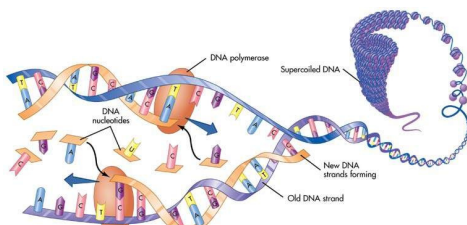


## Altered Cellular Tissue

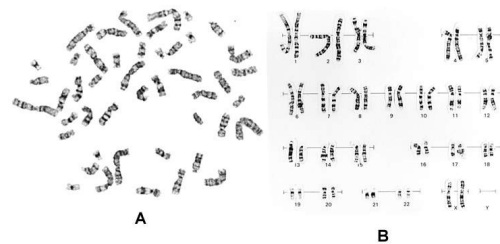
## Heredity

- DNA
  - Chromatin
  - Chromosome
  - Karyotype
- Protein Synthesis
  - Transcription
  - Translation

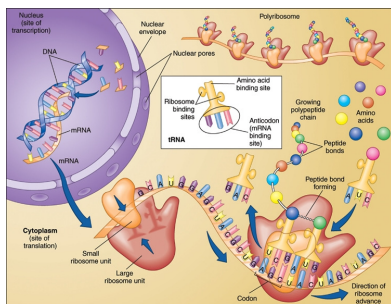
## DNA Replication



## Karyotype/Chromosomes



## Protein Synthesis



## Genetics

- GENES MAKE PROTEINS!!!!!!!
- Gene
  - Autosomal
  - X-linked
  - Y-linked
- Inheritance
  - Mendelian
    - Chromosomal Abnormalities
    - Polygenic
    - Multifactorial
  - Non-Mendelian

## Mendelian Genetic Disorders

- Autosomal Dominant:
  - Presence of the protein causes disease
- Autosomal Recessive
  - Lack of the protein causes disease
- Sex linked
  - X-linked
  - Y-Linked

## Autosomal Dominant: Huntington's Disease

		Affected parent	
		D	d
Affected parent	D	DD Homozygous affected (usually rare)	Dd Heterozygous affected
	d	Dd Heterozygous affected	dd Homozygous normal

		Normal parent	
		d	d
Affected parent	D	Dd Heterozygous affected	Dd Heterozygous affected
	d	dd Homozygous normal	dd Homozygous normal

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## Autosomal Recessive: Cystic Fibrosis, Sickle Cell Anemia, PKU

	D	d
D	DD Homozygous normal	Dd Heterozygous carrier
d	Dd Heterozygous carrier	dd Homozygous affected

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## X-linked Recessive: Hemophilia, Color Blindness

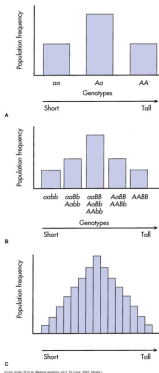
		Mother	
		X <sup>H</sup>	X <sup>h</sup>
Father	X <sup>H</sup>	X <sup>H</sup> X <sup>H</sup>	X <sup>H</sup> X <sup>h</sup>
	Y	X <sup>H</sup> Y	X <sup>h</sup> Y

		Mother	
		X <sup>H</sup>	X <sup>h</sup>
Father	X <sup>h</sup>	X <sup>H</sup> X <sup>h</sup>	X <sup>h</sup> X <sup>h</sup>
	Y	X <sup>H</sup> Y	X <sup>h</sup> Y

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- Polygenic Inheritance
  - Each allele produces larger amounts of the protein



## Multi-Factorial: CAD

- Genetic
  - LDL receptors
  - Liver Function
  - Clotting
  - Inflammation Disposition
  - Blood Pressure
- Environmental
  - Age
  - Smoking
  - Exercise
  - Diet
  - Blood Pressure

## Inheritance

- Genotype vs phenotype
- Penetrance
- Variable expressivity
- Lethal allele

## Environmental Congenital Disorders

- Teratogens
  - Alcohol
  - Cigarettes
  - Medications
  - Radiation
- Infections
  - Toxoplasmosis
  - Other: hepatitis B, syphilis, herpes zoster
  - Rubella
  - Cytomegalovirus (CMV)
  - Herpes simplex virus

## Screening and Treatment

- Genetic Counseling
- Fetal Diagnostics
- Newborn screening
  - FL: PKU, hypothyroidism, galactosemia, hemoglobinopathies
- Management of Phenotype
- Dietary Modification
- Replacement
- Surveillance
- Surgery
- Gene therapy

## Worldview Issues

- Health/Disease
- Genetic diseases