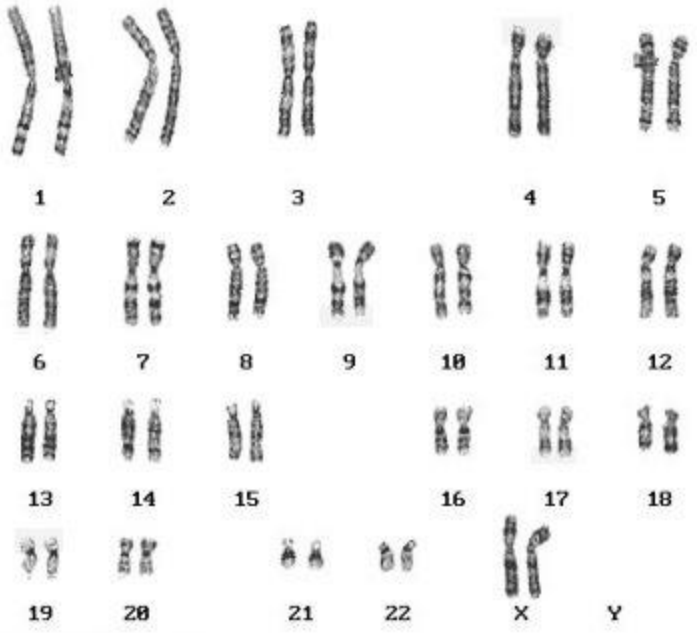
MISTAKES IN MEIOSIS: GENETIC DISORDERS

SBI 3C: NOVEMBER

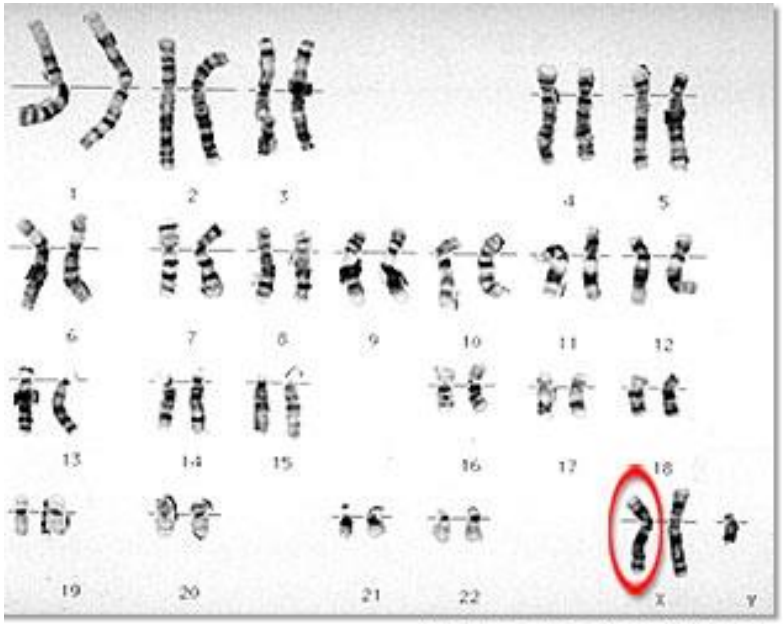
KARYOTYPES

- Test to examine chromosomes
- Can help identify genetic problems by:
  - Counting the number of chromosomes
  - Looking for structural changes in chromosomes
- The test can be performed on almost any tissue:
  - Amniotic fluid
  - Blood
  - Bone marrow
  - Tissue from the placenta



KLINEFELTER SYNDROME

- Not inherited
- Genetic Changes:
  - Boy with extra copy of X chromosome (XXY)
- Problem:
  - Nondisjunction causes polysomy
- Symptoms:
  - Low levels of testosterone
  - Breast development
  - Decreased body and facial hair
  - Abnormal sexual development
  - Learning disabilities



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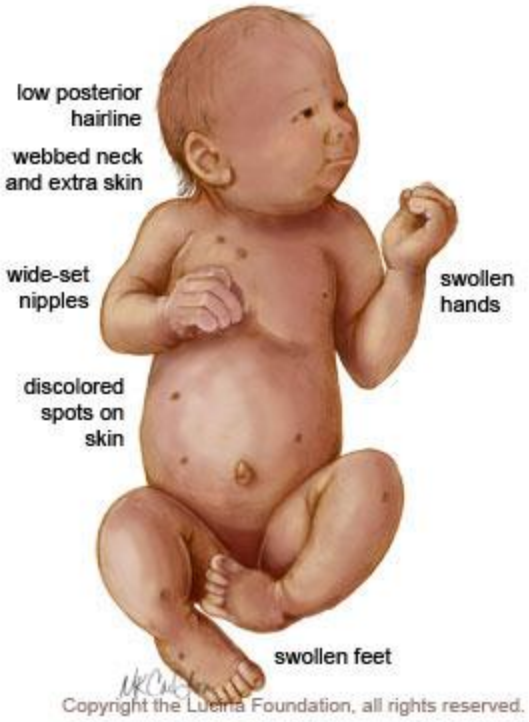
TURNER SYNDROME

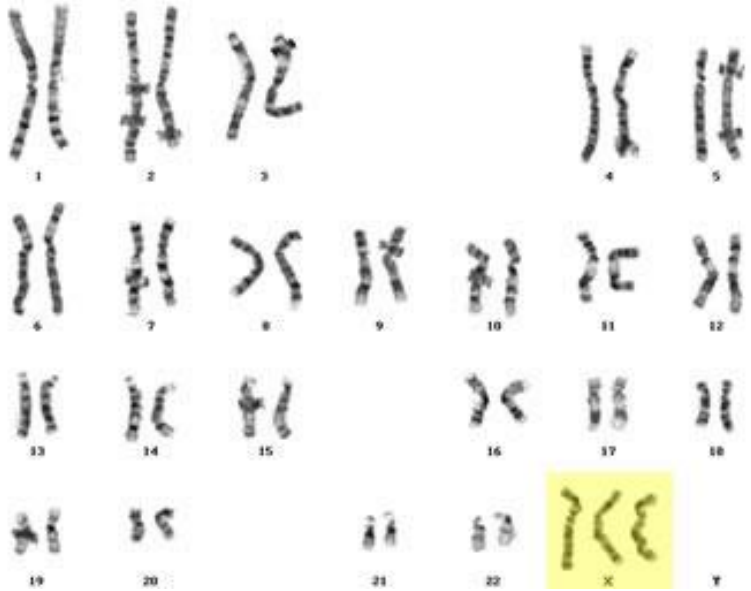
- Not inherited
- Genetic Changes:
  - Missing/damaged X chromosome (X0)
- Problem:
  - Monosomy caused by nondisjunction
  - Deletion or inversion
- Symptoms:
  - Shorter
  - Infertile
  - Webbed neck
  - Skeletal abnormalities
  - Heart defect
  - Kidney problems

Note - cannot have Y0, fetus will not survive



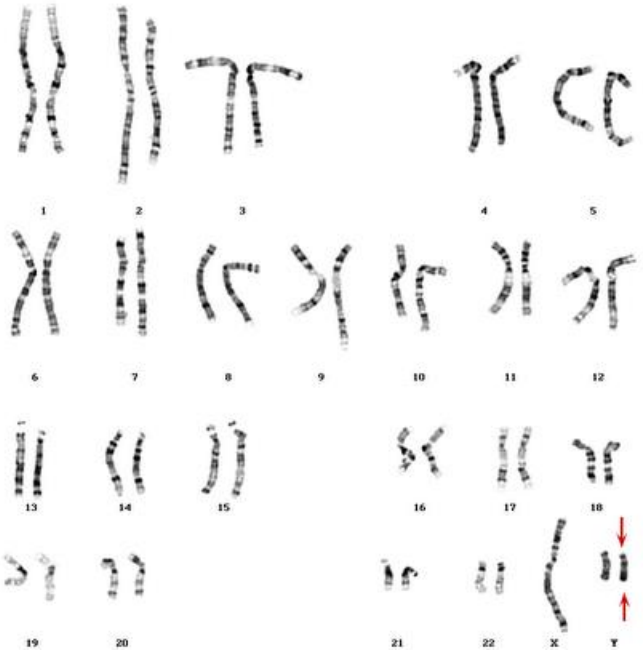
TRIPLE X

- Not inherited
- Genetic Changes:
  - Female with 3 X chromosomes (XXX)
- Problem:
  - Nondisjunction causes polysomy
- Symptoms:
  - Taller
  - Increased learning disabilities
  - Normal otherwise



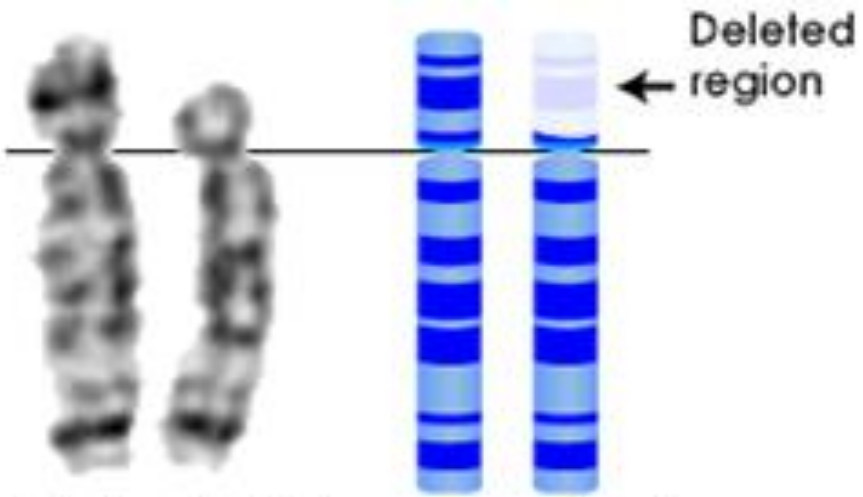
47 XYY

- Not inherited
- Genetic Changes:
  - Extra Y chromosome in males (XYY)
- Problem:
  - Nondisjunction causes polysomy
- Symptoms:
  - Taller
  - No physical problems
  - Some risk of learning disabilities



CRI-DU-CHAT

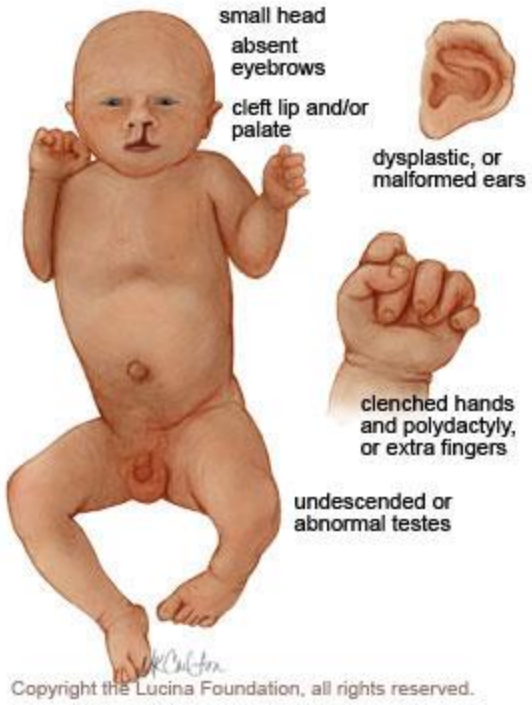
- 90% not inherited, 10% from unaffected parent
- Genetic Changes:
  - Piece of chromosome 5 is missing
- Problem:
  - Deletion
- Symptoms:
  - Cry like a cat
  - Mental retardation
  - Small head
  - Weak muscles



Cri-du-chat Chromosome 5 pair

PATAU SYNDROME

- Generally not inherited, sometimes can be
- Genetic Changes:
  - Additional DNA from 13 (usually extra copy)
- Problems:
  - Polysomy due to nondisjunction
  - Translocation (partial trisomy of 13)
- Symptoms:
  - Sever retardation
  - Small eyes
  - Cleft lip/palate
  - Don't often live past infancy



EDWARDS SYNDROME

- Can be inherited or not
- Genetic Changes:
  - Extra DNA from chromosome 18
- Problem:
  - Trisomy/polysomy of 18 due to nondisjunction
- Symptoms:
  - Low birth weight
  - Abnormal head, jaw, mouth
  - Retardation
  - Only 5-10% survive 1st year

WILLIAMS SYNDROME

- Can be rarely inherited but generally not inherited
- Genetic Changes:
  - Deletion of 6 genes on chromosome 7
- Problem:
  - Deletion
- Symptoms:
  - Mild to moderate retardation
  - Cardiovascular problems
  - distinct facial features