Name: __________________________

Physical Anthropology Lab # 5

Medical Genetics: Dermatoglyphics, Karyotyping

OBJECTIVES: To become acquainted with two of the methods of medical genetics and physical anthropology, dermatoglyphics and karyotyping.


INTRODUCTION:

The application of genetic knowledge to the prognosis and diagnosis of inherited disorders is called genetic counseling. The term “prognosis” as used by medial geneticists refers to the probability that an individual will show an inherited disorder. The majority of people who seek genetic counseling do so after the birth of a defective child. Human geneticists have four methods by which they are able to diagnose an inherited disorder and offer a prognosis: (1) medical examination and laboratory test, (2) pedigrees, (3), dermatoglyphics, and (4) karyotyping.

We have already covered pedigree analysis in a previous lab. In this lab, we will focus on the latter two methods used in medical genetics. Physical anthropologists are also interested in these two methods for the study of human variation and evolution.

DERMATOGLYPHICS

Because chromosome disorders often result in characteristic combination of patterns of the ridged skin of the hands and feet, dermatoglyphics is important to medical geneticists. Often, in fact, a diagnosis of a chromosome disorder can be made without the intrusive procedure of drawing blood from a patient for a karyotype. For example, 60% of individuals with Down's Syndrome have ulnar loops on all fingers and those with Turner's Syndrome often have unusually large ridge patterns on their fingers. Because of this association between dermatoglyphics and chromosomal disorders, finger and palm printing is often part of the routine examination of a medical geneticist.

KARYOTYPING

Because some disorders and deformities have been traced to aneuploidy, human geneticists assemble and analyze human karyotypes. A karyotype is the chromosomal make-up of a species in which the chromosomes are arranged according to size, location of centromere, and banding patterns. The intense study of karyotypes had been possible only in recent times because of the complex laboratory techniques used to study chromosomes. The first human karyotypes to be studied came from incised gonads. It was not until 1956 that the correct number of human chromosomes was known. In the 1960's, techniques became available which allowed researchers to use while blood cells undergoing mitosis for karyotyping. In the early 1970's, banding techniques became available. Today, both white blood cells and cells from an unborn fetus which float in the
amniotic fluids can be cultured, the chromosomes harvested, and a karyotype prepared and analyzed. Once the chromosomes have been prepared for karyotyping, they are photographed, printed, cut out, and assembled into a karyotype.

The attached figure is an enlargements of what you would see if you looked into the microscope after the cells of a normal male have been prepared. The numbers have been added to assist in sorting out this karyotype.

The chromosomes are arranged into groups according to their length and positions of the centromeres:
- Group A chromosomes are long with the centromeres in the center.
- Group B are long chromosomes with the centromere located slightly off-center.
- Group C chromosomes are moderate in length with the centromeres in the center.
- Group D are moderate in length with centromeres toward one end.
- Group E chromosomes are small with the centromeres in the center or slightly off-center.
- Group F are small with centrally located centromeres.
- Group G chromosomes are small with centromeres located toward the ends.

Individual chromosomes are recognized according to their particular banding pattern, which is shown in the attached figure. Unless there has been an inversion (where part of the chromosome turns upside down) or a translocation (where a piece of one chromosome transfers to another chromosome), the bands on the chromosomes are the same in each person. In fact, homologous chromosomes can be recognized among closely related species, thereby tracing the evolution of karyotypes.

**EXERCISES:**
1. Make a good set of dermatoglyphic prints for each of your fingers using the material provided.
2. Complete Exercise 13.5 on pp. 341-342 of the lab manual.
3. Using the schematic provided (Human Karyotype Form), cut out and assemble the human chromosomes that you have been given in order to construct a karyotype. Include the number of chromosomes, sex of subject, and type of disorder or if normal on the same form.