KARYOTYPES

WHAT CAN THEY TELL US?
KARYOTYPE 1
KARYOTYPE 2
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
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.
WRITE THE FOLLOWING DOWN:

- Karyotypes can be used to find gender (male or female) and genetic abnormalities.
  - Monosomy = One chromosome
  - Trisomy = Three Chromosomes
Can you identify where the abnormality is?

Kleinfelter's Syndrome
NORMAL FEMALE KARYOTYPE
NORMAL MALE KARYOTYPE

Normal Male
Abnormalities are usually due to mutations. Mutations are changes in the DNA or chromosomes. These changes occur during meiosis.
Genetic Disorders

**WARNING:** SOME IMAGES MAY BE DISTURBING. USE CAUTION. YOU ARE EXPECTED TO SHOW MATURITY LOOKING AT THESE DISORDERS.
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
NOW IT IS YOUR TURN TO BE A DOCTOR