

## **Syllabus Material for SCCGS, June 2013**

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**GENEALOGY BEYOND THE Y CHROMOSOME:  
AUTOSOMES EXPOSED**  
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**THE BASICS**

Traits are determined by Genes.  
Genes are located on Chromosomes.  
Chromosomes are composed of DNA

DNA is a sequence of base pairs A, C, G, and T

Each cell contains 23 pairs of chromosomes – namely 22 numbered pairs called autosomes and two sex chromosomes, X and Y. Females have two X chromosomes whereas males have one X and one Y chromosome.

Mitochondria are the energy bars of the cell and contain mitochondrial DNA (mtDNA)

**CLASSICAL GENETIC GENEALOGY – A REVIEW**

**Y-Chromosome**

Passed from father to son through the generations. Can be used to trace direct male lineages. Can find recent ancestors as well as very distant ancestors.

**MtDNA**

Passed from mother to all her children through the generations. Can be used to trace direct female lineage. Used mainly to find very distant ancestors.

**AUTOSOMES**

Passed from both parents to all their children. Should be able to allow us to trace all lineages, but how?

**IT STARTED WITH MENDEL**

First law: Segregation of Characteristics  
You inherit one gene randomly from each parent

Second law: Independent Assortment  
You inherit each gene independently of the other genes

Both of these laws were derived by selective matings and taking statistics on the offspring. And for the traits Mendel chose, the statistics verified both laws.

## **THEN CAME CHROMOSOMES**

Mendel didn't know about chromosomes. But once they were discovered, it would seem that genes on the same chromosome would be inherited together and Mendel's second law would fail. And, indeed, when such genes were tested, they were inherited together nearly all the time. But occasionally they were not inherited together, suggesting that something else was coming into play. And that something else is crossover.

## **CROSS-OVER**

Sex cells (sperm and egg) contain only 23 chromosomes, not 23 chromosome pairs. When sex cells are produced, they get one of the two chromosomes of each pair from the cell that made them. But rather than getting the chromosome in tact, they get a blending of the two chromosomes of each pair. This is because the two chromosomes in the pair actually cross over at random points when the sex cell is being produced. And this cross over is what caused two genes on the same chromosome to occasionally get separated.

## **CENTIMORGANS**

The further apart two genes (or other markers) are on a chromosome, the more likely they are to get separated. So the probability of separation is related to distance along the chromosome. We can therefore measure distances by these probabilities. If two markers are separated 1% of the time, they are said to be at a distance of 1 centimorgan apart.

## **WHAT CAN WE DO WITH CENTIMORGANS**

Map the Genome  
Find your Ethnicity  
Find your Cousins

## **MAPPING THE GENOME**

Unraveling the human genome involves:

1. Determining which genes are on which chromosomes and where (mapping).
2. Determining the "normal" DNA code for each gene and then determining the DNA variations (sequencing)

We will focus on the mapping

## **DETERMINING WHICH GENES ARE ON WHICH CHROMOSOMES**

1. Genetic mapping using crossover frequencies
2. Physical mapping using modern biology techniques

We will focus on genetic mapping

To do so, we make a large list of traits.

For each pair of traits, determine the frequency of separation

The smaller the frequency, the closer the genes

## **FINDING YOUR ETHNICITY**

Examining the autosomes can tell what mixture there is of certain ethnicities, such as European, East Asian, Sub-Saharan African, and Native American. Requires having a good estimate of the DNA sequence of these populations. Even small error in the estimates can result in large errors in determining the mixture.

## **FINDING YOUR COUSINS**

Follow the autosomes from generation to generation. At each generation the autosomes get blended. The maximum length of an unblended sequence of DNA decreases with each successive generation. Two descendants of the same ancestor will share some of these unblended sequences, but the further removed they are from the common ancestor, the smaller will be the common sequences. So the maximum length of the common sequence (in centimorgans) can be used to determine what level cousins they are.