

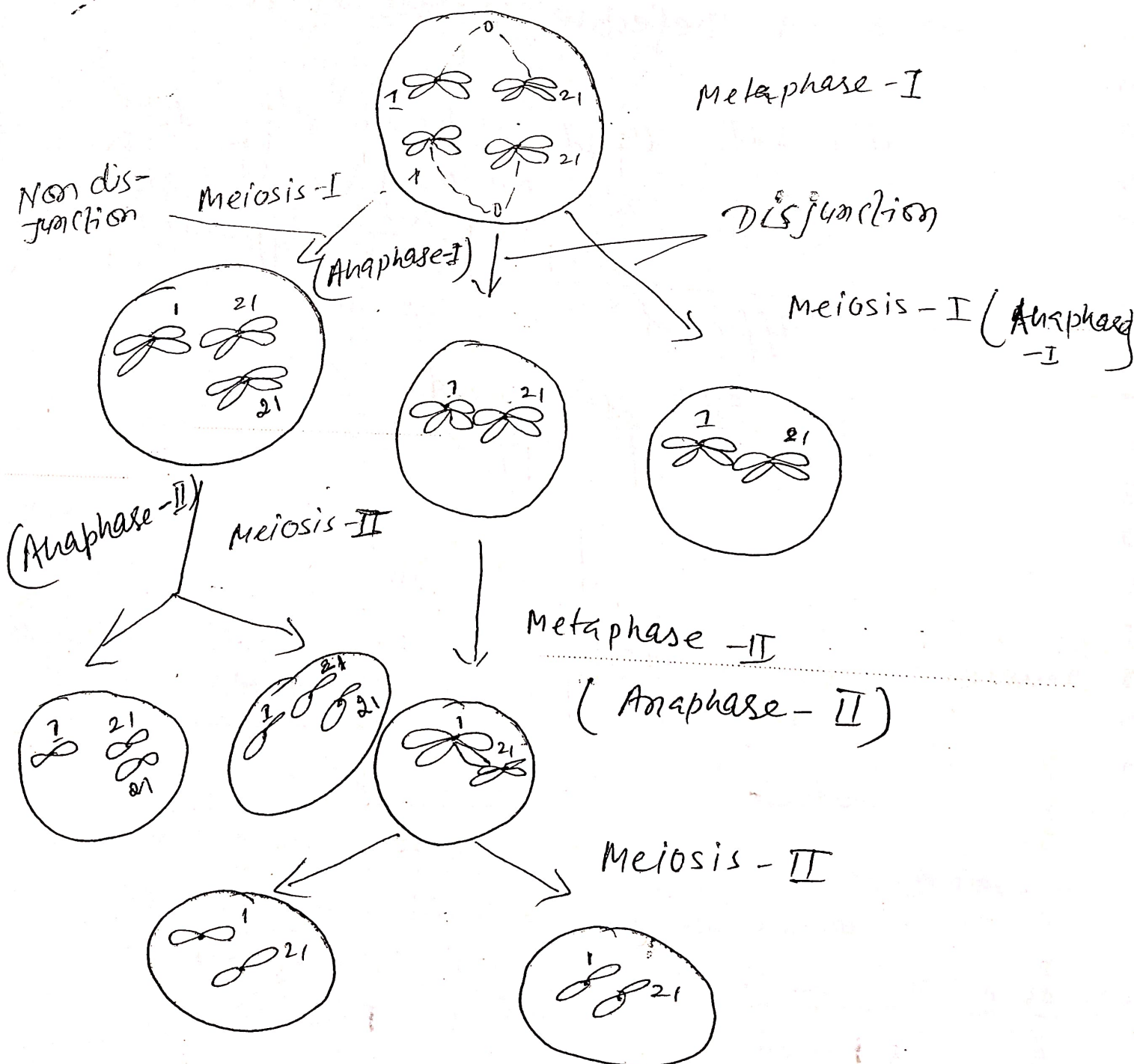
MSc-6
sems

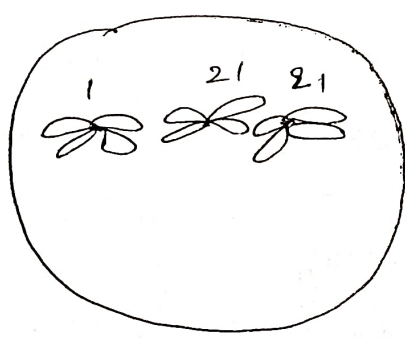
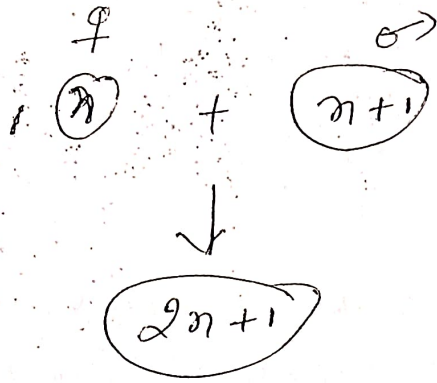
Chromosomal
~~Chromosome~~ Aberration

→ Numerical Aberration → Aneuploidy ($\frac{2n+1}{\downarrow}$, $\frac{2n-1}{\downarrow}$)
Trisomy Monosomy

1. Down syndrome

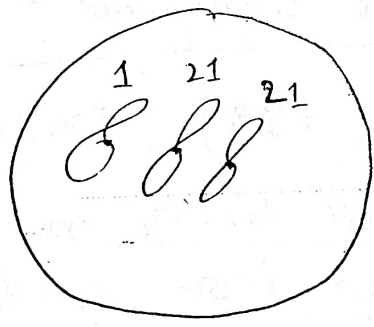
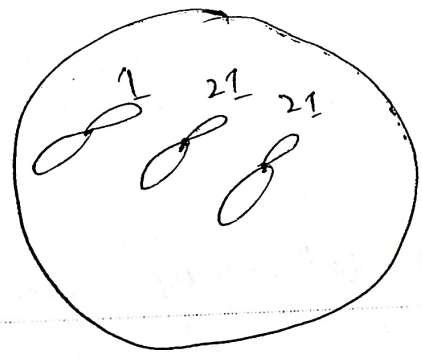
- 47, +21
- Trisomy for #21
- Due to Non disjunction





Metaphase - II

meiosis (Anaphase - II)



→ Short height, epicanthic fold (^{eye} eye half open),
characteristic facial feature, reduced intelligence
 etc

2. Turner syndrome

- 45, XO
- Monosomy
- Adult women with no ovary, sterile female with limited secondary sexual characteristic.
- SRX gene on Y-chromosome present on short arm.

3. Klinefelter syndrome

- 47, XXY ; Trisomy
- The phenotypically male but with some female secondary sexual characteristic, and subjected to Barr body test.

4. Patau's syndrome

- (47, +13) ; Trisomy
- Small brain, mental deficiency, development retardation, cardiac problem etc.

5. Edward's syndrome

- (47, +18)
- These occurs most often among infants of older women. children defectiveness in many organ short sternum. Horse shoe shape kidney.

4