frequency. Several million people in the United States have an I.Q. of about 70 or lower. A large number of these cases are of hereditary origin. The feeble-minded become juvenile delinquents, problem children, and cases for public charity and relief. They breed early and often so tend to increase their kind. The end result is a rapidly mounting bill of their care and for dealing with the crimes that they commit.

Many organizations and persons are now interested in eugenics, a field of endeavor which seeks to better the human race by applying the principles of genetics. Since it is not possible to control the matings between human beings, attempts are made to determine the facts of human heredity, to educate the general public on the effects of good and bad matings, and to encourage legislation that will prevent matings between obviously defective persons. Attempts are also made to improve the environmental conditions under which human beings develop and live so as to give the best possible expressions to the genetic constitutions present in each individual. No measurable aid in human genetics will be possible until much fuller records on human inheritance are available.

CHAPTER IX

CONCLUSION

The result of this study revealed many facts which can be used by the teacher in teaching biology. It seems evident that everyone should have some practical knowledge about heredity, especially human heredity. Because this knowledge will not be attained at home, it is left up to the teacher of biology to supply this information.

The study did reveal many demonstrations, illustrations, and experiments that can be performed by the teacher and pupils which would help the pupil understand the principles and phenomenon of heredity.

Many models and charts can be made by the students and teacher which will be very helpful as teaching aids in teaching different principles. Plaster of Paris models, charts, and polyethylene beads can be used to illustrate the different phases of cell division, types of offsprings expected from a particular cross, chromosomes, genes, crossing over, and many other phenomenon that occur in heredity.

Recognizing the fact that human heredity is one of the most interesting phases of the study of heredity to most students, the teacher can do much to interest the students by using the inheritance of human characteristics to illustrate many genetic principles. With this in mind this study has been presented.

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VITA

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Report: A SYLLABUS FOR A UNIT ON HUMAN HEREDITY FOR SECONDARY BIOLOGY

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Scope of Study: The purpose of this study is three fold: (1) to provide a general knowledge of a few selected human hereditary characteristics that may be easily communicated to high school students; (2) present a general guide which may serve as resource material for teachers, and (3) an attempt to provide illustrations, demonstrations, and sufficient reference for the teacher to proceed to further studies. Throughout the study hereditary characteristics in man have been used to illustrate basic principles of heredity. Many class room demonstrations, their use and effect, have been considered.

Findings and Conclusions: A thorough understanding of heredity must be left to the expert, however the result of this study revealed many facts, demonstrations, and experiments which can be used by the teacher in teaching biology. Plaster of Paris models, charts, and polyethylene beads can be used to illustrate many principles and phenomenon which occur in heredity.

Recognizing the fact that human heredity is one of the most interesting phases of the study of heredity to most students, the teacher can do much to interest the student by using the inheritance of human characteristics to illustrate many genetic principles. With a few general principles and some facts, the high school teacher can do much toward enlightening the future generation in regards to its own welfare with respect to heredity.

ADVISER'S APPROVAL