Non-disjunction Disorders
Sex Chromosomes

• Sex Chromosomes are X and Y
• **Autosomes** – other chromosomes that are not sex chromosomes
• Females have two X chromosomes and males have one X chromosome and one Y chromosome
HUMAN CHROMOSOMES

a) Telomere

Centromere

Chromatid

Abnormal Meiosis: Nondisjunction

- Mistakes can occur in meiosis
- Nondisjunction happens when two homologous chromosomes move to the same pole during meiosis
- 1 daughter cell gets an extra chromosome (24 chromosomes) and 1 daughter cell has one less (22 chromosomes)
- **Aneuploidy** is defined as an abnormal number of chromosomes
• When a haploid with 24 chromosomes joins with a haploid with 23 chromosomes, then the diploid cell has 47 chromosomes instead of 46. This results in a pair of chromosomes that have 3 instead of 2 (trisomy)
When a haploid with 22 chromosomes joins with a haploid with 23 chromosomes, then the diploid cell has 45 chromosomes instead of 46. This results in a pair of chromosomes that have 1 instead of 2 chromosomes (monosomy).
Nondisjunction Disorders

- **Down Syndrome**
- Most common nondisjunction disorder
- 95% of people with Down Syndrome have an extra chromosome in pair 21. The person has too much genetic information.
- Common traits
  - round, full face
  - enlarged, creased tongue
  - short in height
  - large forehead
• 1 in 800 babies have Down Syndrome
• Chance of having a baby with Down Syndrome increases with age of mother. When mother is in her 40’s, she has a 1 in 40 chance of having a baby with Down Syndrome
Female Human Karyotype Chart
Human Male Karyotype Chart
Human Male with Down Syndrome
Turner’s Syndrome

- A monosomic disorder such that a female has only 1 X chromosome.
- When an egg with no X chromosome is fertilized a zygote with 45 chromosomes is produced.
- The person appears female, but does not develop sexually.
- The person is usually short and has a thick neck.
- 1 in 5000 females are born with this syndrome and usually most babies with Turner’s Syndrome are miscarried by the 20\text{th} week of pregnancy.
Karyotype from a female with Turner syndrome (45,X)
Short stature
Low hairline
Shield-shaped thorax
Widely spaced nipples
Shortened metacarpal IV
Small finger nails
Brown spots (nevi)

Characteristic facial features
Fold of skin
Constriction of aorta
Poor breast development
Elbow deformity
Rudimentary ovaries
Gonadal streak (underdeveloped gonadal structures)
No menstruation
Klinefelter’s Syndrome

- Syndrome where the male gets two X chromosomes and a Y chromosome.
- Appear male at birth, but as they enter puberty they begin producing high levels of female sex hormones.
- Tendency to develop breasts, no facial hair and higher voice
- People with Klinefelter’s Syndrome are sterile
- 1 in 1000 babies
Karyotype from a male with Klinefelter syndrome (47,XXY)
Frontal baldness absent
Tendency to grow fewer chest hairs
Breast development
Female-type pubic hair pattern
Small testicular size
Poor beard growth
Narrow shoulders
Wide hips
Long arms and legs
Chromosomal Abnormalities

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Patau’s – Trisomy 13
Edward’s Syndrome – Trisomy 18