

Human Sex Chromosomal Aberrations

**Presented by
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Human Sex Chromosomal Aberrations

- 1. Klinefelter's Syndrome**
- 2. Turner's Syndrome**
- 3. Down's Syndrome**



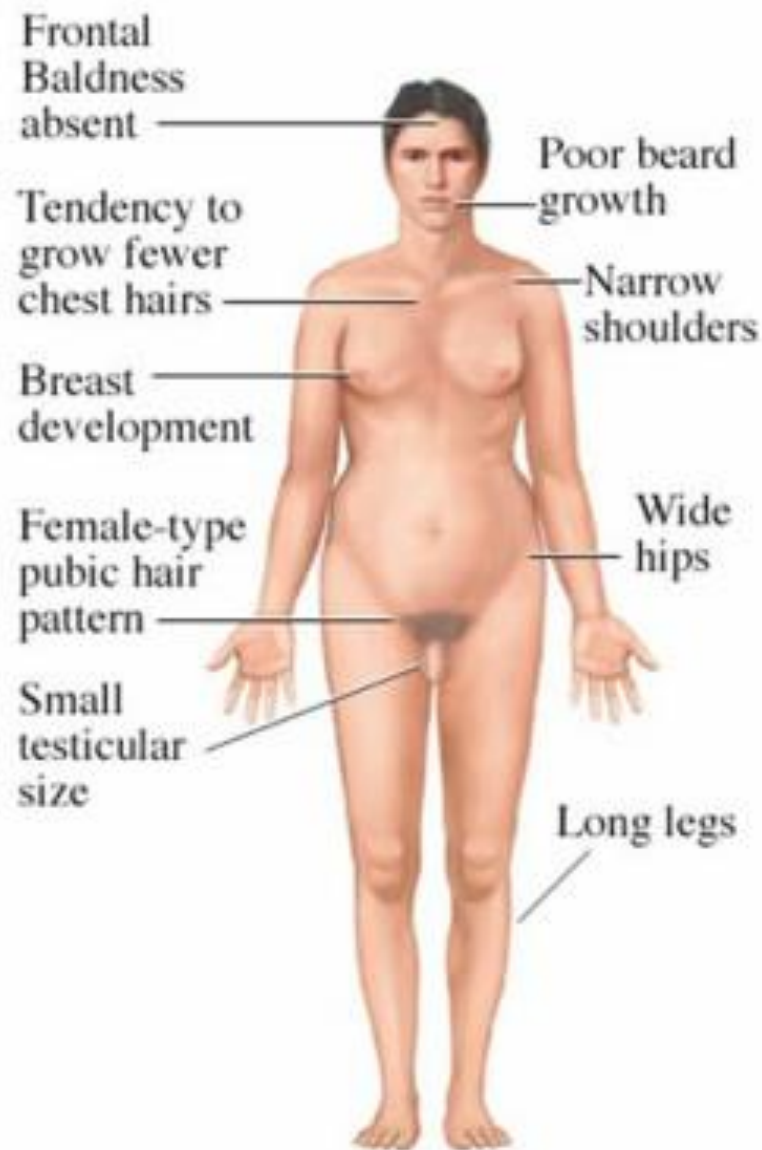
Klinefelter's Syndrome

- 1/1000 males
- Post-pubertal testicular failure
 - Small testes, azospermia
 - Infertility, variable hypogonadism
 - Social pathology
- At least 2 X's and one Y in at least some cells (usually 47, XXY)

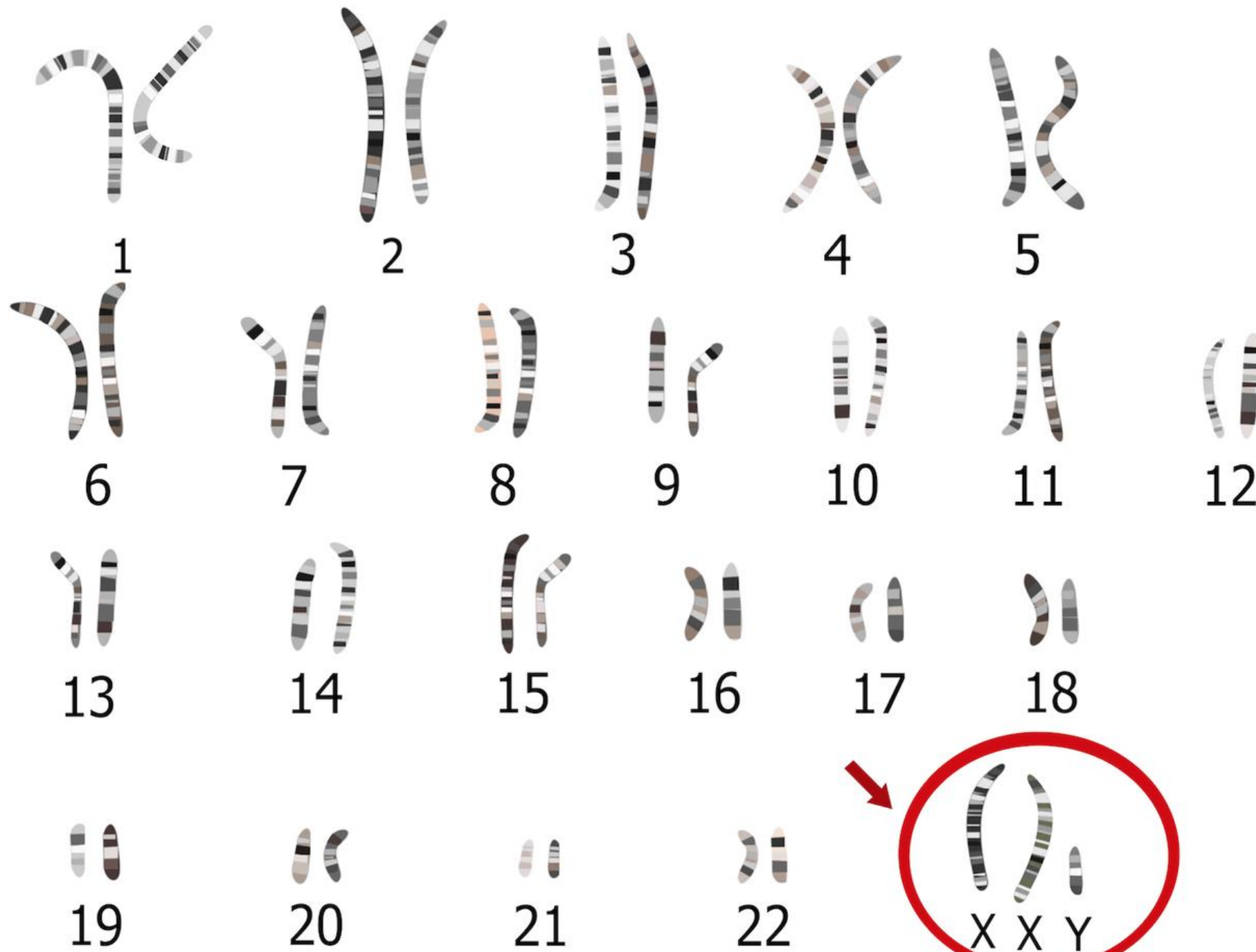


HARRY F. KLINEFELTER
1912-1990

Klinefelter syndrome

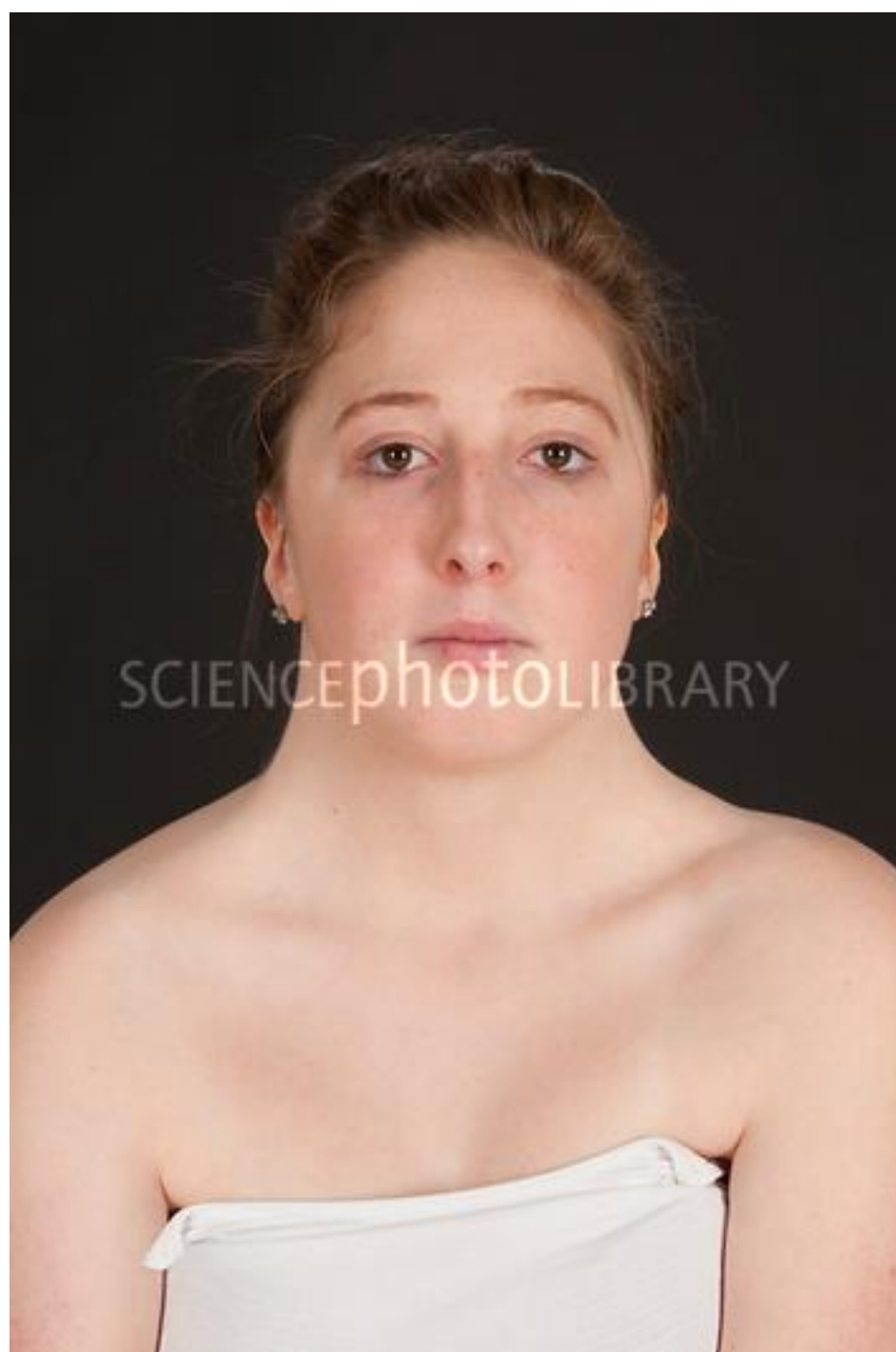


- **Lower IQ than sibs**
- **Tall stature**
- **Poor muscle tone**
- **Reduced secondary sexual characteristics**
- **Gynaecomastia (male breasts)**
- **Small testes/infertility**



2. Turner Syndrome

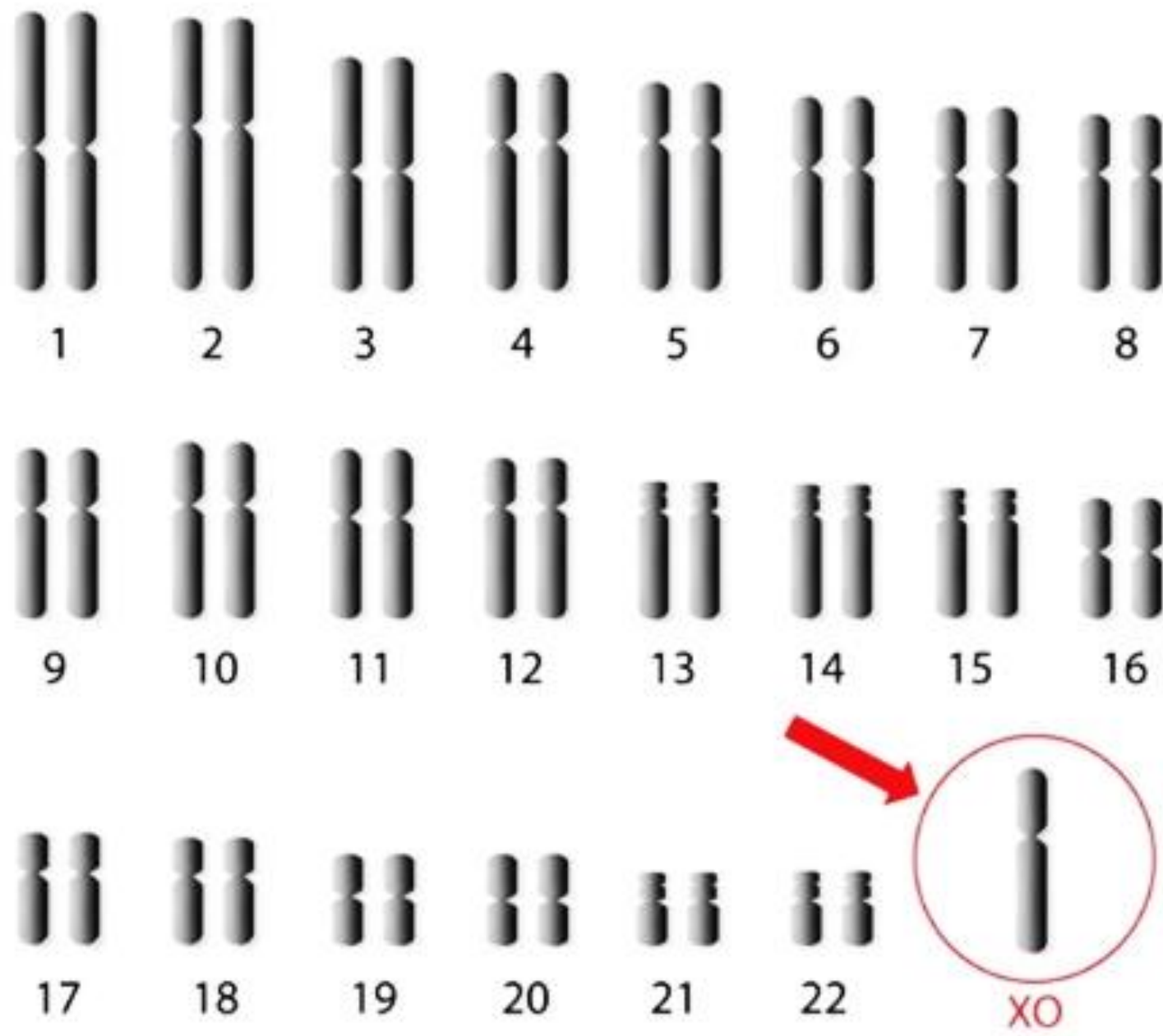
- **1/5,000 females**
- **Gonadal dysgenesis, disappearance of eggs**
- **Sexual immaturity**
 - **Amenorrhea**
 - **Infertility**
- **Short stature**
- **Somatic abnormalities**
- **Something wrong with the second X in some cells**



Turner syndrome



Turner's Syndrome



Why Turner Syndrome

- **Males don't have Turner syndrome**
- **Hypothesis: X-linked genes that**
 - **1. escape inactivation**
 - **2. have Y-chromosome homologs**

3. Down Syndrome

- Growth retardation
- Mental retardation—variable
- Somatic abnormalities
 - Abnormal facies (flattened face and occiput, large tongue, small ears, epicanthal folds)
 - Congenital heart disease (40%)—AV canal
 - GI abnormalities (5%)—duodenal stenosis
 - Leukemia (10 to 20 x increased risk)
 - Ear infections
 - Thyroid problems
 - Premature aging (cataracts, Alzheimer Disease)

Down syndrome



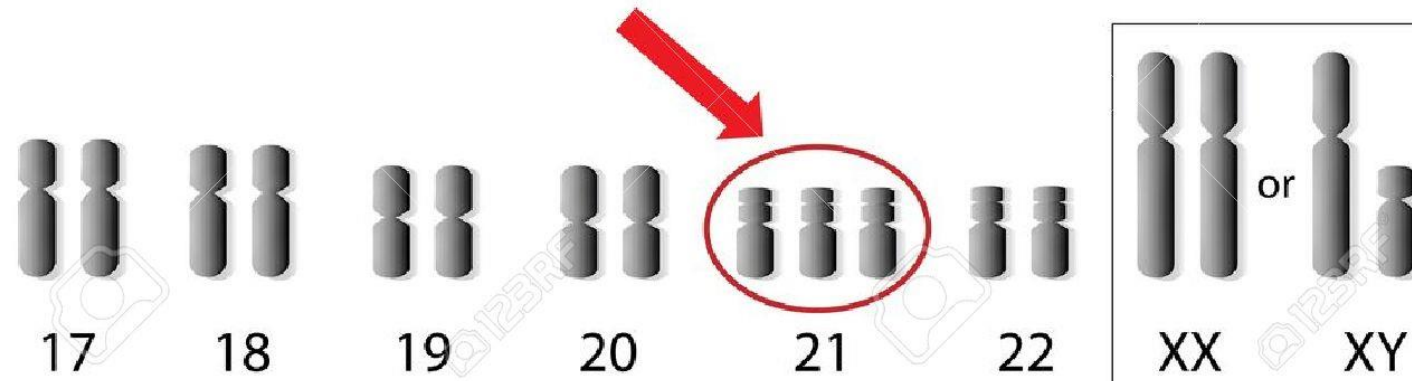
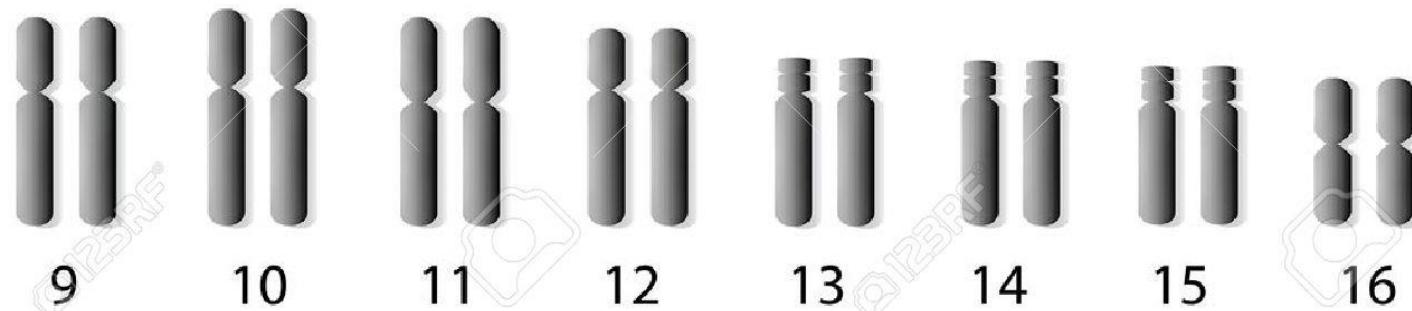
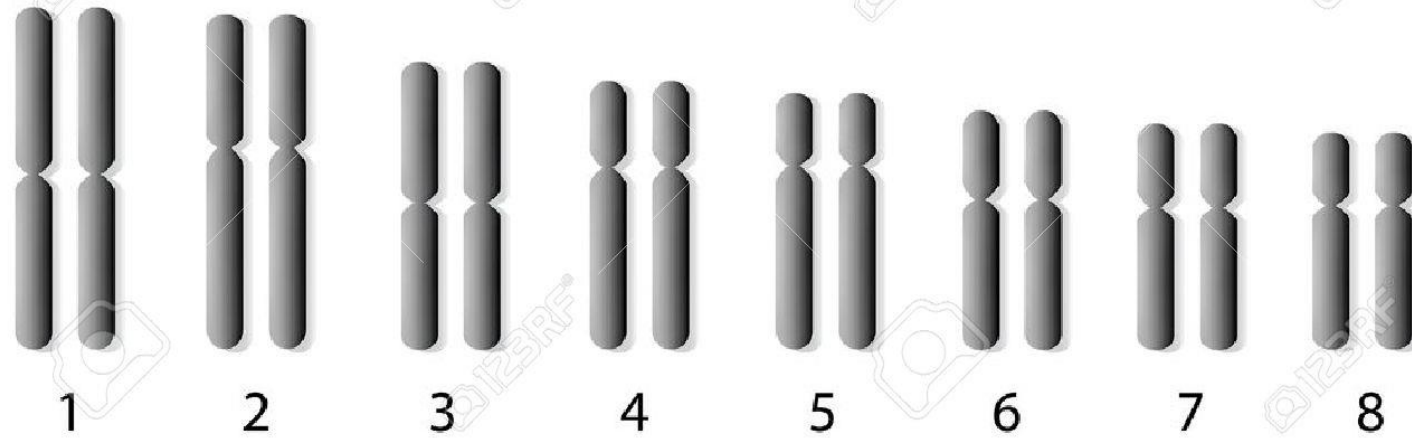
Down syndrome - Cytogenetics

Trisomy 21

95%

- Maternal nondisjunction in 95%
- 80% of these are MI (maternal age effect)

Down Syndrome - Trisomy 21



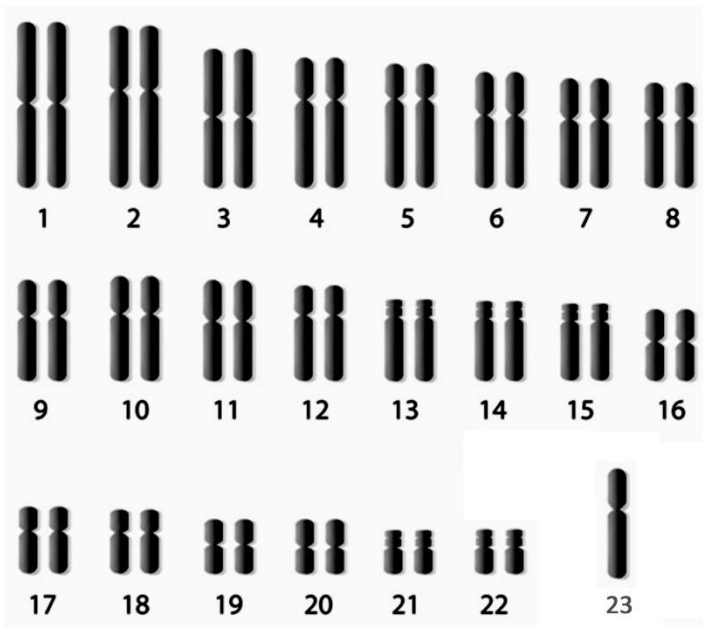
Assessment Test



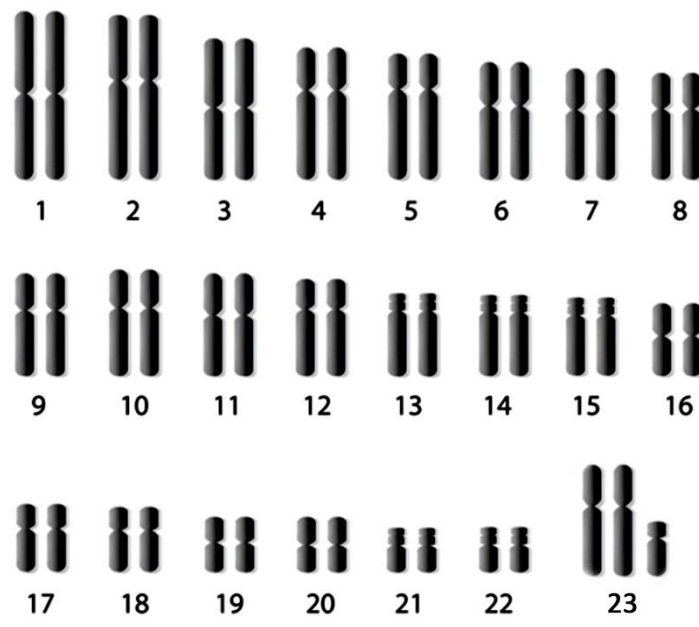
**Match the correct Phenotype
with its Genotype**



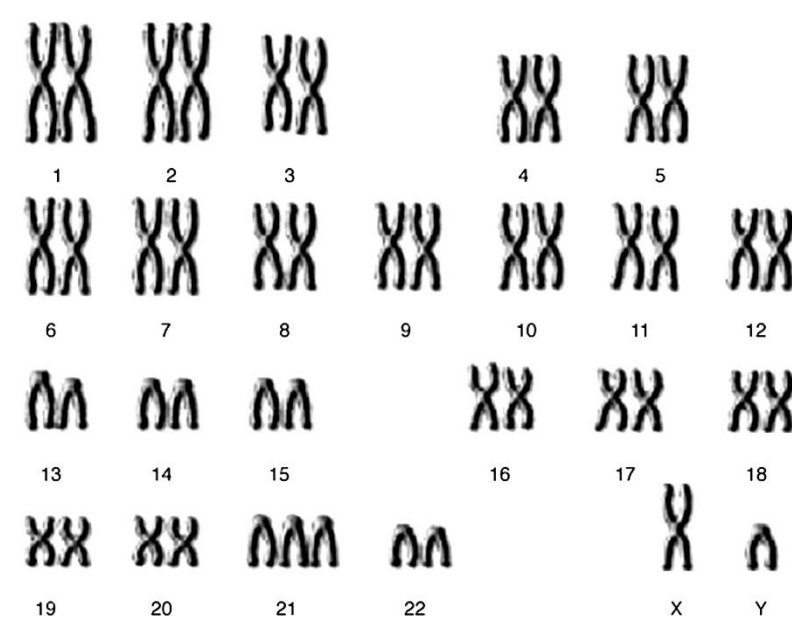
A

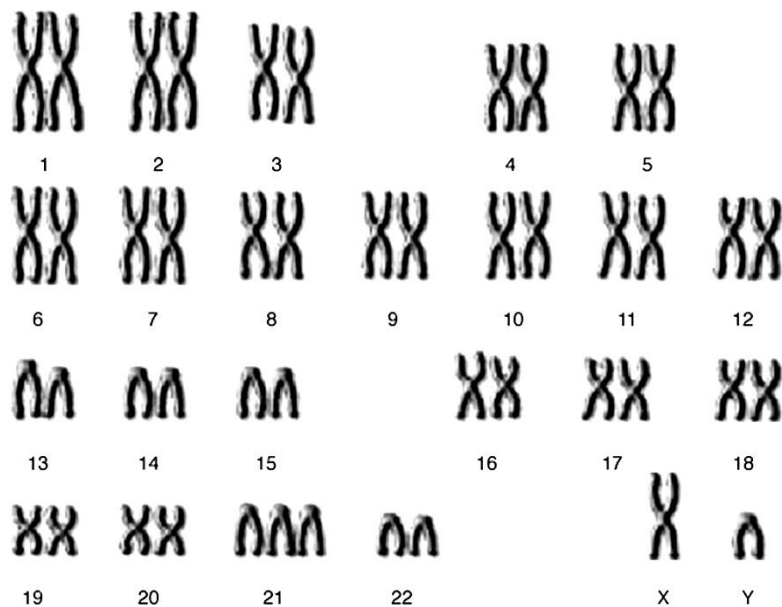
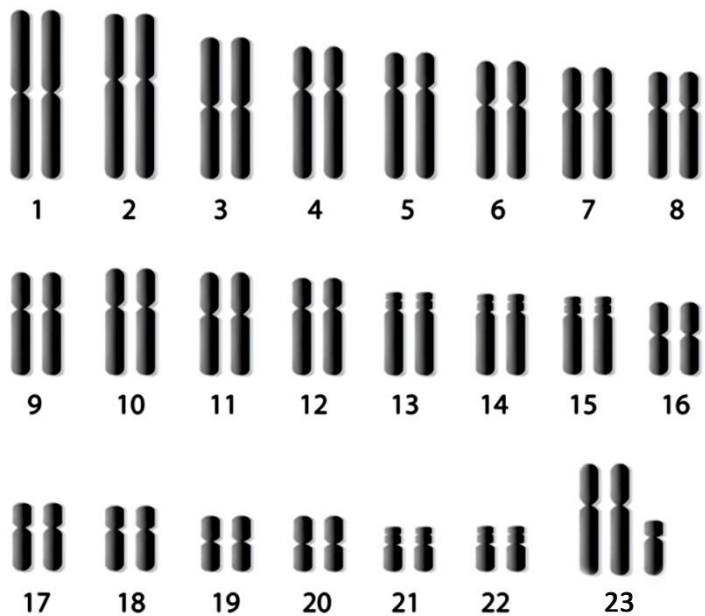
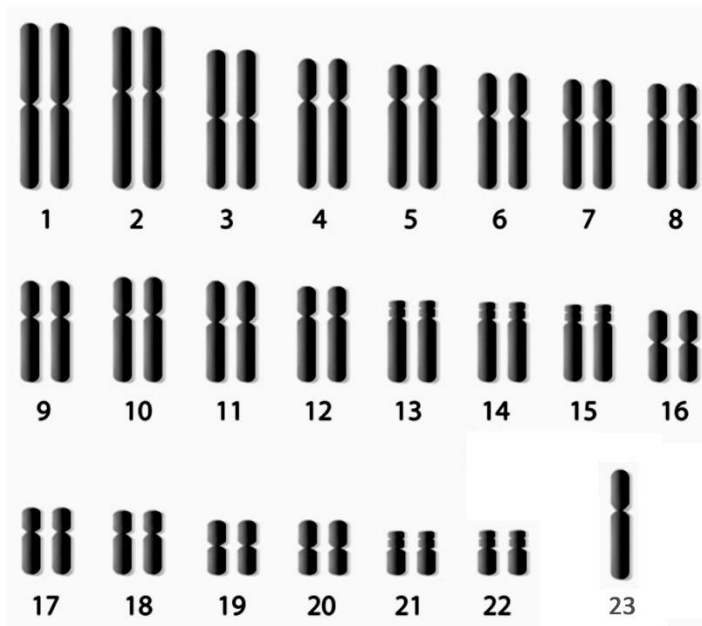


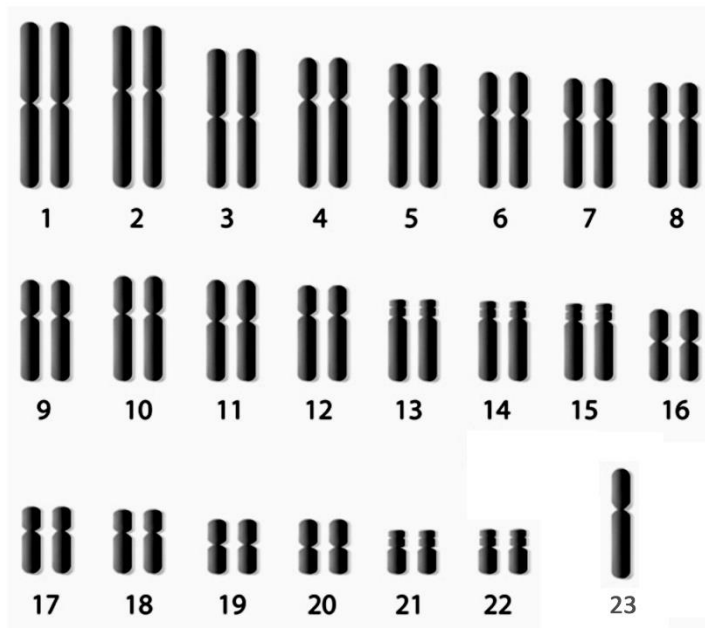
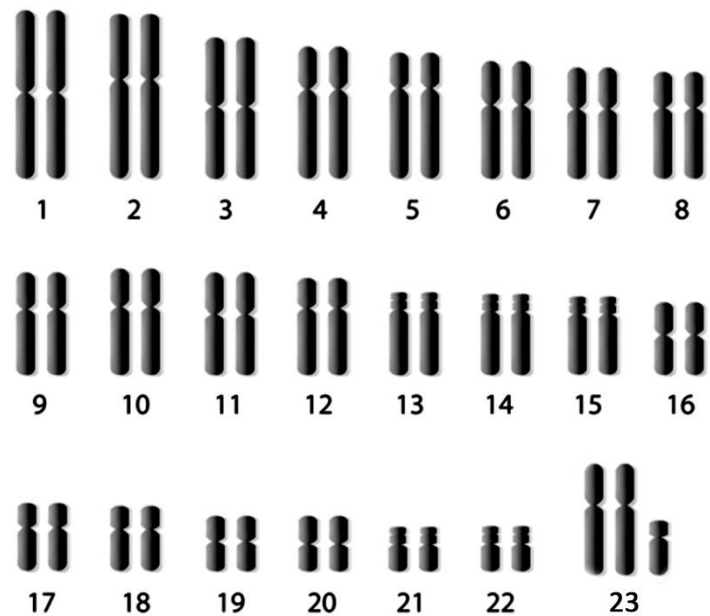
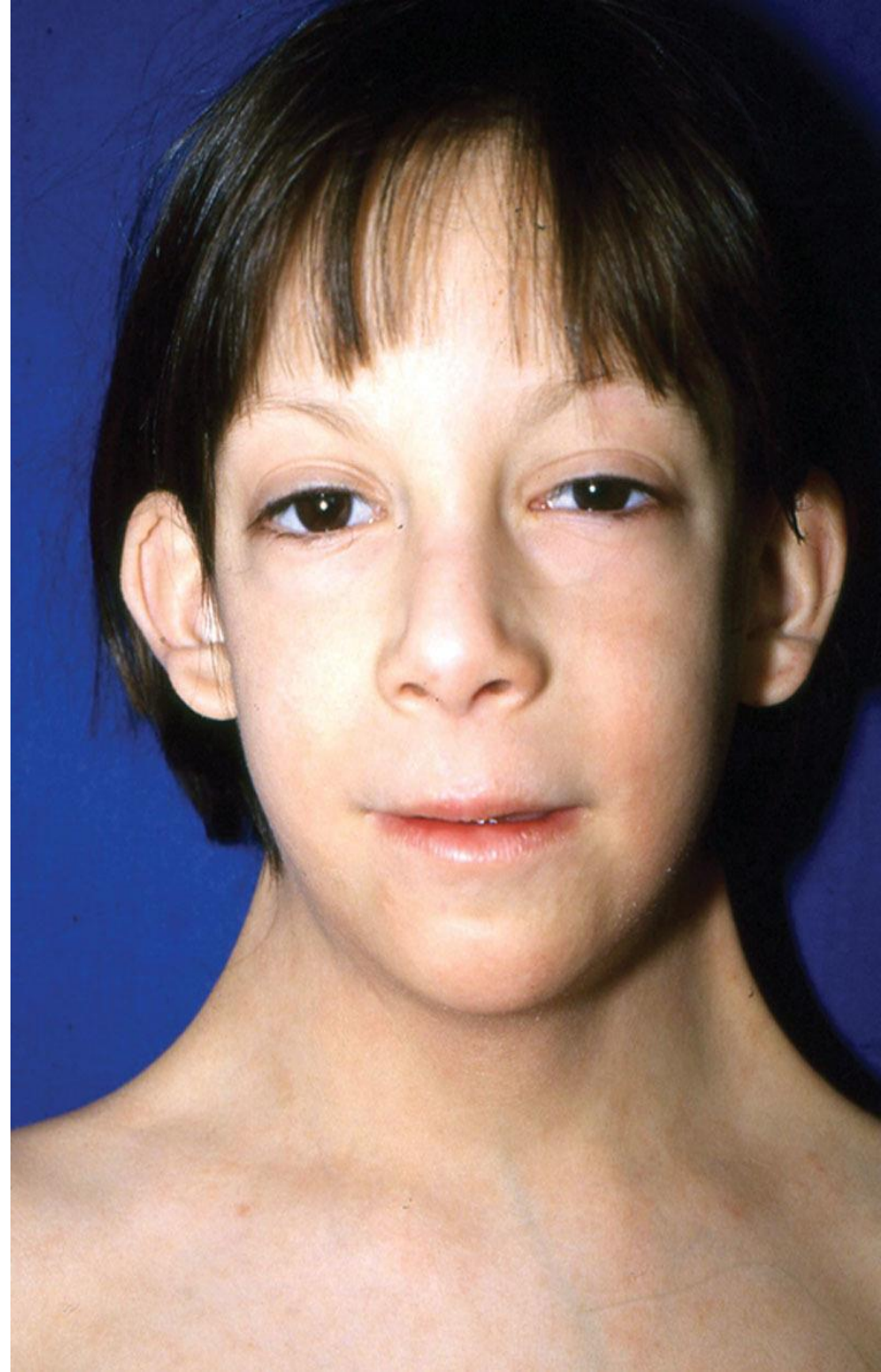
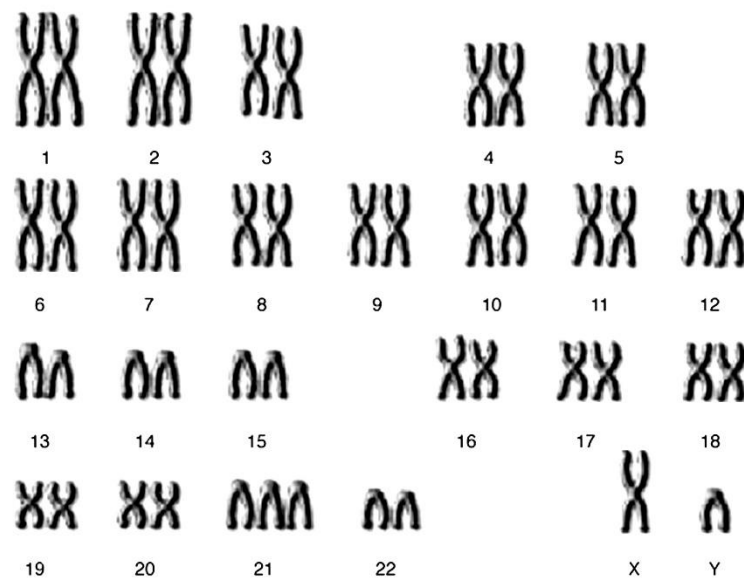
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C



A**B****C**

A**B****C**

Assignment – Compare & Contrast

Disease	Occurrence	Symptoms	Causes	Treatment/ Remedy
Klinefelter				
Turner				
Down				