FADING STRENGTH, UNBREAKABLE TIES : A LEGACY OF MUSCLE LOSS

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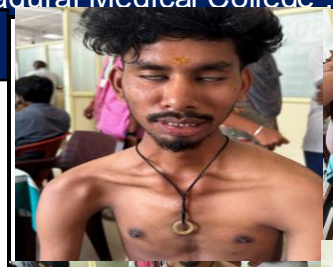
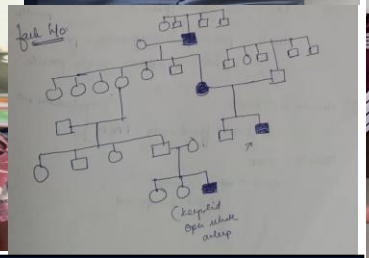
INTRODUCTION

Facioscapulohumeral dystrophy (FSHD) is a genetic myopathy causing progressive muscle weakness and wasting. It has a distinct clinical pattern, initially affecting facial, periscapular, and humeral muscles, followed by the trunk and lower limbs in an often-asymmetrical manner. Severity and progression rates vary greatly. FSHD is inherited as an autosomal dominant trait, though sporadic cases occur, and is caused by two genetic pathways converging on a single mechanism. Extramuscular involvement such as hearing impairment, retinal vasculopathy, and cardiac involvement are more common in infantile cases. Treatment is largely supportive.

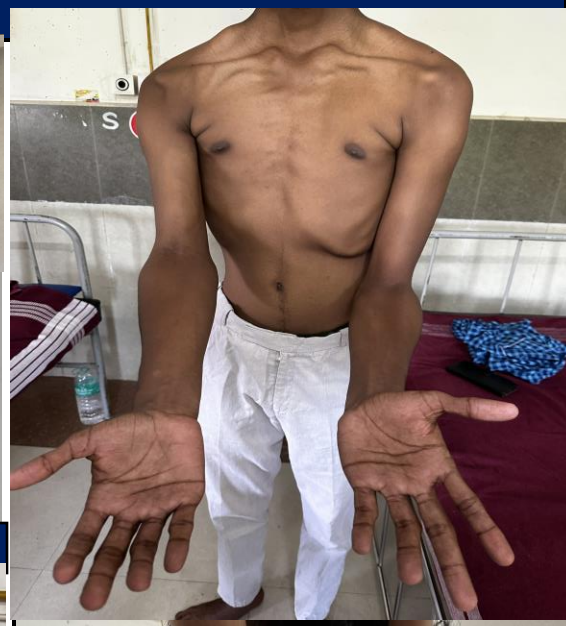




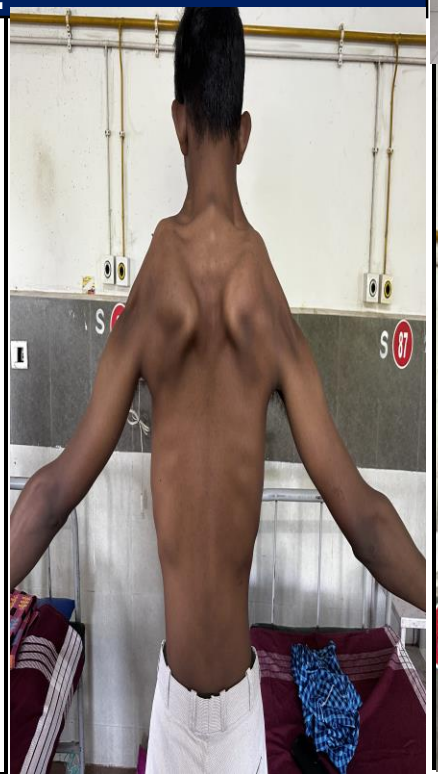
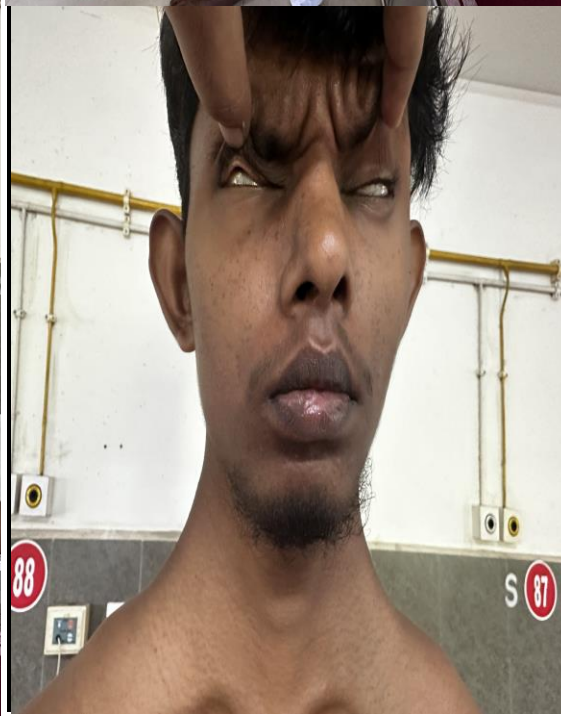
CASE 1

22Y/M presented with insidious onset gradually progressive asymmetrical {right>left} weakness of both upper limbs {proximal} for the past 3 years, with difficulty in raising neck from the pillow, and blowing cheeks, closing eyes tightly . He also has difficulty in turning from side to side in bed. No no sensory disturbance, no weakness of both the lower limbs.A detailed family history showed there was a strong family history with the mother having similar illness and maternal grandfather also had similar weakness but were unnoticed due to the trivial nature and none of them were evaluate or treated . He had undergone surgery for left inguinal hernia 10yrs back, right inguinal hernia in 2021.Birth history, developmental history were normal. Fetal movements were felt normally by the mother. A detailed neurological examination there was selective wasting of scapular and facial muscles with winging of scapula, polyhill sign,pop eye arm appearance, and prominent anterior axillary folds.There was bifacial weakness, lumbar lordosis and positive beevors sign.Sensory ,autonomic, cerebellar function were normal. Laboratory studies showed mildly elevated CPK, and EMG demonstrated mild myopathic potentials. Genetic testing for D4Z4 was initiated.



CASE 2

This pt a 19/M presented with complaints weakness of both upper limbs, proximal R> L , for the past 1yr, associated with wasting of shoulder girldle muscles,no neck muscle/ trunk weakness , There was bifacial weakness with difficulty in closing both eyes against resistance difficulty in blowing the cheeks and , polyhill sign, popeye arm appearance with normal sensory, autonomic higher mental and cerebellar function . With no relavent family history. Normal birth and development. Cpk was mildly elevated , Emg showed myopathic potentials ,Genetic testing sent. Genetic counseling given

Discussion

FSHD remains an underrecognized condition in the Indian context, where limited access to specialized diagnostics can delay identification. Hallmark signs—such as facial diplegia, scapular winging, and selective shoulder girdle muscle wasting—should prompt early suspicion. The value of thorough clinical evaluation and family history cannot be overstated, especially when molecular testing is pending or unavailable.

CONCLUSION

Early diagnosis allows timely genetic counseling, family screening, and future access to targeted therapies such as DUX4-inhibition strategies. Greater clinical awareness and improved diagnostic infrastructure are essential to improve outcomes for patients with hereditary myopathies like FSHD.

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