POSTER PRSENTATION FOR WORLD HEATH CONFERENCE 2015

Title: A rare case of Fascio Scapulo Humoral Dystrophy

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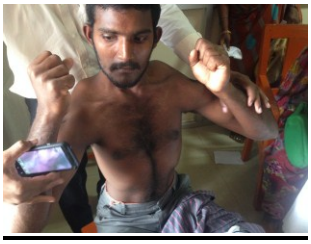
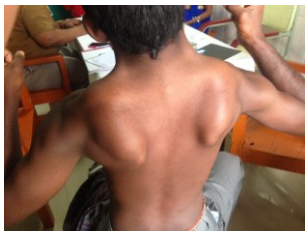
Abstract Category: Select only one: Neurology





Preferred Mode of Presentation: Poster

Abstract Text

Background: Facioscapulohumeral muscular dystrophy is a usually autosomal dominant inherited form of muscular dystrophy that initially affects the skeletal muscles of the face (facio),scapula (scapulo) and upper arms (humeral). FSHD is widely stated to be the third most common genetic disease of skeletal muscle the with prevalence as 4/100,000. Symptoms may develop in early childhood and are usually noticeable in the teenage years with 95% of affected individuals manifesting disease by age 20 years. A progressive skeletal muscle weakness usually develops in other areas of the body as well; often the weakness is asymmetrical. In more than 95% of known cases, the disease is associated with contraction of the D4Z4 repeat in the 4q35 subtelomeric region of Chromosome 4. Life expectancy is normal, but up to 20% of affected individuals become severely disabled.Unfortunately, no effective treatments currently exist for FSHD. However, supportive measures involving physical therapy and the use of orthotics may aid in improving function and mobility.

Case Report: A 25 year old male patient presented to neurology OPD with complaints of progressive weakness of both upper limbs since 2 years. Weakness is in the form of difficulty in lifting shoulders above the head, difficulty in taking food nearer to mouth .He can able to mix the food , buttoning and unbuttoning the shirt & able to move upstairs,get up from squatting position,fro bed & holding chapplas.He had difficulty in squeezing liquids with a straw.No History of swaying to one side. He is not an alchohalic, diabetic or hypertensive.His birth history is normal. He brought up in non consanguineous marriage.No significant family history.

On examination no pallor , jaundice. Vitals stable. Neurological examination revealed normal mentation,no cranial nerve palsies,bilateral facial muscle wasting present,Proximal muscle weakness of upper limb grade 3/5 present ,with atrophy of neck muscle , prominent scapula more on right side with gross wasting of pectoralis and rhomaboidus muscle on both side sparing deltoid muscle. Remaining neurological examination and other sytems were normal.

Laboratory tests of liver, renal,thyroid were normal with elevated serum creatinine kinase levels. Electrodiagnostic testing of the upper and lower extremities resulted in a normal nerve conduction study (NCS). However, electromyography (EMG) did demonstrate findings consistent with a myopathic disorder.Genetic analysis not done.

Finally **Fascio Scapulo Humoral Dystrophy diagnosis was made.**

Patient was treated symptomatically,but weakness neither improved nor progressed and he was regular follow up.

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