GENETIC DISORDERS

- Canavans (3:16)
  - https://www.youtube.com/watch?v=UjXEaAlAYiLI
- Congenital insensitivity (1:00)
- Williams Syndrome (3:40)
  - https://www.youtube.com/watch?v=BlEJkhZcSVQ
- Angelman Syndrome (3:45)
  - https://www.youtube.com/watch?v=U5J0kvFSTtA
- Rohhad.Net (4:20)
  - https://www.youtube.com/watch?v=mzUJcsMKu5Q
- XYY Syndrome (4:00)
  - https://www.youtube.com/watch?v=6BsxLnlL9ok
- Larsen Syndrome
  - https://www.youtube.com/watch?v=k9Ts8jXw5ml
- Sanfilippo Syndrome Type C (3:00)
  - https://www.youtube.com/watch?v=XikZm0l_tEM
- Hallermann-Streiff Syndrome (7:00)
  - https://www.youtube.com/watch?v=mMzKvVtrC6U
KARYOTYPES

WHAT CAN THEY TELL US?
KARYOTYPE:
In your own words, explain what a karyotype is.
WRITE THE FOLLOWING DOWN:

• Karyotype = a picture of chromosomes
• Humans have 23 pairs
• Pairs #1 – 22 are autosomal chromosomes
• Pair #23 are the sex chromosomes
Nondisjunction = chromosomes do no separate correctly during meiosis.
A typical human cell will have chromosomes in pairs.

- **Monosomy** = One chromosome
- **Trisomy** = Three Chromosomes

- **Karyotype** = AKA chromosomal analysis = shows the number and appearance of chromosomes in an eukaryotic cell
- Karyotypes can determine the gender and chromosomal mutation.
- A typical human has **23 pairs** of chromosomes for a **total of 46** chromosomes.
KARYOTYPE

• What two things does a karyotype tell you?
NORMAL FEMALE KARYOTYPE

Normal Female
NORMAL MALE KARYOTYPE

Normal Male
KARYOTYPES

• Can you identify where the abnormality is?
• How many autosomal chromosomes are there?
• How many sex chromosomes are there?
• Is this a male or a female?

Kleinfelter's Syndrome
**KARYOTYPES**

- Can you identify where the abnormality is?
- How many autosomal chromosomes are there?
- How many sex chromosomes are there?
- Is this a male or a female?
WRITE THE FOLLOWING DOWN:

• Abnormalities are usually due to mutations during meiosis

• Mutations are changes in the DNA or chromosomes
TURNERS SYNDROME

• 1 in 2,500 - 5,000 births
• 45 chromosomes
• X only
• #23

Monosomy
Nondisjunction

Can you identify where the disorder is?
TURNERS SYNDROME

- 1 in 2,500 – 5,000
- 96-98% do not survive to birth
- No menstruation
- No breast development
- No hips
- Broad shoulders and neck
JACOB’S SYNDROME

- 1 in 1,000 – 2,000 births
- 47 chromosomes
  XYY only
- #23 Trisomy
  Nondisjunction

Can you identify where the disorder is?
JACOB’S SYNDROME

- 1 in 1,000 to 2,000 males
- Normal physically
- Normal mentally
- Increase in testosterone
- More aggressive
- Normal lifespan
KLINEFELTER’S SYNDROME

- 1 in 1,000 to 2,000 births
- 47 chromosomes
  - XXY only
- #23 Trisomy
  - Nondisjunction

Can you identify where the disorder is?
KLINEFELTER’S SYNDROME

• 1 in 1000-2000 births
• Scarce beard
• Longer fingers and arms
• Sterile
• Delicate skin
• Low mental ability
• Normal lifespan
TRIPLE X SYNDROME

- 1 in 1,000 - 2,500 births
- 47 chromosomes
  - XXX only
- #23 Trisomy
  - Nondisjunction

Can you identify where the disorder is?
TRIPLE X SYNDROME

- 1 in 1000 – 2500 females
- Normally physically with minor deformities (curved pinky, flat feet)
- Normal mentally, minor disability issues like dyslexia
- Early development, taller than normal, weak muscle tone
- Short lifespan
DOWN SYNDROME

#21 Trisomy
- Nondisjunction
- 1 in 1,250 births
- 47 chromosomes
- XY or XX

#14/21 Translocation
- 1 in 31,000 births
- 46 chromosomes
- XY=97%
- XX=3%

Can you identify where the disorder is?
DOWN SYNDROME

- 1 in 1,250 births
- Short, broad hands
- Stubby fingers
- Rough skin
- Impotency in males
- Mentally challenged
- Small round face
- Protruding tongue
- Short lifespan
EDWARD’S TRISOMY SYNDROME

- 1 in 4,400 births
- 47 chromosomes
- XX=80%
- XY=20%
- #18 Trisomy nondisjunction

Can you identify where the disorder is?
EDWARD’S TRISOMY SYNDROME

• 1 in 4,400
• Small head
• Mentally challenged
• Internal organ abnormalities
• 90% die before 5 months of age
PATAU’S TRISOMY SYNDROME

- 1 in 14,000 births
- 47 chromosomes XY or XX
- #13 Trisomy Nondisjunction

Can you identify where the disorder is?
PATAU’S TRISOMY SYNDROME

- 1 in 14,000
- Small head
- Small or missing eyes
- Heart defects
- Extra fingers
- Abnormal genitalia
- Mentally challenged
- Cleft palate
- Most die a few weeks after birth

polydactyly
NOW IT IS YOUR TURN TO BE A DOCTOR