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Case Study

Duchenne Muscular Dystrophy is an Inherited Neuromuscular Disorder

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Introduction

Duchenne Muscular Dystrophy is an inherited neuromuscular disorder. It is a genetic condition which affects the muscles and cause progressive muscle weakness. It is a serious condition which starts in early childhood and leads to weakness of muscle which is not noticeable at birth, even though the child is born with the gene which causes it. It is a most common type of muscular dystrophy in which the weakness develops gradually and usually noticeable by the age of three and the symptoms are mild at first but become more severe as the child gets older.

Mechanism of injury

Duchenne Muscular Dystrophy cause is a mutation in the gene on the 23rd, or X, chromosome that encodes the 427-kDa cytoskeletal protein dystrophin which affects the muscles and that results in the failure of the body for the production of a functional muscle protein dystrophin and people with DMD have a shortage of dystrophin in their muscles. The lack of dystrophin leads to muscle fibre damage and a gradual weakening of the muscles.

Case Study

SukhRam Singh a child of 8 year old male, from Phagwara was assessed on 18-02-2020 at NHS Hospital. Patient was complaining of difficulty in walking, running, and climbing stairs and weakness of lower limb since 4 years. Patient had a history of lack of dystrophin protein due to which patient is having difficulty in walking, climbing stairs, running having frequent falls while walking and progressive muscle weakness which starts proximally in lower extremity then moves to upper extremity (from proximal to distal). Patient parent went to Vishwakarma Hospital Phagwara with his child for his medical treatment 3 year before. Now his parents come to NHS Hospital for his medical and physiotherapy treatment since 1 month he is admitted in NHS Hospital. Patient was not known case of hypertension, thyroid. Patient had no surgical history. According to Kuppu Swami Socioeconomic Scale patient was cooperative and his family member can afford his treatment. His vitals are normal. In Pain Evaluation while checking for pain the Onset of pain in patient was gradual, type of pain was sharpshooting, aggravating factor for the patient was walking and climbing stairs, and relieving factor was rest for the patient. According to VAS and NPRS scale the rating for pain was 8/10 by seeing his facial expression. On Observation while observing the patient built was mesomorphic, he was in sitting posture, he has a waddling gait, calf enlargement and lumbar lordosis which disappears on sitting, skin texture was normal. Due to the weakness of the proximal muscles of the lower limb the patient uses his hands and arms to walk up their own body from a squatting position due to lack of hip and thigh muscle strength it presents of Gower's sign. On Palpation while checking for palpation patient was having muscle wasting of bilateral lower limb, patient was non-febrile, hypotonicity was present on bilateral lower extremity, pseudohypertrophy of calf muscle, pressure sore was absent, and swelling was absent. On Examination while doing examination patient's Range of Motion of shoulder flexion was decreased, hip flexion is increased due to weak hip extensors, ankle dorsiflexion was decreased from normal and ankle plantar flexion was increased, while checking for Manual muscle testing grade was 2- (patient was able to move the joint but unable to complete full range of motion), Balance and Coordination of the patient was affected. In Investigation while investigating the report of patient of CBC Creatine Kinase level was increased, EMG, Muscle biopsy lack of dystrophin protein is noticed. Differential Diagnosis for this patient could be Becker's Muscular Dystrophy, Spinal cord lesion, Motor Neuron Disease. Provisional Diagnosis for this patient was Duchenne Muscular Dystrophy. Physiotherapy Diagnosis for this patient was progressive muscle weakness, limited range of motion, respiratory muscle weakness, gait deformity/abnormality. Short term goal for this patient is to reduce pain, to increase respiratory muscle strength, to increase range of motion, to increase the muscle strength. Long term goal for this patient was to increase muscle strength, to improve the balance and coordination of the patient, to improve the gait of the patient, to make the

patient independent, to improve the patient ADL. As there are no cure of DMD the treatment was symptoms based by multidisciplinary team management (MDT). Medical Management patient was taking NSAIDS (ibuprofen) and Corticosteroid (prednisolone) for maintaining the muscle strength. Physiotherapy Intervention/ Rehabilitation for this patient was preventing secondary complications like contracture, DVT etc by asking to do ankle pump movement and providing night time ankle splint for support or ankle foot orthosis for preventing contracture. Respiratory muscle strengthening training by doing deep breathing exercises and spirometry exercise by inhale and exhale process. Passive ROM exercise, Active assisted ROM exercise, Active resisted ROM exercise, Isometric exercise, Regular Stretching exercise of ankle knee and hip, Strengthening exercise for increasing muscle strength of both upper and lower extremities, Resistance training by using theraband etc, Frenkel exercises, Balance training (static and dynamic balance training), Coordination training (equilibrium and non-equilibrium coordination training), Gait training by using parallel bars, mirror therapy, swissball training etc. Splinting and Orthotic intervention and education for the minimization of risk of fall and regain the ambulation, Functional tasks and strategies for improving the activity of daily living of the patient like self feeding etc and making the patient independent.

ICF model

- Impairment
  - Weakness
  - Muscle shortening
  - Contracture
  - Gower's sign
  - Waddling gait
- Activity limitation
  - Difficulty in walking, running
  - Difficulty in climbing stairs
  - Problem with balance
  - Problem with walking on different surfaces
  - Problem in walking around obstacles
  - Participation restriction
  - Reduced participation in physical activity
  - Decreased participation in sports and exercises
- Environmental factors
  - Home environment
  - School and Community environment
  - Requirement for gait aids and mobility devices
  - Indoor and outdoor environment
  - Walking surfaces and changing in levels
  - Stairs and herbs
  - Cooperative Family and Society members
- Personal factors
  - Age
  - Gender
  - Disease severity
  - Perception of self problem and disability [1-8].

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