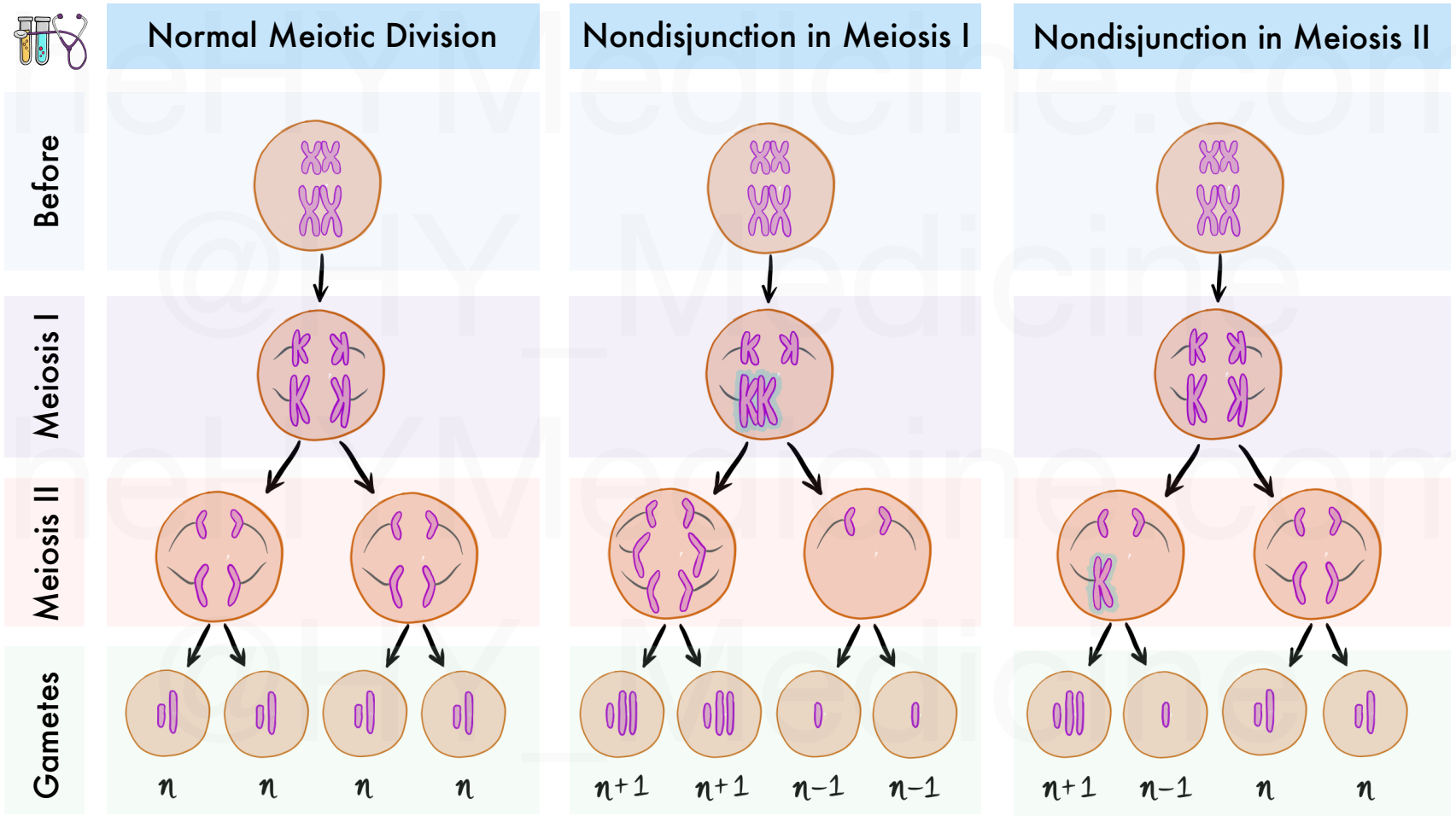

Autosomal Trisomy

Trisomy 21 - Down syndrome

- Most common live-birth autosomal trisomy
- Intellectual disability, depressed nasal bridge, single palmar crease, short stature, epicanthal folds, gap b/w first 2 toes, congenital heart defect (CHD)
- β2 Amyloid gene is on chromosome 21 → early onset Alzheimer's

Trisomy 18 - Edward Syndrome

- Second most common live-birth autosomal trisomy
- Rocker bottom feet, clenched fists, micrognathia, microcephaly, CHD
- Death usually occurs within the first year of life

Trisomy 13 - Patau Syndrome

- Polydactyly, cleft lip/palate, micropthalmia, microcephaly, cardiac anomalies
- Death usually occurs within the first year of life

Sex Chromosome Aneuploidy

Turner Syndrome (45, X)

- X chromosome monosomy; no barr-body (no inactivated x chromosome)
- Short stature, webbed neck, amenorrhea, gonadal dysgenesis ('streak ovaries')
- Bicuspid aortic valve & horseshoe kidney
- ↓ estrogen → ↑ FSH & LH

Klinefelter syndrome (47, XXY)

- Presence of barr-body (presence of inactivated x chromosome)
- Gynecomastia, female distribution of hair, and testicular atrophy → infertility
- Abnormal leydig cell → ↓ testosterone → ↑ LH → ↑ estrogen