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Duchenne Muscular Dystrophy

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CLINICAL HISTORY:

9 year old male child with progressive weakness of both lower limbs since 5 years and both upper limbs since 2 years.

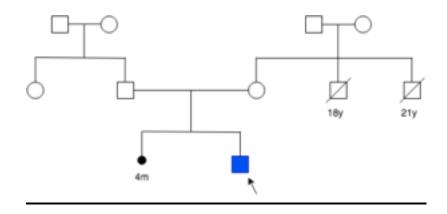
Since 5 years: Difficulty in getting up from sitting and squatting position and difficulty in climbing stairs

Since 2 years: Difficulty in lifting arms overhead, difficulty in mixing food, writing and tripping of feet while walking.

At present: Increased tendency to fall, unable to walk and unable to lift arms.

Milestones till 5 years normal, after which motor milestones started regressing.

Family history: Two of the child's maternal uncles died at the age of 18 years and 21 years with similar kind of clinical picture.



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]}

ACKNOWLEDGEMENTS: None

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