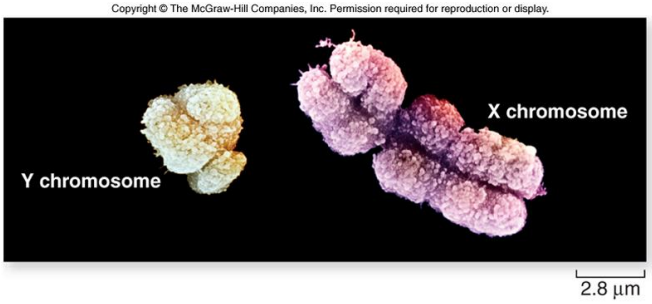
Chromosomes, Mapping, and the Meiosis-Inheritance Connection

Chapter 13



Chromosome Theory

Chromosomal theory of inheritance

- developed in 1902 by Walter Sutton
- proposed genes present on chromosomes
- based on observations that homologous chromosomes pair with each other during meiosis
- supporting evidence with work on fruit flies

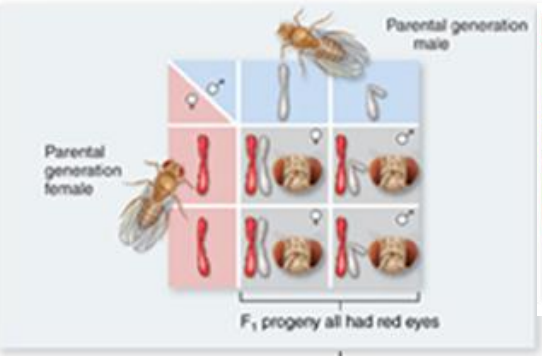
Chromosome Theory

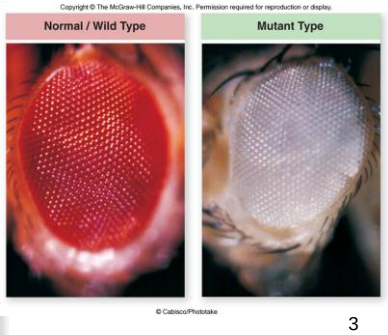
T.H. Morgan: mutant white-eyed *Drosophila*

- red-eyed female x white-eyed male →

  F_1 generation of all red eyes

- Morgan concluded red eyes dominant





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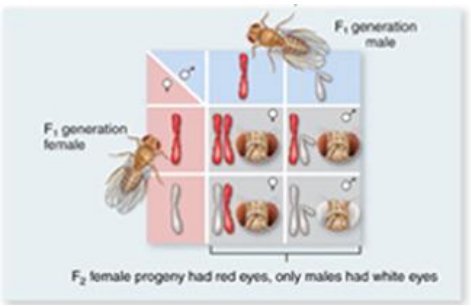
Chromosome Theory

Morgan crossed F_1 females X F_1 males

-F_2 generation contained red and white- eyed flies but *all white-eyed flies male*



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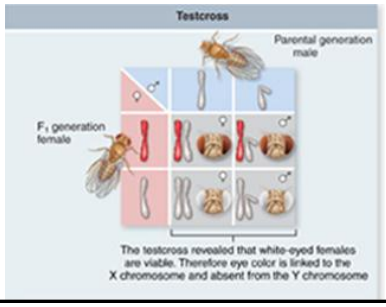
Chromosome Theory

-Morgan testcross of F_1 female with white-eyed male showed viability of white-eyed females

-eye color gene linked to X chromosome



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Sex Chromosomes

Sex determination in *Drosophila* based on # of X chromosomes

2 X chromosomes = female

1 X & 1 Y chromosome = male

Sex determination in humans based on presence of Y chromosome

2 X chromosomes = female

having a Y chromosome (XY) = male

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Sex Chromosomes

In many organisms, Y chromosome greatly reduced or inactive

genes on X chromosome present in only 1 copy in males

sex-linked traits: controlled by genes present on the X chromosome

Sex-linked traits show inheritance patterns different than those of genes on **autosomes**

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TABLE 13.1 Sex Determination in Some Organisms

	Female	Male
Humans, *Drosophila* 	XX	XY
Birds 	ZW	ZZ
Grasshoppers 	XX	XO
Honeybees 	Diploid	Haploid

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Sex Chromosomes

Dosage compensation ensures an equal expression of genes from sex chromosomes even though females have 2 X chromosomes & males have only 1 X

In each female cell, 1 X chromosome inactivated & highly condensed into a **Barr body**

Females heterozygous for genes on X chromosome → **genetic mosaics**

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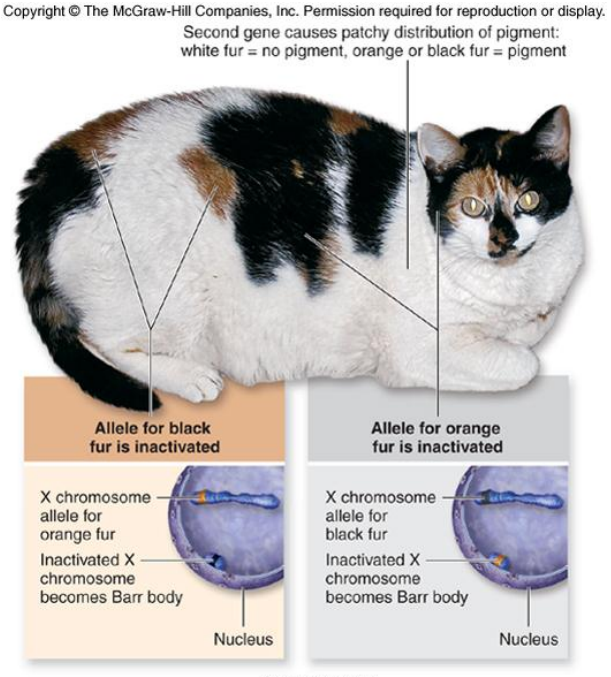

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Chromosome Theory Exceptions

Mitochondria & chloroplasts contain genes

-traits controlled by these genes do not follow chromosomal theory of inheritance

-genes from mitochondria & chloroplasts often passed to offspring by only one parent

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Chromosome Theory Exceptions

Maternal inheritance: uniparental (one-parent) inheritance from mother

mitochondria in zygote from egg cell; none comes from sperm during fertilization

in plants, chloroplasts often inherited from mother, although is species dependent

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Genetic Mapping

Early geneticists realized they could obtain information about distance between genes on a chromosome → **genetic mapping**

Type of mapping based on genetic recombination (**crossing over**) between genes

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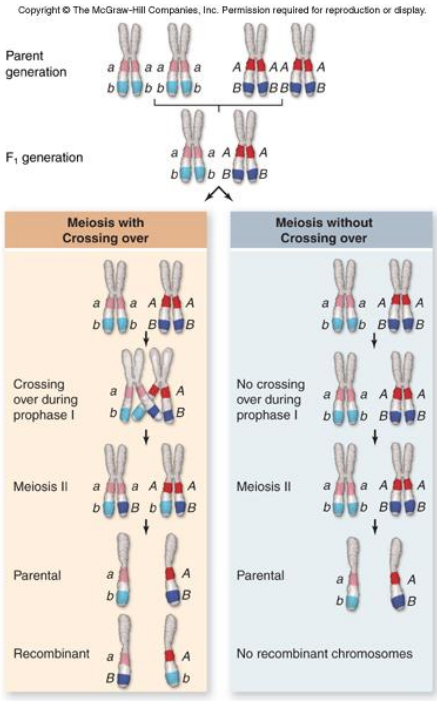

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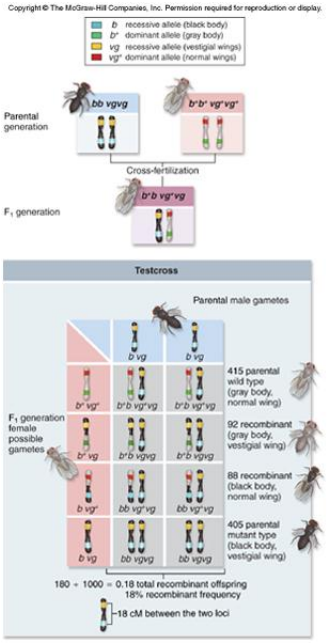

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Human X-Chromosome Gene Map

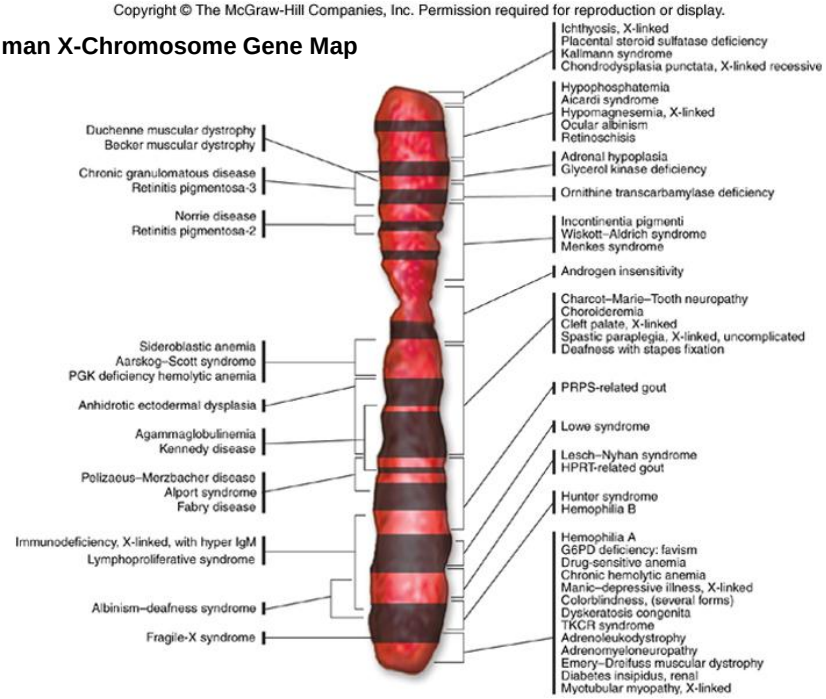

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TABLE 13.2 Some Important Genetic Disorders

Disorder	Symptom	Defect	Dominant/ Recessive	Frequency Among Human Births
Cystic fibrosis	Mucus clogs lungs, liver, and pancreas	Failure of chloride ion transport mechanism	Recessive	1/2500 (Caucasians)
Sickle cell anemia	Blood circulation is poor	Abnormal hemoglobin molecules	Recessive	1/600 (African Americans)
Tay–Sachs disease	Central nervous system deteriorates in infancy	Defective enzyme (hexosaminidase A)	Recessive	1/3500 (Ashkenazi Jews)
Phenylketonuria	Brain fails to develop in infancy	Defective enzyme (phenylalanine hydroxylase)	Recessive	1/12,000
Hemophilia	Blood fails to clot	Defective blood-clotting factor VIII	X-linked recessive	1/10,000 (Caucasian males)
Huntington disease	Brain tissue gradually deteriorates in middle age	Production of an inhibitor of brain cell metabolism	Dominant	1/24,000
Muscular dystrophy (Duchenne)	Muscles waste away	Degradation of myelin coating of nerves stimulating muscles	X-linked recessive	1/3700 (males)
Hypercholesterolemia	Excessive cholesterol levels in blood lead to heart disease	Abnormal form of cholesterol cell surface receptor	Dominant	1/500

Color-blindness: sex-linked disorder males 1/76 or 10% (Americans)
X-linked recessive

Afflicted female X^cX^c Carrier female X^CX^c

Afflicted male X^cy

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Human Genetic Disorders

Some human genetic disorders caused by altered proteins

altered protein encoded by a mutated DNA sequence

altered protein does not function correctly, causing a change to phenotype

protein can be altered at only a single amino acid (e.g. sickle cell anemia)

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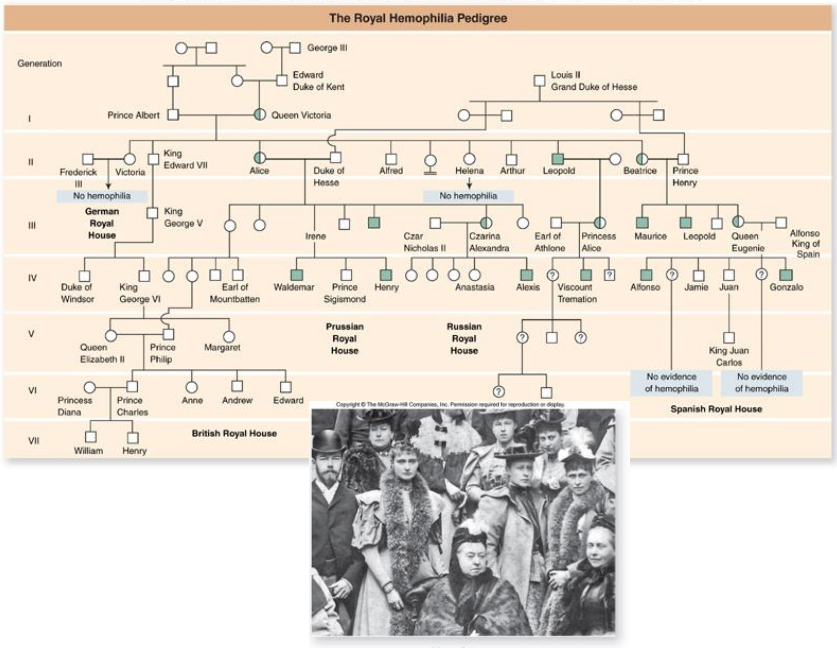
Hemophilia

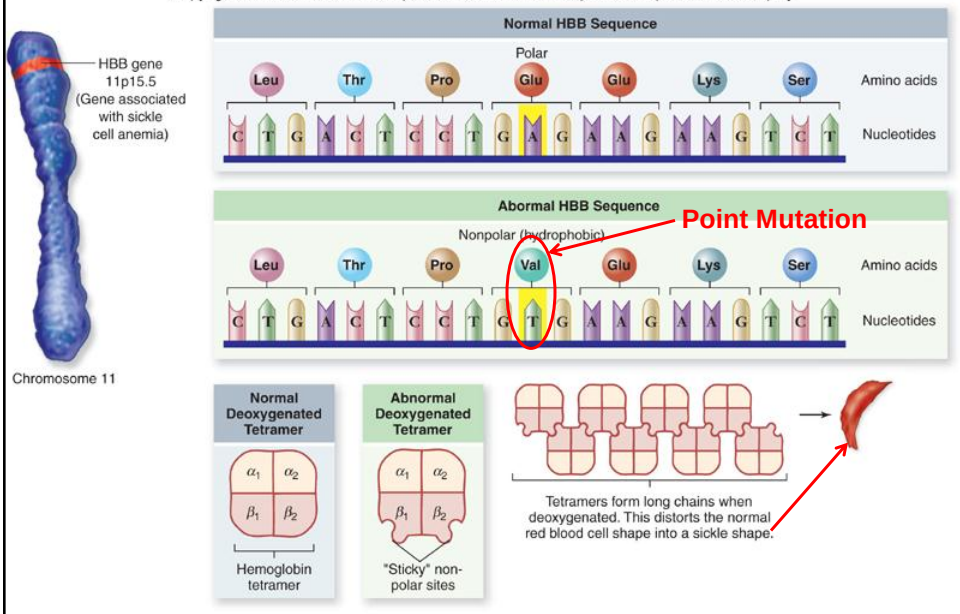


Mutations

Glitch in DNA code/codon causing different gene product (phenotype)

1) bad mutations
2) good mutations
3) neutral mutations

- **Point mutation**-single glitch
- **Nondisjunction**- unequal # of chromosomes
- **Chromosomal Aberration**
  - 1) deletion: piece of chromosome breaks off & lost
  - 2) inversion: breakage at the chromosomal ends followed by 180º rotational flip of chromosome
  - 3) translocation: piece of chromosome breaks off & attaches to a different chromosome



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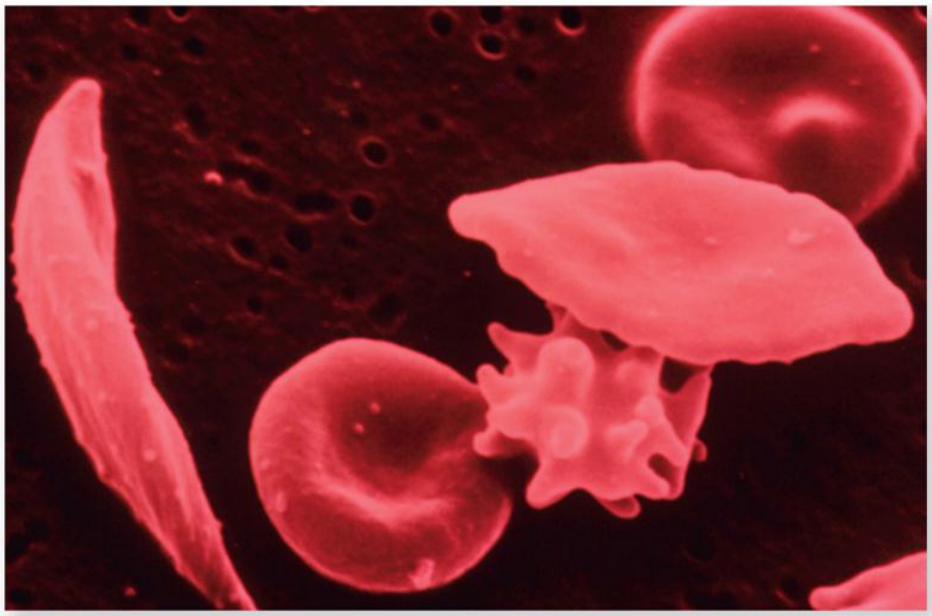

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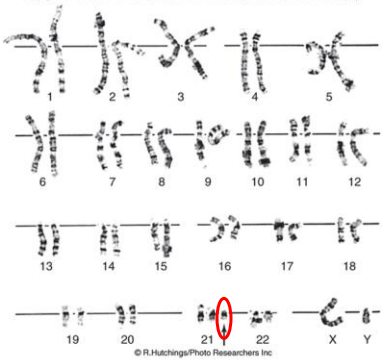
Human Genetic Disorders

nondisjunction meiosis can create gametes with one too many or one too few chromosomes

fertilization of these gametes creates **trisomic** or **monosomic** individuals

Down syndrome: trisomy of chromosome 21



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Human Genetic Disorders

Nondisjunction of sex chromosomes can result in:

XXX triple-X females

XXY males (Klinefelter syndrome)

XO females (Turner syndrome)

OY nonviable zygotes

XYY males (Jacob syndrome)

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Triple X Females

Slightly lower intelligence/clumsiness/particular problems with verbal skills
Tallness, small/long head, prominent forehead/small feet & hands/reduced muscle tone
Sometimes menstrual irregularities/infertility

Klinefelter Syndrome Symptoms

Sparse pubic, facial, & body hair
Underdeveloped muscles/enlarged breasts (gynecomastia).
Taller than other males in family/long legs, narrow shoulders & wide hips

Turner Syndrome Symptoms

Young infants include:
Swollen hands & feet/wide & webbed neck
Older females:
Absent/incomplete development at puberty/sparse pubic hair & small breasts
Broad, flat chest shaped like a shield/short height
Drooping eyelids/dry eyes
Infertility/vaginal dryness/no periods (absent menstruation)

Jacobs Syndrome

↑testosterone, acne, tall, behavioral problems (aggression), learning & speech disability

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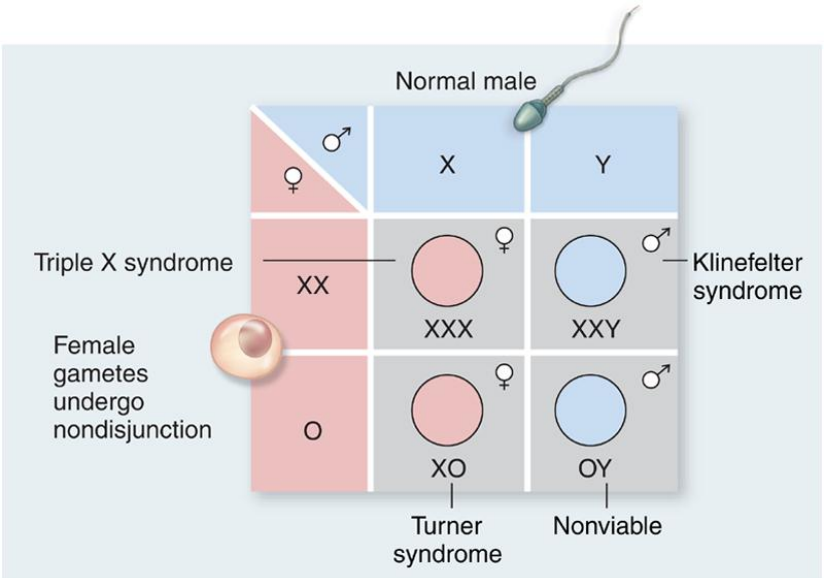

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Human Genetic Disorders

genomic imprinting occurs when phenotype exhibited by particular allele depends on which parent contributed allele to offspring

specific partial deletion of chromosome 15:

Prader-Willi syndrome: father chromosome

respiratory distress, mild mental retardation, obesity, short stature & obsessive-compulsive behavior

Angelman syndrome: mother chromosome

developmental delay, severe mental retardation, hyperactivity, aggressive behavior & inappropriate laughter

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Human Genetic Disorders

Genetic counseling: use pedigree analysis to determine probability of genetic disorders in offspring

Some genetic disorders can be diagnosed during pregnancy

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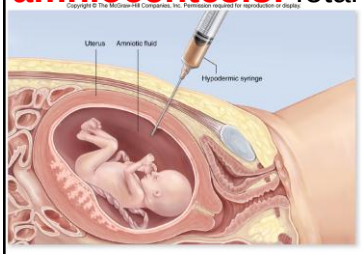
amniocentesis: fetal cells from amniotic fluid for examination



15th-20th week of pregnancy

Patau syndrome (trisomy 13)
cleft lip/palate, microcephaly, microphthalmia, rocker-bottom feet, scalp defects (cutis aplasia) hernias
Edwards syndrome (trisomy 18)
2nd most common after Down
heart abnormalities, kidney malformations & other internal organ disorders
Down syndrome (trisomy 21)
Fragile X syndrome
mental retardation/speech delays/heart problems
large ears/jaws/testes/prominent foreheads
Rare, inherited metabolic disorders
Neural tube defects
anencephaly & spina bifida

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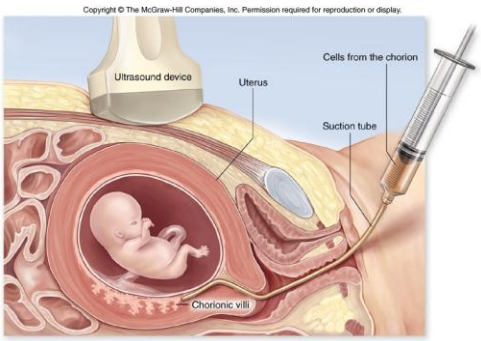
chorionic villi sampling (CVS): placental cells examination



10-12 weeks of pregnancy

1) Tay-Sachs disease
chromosome #15
neurological disease
⬇ hexosaminidase A breaks down ganglioside GM2
~1/27 of Ashkenazi Jewish pop. carries Tay-Sachs gene
2) Hemophilia
3) Down syndrome

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