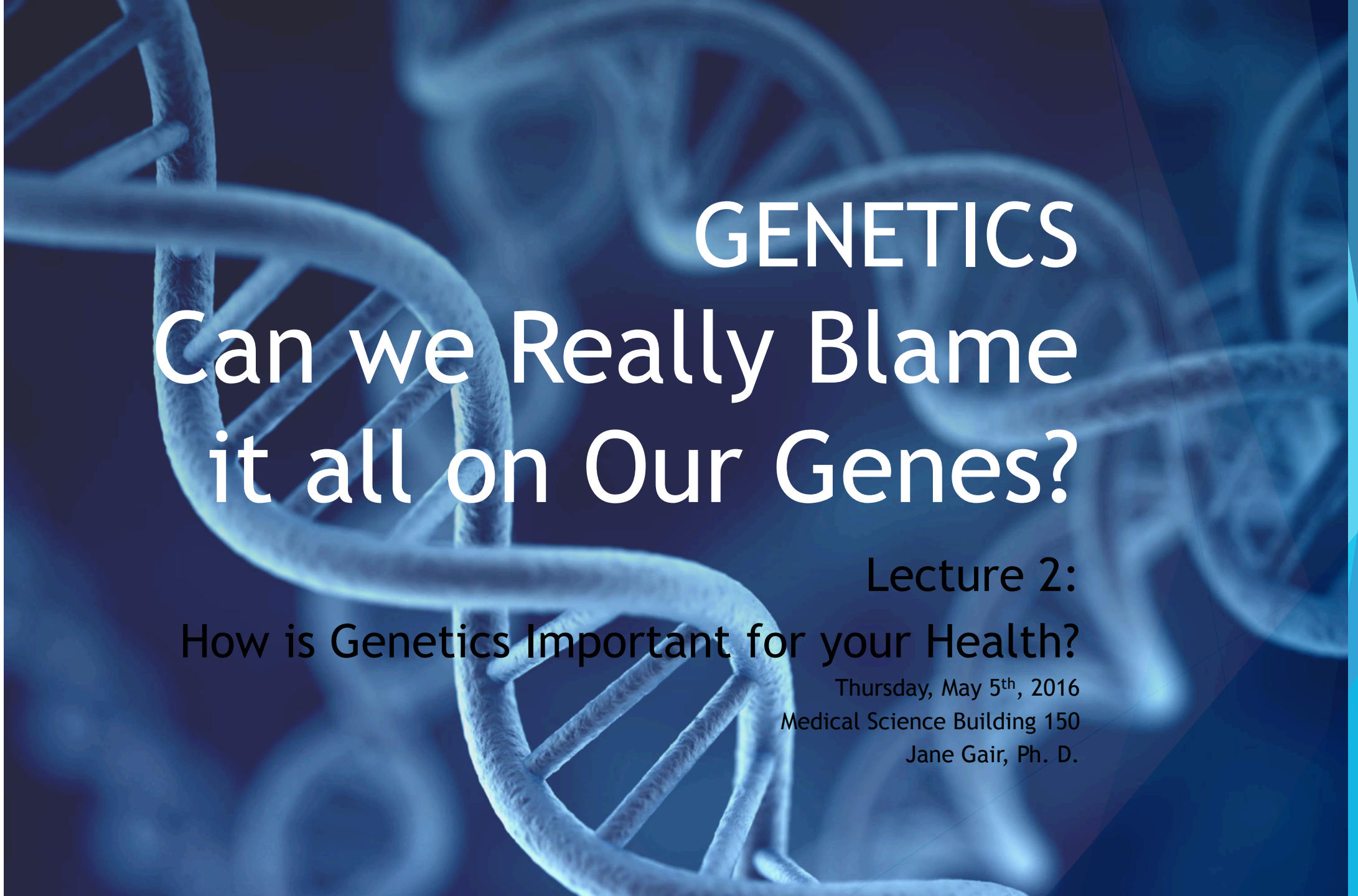


GENETICS
Can we Really Blame it all on Our Genes?

Lecture 2:

How is Genetics Important for your Health?

Thursday, May 5th, 2016

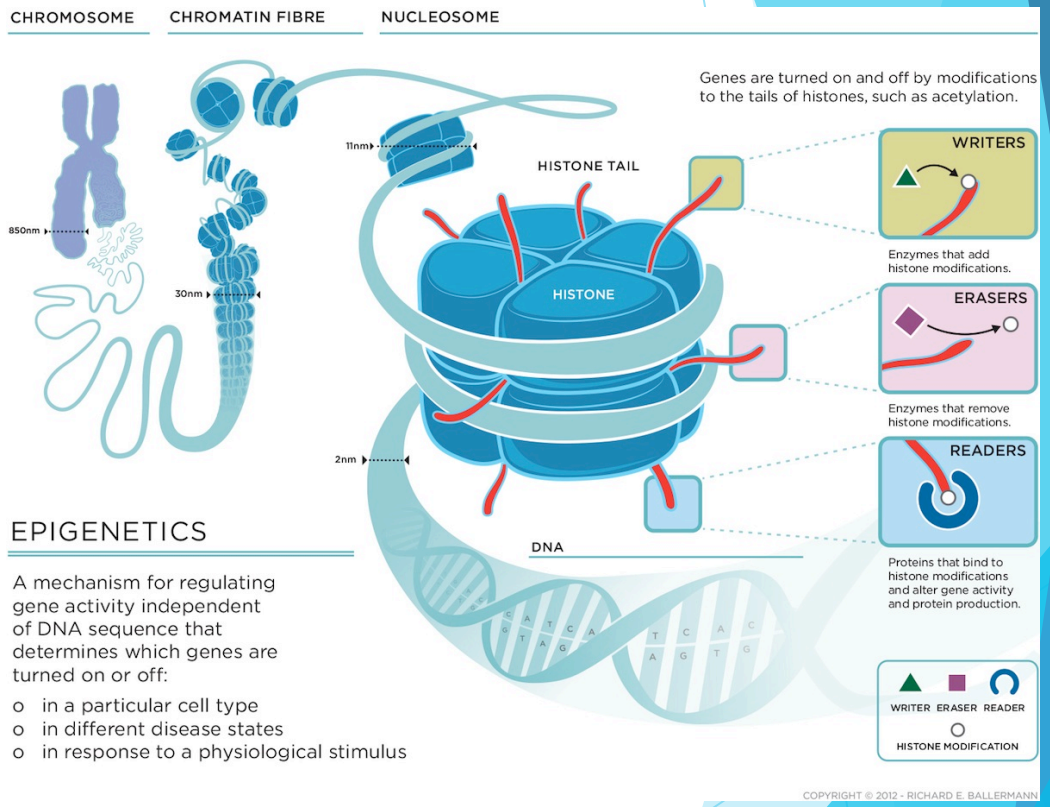
Medical Science Building 150

Jane Gair, Ph. D.

An Introduction to Genetics (Lecture 1)

CONTINUATION FROM LAST WEEK

- **EPIGENETICS**
  - **Study of cellular and physiological phenotypic variation in the lifespan**
  - **Modification to genes through alteration in histone elements by proteins**



An Introduction to Genetics (Lecture 1)

Epigenetics- Video
https://www.youtube.com/watch?v=AvB0q3mg4sQ

- **APPLICATION OF GENETICS**
  - **GENOMICS**
    - **Analyzes genome sequences to study structure, function, and evolution of genes and genomes**
  - **PROTEOMICS**
    - **Identifies proteins present in cells in certain conditions and studies post-translational modifications, locations and their interactions**
  - **BIOINFORMATICS**
    - **Stores, retrieves, and analyzes data generated by genomics and proteomics**

- **BIOTECHNOLOGY**
  - **Genetic modification of crop plants for:**
    - **Increasing herbicide, insect and viral resistance**
    - **Nutritional enhancement**

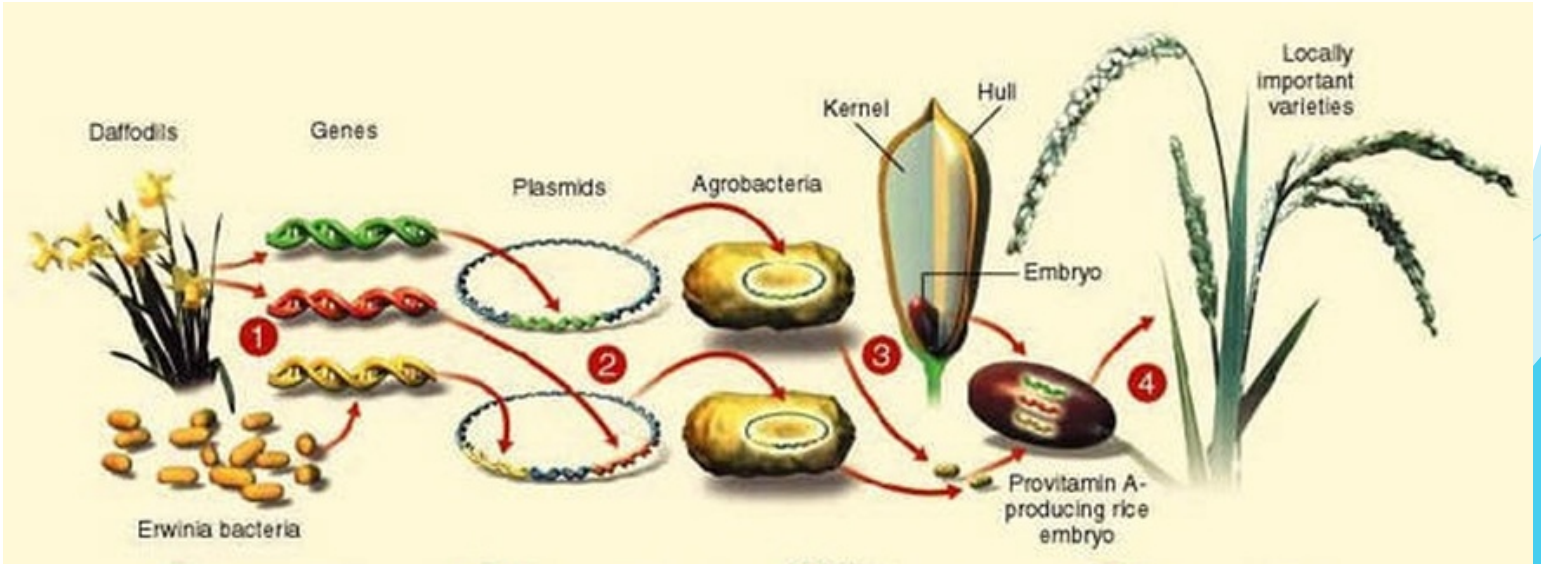
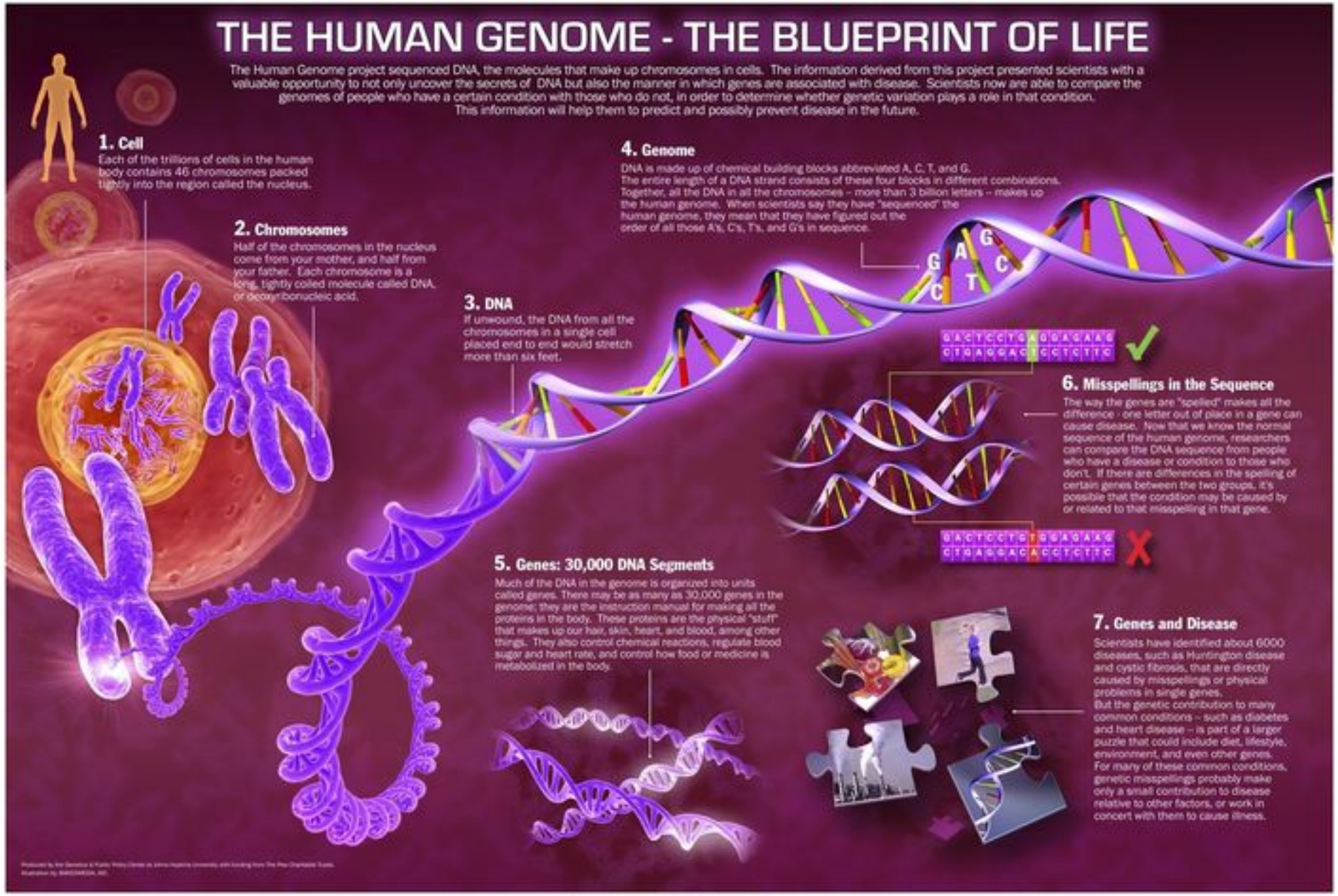


TABLE 1.1

Some Genetically Altered Traits in Crop Plants

Herbicide Resistance	
	Corn, soybeans, rice, cotton, sugarbeets, canola
Insect Resistance	
	Corn, cotton, potato
Virus Resistance	
	Potato, yellow squash, papaya
Nutritional Enhancement	
	Golden rice
Altered Oil Content	
	Soybeans, canola
Delayed Ripening	
	Tomato

Table 1.1

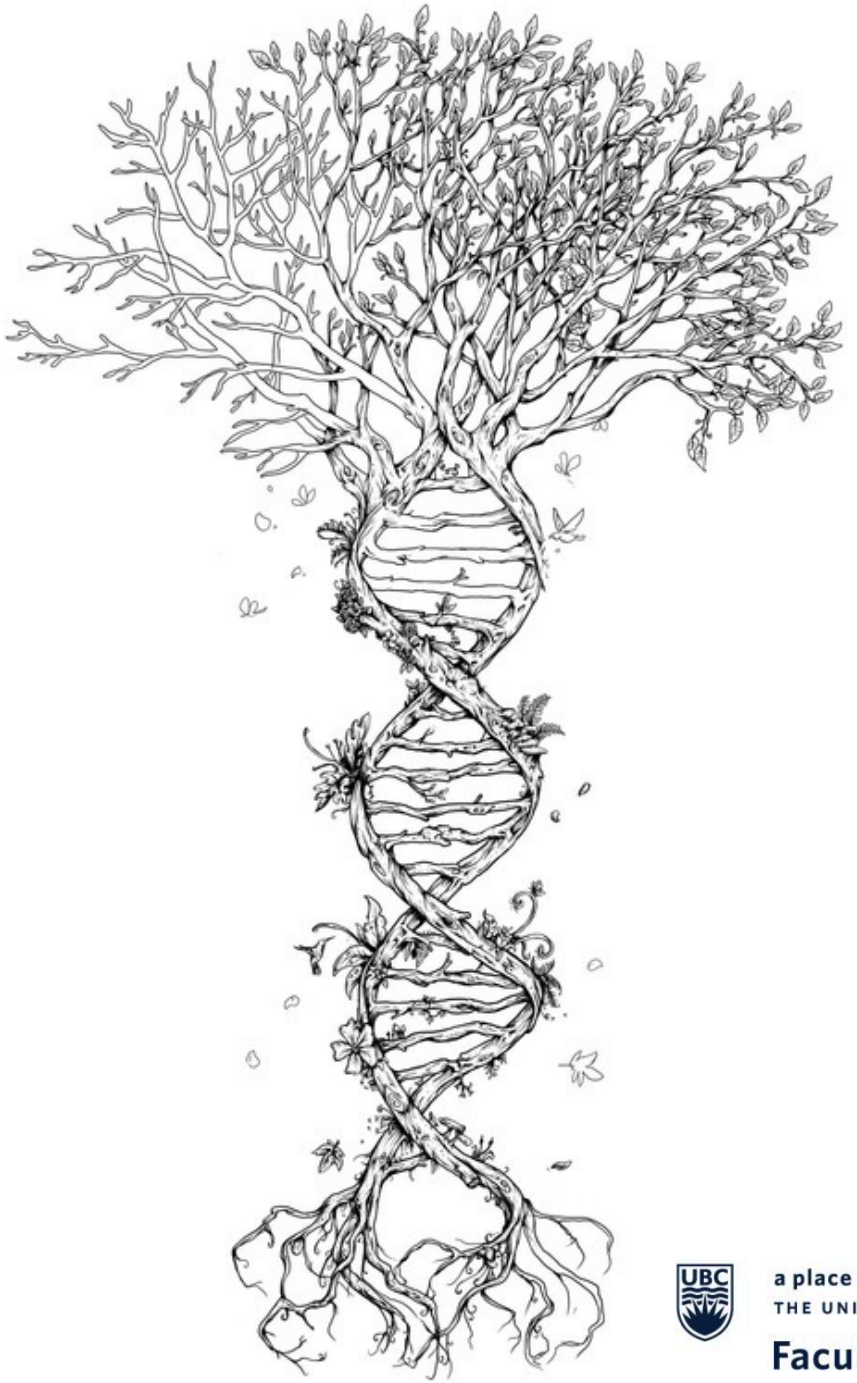


www.Infographicality.com

GENETICS: Can we Really Blame it all on Our Genes? Series Overview

- SUMMARY
  - Genetics is a very broad field relating to every living being on earth
  - Provides guidelines that aid in determining the makeup of an individual
  - Differentiation in genes, chromosomes, and other genetic elements all contribute to creating a unique organism
  - Genetics is not only useful in natural settings but can also be manipulated to serve certain needs, as seen in agriculture

QUESTIONS?



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Faculty of Medicine

University of Victoria

GENETICS: Can we Really Blame it all on Our Genes? Series Overview

- WEEK 1 (April 28th, 2016):
  Introduction to Genetics

- **WEEK 2 (May 5th, 2016):**
  How is Genetics Important for your Health?

- WEEK 3 (May 12th, 2016):
  Understanding the Genetics of some Common Diseases and Disorders

- WEEK 4 (May 19th, 2016):
  How Medicine can work with your Genetics to Improve your Care

OVERVIEW: How is Genetics Important for your Health? (Lecture 2)

- Summary from last week
- Genes, gene mutations, causes of mutations and onset of disease.
- Description of Pedigrees
- Family trees and how some diseases run in families
- Importance of Family History taking in medicine
- Impact of diet, exercise and lifestyle choices (e.g. Smoking) on genes, aging and overall health

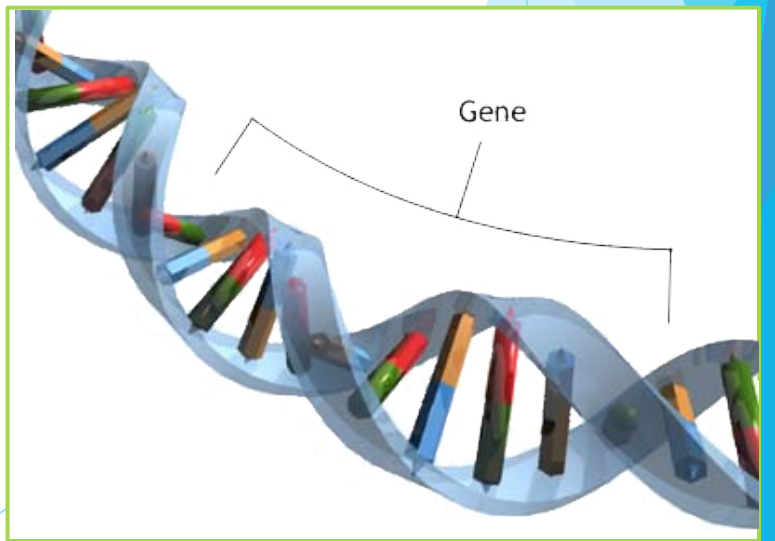
How is Genetics Important for your Health? (Lecture 2)

Recap from last week- An Introduction to Genetics- Video
https://www.youtube.com/watch?v=bVk0twJYL6Y

Genome
BritishColumbia

How is Genetics Important for your Health? (Lecture 2)

- **GENES**
  - **Fundamental units of genetic material/ information**
  - **DNA represents the sequence coding a polypeptide**
  - **~20,000-25,000 human protein-coding genes**



How is Genetics Important for your Health? (Lecture 2)

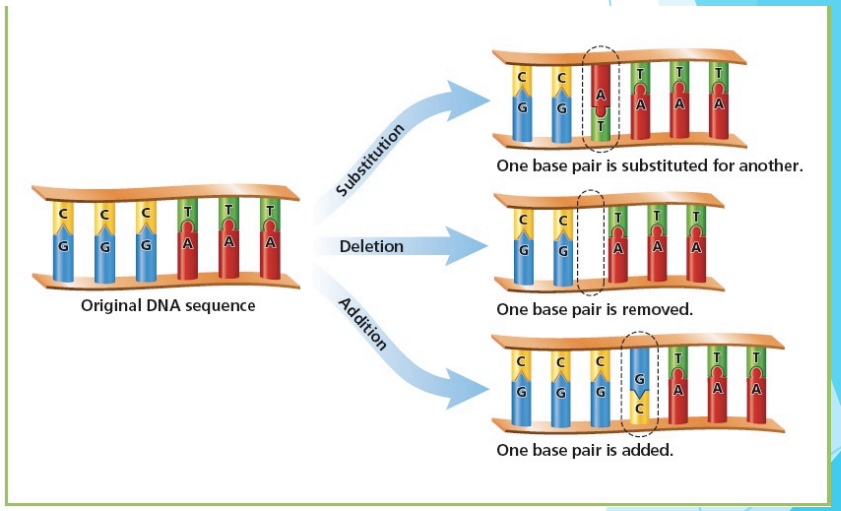
- **MUTATIONS**
  - **Permanent alteration of the nucleotide sequence of the genome**
  - **Results from damage to DNA that is not repaired**
    - **Errors in replication process**
    - **Insertion/deletion of segments of DNA**
  - **May or may not produce observable changes in the phenotype of an organism**

How is Genetics Important for your Health? (Lecture 2)

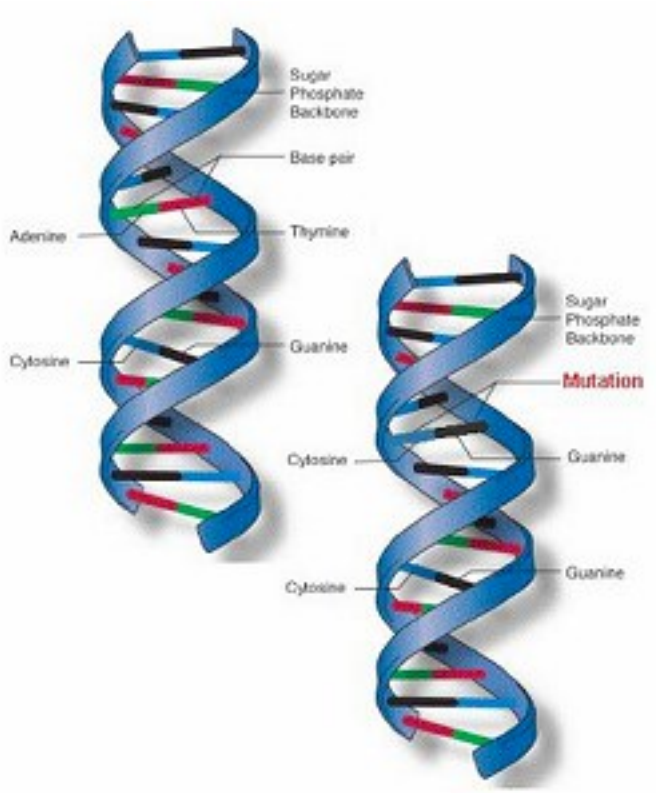
- Two types of mutations
  - GENE MUTATIONS
    - Changes in a single gene
  - CHROMOSOMAL MUTATIONS
    - Changes in whole chromosomes
- GERM MUTATIONS
  - Effect reproduction, heritable
- SOMATIC MUTATIONS
  - Effect the body (NOT heritable)

How is Genetics Important for your Health? (Lecture 2)

- **POINT MUTATIONS**
  - **SUBSTITUTIONS**
    - **One base is changed to a different base**
  - **INSERTION**
    - **One base is inserted from the DNA sequence**
  - **DELETION**
    - **One base is removed from the DNA sequence**



GENE MUTATIONS

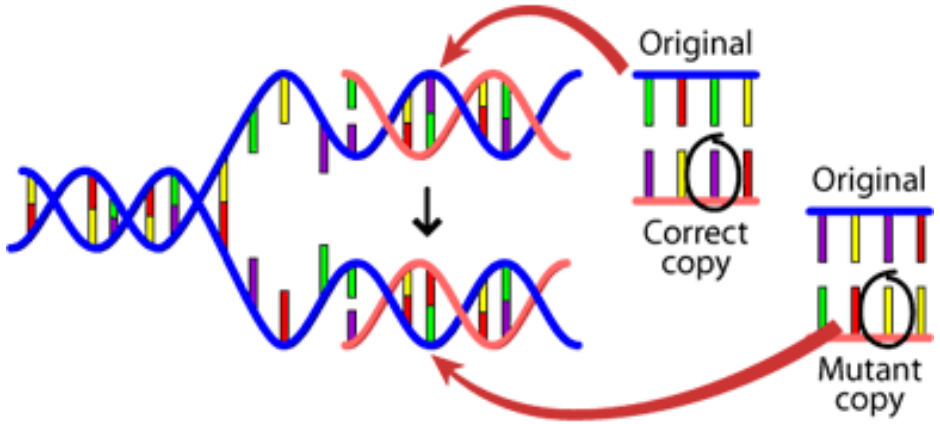


How is Genetics Important for your Health? (Lecture 2)

An Introduction to Genetic Mutations – Video (5 mins)
https://www.youtube.com/watch?v=g02RnGXCXrQ

How is Genetics Important for your Health? (Lecture 2)

- **SOURCES OF MUTATIONS**
  - **ENVIRONMENTAL FACTORS**
    - **Sunlight**
    - **Radiation**
    - **Smoking**



  - **ENDOGENOUS FACTORS**
    - **Errors in replication**
    - **Toxic bi-products from cellular metabolism**

How is Genetics Important for your Health? (Lecture 2)

- **Not all mutations are bad…**
  - **Mutations are the main driving factor for EVOLUTION**
  - **RESISTANCE to pathogens**
  - **TOLERANCE to cold**
  - **Need little SLEEP (mutation in DEC2 gene)**
  - **Super-DENSE bones**
  - **Crystal- Clear underwater VISION**

How is Genetics Important for your Health? (Lecture 2)

- DISEASE
  - Abnormal condition
  - Disorder of a structure
  - Disorder of a function

  - Usually has symptoms and signs

- PATHOLOGY
  - Study of diseases

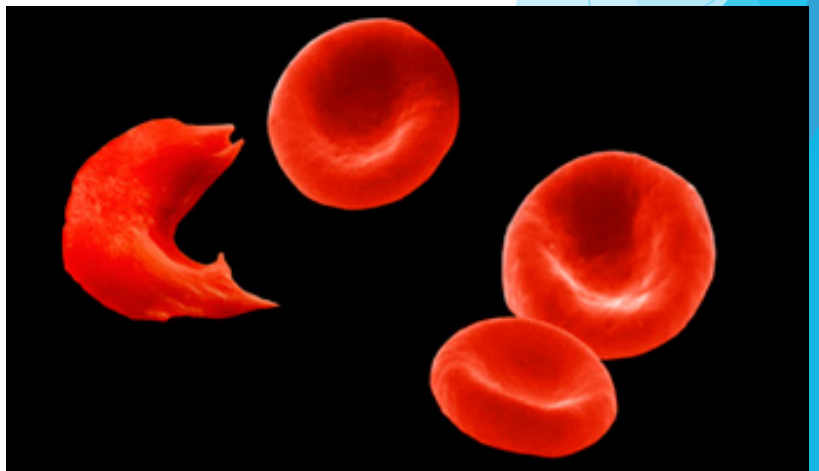
How is Genetics Important for your Health? (Lecture 2)

- **SUSCEPTIBILITY TO DISEASE**
  - **easily affected, influenced, or harmed by something**

- **IMMUNITY**
  - **Balanced state of adequate biological defenses to fight disease, while having adequate tolerance to avoid allergy and autoimmune disease**

How is Genetics Important for your Health? (Lecture 2)

- Disorders Caused by Individual Genes
  - Sickle Cell Disease
    - Severe hereditary form of anemia
  - Cystic Fibrosis
    - Affects exocrine glands
    - Causes production of abnormally thick mucus
    - Blocks pancreatic ducts, intestines, and bronchi
    - Results in respiratory infection



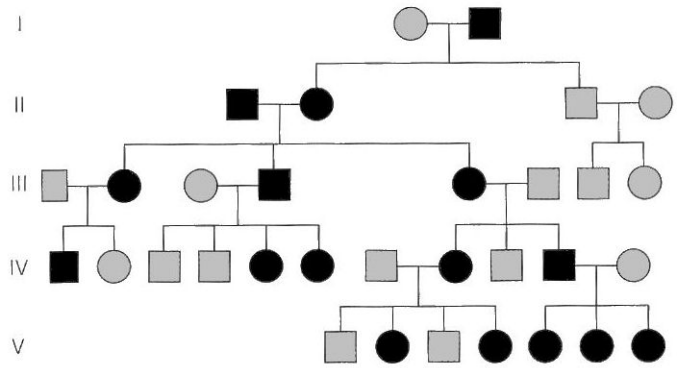
- **Disorders Caused by Individual Genes (Cont'd)**
  - **Huntington Disease**
    - **Progressive brain disorder that causes uncontrolled movements, emotional problems and loss of cognition**
    - **Caused by CAG repeats**
  - **Hemophilia**
    - **Group of hereditary genetic disorders that impair body's ability to control blood clotting**
    - **Hemophilia A is the most common form**

How is Genetics Important for your Health? (Lecture 2)

Five Amazing Genetic Human Mutations- Video (5 mins)
https://www.youtube.com/watch?v=kSqXjqIclAI

How is Genetics Important for your Health? (Lecture 2)

- Pedigree
  - Document to record ancestry
  - Used by doctors (genealogists) to study family lines
    - Ancestry chart
  - Selective breeding of animals
    - Breed registry



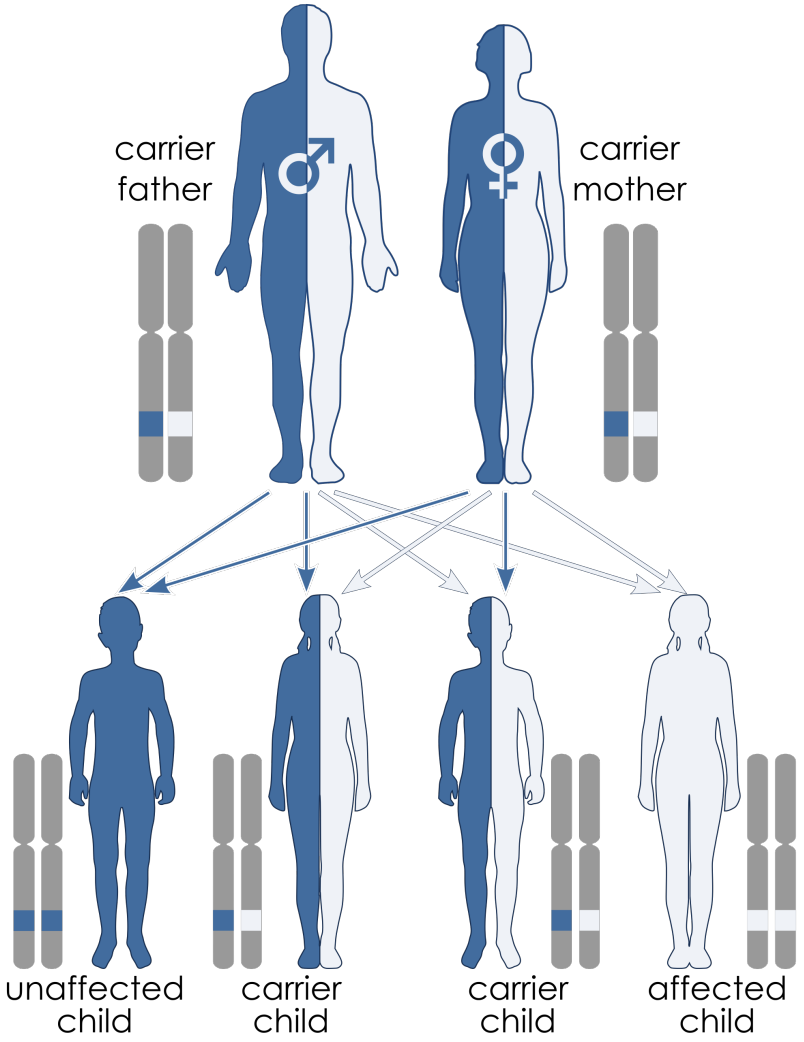
How is Genetics Important for your Health? (Lecture 2)

- **Pedigrees and Family trees**
  - **Analysis used to determine the pattern of inheritance of traits in humans**
- **Mode of inheritance in humans**
  - **Autosomal Recessive**
  - **Autosomal Dominant**
  - **X-linked Recessive**
  - **X-linked Dominant**
  - **Sex-influenced and sex-limited autosomal traits**
  - **Mitochondrial inheritance**

How is Genetics Important for your Health? (Lecture 2)

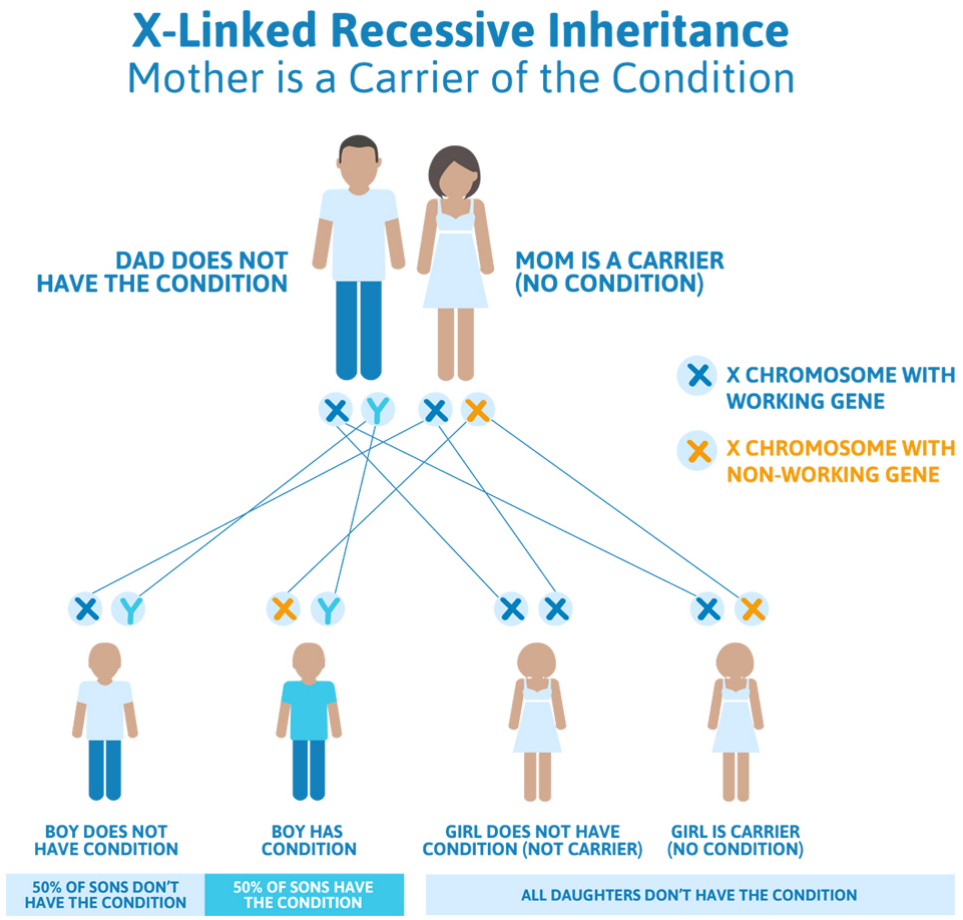
- **Autosomal Recessive inheritance**
  - **Traits not expressed in heterozygotes**
  - **Often skip a generation**
  - **Equal proportion of males and females affected**
  - **Rare traits often found in consanguineous marriage (union between related individuals)**
  - **CYSTIC FIBROSIS, SICKLE-CELL ANEMIA are examples of autosomal recessive diseases**

Autosomal recessive inheritance



How is Genetics Important for your Health? (Lecture 2)

X-linked Recessive – Video (3:41 mins)
https://www.youtube.com/watch?v=Vdam8pKhRNo



- **SEX-INFLUENCED TRAITS**
  - **Autosomal traits (not carried on sex chromosomes) that are influenced by sex**
  - **Males require one recessive allele while females need two recessive alleles to show the same trait**
  - **Example: Baldness**

Genotype	Female	Male
$B^+ B^+$	Bald	Bald
$B^+ B^-$	Full hair	Bald
$B^- B^-$	Full hair	Full hair

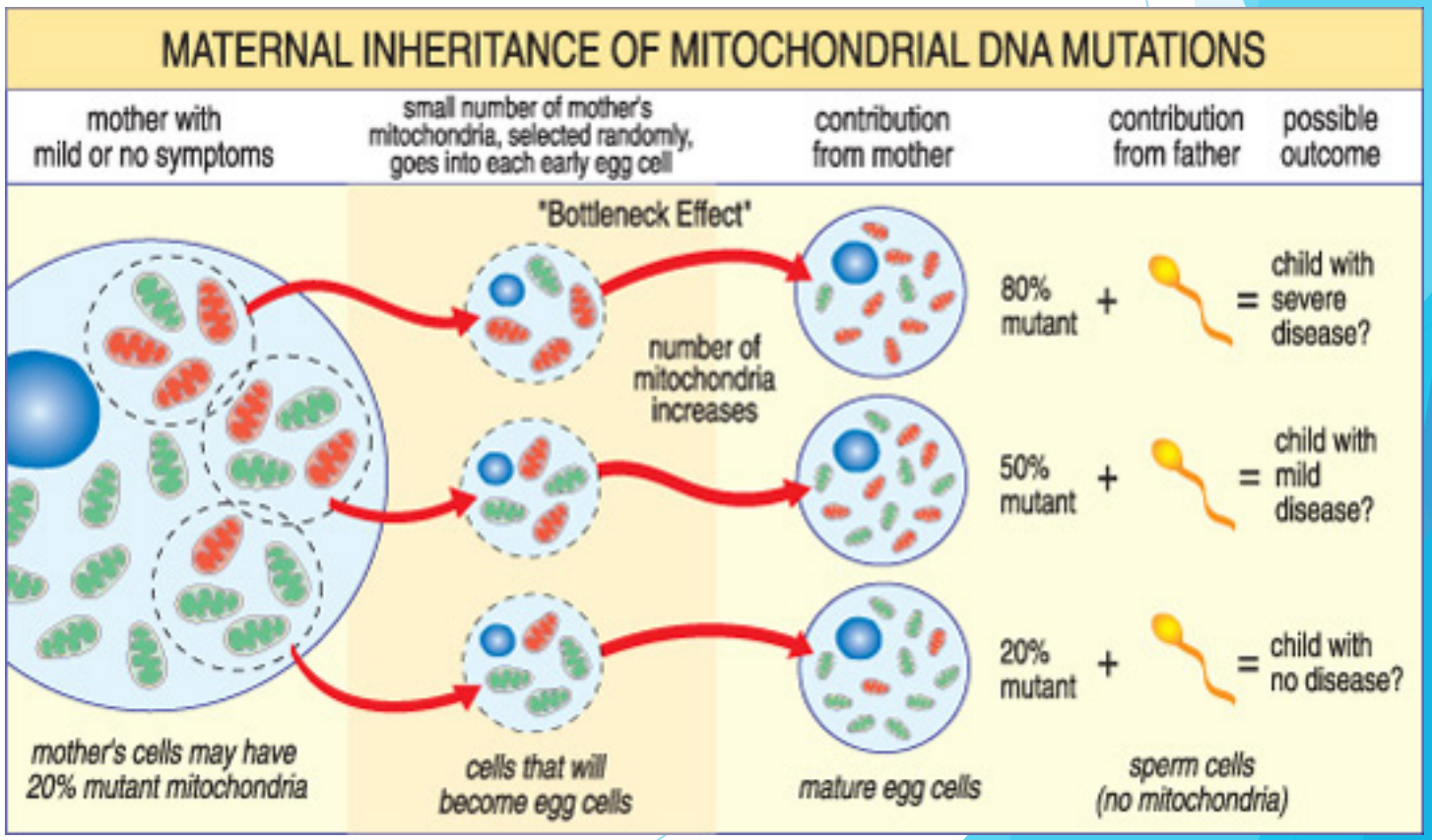
How is Genetics Important for your Health? (Lecture 2)

- **SEX LIMITED TRAITS**
  - **Autosomal trait**
  - **Trait that is expressed in one of the sexes**
  - **Examples:**
    - **Beard growth**
    - **Milk production**
    - **Plumage patterns in male and female birds**

How is Genetics Important for your Health? (Lecture 2)

- Mitochondrial Inheritance
  - Autosomal Recessive Inheritance
  - Nuclear DNA in mitochondria inherited from both parents

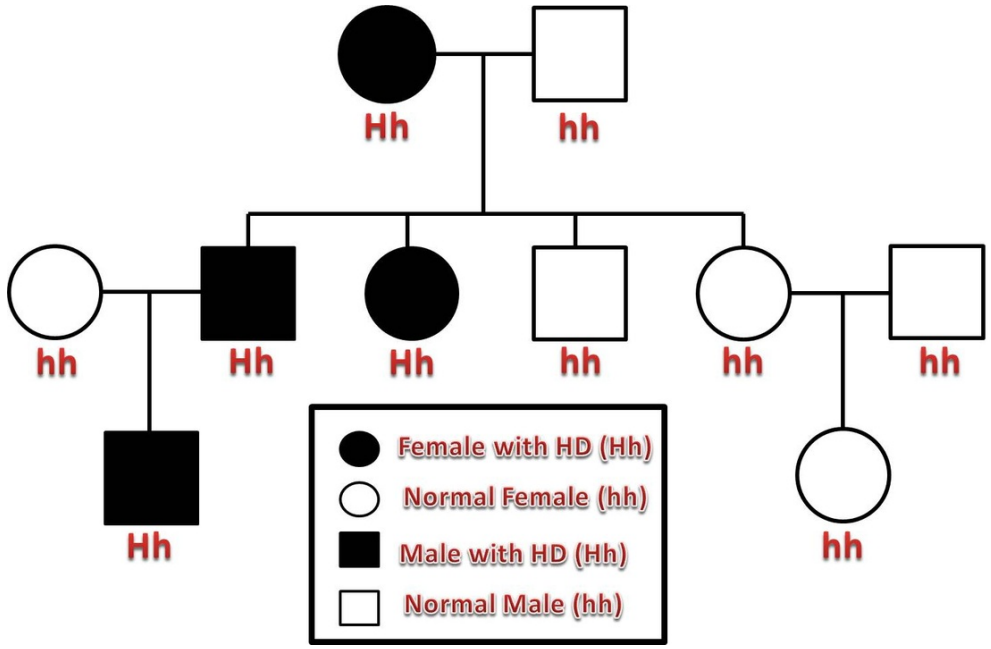
- Passed on if BOTH mother and father are carriers



How is Genetics Important for your Health? (Lecture 2)

HUNTINGTON DISEASE - Video **(2:10 mins)**
https://www.youtube.com/watch?v=JL9Y3P870jU

PEDIGREE:

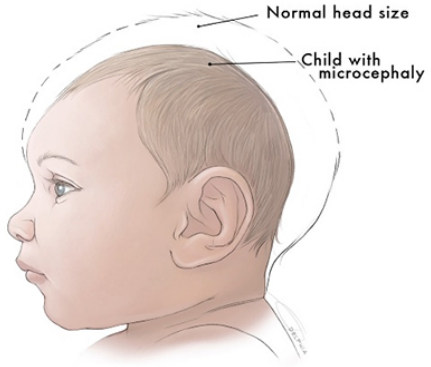


Huntington Disease (Continued)

- Results from a dominant mutation
- All heterozygotes develop the neurological disease (50% Probability Risk)
- Affects patients in their middle age
- Offspring have 50% probability of inheriting disease allele
- Block of huntingtin cleavage by caspase IX will treat Huntington disease (cleaved peptides are highly neurotoxic)

Inheritance of Zika Virus- Present day disease concerns

- Heritability of Zika virus not fully understood yet
- Known to cause disruptions in fetal development
- Instances of Microcephaly reported
- Zika Virus: Risk higher than first thought (Video from May 2nd, 2016) http://www.bbc.com/news/world-latin-america-36184799



How is Genetics Important for your Health? (Lecture 2)

- **Importance of Family Medical History Records**
  - **Powerful screening tool**
  - **Allows for faster diagnosis of genetic diseases**
  - **Should be updated each visit**
  - **Family History for Prenatal Providers**
    - **Address family history**
    - **Improved health for female patient, fetus and family**
  - **Helps investigate genetic predisposing factors that are associated with the health of the individual**

- **Importance of Family Medical History Records**
  - **Should include at least three generations**
  - **Questions include:**
    1. **General information (names and birthdates)**
    2. **Family's origin or racial/ethnic background**
    3. **Health Status**
    4. **Age at death and cause of death of each member**
    5. **Pregnancy outcomes of the patient and relatives**
  - **Formation of Pedigrees to analyze possible risk factors (certain characteristics of the individual that ↑ likelihood of disease)**

How is Risk Calculated?

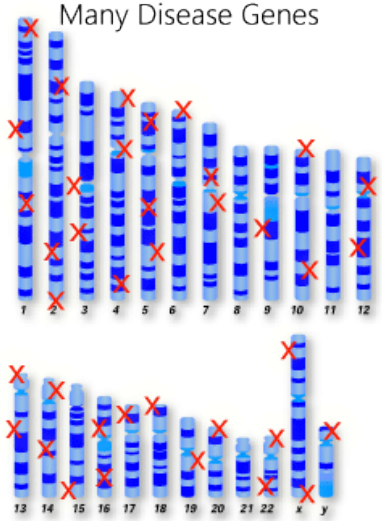
Simple Disease

Risk is easy to calculate for rare disorders caused by a single gene.

Single Disease Gene

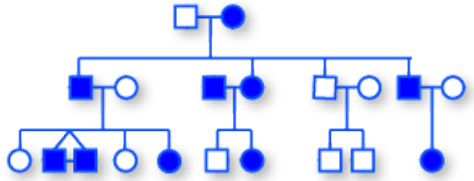
Complex Disease

Many Disease Genes



But for complex diseases that are influenced by multiple genes, risk is much more difficult to calculate.

Complex Disease Family Pedigree



Risk must be estimated based on observation of data collected from large families affected by these diseases.

How is Genetics Important for your Health? (Lecture 2)

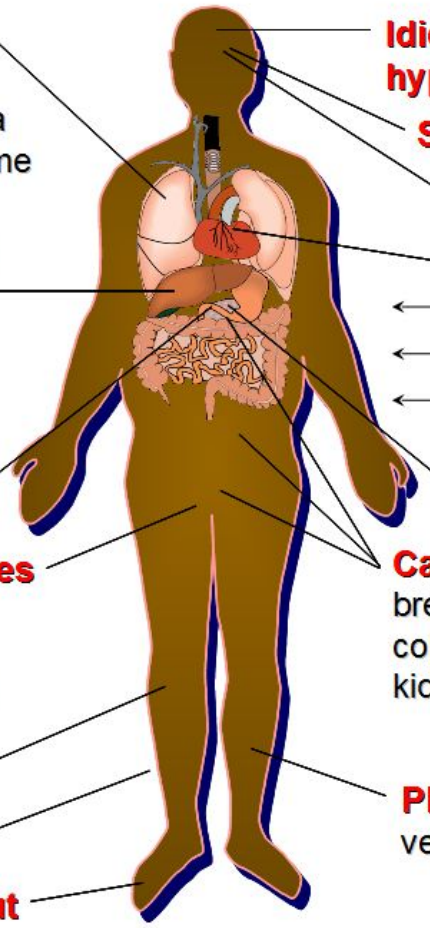
- **Record-keeping strategies are becoming more advanced**
  - **23andMe project reports genetic health, traits and ancestry for individuals for less that $300 CAD**

  - **The Genographic project by National Geographic is tracing human history**
    - VIDEO
    - https://www.youtube.com/watch?v=MdTCj9tC1Pw

How is Genetics Important for your Health? (Lecture 2)

- Diseases are not only caused by genetics
  - DIET can alter predisposition to impair health
    - Provides nutrients that the body requires
    - Lack of well-nourishing diet leads to increased risk from chronic disease
  - EXERCISE
    - Works synergistically with poor diet
    - Sedentary lifestyle in combination with diet and high stress are the main causes for obesity and type 2 diabetes in North America

Medical Complications of Obesity



Pulmonary disease
abnormal function
obstructive sleep apnea
hypoventilation syndrome

Nonalcoholic fatty liver disease
steatosis
steatohepatitis
cirrhosis

Gall bladder disease

Gynecologic abnormalities
abnormal menses
infertility
polycystic ovarian syndrome

Osteoarthritis

Skin

Gout

Idiopathic intracranial hypertension

Stroke

Cataracts

Coronary heart disease

Diabetes

Dyslipidemia

Hypertension

Severe pancreatitis

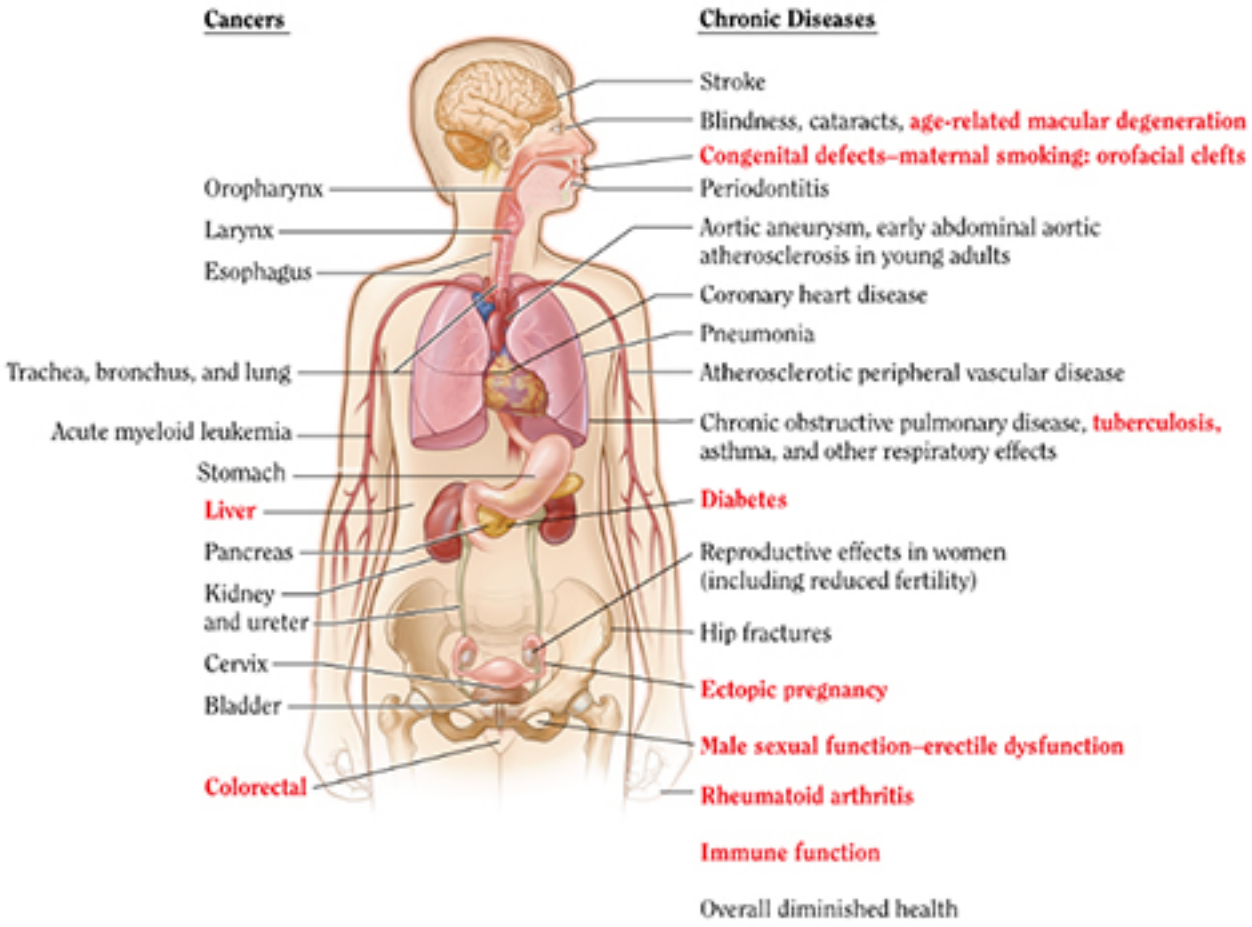
Cancer
breast, uterus, cervix
colon, esophagus, pancreas
kidney, prostate

Phlebitis
venous stasis

How is Genetics Important for your Health? (Lecture 2)

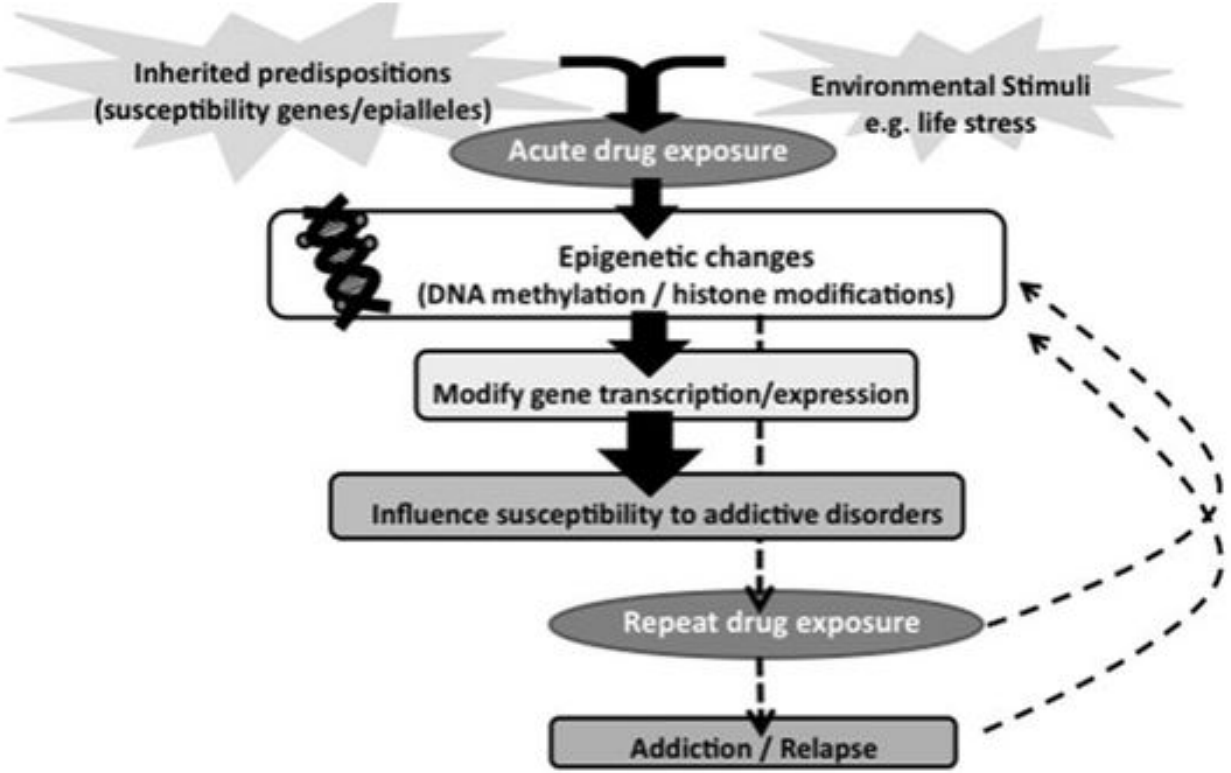
- **LIFESTYLE CHOICES**
  - **Proper diet and exercise are crucial for good health, but other factors also influence health**
  - **Introducing substances into your system can drastically impact health**
    - **Smoking ↑ risk of many fatal diseases**
    - **LACK OF SLEEP can lead to obesity, diabetes, heart disease and hypertension**

Impact of Smoking on Health



How is Genetics Important for your Health? (Lecture 2)

- **ADDICTION INHERITANCE**
  - Are Drug Addicts Born Susceptible? -Video (3 mins)
  - https://www.youtube.com/watch?v=dvnJhtw15HA

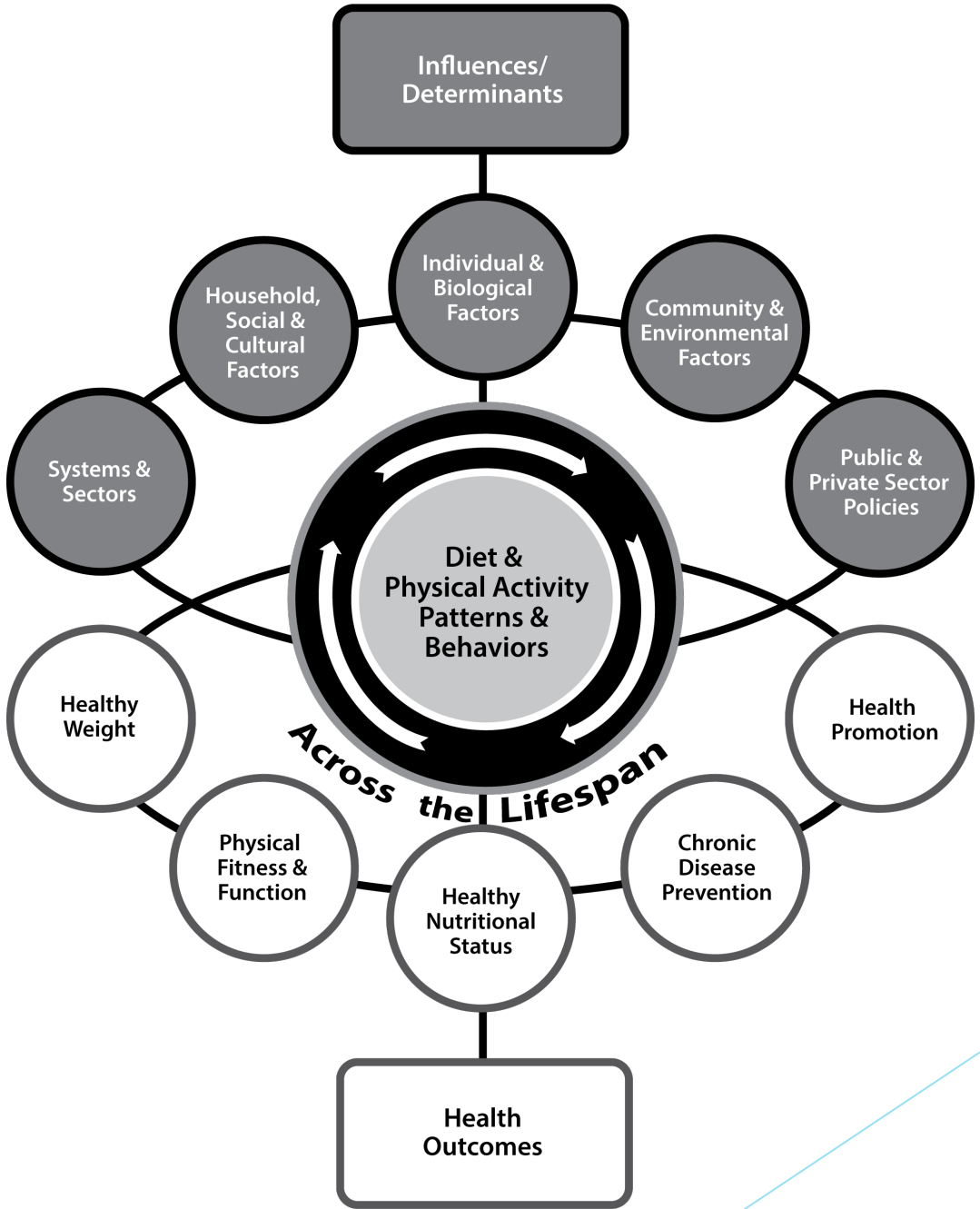


How is Genetics Important for your Health? (Lecture 2)

- **A healthy life involves all factors discussed (exercise, diet and lifestyle choices)**

- **Also requires a balance of nourishing aspects: physical, social, emotional, mental and spiritual engagements.**

Diet and Physical Activity, Health Promotion and Disease Prevention at Individual and Population Levels across the Lifespan



- SUMMARY
  - Mutations can be beneficial for an organism, but can also cause changes that lead to abnormalities and disease
  - Family history and formation of pedigrees allows for an analysis of possible risk factors
  - Genetic, as well as environmental factors contribute to the alteration of genes and overall health
  - Different forms of inheritance results in disease phenotype but sometimes traits are silenced in some generations (reiterates the importance of family history-taking)

Upcoming Let's Talk Science MEDS Seminar

LIVE AND LET DIE:
An explanation of physician-assisted suicide in Canada

Let's talk science with med students at UVic

- What does the Supreme Court of Canada 2015 ruling say?
- What does the current proposed legislation say?
- Which other countries have a physician assisted suicide law?

Join first-year Island Medical Program students **Sergiy Shatenko, Samuel Harder** and **Andrew Watters** to learn the answers to these questions.

Tuesday, May 17 @ 7:00 pm
Medical Sciences Building Room 150

Refreshments available | This is the latest in a series of medical student presentations
More information: Dr. Jane Gair - jgair@uvic.ca

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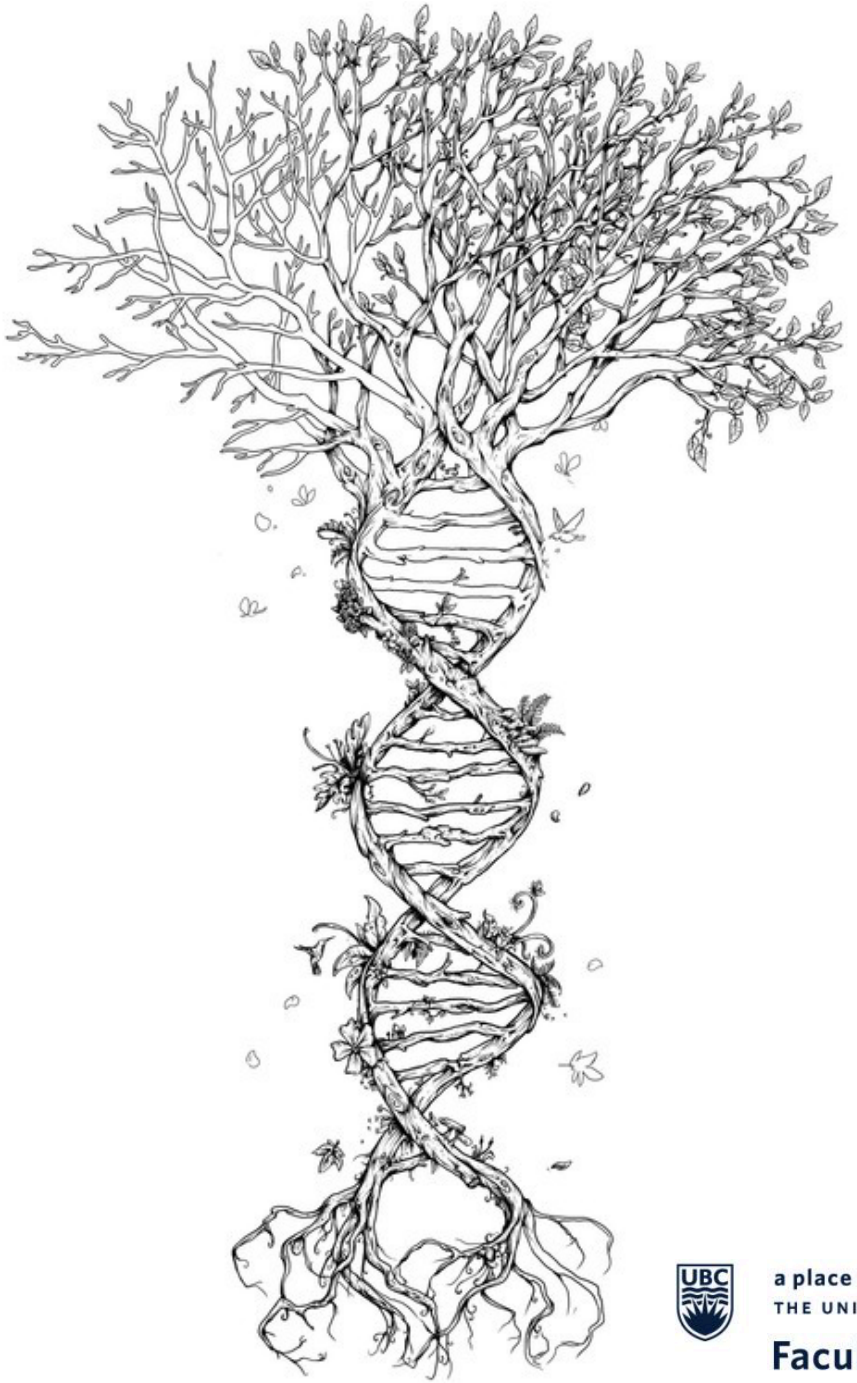
QUESTIONS?



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