Human Genetics (50-233)

Instructor: Carl Hansen

Office Hours: M 10:00 am – 11:30 am
W 10:00 am – 11:30 am
TH 9:00 am - 11:00 am
and by appointment

Office : 123 Hartline Science Center

Lecture: **MWF, 9-9:50 am, 86 HSC.**

Required text: **Human Genetics**, Ricki Lewis

Website: **http://facstaff.bloomu.edu/chansen/**

Course Objectives:

1. To provide an understanding of the basic principles of genetics.

Classical genetics

Passage of traits within families

Meiosis, Mendel's laws, sex linkage, cytogenetics

Molecular Genetics

Study of DNA, RNA and proteins

Gene expression, mutation, cloning and genomics

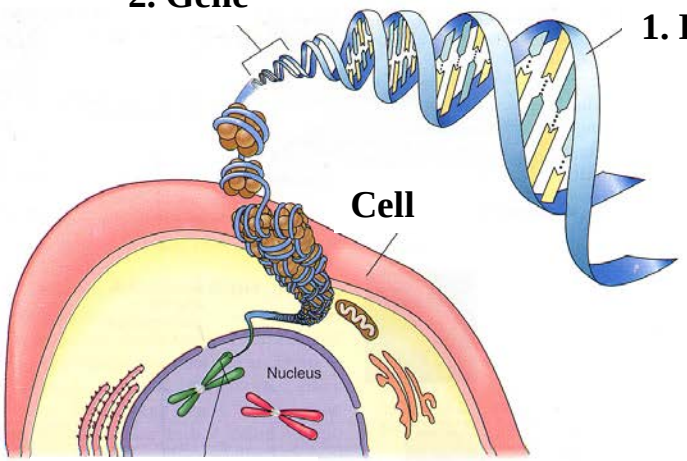
Population/Evolutionary Genetics

Study of allele frequency within the population

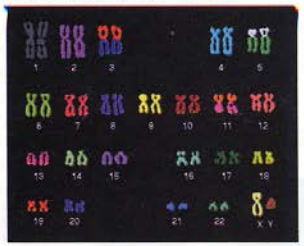
Quantitative genetics, Hardy-Weinberg equilibrium, evolution and speciation

2. Gene

1. DNA

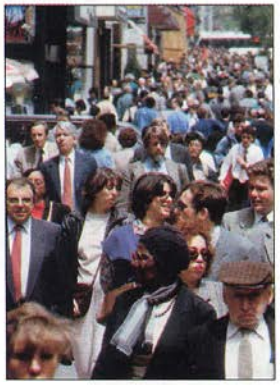


4. Genome



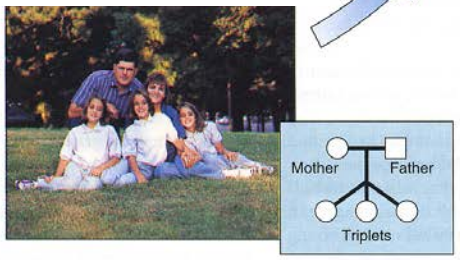
3. Chromosome

7. Population



5. Individual

6. Family (pedigree)



Course Objectives:

2. To provide an understanding of the genetic basis of human disease and current approaches to treatment and prevention of genetic disorders.

3. To provide an awareness of the interaction between genetic and environmental factors underlying birth defects and the development of human disease.

Mendelian Disorders

Disorder	defective protein	phenotype
Achondroplasia	growth factor receptor	dwarfism
Cystic fibrosis	chloride transporter	lung, digestive pathology
Duchennes muscular dystrophy	dystrophin	loss of muscle function
Hemophilia	Factor VIII	clotting defects
Familial Hypercholesterolemia	LDL receptor	high cholesterol early heart disease

Multifactorial Disorders

Breast Cancer	
Biopolar affective disorder	- manic depression
Dyslexia	
Diabetes mellitus	- adult onset & obesity
Hypertension	
Neural tube defects	- folic acid
Schizophrenia	- 1% of pop affected
Seizure disorders	

Course Objectives

4. To relate the study of human genetics to major advances in molecular genetics and biotechnology.

5. To relate human genetics to current issues in genetic screening, genetic counseling and genetic engineering.

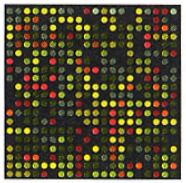
Trait	Risk
Addictive behavior	Greater than general population
Lung cancer	Greater than general population
Colon cancer	Less than general population
Alzheimer disease	Less than general population

Mackenzie's Genetic Profile

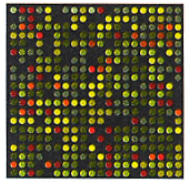
Trait	Risk
Cystic fibrosis	100% diagnosis
Blood serum cholesterol level	Greater than general population

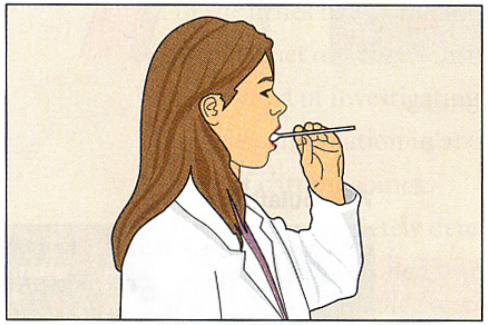
Laurel's Genetic Profile

Step 4: Results calculated, communicated

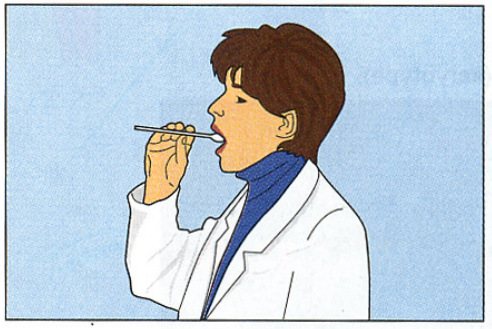


Step 3: Sample DNA applied to personalized DNA chips





Step 2: Provide DNA sample



Step 1: Research and record family history

Mackenzie

Laurel

Grade for the course will be based on four exams, case study presentations and a research paper.

Exam I	Jan 31, 2003	15 % of final grade
Exam II	Feb 24, 2003	17.5 % of final grade
Exam III	April 7, 2003	17.5 % of final grade
Exam IV (comprehensive final)	May 5, 2003	25 % of final grade
Research Paper	April 16, 2003	10 % of final grade
Case Study Presentations		15 % of final grade

Case Study Presentation Groups

Presentation Groups - 3-5 students.

Presentations - last 10-15 min of each class

- individuals from different groups will be asked to present assigned case studies to the class.

Group grade will be based on content, organization, and understanding of information presented during the oral presentation.

Each presentation will receive a 0, √-, √, or √+ (equivalent to 0, 60, 80 or 100). Final presentation grade for each group member will be the average of the grade received for members of the group.

Case study assignments correspond to the book chapter assignments and are to be completed by the chapter's lecture date.

Research Paper.

Each student is required to submit a short library research report (4-5 pages) on a human disease or genetic syndrome.

Topic to be covered must be approved by instructor. Students are welcome to ask for help in selecting a topic.

The purpose of the report is to allow you to learn about gathering, digesting and disseminating information regarding the genetic basis of a disease.

You will be graded on clarity, completeness and accuracy.

Acceptance of a paper after April 1, 2003 will only occur with documentation of an exceptional extenuating event.

What is Genetics?

Genetics is the study of heredity and its variation.

Genetic information is transmitted at several levels

DNA
Genes
Chromosomes
Genome
Individual
Family
Population

Genes

- are the basic unit of inheritance.
- are composed of DNA (deoxyribonucleic acid).
- direct the formation of proteins.

Different versions of the same gene are called alleles.

Alleles result from the process called mutation.

The genome

is the complete set of genetic information characteristic of the organism.

The genome includes:

- all of the genes present in an organism and
- other DNA sequences that do not encode genes

The human genome

- consists of 3 billion base pairs of DNA
- includes 28,000 to 34,000 genes
- is organized as 23 pairs of chromosomes

Individuals

carry two alleles of each gene.

Genotype is the combination of alleles that an individual possesses.

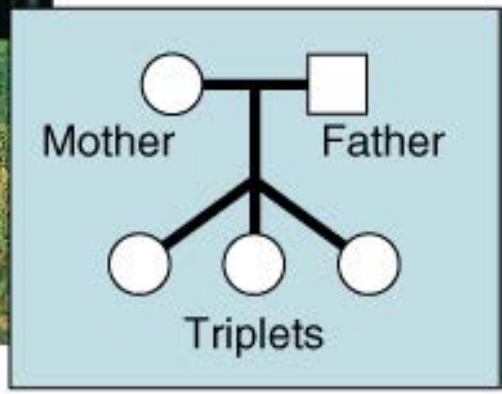
Phenotype is the visible trait that results in a particular genotype.

The family

Inheritance of traits can be observed in families.

A pedigree indicates the structure of a family schematically.

6. Family (pedigree)



A population

is a group of interbreeding individuals who possess a particular collection of alleles or gene pool.

Between species

Comparison of DNA sequences indicates the amount of similarity between two species.

98% of human DNA sequences are shared with chimpanzee.

Many genes present in humans are also present in mice, fish, fruit flies, yeast, and bacteria.

Human to human, we are very similar

On average two random people share the same DNA sequence in 99.9% of their genome.

Studies of variation among humans indicates humans arose in Africa and migrated across the globe with relatively little change.

We are also very different

On average two random people differ at 3 million base pairs (approximately one nucleotide of every thousand).

Traits can be determined predominantly by one gene.

Mendelian traits result from variation in alleles of one gene.

Traits can be determined by multiple genes.

Polygenic traits result from variation in several genes.

Traits can be determined by genes and environment.

Multifactorial traits result from effects of one or more genes and the environment.

Genetic risk

Absolute risk	the probability that an individual will develop a condition or trait
Relative risk	the likelihood that an individual from one group will develop a condition in comparison to another group (usually the general population)
Empiric risk	risk determined by observing incidence of a trait in the population
Risk factor	a situation that alters incidence of a disease (or trait)

History of understanding inheritance.

12,000-10000 BP (before present)

- **Domestication of animals**
- **Development of farming and plant-breeding**

2,000BC - Artificial fertilization of date palms

Pre-Darwinian Theories of Inheritance

Blended inheritance -the traits of parents average-out in the offspring.

-

Acquired characteristics -the experiences of the parents effect the offspring. Jean-Baptiste Lamarck (1744-1829)

Pangenesis –particles within our bodies know as "gemmules" carried the instructions to form new organisms.

- **Scientifically tested in 1871 by Francis Galton**

1866 - Gregor Mendel

“ Experiments in Plant Hybrids”

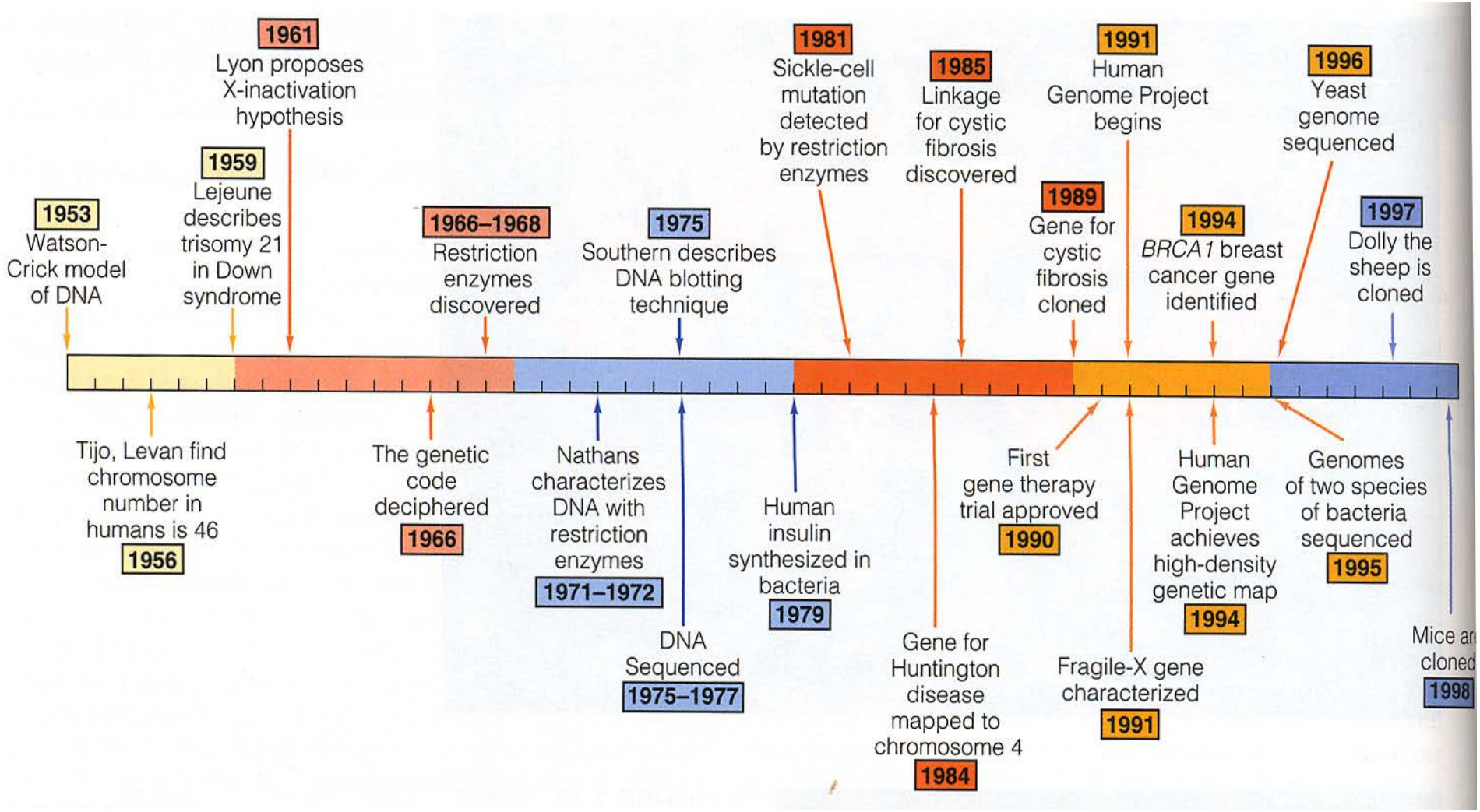
- established the basics rules of inheritence

- laid basically unnoticed till 1900

1900 – three biologists working independent – Hugo DeVries, Carl Correns and Ericj von Tschermak – rediscovered Mendel landmark paper

11900-1944 genetic flourished

- chromosome theory – they are linear arrays of genes


1953
Watson-Crick model of DNA
1956
Tijo, Levan find chromosome number in humans is 46
1959
Lejeune describes trisomy 21 in Down syndrome
1961
Lyon proposes X-inactivation hypothesis
1966
The genetic code deciphered
1966–1968
Restriction enzymes discovered
1971–1972
Nathans characterizes DNA with restriction enzymes
1975
Southern describes DNA blotting technique
1975–1977
DNA Sequenced
1979
Human insulin synthesized in bacteria
1981
Sickle-cell mutation detected by restriction enzymes
1984
Gene for Huntington disease mapped to chromosome 4
1985
Linkage for cystic fibrosis discovered
1989
Gene for cystic fibrosis cloned
1990
First gene therapy trial approved
1991
Human Genome Project begins
1991
Fragile-X gene characterized
1994
BRCA1 breast cancer gene identified
1994
Human Genome Project achieves high-density genetic map
1995
Genomes of two species of bacteria sequenced
1996
Yeast genome sequenced
1997
Dolly the sheep is cloned
1998
Mice are cloned