CBL
LOCOMOTOR MODULE

LEARNING OBJECTIVES

By the end of this session students should be able to

1. Recognize the causes of weakness in the limb.
2. Identify modalities of investigation of muscular disorders.
3. Recognize the genetic basis of muscle diseases.
4. Understand the finding of Electrophysiology (EMG / NCVs).

CASE SCENARIO

Parents of a six year old boy have noticed that the child has gradually started walking on toes, falls frequently and has difficulty in getting up from the ground when he falls. His parents are afraid that as two of his maternal uncles had similar onset of illness, became bed bound by ages around ten years and died as teenagers, the child will suffer the same problem. On examination of lower limbs, unexpectedly, the child has bulkier and stronger looking calves than the rest of his body built.

However, his thigh muscles are weak, that he can raise his legs against gravity but not against resistance. His CPK (Creatinine Phosphokinase) is 7900 IU / ml. His electromyography shows short polyphasic action potentials with normal nerve conduction studies.

Questions:

1. Why is it difficult for the patient to get up from the ground when he falls?
2. Why the calf muscles are appear enlarged?
3. Why was his CPK elevated?
4. What other conditions can present like this disease?
5. Looking at the history in the family (Family history) what seems to be the mode of inheritance of this disorder?
6. Why is the parents' concern justified?
7. How can you counsel the parents regarding future pregnancies and marriages in family?