

Structural Abberation

MSC-6
SEM-3

- Deletion
- Translocation

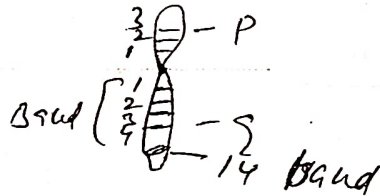
Deletion

1. Cri-du-chat syndrome

- chromosome 5 at short arm (P)
- Cat like cry - Abnormal development of ~~eye~~ ^{Larynx}
- small head, eye with epicanthic fold, mental retardation, characteristic special feature.

2. Retinoblastoma (RB)

- RB gene on chromosome no-13 and deletion in long arm (Q) of band no-14.



- $RB^+ \quad RB^+ \rightarrow \text{Normal}$
- $RB^+ \quad RB^- \rightarrow \text{Normal}$
- $RB^- \quad RB^- \rightarrow \text{Retinoblastoma}$

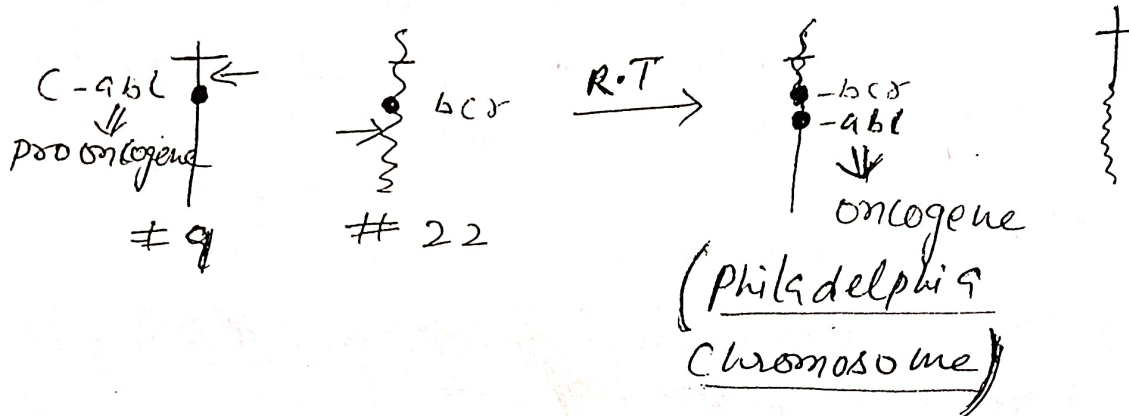
- RB gene is responsible for cell cycle regulation but mutant RB gene lead to ~~uncontrolled~~ uncontrolled cell division which lead to cancer.

3. Wilson's Tumor

- > Deletion in chromosome no-13 in short arm of band no-14.
- > leading to embryonic kidney tumor.

Reciprocal Translocation

(i) ~~chronic~~ chronic Myeloid Leukemia (Acute lymphoblastic Leukemia)



(2) Burkitt's lymphoma :

- > B-cell tumor
- > Common in people of Africa

