A CASE REPORT- EMERY DREIFUSS MUSCULAR DYSTROPHY.

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Abstract:
Emery-Dreifuss Muscular Dystrophy is a rare disease characterised by early contractures especially in the neck, elbows and ankles, slowly progressive muscle weakness more prominent in humeroperoneal region. Age of onset between 5 and 15 years with peculiar cardiac problems in the form of conduction disturbances followed by death in some cases and need for a permanent cardiac pacemaker in others1,2.

Keyword: Emery-Dreifuss Muscular Dystrophy, muscle contractures, muscle weakness, cardiac conduction disturbances.

CASE REPORT: A CASE OF EMERY – DREIFUSS MUSCULAR DYSTROPHY CASE REPORT:
A 11 years old boy presented with difficulty in walking and difficulty in using upper limbs for carrying objects of 3 years duration.

He walked with tip toes over the floor, was unable to extend the elbows fully and had difficulty in turning and bending the head. No history of cardiovascular symptoms.

He was born to non-consanguinous parents. Family history of similar illness present in the sibling and in his mother. Her mother showed similar problems of mild contractures of muscles of neck, elbows and Tendo Achilles. His elder brother had similar illness started at 8 years of age and expired at the age of 13 years. After the death of their first male child only, parents brought their second male child only, parents brought their second male child [This patient] for neurological was examined clinically and found to be normal and there was no subtle signs of muscle weakness or muscle contractures. Female sibling serum CPK level was within normal limit [126IU/L]. Our patient is the third child and is suffering from muscle contractures for the past 3 years.
On examination, patient higher mental functions were normal. All cranial nerves were normal. No ptosis or ophthalmoparesis or facial weakness or bulbar weakness was noted. Examination of spinomotor system revealed mild wasting of scapular muscles, biceps and small muscles of hands. Muscle contractures were noted in posterior neck muscles, elbow and Tendo Achilles with tip toes walking. Muscle tone was decreased. Muscles of all four limbs were weak, proximal weakness more than distal weakness. Plantar response was flexor. Deep tendon reflexes were sluggish in upper limbs and absent in lower limbs. Sensory system, cerebellum, bladder and bowel functions were normal. Examination of cardiovascular system was normal. Pulses were regular, no

LABORATORY INVESTIGATIONS:
Serum CPK (Total) level showed value of 1573 IU/L (10 times of normal value). Basic hematological and biochemical investigations were normal. 12 lead Electrocardiogram was taken and was within normal limit. Cardiologist opinion sought and echocardiogram was done which revealed normal cardiac function. EMG study showed the myopathic pattern.
Muscle biopsy done from Right Gastrocnemius muscle revealed features of muscle fibre degeneration and regeneration. There is considerable fibre size variation, with scattered hypertrophic and hypercontracted fibres in addition to small, rounded, regenerating fibres suggestive of muscular dystrophy.

CASE DISCUSSION:
In our patient important clinical findings observed were proximal muscle weakness suggestive of muscle disease, prominent muscle contractures in neck elbow muscles and Tendo Achilles and positive family history of similar illness in the mother and
in elder sibling who expired recently. Above three clinical features i.e., of Tendo Achilles present but there is no proximal weakness more than distal contracture of neck and elbow muscles. Scapular winging occurs with whole of positive family history are present in following the medial scapular border jutting back clinical conditions. They are Emery Dreifuss Muscular Dystrophy, Duchenne and Becker Muscular Dystrophy and Spinal Muscular Atrophy.

In Emery Dreifuss Muscular Dystrophy, there is prominent contractures of neck, elbow muscles and Tendo Achilles along with proximal muscle weakness, cardiac conduction abnormalities and positive family history. In our case, at present there is no cardiac conduction abnormalities. The cardiac symptoms usually appear between third and fifth decades of life in EDMD. Because of non-availability of Holter monitoring in our center, we advised the patient to attend higher center for Holter monitoring to look for cardiac conduction abnormalities and to report immediately if patient experiences any palpitation or syncope etc. In Duchenne and Becker Muscular Dystrophies, proximal muscle weakness more than distal muscle weakness is present. Recently, criteria to establish diagnosis of EDMD have been postulated. 1. Early contractures of Achilles, elbows and spine. 2. Slow progression of muscular atrophy and bilateral symmetric muscle weakness prominent in humero-peroneal muscles. 3. Abnormality of cardiac conduction and/or another cardiomyopathy evidence. 4. Muscle biopsy revealing myopathic aspects or muscle dystrophy. 5. Pedigree consistent with X linked inheritance.

The most common form of EDMD is X-linked, but there are other genotypes that can cause the EDMD phenotype. X-linked EDMD is caused by mutation in a gene (STA) located on chromosome Xq28, which encodes for the protein emerin. Autosomal dominant EDMD is caused by mutations in the lamin A/C gene. Other forms of autosomal dominant EDMD have mutations in the genes that encode for nesprin-1 and nesprin-2. Emerin, Lamin A/C, naeprin-1 and nesprin-2 are nuclear envelope proteins expressed in skeletal, cardiac and smooth muscle fibres as well as skin cells. Mutations in these genes result in the disorganization of the nuclear lamina and heterochromatin. In EDMD, muscle biopsy may reveal dystrophic or non specific myopathic features, but EMG can show mixed neurogenic and or myogenic patterns for some unknown reasons as Rowland had suggested. In several
families and sporadic cases mentioned in literature, the patterns in EMG or muscle biopsy or both are compatible with myopathy. Although EDMD is not such a disabling disorder physically as DMD and BMD, the combination of progressive muscle contractures and muscle weakness and in particular, the serious cardiac complication result in considerable morbidity and at times mortality, so that accurate identification of carrier status and prenatal prediction by using closely linked probes should be of help to family members.

REFERENCES: