

# FOUNDATION MODULE

## CBL-03 SEMESTER-I WEEK-07, (Batch 2016-17) SMC

### CASE

- 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 palmer crease.



EEG: Recurrent Seizure discharges.

- ECG: Prolonged QRS complex.
- Blood sugar: 205mg/dl
- All other investigations were normal.

**Q1 : What is this condition?**

**Q2: Why other siblings are not affected?**

**Q3 : Which part(s) of the cell is involved, how and when does this disorder happen?**

**Q 4: What Organs/systems are involved in this condition?**

**Q5: Name some other conditions caused by Abnormalities occurring at this level.**

