C A S E

Enabling Objective:

By the end of the Module II, student will be able to:

- Correlate the changes in structure and function of the different cell parts in health and in disease states.
- Name few clinical common conditions associated with abnormal structure and function of different cell parts.

Learning Objectives:

1. Structure, function and different parts of cell.
2. Importance of cell membrane in regulating various function and defects in disease states.
3. Importance of cell nucleus in defining cell function and determining phenotype and genotype of the species.
4. Structure, number and types of chromosomes.
5. Effects of Chromosomal anomalies on gross appearance and physiological functions of human body
6. Clinical feature of this syndrome and name it.
7. Social and ethical issues associated with this syndrome.
8. Other syndromes of numerical and structural chromosomal abnormalities.

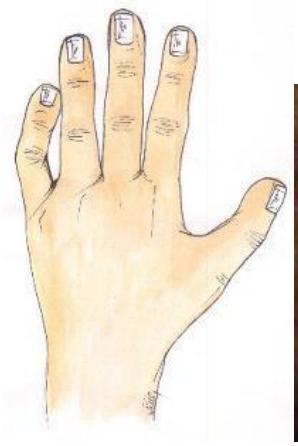
Parents of a 5-Year-old boy present to a clinic of a tertiary care hospital with complains that the child has not yet started walking unsupported or speaking, & has looks very different from his siblings. On asking parents also report delayed sitting, & that he was also a slow learner. He could not feed himself & was not yet toilet trained also. He was born about 10 year later than their last child when mother's age was 45 years.

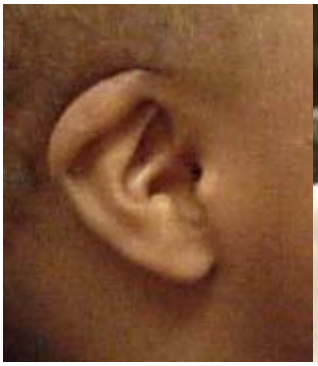
All other family members were normal.

On Examination:

He appeared shorter for his age and plump. He had a small head; tongue appeared larger because it appeared to be protruding out of the mouth all the time. He had depressed nasal bridge and small ears with folded ear pinna. He could barely talk but could communicate with gestures. Hands were small with fifth finger bent towards the fourth and there was only single palmar crease.

His investigations showed:





EEG: Recurrent Seizure discharges.

ECG: Prolonged QRS complex.

Blood sugar: 205mg/dl

All other investigations were normal.

Questions:

1. What is this condition?
2. Why other siblings are not affected?
3. Which part(s) of the cell is involved, how and when does this disorder happen?
4. What Organs/systems are involved in this condition?
5. Name some other conditions caused by abnormalities occurring at this level.
6. Can you explain the abnormal results of investigations on basis of abnormal functioning of single part of a cell?
7. How does this condition affect the life expectancy & functional status of a person?
8. What social issues are faced by the child & the parents?
9. What is the earliest time when this condition can be diagnosed & what ethical issues are associated with diagnosis at this time?