

[6] Cystic Fibrosis :- It is caused due to a recessive gene present on chromosome no. 7.

It causes failure of chloride ion transport mechanism. It results clogging of mucus in lungs, liver and pancreas showing anomalies.

[7] Huntington's Disease :- This disease does not appear till the age of 25-55. The patient is affected by disorder of muscle movements. Gradual degeneration of brain tissue takes place in middle ages. It is due to production of inhibitor of brain cell metabolism. It is a genetic disease. It is caused due to genes  $p$  dominant genes present on chromosome no. 4.

[8] Brachydactyly :- It is a genetic disease caused by a dominant gene.

In the affected child middle joints of the digits in hand and feet remain rudimentary and it gets fused with adjacent joints, so that causing shortening of fingers and toes.

Q1 PTC Taster :- Phenyl thio carbamide (PTC)

It is a chemical. It tastes bitter to some person, where as others do not feel it. The taster gene is a dominant gene. So that taster persons may be homozygous (TT) or heterozygous (Tt), but the non-tasters are always homozygous (tt) i.e. for the recessive gene (t).  $\rightarrow$  non-taster.

### [10] Cat-cry Syndrome (Cri du chat) → [14]

is a genetic disease caused due to ~~pa~~ deletion (loss partial loss) of short arm of chromosome no. 5.

The effects are seen in early childhood. The patient has small epiglottis and larynx and cries like a cat.

The face of child is like moon, head is small. ~~The~~ It suffers from retarded growth both mental and physical.

### [11] Wolf Hirschhorn Syndrome → [15]

It is a genetic disease caused due to deletion (partial loss) of short arm of chromosome no. 4.

The patient shows Microcephaly, cleft palate, wide set eyes, epicanthus, low set ears, Cryptorchidism

### [12] Retinoblastoma: It is a genetic disease,

caused due to deletion (partial loss) in short chromosome no. 13. as they have very high risk of this disease.

The patient shows malignant tumor of eyes.

[14] It is caused due to deletion (partial loss) in the long arm of chromosome no. 8.

The patients suffer facial alteration abnormalities in skeletal and ophthalmic and profound mental retardation.

[15] Caused due to translocation of a segment between 13 chromosome no. 13 - 21.

The affected child suffers from delayed speech and I.Q.

[16] Granulocytic Leukemia: It is a genetic disease caused due to translocation of a segment of chromosome no. 22 to chromosome no. 9.

The patient suffers increased proliferation and accumulation of the granulocytes.