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Case Report

An uncommon variant of rare type of muscular dystrophy

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ABSTRACT

The muscular dystrophies are a group of hereditary degenerative diseases characterised by progressive myopathy. Emery-Dreifuss muscular dystrophy (EDMD) is a rare genetically heterogenous type of muscular dystrophy characterized by early contractures (especially in the neck, elbows and ankles), slowly progressing muscle weakness more prominent in humeroperoneal region, onset in early childhood and cardiac problems. Emery-Dreifuss muscular dystrophy is commonly inherited in an X linked recessive pattern and rarely autosomal dominant inheritance or autosomal recessive fashion. Here we report a case of autosomal recessive type of Emery-Dreifuss muscular dystrophy from our hospital.

Keywords: Emery dreifuss muscular dystrophy, Contractures, Autosomal recessive

INTRODUCTION

The worldwide incidence of Emery-Dreifuss muscular dystrophy is 1 in 1,00,000.[1] Emery-Dreifuss muscular dystrophy is a rare genetically heterogeneous, familial myopathy characterised clinically by early contracture of joints of limbs and neck extensors, predominant humeropelvic weakness, cardiomyopathy and disease of conduction system of heart[2]. Emery-Dreifuss muscular dystrophy is commonly inherited in a X linked recessive pattern[3] and rarely autosomal dominant inheritance[4] or autosomal recessive fashion.[5] Here we report a case of autosomal recessive type of Emery-Dreifuss muscular dystrophy admitted in department of General Medicine of our hospital.

CASE REPORT

A 25 years old male was admitted on 2nd March 2013 with history of difficulty in walking since 12 years and breathlessness on exertion since 2 years. Patient was apparently normal up to 13 years of age, then he noticed that both his heels were not touching the ground, making him to walk on his toes. Patient gave history of difficulty in gripping footwear. After 1 year he noticed difficulty to get up from squatting position. After 5 years of onset of weakness of lower limbs he noticed weakness of both upper limbs in the form of difficulty in combing his hair. However there was no history suggestive of distal muscle weakness of both upper limbs. Presently he walks without support on his toes. There was no history suggestive of higher mental, cranial nerves, sensory, bowel and urinary bladder dysfunction or convulsions. Past 2 years he noticed insidious onset of breathlessness on physical exertion, not associated with orthopnoea or paroxysmal nocturnal dyspnoea. He gave no history of palpitations, chest pain or syncopal episodes. His developmental mile stones were normal. He was born of a consanguineous marriage couple, his father had 4 children and he has twin sister as shown in pedigree chart (Figure 1). His elder brother and his twin sister had similar abnormal clinical features. The patient's mother

revealed that her maternal uncle had similar abnormal clinical features and died at his age of 15 years.

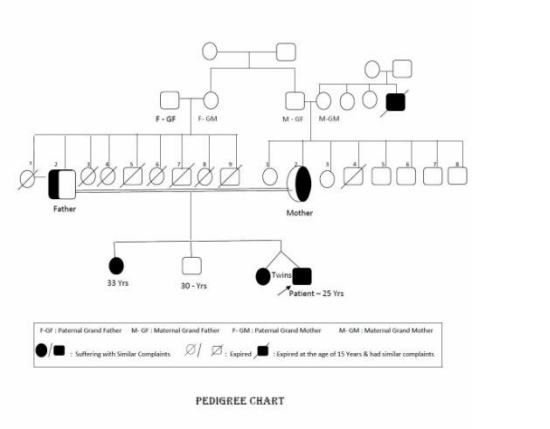

Figure 1: Pedigree chart; note that both twin sister and elder brother of the patient has similar problems.

On general physical examination, patient was moderately built and poorly nourished, conscious oriented, pulse was 81 per minute, regular in rhythm, normal volume and character with no radio femoral delay. Blood pressure was 110/70 mm of Hg, normal pulse pressure and oral temperature was 98° F. He had no pallor. There was no pedal oedema, clubbing or cyanosis.

On systemic neurological examination: No neurocutaneous markers. Higher mental functions were normal. There were no signs of cranial nerve dysfunction. Motor system examination revealed severe wasting of shoulder girdle muscles, biceps brachii, winging of both scapulae, distal part of both legs, flexion contracture of both elbows and Achilles tendons with decreased range of motions in both elbows and both ankles (Figure 2). There was hypotonia in both lower limbs. There was no wasting of post cervical muscles and contracture of neck muscles. There was no limitation of neck flexion. Power in proximal group of muscles of both upper and lower limbs was grade 4 /5 (medical research council grade). Gower sign was present. Deep tendon reflexes were absent. Plantar response was flexor in both lower limbs. There were no bowel and bladder dysfunction. Sensory system examination was normal. Patient showed wide based gait with tip toe walking.

Cardiovascular, respiratory and abdominal systemic examination was unremarkable.

Examination of spine revealed no scoliosis or contracture of spinal muscles.

Laboratory examination revealed Hb% of 12.4 gms% (N: 11 to 18.8 gms/dl), total leucocyte count 8,100/µL (N: 4000 to 11,000/µL), platelet count 3,10,000/µL (N:1,50000 to 4,00,000/µL). Peripheral blood smear showed normocytic, normochromic red blood cells. Random blood glucose was 108 mg/dL (N: 70 to 140 mg/dL). Creatinine phosphokinase was 300 (N: 38 to 174). Chest X ray posterior anterior view was normal. Pulmonary function test revealed mild restrictive lung defect.

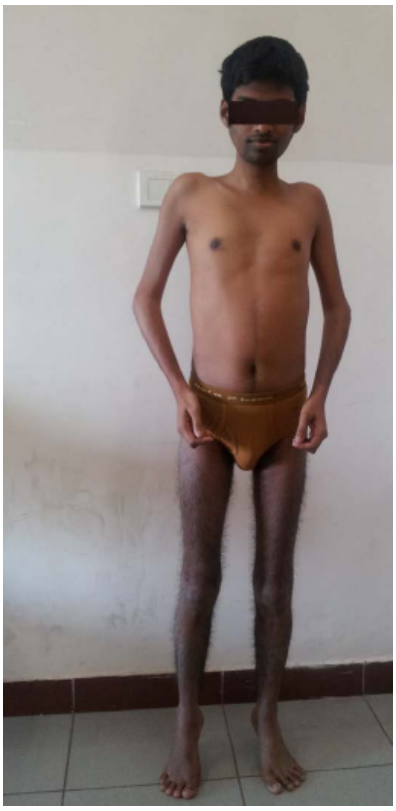

Figure 2: The Male patient. Note the contracture of both elbows and ankles with wasting of biceps brachii and shoulder girdle muscles.

Electrocardiogram revealed sinus rhythm with features of left ventricular hypertrophy by voltage criteria (Figure 3). However 2D echocardiogram was reported normal. Holter monitoring showed sinus rhythm throughout with no ectopics or arrhythmias.

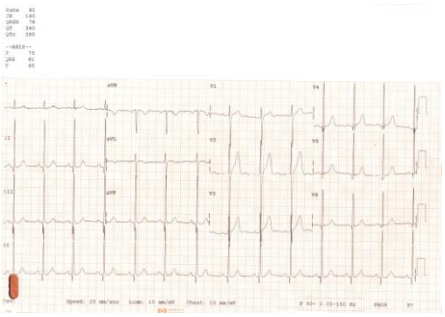

Figure 3: Electrocardiogram showing left ventricular hypertrophy.

ENMG revealed normal insertional activity, no abnormal spontaneous discharges, polyphasic low amplitude compound muscle action potential incomplete interference, all suggestive of myogenic pattern.

Biopsy done from left gastrocnemius muscle showed extensive replacement of interstitium by fibrous tissue with adipocytic infiltration, scattered degenerated myocytes with aggregation of nuclei consistent with myopathy.

A diagnosis of Autosomal recessive Emery dreifuss muscular dystrophy was made in view of early onset of contractures, symmetrical slow progressive less profound weakness predominantly involving scapulohumeropelvicperoneal muscles with wasting and pedigree compatible with autosomal recessive inheritance.

Patient was advised physiotherapy, tendon release orthopaedic surgery and he and his siblings were advised to undergo ECG monitoring annually. All the siblings and the family were counselled that the recurrence rate of this muscular disorder is 25% in the next generation and to avoid consanguineous marriages in future.

DISCUSSION

Emery-Dreifuss muscular dystrophy is commonly inherited in an X linked recessive pattern[3] and rarely autosomal dominant inheritance[4] or autosomal recessive pattern[5]. It is characterized by the triad of: a) Early contractures, often before there is any significant weakness of the Achilles tendons, elbows and postcervical muscles subsequently leading to limitation of neck flexion and later forward flexion of the entire spine becomes limited. b) Slowly progressive muscle wasting and weakness with a distinctive humero-peroneal distribution (i.e. proximal in the upper limbs and distal in the lower limbs) early in the course of the disease, later extending to the proximal limb girdle musculature. Weakness of muscles are rarely profound. c) Cardiac conduction defects (ranging from sinus bradycardia, prolongation of the PR interval on electrocardiography to complete heart block).[6]

Our patient presented with contractures of both elbows, both ankles since the age of 12 yrs with wasting of muscles of shoulder girdle, scapular muscles, biceps brachii and pelvic girdle muscles with relatively minimal weakness, no cardiac conduction defect as evaluated by 24 hours holter monitoring with family history of born to consanguineous marriage couple, similar clinical features in his twin sister and brother but his father and mother being asymptomatic, suggestive of autosomal recessive inheritance. However Electrocardiogram revealed Left ventricular hypertrophy by voltage criteria even though Echocardiogram was normal. Few authors[7] have reported that all patients with Emery-Dreifuss muscular dystrophy will not have cardiac involvement. Cardiac dysrhythmias occur more frequently in late 20 or early 30 year olds and our patient is aged 25 yrs, probably he may develop cardiac conduction defect in future[8]. Our patient had characteristic clinical picture suggestive of EDMD with clear evidence of non X linked autosomal inheritance probably autosomal recessive.[5]

Breathlessness in our patient is due to weakness of muscles of chest as evidenced by restrictive pattern on pulmonary function test. X linked type of EDMD is associated with deficiency of protein called emerin found in inner nuclear membrane of all cells. Emerin is present in Autosomal inherited type of EDMD.[9]

X linked type of EDMD is due to mutation in STA gene on chromosome Xq28 and autosomal type of EDMD is due to mutation in LMNA gene on chromosome 1q11-q23.[9] Unfortunately we could not do Immunohistochemistry of biopsied muscle and genetic mutational analysis as the patient refused to undergo further investigation.

Duchenne muscular dystrophy, Becker muscular dystrophy, Rigid spine syndrome and Limb girdle muscular dystrophy type 1 B (LGMD1B) was considered as differential diagnosis. EDMD was distinguished clinically from the Duchenne and Becker forms by absence of pseudohypertrophy of skeletal muscle, early contracture of the elbow and ankles and normal to moderately raised serum creatinine kinase (CK) level.[8] In Duchenne muscular dystrophy, serum CK is elevated 10 times greater than normal in relation to age[10]. Elevated serum CK is usually more than 5 times than normal range in relation to age[10] but lower in Becker muscular dystrophy than with Duchenne muscular dystrophy.[8] Moreover Duchenne muscular dystrophy is X linked recessive, Rigid spine syndrome is autosomal recessive and is characterised by rigidity of spine[11] but our patient did not have involvement of spine. LGMD1B is characterised by mild contractures and slow progressive weakness of proximal muscles.[12]

CONCLUSION

A 25 year male presented with early contractures of ankle, elbows, scapuloshoulder pelvic muscle weakness with wasting, no cardiac conduction defect with autosomal recessive inheritance suggestive of autosomal recessive EDMD. Clinicians should be aware of this clinical entity as these patients develop cardiac conduction defects and sudden cardiac death. Hence prophylactic pacemaker should be considered to prevent sudden cardiac death.

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