Original Research Paper Commerce

Case Report of Duchenne Muscular Dystrophy

Dr.Ramesh Nigade	Department of paediatrics,D.Y. Patil medical college hospital, kadamwadi,Kolhapur,Maharashtra -416003
Dr.P.M. Khare	Department of paediatrics,D.Y. Patil medical college hospital, kadamwadi,Kolhapur,Maharashtra -416003
Dr.Vikas Sapehia	Department of paediatrics,D.Y. Patil medical college hospital, kadamwadi,Kolhapur,Maharashtra -416003
Dr.Arijit Sen	Department of paediatrics,D.Y. Patil medical college hospital, kadamwadi,Kolhapur,Maharashtra -416003

ABSTRACT

in the present paper an attempt has been made to analyse the role of muscular biopsy in duchenne muscular dystrophy.

KEYWORDS

INTRODUCTION:- Duchenne muscular dystrophy is the most common hereditary neuromuscular disease affecting all races and ethnic group. Its characteristic clinical features are progressive weakness, intellectual impairment, hypertrophy of the calves, and proliferation of connective tissue in muscle. The incidence is 1:3,600 liveborn infant boys. This disease is inherited as an X-linked recessive trait. The abnormal gene is at the Xp21 locus and is one of the largest genes.

In the present paper an attempt has been made to analyse the role of muscular biopsy in duchenne muscular dystrophy.

CASE REPORT:- A 8yr old male child resident of Kolhapur presented with complaint of difficulty in walking and running and difficulty in standing from sitting position. patient was apparently normal 8 months back than his parents notice that his child is not walking properly and difficulty while walking which is gradual in onset and progressing and at 7 year of age parents noticed that he is not able to walk properly and after walking for some distance patient falls and without support patient is not able to stand from sitting position. With all these complaints patient presented in opd in D. Y . PATIL HOSPITAL KOLHAPUR for further management. 2nd degree Consanginous marriage between the parents and patient is first baby. Immunisation is complete as per the age. No gross milestone delay.

On examination higher mental function are normal, cranial nerves are normal and motor examination shows hypertrophy of calf muscle in bilateral lower limb and lower limb shows power 4 in distal muscle no fasciculation is present in tongue deep tendon reflexes are absent gower sign is positive. Patient was admitted and blood haemogram,serum electrolyte done which are in normal limit creatine phosphokinase level were raised 6487.3 IU/L and to confirm the diagnosis muscle biopsy was planned and done sample taken from calf muscle**, muscle biopsy shows hypertrophied skeletal muscle bundle with proliferation of intervening fibrous stroma areas of regeneration of muscle fibres noted**.

Discussion:- Duchenne muscular dystrophy is the most common hereditary neuromuscular disease affecting all races and ethnic group. Its characteristic clinical features are progressive weakness, intellectual impairment, hypertrophy of the calves, and proliferation of connective tissue in muscle. Muscle biopsy is diagnostic and confirmatory test.

Conclusion:- Duchenne muscular dystrophy is the most common hereditary neuromuscular disorder and is inherited in an X linked recessive manner. Raised CPK level and muscle biopsy are cheap and easily available test for the confirmation diagnosis of duschenne muscular dystrophy.