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Duchenne Muscular Dystrophy

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EXAMINATION AND INVESTIGATIONS:

General Physical Examination, Vitals, and Anthropometric measurements are normal.

On Head to toe examination: Hypertrophy of calf muscles, Exaggerated lumbar lordosis,

Protuberant abdomen and Scoliosis seen.

No baldness, cataract, telangiectasia, high arched palate, syndactyly/ polydactyly, pectus carinatum/excavatum were found.

CNS Examination:

Higher Mental Functions:

Cranial Nerves: Normal

Motor System: Power is 3/5 at Shoulder Joint and 4/5 at Elbow, Wrist, Hip, Knee and Ankle Joint.

Reflexes: Normal

Sensory System: Normal

Gait: Waddling

Gower's sign: Present **

CVS, RS, PA – NAD **



CBC: Normal

LFT: Normal

Urine Analysis: Normal

ECG: Normal

CK-NAC: 14000 U/L

Calcium: 9.9 mg/dl

Phosphorus: 5.6 mg/dl

Alkaline Phosphatase: 419 U/L

Nerve Conduction study: Normal

ECHO: Normal

Clinical Exome Sequencing Report:

A hemizygous two base pair deletion in exon 35 of the DMD gene was detected.

FINAL DIAGNOSIS:

Duchenne Muscular Dystrophy

Treatment Given:

Prednisolone 1mg/kg

Physiotherapy and exercise

Psychosocial

Rehabilitation

Genetic Counselling

Yearly Cardiac follow-up

DISCUSSION:

Duchenne muscular dystrophy

- X-linked inherited neuromuscular disorder

- Mutation in DMD gene – chromosome xp21

- Deficient or defective synthesis of dystrophin protein. ^[2]

- Prevalence – 1 in 3500 males. ^[1]

- Presents between 3 to 5 years of age

- Delayed motor milestones, progressive muscle weakness from proximal to distal, frequent falls with difficulty in running and jumping. ^[2]

- Clinical Examination: Calf muscle hypertrophy (gastrocnemius pseudohypertrophy, lordotic posture, waddling of gait, and poor hip excursion during running.

- Develop associated respiratory and cardiac ailments. Absence of regular monitoring or supportive care, young men with DMD typically die in their late teens and early 20.

Recent Advances:

- Ataluren: Produces a dystrophin which is smaller but functional.
- Studies on modulation of other muscular proteins like myostatin and utrophin .
- Nano Particles as delivery system for DMD therapy^{[1][2]}

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