

Curriculum Units by Fellows of the Yale-New Haven Teachers Institute 1990 Volume VI: Genetics

# Parents are the Key A Curriculum Unit for Modern Genetic Study

Curriculum Unit 90.06.03 by Lee Hotchkiss-Durward

The old saying goes, "you can pick your friends, but not your relatives." If you are here with no visible defects and have healthy parents and grandparents, you should consider yourself lucky. However, it is estimated that each of us carriers at least four to eight harmful hidden genes that we may pass on to our children or grandchildren. The large percentage of babies are born healthy. But the chances of a family having a child born with a genetic disease are great enough so that all potential parents should understand the basic facts of genetic diseases. This unit is planned to introduce high school students in the classes of biology, health, anatomy, physiology, parenting, and sociology to some basic genetics and to the rapidly growing fields of genetic medicine and genetic engineering. Students of today are taught to take responsibility and to have concern for their prospective children. In addition, today's student. should be able to to read and assimilate the myriad of articles in newspapers, magazines, books, television, and radio that assume knowledge of genetics. As educators, I feel that it is our duty to help our students explore the facts and to be ready to make informed opinions concerning personal, social, and political decisions, while understanding the ethical implications involved.

This curriculum will explore the scientific aspects of genetics before applying this information to personal use. In this unit there will be information on how to introduce students to the causes of genetic disorders by examining the relationship of genes, chromosomes, and inherited traits. Also to be investigated will be traits carried by sex chromosomes as well as the effects of missing or extra chromosomes.

Although some genetic diseases are still a mystery, medicine has developed the ability to detect and diagnose many of them. The medical practice of genetic testing in obstetrical care will be explained so that students will be prepared to ask knowledgeable questions if they are ever in a position to do so. As parents of the future, the student should be made aware of the genetic screening a newborn receives. Students can be helped to make decisions for themselves with help from counseling and education.

How can we know whether or not there is a possibility of a genetic problem? The Watson-Crick model of DNA and the theory that a segment of DNA, the gene, controls the production if a polypeptide chain have helped to make inherited diseases understandable. Since the time of this revelations, medicine has made great strides in discovering practical applications of genetics. Because of these recent advances in medicine, it is now possible to prevent problems by becoming aware of the risk factors. Here is a sample of these risk factors:

1. If your family has a history of genetic disease.

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- 2. If you have had one child with genetic disease of birth defect.
- 3. If you are pregnant and are over the age of thirty-five or under the age of fifteen.
- 4. If you or the other parent has a history of a genetic disease.
- 5. If you have been exposed to excessive radiation.

6. If you are a substance user. A genetic counselor can help to interpret medical knowledge of genetic defects and explain results of recently developed technique of prenatal diagnosis and carrier detection, all of which helps to lessen fears. If you are planning to have a child, and might be at risk, the counselor can tell you the chances of your passing along a genetic disease to your child. Now there is enough medical technology to detect carriers of genetic disorders, even though the client may not display physical symptoms. prenatal testing can recognize certain genetic disorders at birth. Upon diagnosis of specific disorders, usually metabolic and genetic in origin, special treatment can help, but never cure a genetic disorder. For instance, one gene normally controls the production of a liver enzyme which converts the protein segment phenylalanine into tyrosine. In the birth defect phenyleketonuria (PKU), the gene involved in this function fails to direct the manufacture of this enzyme. This irregularity in metabolism produces mental deficiency in the child who appears normal at birth, but eventually becomes seriously retarded. PKU can now be detected at birth, as all newborns are screened for this disorder. Fortunately, the symptoms of this disease may be prevented by strict adherence to a diet, and the subject can go on the lead a normal life.

Genetic counselors are often physicians with expert knowledge of genetics. Not all physicians can qualify, one must have special training. Heretofore, some physicians were more concerned with other aspects of medicine and have not had the thorough training and experience in the rapidly developing science of genetics. There are also people trained in genetics who work with physicians, in an exponentially expanding career known as Genetic Counseling.

There is a need for people to understand the mechanism of heredity. Unfortunately, some are overwhelmed upon learning that their smiling, lovable child will remain in a child-like mentality for the rest -of its life. The reason for this is, one extra chromosome which manifests itself at the time of conception. A parent who is not aware of what a chromosome is, would feel helpless and fearful of how such a disorder was caused, and whether or not is could be prevented in the future. The younger generations of today will be educated in the field of genetics, understanding the difference between congenital and hereditary defects.

Knowledge will help people to accept and to understand the possibilities that can occur when sperm meets egg. It is a question of numbers, integrity, and quality. All human cells normally have forty-six chromosome in twenty-three pairs of two. In 1959, it was discovered that a person with Down Syndrome had forty-seven chromosomes. This finding was the beginning of medical interest and involvement in chromosomes. The fact that Down Syndrome was associated with an abnormal amount of chromosomal material stated the science of cytogenetics, a science that studies the structure and number of the chromosomes.

Chromosome study begins with photographing through the microscope of the chromosomes from a single cell. The mis-array of chromosomes is paired, arranged by size, and counted. Twenty-three pair in size from largest to smallest. Three chromosomes (1-22) are called autosomal chromosomes. The twenty-third pair of chromosomes is designated a X and Y. If a cell has two Xs in the chromosomal material, then the child is a girl.

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If in the nucleus, where chromosomal material is found, there is an XY, then the child is a boy. The technique for identifying these chromosomes that have enlarged for viewing the microscopic picture is called karyotype. A Down Syndrome patient has an extra number twenty-one chromosome and is referred to as trisomy 21. The introductory page shows what the microscopic picture of chromosomes might look like.

All cells in our body, regardless of their specialized function, poses similar basic parts. The nucleus of the cell is the center for the important functions of life. It also contains the traits inherited from our parents, in which we in turn will pass along to any children in the future. Actually, we only pass along half of our chromosomes.

The important component of chromosomes is DNA, almost a household work, whose chemical name is based on that which makes it up: deoxyribonucleic acid. The chromosomes are composed of genes. So in each pair of chromosomes we have the duplicated genes that are derived from our mother in addition to the duplicated genes that came from our father.

Size alone is not the only way to pair up the chromosomes. Using the newest techniques and staining methods, we can distinguish horizontal bands on every chromosome. These techniques make is possible to detect small defects and also to show, like fingerprints, the unique chromosomal picture for every individual. This is not true for identical twins as they come from a singe zygote; therefore, their chromosomal pictures are alike.

In the maturing of the sperm and the egg, each must lose half of the forty-six chromosomes. This process is collectively called meiosis. This process will be demonstrated in one of the activities. When fertilization occurs, the parents' twenty-three chromosomes are untied so that the zygote of fertilized egg will resume the correct number of chromosomes for our species. It is during the process of meiosis that there could be an exchange of genes between homologous, or two chromosomes that contain genes for the same trait. This is called crossing-over, during which the chromatics, or replicated chromosomes, become entangled during the tetrad sequence. The sequence includes four chromatics of a homologous pair of chromosomes that form during meiosis. It is the exchange of genetic material the adds variety to life.

We start life wit forty-six chromosomes contained in the nucleus of the cell. The cell then divides by a process known as mitosis which produces two identical daughter cells. Unfortunately, mitosis can go awry, or the egg or sperm, could already be abnormal before fertilization. Most all recognizable chromosomal abnormality can be diagnosed in the fetus early enough to consider the options open to them. During mitosis, some abnormalities arise resulting in either too many chromosomes, or in some cases, too few. Another type of chromosomal abnormality can arise if a chromosome sticks with another during cell division, called nondisjunction. There is a possibility that an environmental factor may be at work to make the extra chromosome stick to another in the case of these disorders. Suggested causes might be X-ray exposure, virus infections, thyroid or diabetes disease of the mother, and the age of the mother.

Sex-chromosomes can have the same king of chromosomal abnormalities with a variation. Klinefelter's syndrome shows a chromosomal picture of a person with an extra X chromosome making him have XXY. Males that are affected are frequently taller than normal and usually cannot manufacture sperm. If breasts are enlarged, males can be given male hormone to help them return to normal size. There can also be a deficiency of sex chromosomes. If a female is born with XO, or missing X chromosome, this female has the abnormality called Turner's Syndrome. These women are usually under five feet, show a webbing of the neck, and their ovaries are underdeveloped.

To talk of genes needs a discussion of DNA. DNA is the chemistry involved in gene formation. The genes are

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located on the structure known as deoxyribonucleic acid. The chemicals direct our life. The chemicals that are involved are sugar (deoxyribose), phosphates, two purine bases, adenine (A) and guanine (G), and two pyrimidine bases; thymine (T) and cytosine (C). Watson and Crick suggested the structure is like a ladder with the bases acting as the rungs of the ladder. The bases must be paired in a very specific way. For example, adenine must always be paired with thymine, and guanine must always pair with cytosine. The pairs are held together by very weak bonds. The bases are attached to the sugar molecule and the sugar is chemically bonded to the phosphate. The job of DNA is to get the correct information that will be transcribed to Ribonucleic acid (RNA). In RNA, uracil (U) replaces thymine and the sugar is ribose in place of deoxyribose. The function of both DNA and RNA is to send messages to make amino acids that eventually become the building blocks of proteins. Three bases constitute a codon, a unit of genetic code to determine which amino acid will become part of the protein chain. And so, the codons are part of the genes. If, for instance, th coding is incorrect and the gene abnormal, then the proper sequence for coding the protein will be abnormal.

The process of gene production occurs by means of the DNA ladder splitting down the center where the weak chemical bonds holding the bases are broken and chemical substances from the cell join to form new strands. Since this paper is concerned with the fate of the genes, only this brief explanation seems necessary. The mechanism of how the proteins are made can be found in any modern biology book. It is also advisable to read the history concerning the early work of Gregor Mendel, who is considered the father of heredity. His work with peas and probability can be transposed to people; especially when considering the fate of dominant and recessive genes.

Some diseases can be inherited from one parent, who is affected, as in Huntington's chorea, caused by a dominant gene that causes a deterioration of the central nervous system. On the other hand, a disease such a cystic fibrosis causes chronic lung infection and thick mucous that results in troubled breathing and digestion. This disease is the effect of two parents who are healthy yet unknowingly carry a recessive gene. Another devastating disease is the brain-destroying disease of Tay-Sachs caused by recessive genes transmitted from both parents. Sickle cell anemia is transmitted to unsuspecting children in the same manner. Unlike dominant inheritance, both recessive genes must be united for the disease to manifest itself. A carrier for the trait will not have the disease. Diseases caused by dominant genes are different, as an individual with just one gene for the dominant disorder will be effected.

The traits each person has are not only determined by the genes, but also by the environment. The environment is involved in the replicating process from the time of conception to the time of death. One must be aware of the environment's effects at all times. Exposure to radiation, certain drugs, could cause chromosomal damage.

The genes we have been concerned with thusfar are the autosomal genes which are shared by both male and female, with a total of two sex chromosomes a piece. These chromosomes contain the genes found on the X and Y chromosomes. Not only do these genes control and determine our sex; there are other traits too. By just size alone, the X chromosome, which is larger, carries more genes. Because female has two X chromosomes, any undesirable trait would have to have both chromosomes showing the disorder. This is not true for the male. Not having enough chromosomal material, a faulty gene on the male X chromosome will be shown. Hemophilia, a bleeding disease due to missing blood factor, is most often transmitted by females to their sons. Such a situation exists for the male in Duchenne Muscular Dystrophy because they have inherited the culprit gene from the mother.

There are other more complex ways to account for our inheritance, and even more that scientists are in the

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process of discovering. Sometimes we can, through our genes, be predisposed to react to our environment. This can, perhaps, explain why some people have a specific reaction to plain aspirin causing a capillary fragility and anemia. Other people have serious reactions to penicillin. There is a partial listing that will, given certain modes of inheritance react to our chromosomes. Luckily in today's world, new techniques and dedicated people are helping to detect some of these genetic problems and are trying to find better treatments to ease the situation caused by these diseases. It should be noted that the March of Dimes has been instrumental in supporting research and disseminating knowledge.

Today new techniques are continually being developed to study molecular biology to allow scientists to manipulate individual genes. This is a new and exciting science of recombinant DNA. The DNA segments can be cut by specific enzymes and a new segment that comes from a different source can be inserted. From this technique, can come wonderful new approaches to pharmacology, as in the production of inteferon, a protein produced by virus-infected cells that protect other cells from becoming virus-infected.

Bacteria have been then helpers. They have the ability to mutate (change genetically), to adapt and to reproduce prolifically. It is hoped that genetic engineering will be the tool that can eliminate or at least alleviate suffering.

Hardly a day goes by with our some reference to something pertaining to genetics. The police use DNA fingerprints to solve a crime, or new gene site has been discovered which might eventually aide in gene therapy. The future will hold many decisions to be made; we should be aware so as to make the best decisions as possible. There is a whole new biotechnological industry that our students should be prepared to be involved with. Genetic literacy should be our goal.

#### The Structure of DNA

The DNA molecule as proposed by Watson and Crick resembled a gently-twisted ladder. The rails of the ladder are composed of alternating units of deoxyribose sugar and phosphate. The rungs of the helical ladder are composed of a pair of nucleotides, held together by weak hydrogen bonds. (Fig. 1) Since DNA does not leave the nucleus, messenger RNA takes out the DNA message for protein synthesis by passing into the cytoplasm and becoming attached to ribosomes. Messenger RNA differs from DNA because RNA contains no thymine, is single-stranded and contains the sugar, ribose. RNA substitutes uracil for thymine. Messenger RNA is synthesized in the nucleus. The base sequence of mRNA is determined by the base sequence of the segments of DNA on which it is formed. (Fig. 2)

When messenger RNA leaves the nucleus it interacts with two other forms of RNA: ribosomal (rRNA) and transfer RNA (tRNA). It is on the site of the ribosumes that proteins are formed by joining amino acids. That is called translation. The order in which the amino acids are arranged is determined by the base sequence (codons) of the mRNA, and thus by the base sequence of the cell DNA.

Fig. 2 Messenger RNA Transcription (From DNAScience-Cold Sprint Harbor)

(figure available in print form)

Note\* An interesting assignment would be to read the history and excitement behind the race to first be credited with the model of DNA. Fig. 1 The Double Helix

Watson-Crick Model

(figure available in print form)

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a. The diagram above shows the nucleotide sequence of a segment of a DNA strand, called a codon. (figure available in print form)

Complementary Base Pairing

a. Adenine always pairs with thymine and cytosine always with quinine. In the nucleotide alphabet on one half of the DNA helix determines the alphabet of the other half.

ANSWERS: GENETICS WORD LIST: GENETICS

(figure available in print form)

Fig. 3

## Crossing Over

During the formation of reducing the number of chromosomes (diploid number) to one half (haploid) in a process of sex cell formation of sperm and egg, homologous pairs of chromosomes undergo synapsis (form tetrads). The process of splitting the number of chromosomes is called meiosis. Sometimes there is an exchange of segments between chromatids (individual chromosomes) of a tetrad. In such an exchange of parts, some of the parts of the chromosomes exchange places. This is called crossingover and it is because of the new linkage that we have variety in life.

Major Source of Genetic Variation

a. Mutation gene A

(figure available in print form)
b. Crossing-over between alleles A and B
(figure available in print form)
Ultrasound image of fetus
(figure available in print form)

Prenatal Screening

- 1. Maternal checking for the Rh factor.
- 2. Maternal Serum Alpha-Fetoprotein Screening.
- a. This blood test measures the amount of alpha-fetoprotein (AFP) levels. Women carrying fetuses with spina bifida (in which the spinal column fails to close during development) and other defects of the central nervous system.
- 3. Ultrasound-imaging can reveal defects in central nervous system, kidney and urinary tract obstructions and in conjunction with echocardiography, physicians can examine heart structure and function. The approximate of the fetus can be determined.
- 4. Amniocentesis—the fluid in the amniotic space checked for chromosomal abnormalities.
- 5. Chorionic villus sampling—(CVS), fingerlike projections of the membrane surrounding the embryo later becoming the placenta. CV cells carry the same genetic information as the developing fetus. The CVS testing can be done during the ninth through eleventh weeks of pregnancy.
- 6. Analysis of fetal cells by using recombinant DNA technology. These tests focus on the structure of the human genome.

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7. For the future, it is hoped that by a blood test alone the same results can be obtained, since there is a chance of some cells of the developing embryo circulating in the mother's blood.

Prenatal Diagnosis (figure available in print form) GENETICDISORDERS

How Recessive Inheritance Works

(figure available in print form)
—March of Dimes

Autosomal Recessive Disorders

Recessive disorders need the presence of a pair of mutant genes (see diagram). The parents are usually healthy carriers, but both carry the same harmful gene then each of their children will have a 1-4 or 25% chance of having the genetic disease. Each child will also have a 25% chance of not having the disease; and each child has a 50% chance of getting only a single defective gene and becoming a carrier.

Recessive Genetic Disorders

- 1. Cystic fibrosis disorder affecting the mucous function.
- 2. Sickle cell disease blood disorder that causes the sickling of the red blood cells. An abnormal form of hemoglobin that can lead to a severe anemia.
- 3. Galactosemialnability to metabolize milk.
- 4. Phenylketonuria (PKU)Essential liver enzyme deficiency.
- 5. Thalassemia-blood disorder
- 6. Tay-Sachs disease—Fatal brain damage

How Dominant Inheritance Works

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(figure available in print form)

#### **AUTOSO-MAL DOMINANT INHERITANCE**

Dominant disorders show a 50% risk that each child born to a couple with one parent having the dominant faulty gene. There is also a 50% chance of a child being fortunate enough not to get the faulty gene. With all the new genetic technology, parents can now find out how the chances worked.

#### Dominant Genetic Disorders

- 1. Huntington's disease—a progressive nervous system degeneration that manifests itself at midlife.
- 2. achondroplasia—a form of dwarfism
- 3. chronic simple glaucomablindness if untreated
- 4. polydactyly—extra fingers or toes
- 5. hypercholesterolemia-high blood cholesterol
- 6. neurofibromatosis—multiple tumors of skin and nerves

Note: For every child born to the same mother and father, the risk of a genetic disease is the same. The percentage risk remains the same.

## Chromosomal Abnormality Disorders

- 1. Down syndrome (Trisomy 21)—Extra chromosome causes mental retardation, increased susceptibility to infection, heart defects and folding eyelids.(karyotype)
- 2. Philadelphia chromosome in leukemia due to a deleted chromosome 22. (karyotype shown in diagram)
- 3. Klinefelter syndrome—shows an extra female sex chromosome (XXY) and is defective in sexual development.
- 4. Turner syndrome—a female with a deleted X chromosome showing only XO. Shows lack of sexual development.
- 5. Fragile X chrosome—most common hereditary cause of mental retardation.

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How X-Linked Inheritance Works (figure available in print form)

Genetic Counseling —March of Dimes

#### Sex-linked Inheritance

A normal female has two X-chrosomes and a normal male his one X and one Y. When the mother carries a faulty gene on one of the X chrosomes, a son has a 50% risk of having the disorder. If the daughter gets the faulty gene, she can become a carrier of the genetic faulty gene. The carriers are usually unaffected by the disease but the mother can transmit the disease to a son.

#### Sex-linked Genetic Disorders

- 1. Hemophilia—defect in the blood-clotting mechanism, that does not make the right protein. Hemophilia A, the most common form of hemophilia, is caused by a deficiency of factor VIII. Remophilia B is caused by a deficiency of factor IX.
- 2. Agammaglobulinemia—lack of immunity to infections.
- 3. Color-blindness—the inability to distinguish colors.
- 4. Duchenne Muscular Dystrophy—An X-linked disorder associated with progressive muscle weakness beginning at about three years of age.

### KARYOTYPE OF A NORMAL MALE

(figure available in print form)
Chromosome Abnormality
(figure available in print form)
—March of Dimes Birth Defects

# Karyotype of Leukemia Patient with Philadelphia Marker

Cells from the bone marrow of patients with chronic myelogenous leukemia have a particular abnormal karyotype in many of the cases. There is a rearrangement of a part Observe arrows in karyotype. This could be helpful in diagnosis, treatment, and monitoring of the disease. This rearrangement is known as the Philadelphia marker.

Other leukemias have shown karyotypic abnormalities. Karyotyping has become a important diagnostic tool.

#### **INHERIITANCE** continued

## Multifactorial Disorders

These disorders make up the most common and probably the least understood of all the groups of inherited diseases. These disorders result from the interaction between one or more genes with the environment. The environment can include not only the actual environment that the mother is exposed to but the direct uterine

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environment of the embryo and fetus.

- 1. Congenital heart lesions
- 2. Neural tubo defects (spins bifida and anencephaly) malformations of brain and spinal cord exposed.
- 3. Juvenile onset diabetes
- 4. Cleft lip or cleft palate
- 5. Clubfoot

Chromosomal Disorders—Down Syndrome (Tranalocation of Chromosome) (figure available in print form)
Translocations and Down Syndrome

The karyotype for Down syndrome does not always consist of 47 chromosomes. Sometimes the extra No. 21 chromosome is linked by translocation to another chromosome. The patient has 46 chromosomes but still has a triple dose of the No. 21 chromosome material and exhibits all the clinical symptoms of Down syndrome. In this karyotype the extra No. 21 chromosome is attached to the top of one of the No. 14 chromosomes.

When this karyotype appears in a child with Down syndrome, the parents' blood should be studied to determine if the translocation is new or inherited. Parents can carry the translocation and themselves be unaffected. With this type of translocation they would have a total of 45, not 46 chromosomes, but would have the normal, balanced dosage of all genetic material. Families with an inherited translocation would be at increased risk for producing more affected offspring.

Down syndrome (shows extra chromosome Trisomy 21)

(figure available in print form)

The chromosomal map of some diseases. The genome project hopes to find specific genes for human disease. (figure available in print form)

Source:Smithsonian, Feb. 1990. James Watson and the search for biology's 'Holy Grail', Stephen S. Hall.

#### **ACTIVITY # 1 A GENETIC SURVEY**

Objective To find out, by means of a survey, of some genetic truths and some myths, how much people understand some basic genetic principles.

# **Procedure**

A. To conduct the survey you will need to get ten people to answer a true false questionnaire based on some truths and some superstitions and false ideas. You will be given the answers.

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B. Collect the data from the entire class. Get the total number of answers after each statement.

# Part 1. Collecting Data True False Total

- 1. Identical twins are always the same sex.
- 2. The mother determines the sex of the child.
- 3. Each parent contributes half the number of chromosomes in the developing embryo.
- 4. Fraternal twins are more closely related to each other than to their siblings.

- 5. The embryo may be affected and the genetic changed by unhappy experiences of the mother.
- 6. An alcoholic mother could produce an alcoholic child.
- 7. More males are color-blind than females.
- 8. Male babies are stronger than female babies.
- 9. A craving for a specific food during pregnancy may cause a birthmark.
- 10. The parent with a more determined nature will contribute the more genes to the child.
- 11. Children born to older parents are usually lacking in vitality.
- 12. Amniocentesis is always performed on everyone who is pregnant.
- 13. A parent may contribute characteristics to the child, which he does not show.
- 14. If a parent loses an arm or leg in a accident, the developing child will be missing a limb.
- 15. Genetic screening gives a good picture of the developing fetus.
- 16. A congenital malformation is sometimes the sign of being punished for wrong doing.
- 17. Ultrasound is very dangerous to the developing embryo.
- 18. Certain inherited traits may be altered by the moon shortly after conception.
- 19. Some hereditary characteristics are controlled by the blood.
- 20. All new borns should be tested for PKU.

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Part 2. Complete the data table from the material you have gathered and evaluated. The survey answers are included.

Part 3. Make a graph of the data. Show the percent of the correct and incorrect answers to each of the questions in the survey. Suggestion; use different colors to distinguish correct and incorrect answers.

Conclusions	After studying the	results, were th	ere any qu	estions where	many	people	were
incorrect. Gi	ve some reasons for	the false answ	ers.				

Part 2. Evaluating and compiling survey data for the class. Survey answers are given below.

(figure available in print form)

# **ACTIVITY #2 Dominant and Recessive Traits**

Objective To visually show the results of dominant traits and recessive traits.

Procedure This exercise works best with an overhead projector.

Materials Pieces of brown and colorless acetate samples. (4)

**Collecting Data** 

- A. Make a large Punnett Square and use the acetate pieces to represent a dominant trait. Record.
- B. Show result of a cross of two each with one recessive gene (colorless). Record.

#### Conclusion

- 1. Explain the results.
- 2. Research some diseases of dominance inheritance and diseases caused by recessive inheritance.

Problem How often do certain human traits manifest themselves?

Procedure Two traits to be studied are the ability to roll one's tongue and how the earlobe is attached. The ability to roll one's tongue is dominant over the inability and will be assigned the capital R. The inability will be designated by the small r. The small r is recessive.

Earlobes are either free or attached to the jaw. The free earlobes are dominant and represented by the capital

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letter E. The attached earlobes are recessive and represented by the small e.

The data will be shown for earlobe attachment in three ways; (a) by a human pedigree chart, (b) by a Punnett square, (c) and by a genotype-phenotype chart.

#### Free

(figure available in print form)
Attached
(figure available in print form)
Rolled tongue
(figure available in print form)
(figure available in print form)

#### Conclusion

- 1. Explain the results of your experiment.
- 2. Try to get so results from family or other friends.

# **ACTIVITY #3 GENETICS**

(figure available in print form)

# Activity #4 What Do Normal and Sickled Cells Look Like?

Facts The function of red blood cells is to carry oxygen. Oxygen is carried by the hemoglobin molecule inside the red blood cell. Red blood cells must move easily through the tiny capillaries.

Purpose To examine and compare slides of normal and sickled red blood cells.

Materials Prepared slides of normal and sickled red blood cells. (Carolina Biological). Microscope.

#### **Procedure**

- 1. Directions will be included to make your own slide or examine the prepared slide of normal red blood cells. -Locate the cells under low and then high power.
- 2. Draw and title several of the normal red blood cells in the space provided below.
- 3. Examine the slide of sickled red blood cells under low and then high power.
- 4. Draw and label in the space below.

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(figure available in print form)

## **Questions**

1. Describe the shape of normal red blood cells
2. Describe the shape of sickled red blood cells
3. Explain why sickled red blood cells do not easily move through capillaries.
4. Discuss the reason why sickled red blood cells do not carry as much oxygen as normal red blood cells

## Homework Find the origin and discovery of the trait and disease.

Note: Carolina Biological has a Biokit (70-4001) that is designed to perform a modern clinical test. Students use samples of their own blood to observe normal blood morphology and to simulate the sickle cell abnormality in crisis. Microscope Slides of Sickle Cell Anemia, (PH 1015) and Sickle Cell and Hemoglobin C. (PH 1016) peripheral blood film showing both of these hemoglobinopathies existing together.

# **Activity #4 Technique for SUSTAINING Blood Smears**

(figure available in print form)

### **Procedure**

- 1. Rub the tip of your ring finger with alcohol. Let dry.
- 2. Make a guick prick with lancet or microlancet.
- 3. Don't squeeze finger. Let one drop ooze out. Touch the second drop near one end of slide (a.).
- 4. Sterilize the puncture with the alcohol.
- 5. Tilt the spreader slide and draw the slide in the position (b) until it touches the drop of blood. The blood will spread in the angle formed by the two slides. Quickly push the spreader slide forward toward the far end of slide (c). Allow the slide to dry.
- 6. Flood slide with Wright's stain but do not let it overflow. *Gently*, after 3 minutes, add some buffer solution (pH7) and the *very gently* blow on slide to mix(e&f). After 1 minute wash of slide in running tap water. Dry.
- 7. Slide should be a pinkish-purple.

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## Audio-Visual Material

Anna: Morguio syndrome, accompanied by Dr. Greta Seashore or some other genetic professional. This video will give incite into how one person, with a genetic disease has adjusted her life and is a productive member of society.

"Chances Choices" Ten scenes describing genetic concepts in the context of one fictional family's medical history. 53 wall St for brochure.

Genetic Concepts Kit, Ca #70, Lab-Aids, Inc. wards, 53 Wall St.

Speakers available as genetic professionals to speak on selected topics in Medical Genetics. (contact Dr. Greta Seashore)

DNA Made Easy—Plastic DNA and RNA building blocks; transcription and translation. Ward's catalog) 53 Wall St.

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