CASE REPORT

Facioscapulohumeral Muscular Dystrophy (Landouzy-Dejerine Type) in a Nigerian Female: A Case Report.

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ABSTRACT

Background: *Muscular dystrophy is not an uncommon entity in Nigerian medical clinics. The facioscapulohumeral type represents a rare variety of the disorder with its own distinctive characteristics but is not expected to have significant cardiac manifestations.*

Method: *The case report of a 17 year old Nigerian female with facioscapulohumeral muscular dystrophy and significant cardiac dysfunction is presented and the relevant literature is reviewed.*

Results: *A 17 year old Nigerian female presented with 18 months history of shortness of breath on mild to moderate exertion, generalized weakness, weight loss and abnormal gait. Leg swelling developed a few weeks prior to presentation. Intrauterine life and early childhood were uneventful while her family history was unremarkable. She had typical features of facioscapulohumeral muscular dystrophy but in addition demonstrated evidence of significant cardiac impairment, which is uncommon and not typically expected in this disorder.*

Conclusion: *It is important for clinicians to comprehensively evaluate every patient presenting with a hitherto "clear "diagnosis in order to unmask unexpected associated clinical details.*

KEYWORDS: Muscular dystrophy; Facioscapulohumeral; Nigerian.

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INTRODUCTION

The term 'muscular dystrophy' represents a range of hereditable degenerative muscular diseases with different genetic and phenotypic backgrounds. Generally uncommon in occurrence, it is regarded as a primary muscle disorder though clinical and electrophysiological evidence had suggested a neurogenic component[1].

Although the clinical features often permit a confident diagnosis to be made, it is necessary that full confirmatory studies including electrophysiological studies and muscle biopsy are performed .The different varieties of muscular dystrophy have involvement of other organ systems to differing extents.

We present a case of facioscapulohumeral muscular dystrophy, eponymously called Landouzy-Dejerine syndrome treated in our center, in order to highlight its occurrence in our locality; to review the aetiopathogenesis and clinical features and appraise the treatment modalities.

Case summary

Miss C.O. is a 17-year old secondary school girl, in SS2. For 18 months prior to presentation, she was noticed to have increasing breathlessness on mild to moderate exertion, weakness, weight loss, and abnormal gait. She had one episode of paroxysmal nocturnal dyspnoea. Amenorrhoea was present for the same period. Pedal swelling, with diurnal variation developed two weeks before presentation. She had been on treatment in another hospital for ventricular septal defect and presented to us for possible surgical closure of the defect.

The period of intra-uterine life and early childhood were un-eventful. There were no educational setbacks. Menarche was at the age of 14years and there was no significant illnesses in the past.

Patient is second out of 6 children in the family, 4 males and 2 females. She is also the second girl. Her parents and siblings are all alive and well. There was no family history of similar illness, or any other illness. The patient does not take alcohol or use tobacco in any form.

Physical examination revealed an ill-looking patient with bilateral pitting leg oedema up to the upper one-third of tibia. Pulse rate at presentation was 90 beats per minute, regular and BP, 120/60 mmHg. Apex beat was located at 6th left intercostal space, mid-axillary line, and there was no heave. Heart sounds S_1, S_2, and S_4 were heard. S_2 was loud and split. There were no murmurs. She had a palpable liver 5cm below the right costal margin which was soft and tender. Lung fields were clear at the time of presentation.

Neurologic examination revealed a sluggish mentation with otherwise normal higher cerebral functions. There was no meningeal irritation. The cranial nerves were intact while the pupils were normal, equal

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and reactive to light. Fundoscopy showed normal disc and vessels. No incoordination was demonstrable. There was generalized wasting, more evident at temporalis muscles and the face, the shoulder (Figs. 1 and 2) and pelvic girdle muscles as well as the small muscles of the hand (thenar and hypothenar also involved). There was 'winging' of the scapulae. (Fig. 2). No fasciculation was seen. Also present were bilateral hypotonia, bilateral areflexia, mute plantar responses and quadriparesis with weakness more proximally. The sensations were spared. A waddling quality was demonstrable to the gait.

Musculoskeletal findings included a narrowed elongated facies, high arched palate, scoliosis of the thoracic spine to the left and pes planus (flat feet with loss of arches).

Full blood count, liver and kidney function tests were normal. Blood group was O positive, and genotype AA.

Serum creatinine kinase was 108.0 IU/L (25.0-192.0), CK-MB 11.3 IU/L (0.0-24.0).

Chest X-ray showed multi-chamber cardiomegaly, with a cardiothoracic ratio of 17:28. The pulmonary conus and left atrial appendage were prominent, but the aortic arch was small. No focal lung lesions seen.

Electrocardiogram showed sinus rhythm, 90 beats per minute, plus one ventricular ectopic beat in lead II. There was bi-atrial enlargement. PR interval was normal at 0.20 seconds with a QRS axis of +90°. Non-specific intraventricular conduction defect was detected in V_1 and there was clockwise rotation. ST segment depression and T wave inversion were seen in leads II, III, aVF, V_4, V_5 and V_6.

Echocardiogram with colour flow Doppler showed dilated left atrium (4.75cm), right atrium and right ventricle (3.05cm). Left ventricular end-diastolic diameter was normal (4.54cm). There were longish and redundant mitral valve leaflets, with closure line near the posterior atrioventricular groove. No valvular regurgitation or stenosis was detected but there was a restrictive pattern of left ventricular diastolic inflow (mainly E wave on trans-mitral pulsed-wave Doppler, velocity 42.7cm/s). Cardiac contractility was slightly reduced (ejection fraction 46.1%, fractional shortening 20.9%). Small pericardial effusion was detected.

Muscle biopsy showed skeletal muscle fibres demonstrating marked variability in fibre diameter, fibre splitting and crowding of internal nuclei of the myocytes. Mild endomysial fibrosis was seen with no inflammatory infiltrate present.

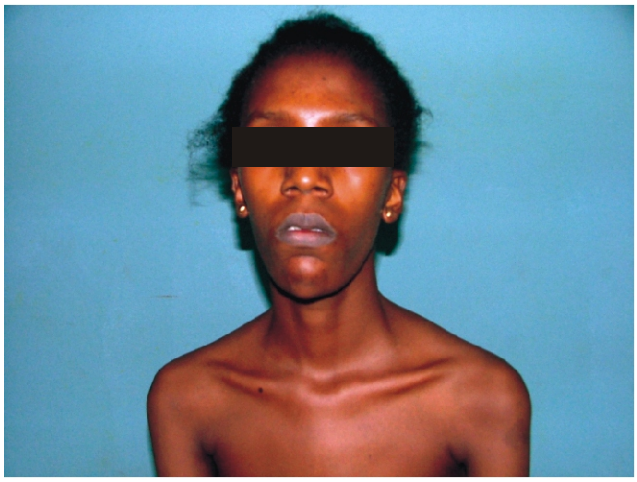

Fig.1. Case of facioscapulohumeral dystrophy showing wasted facial and shoulder muscles and pouting of the lips.

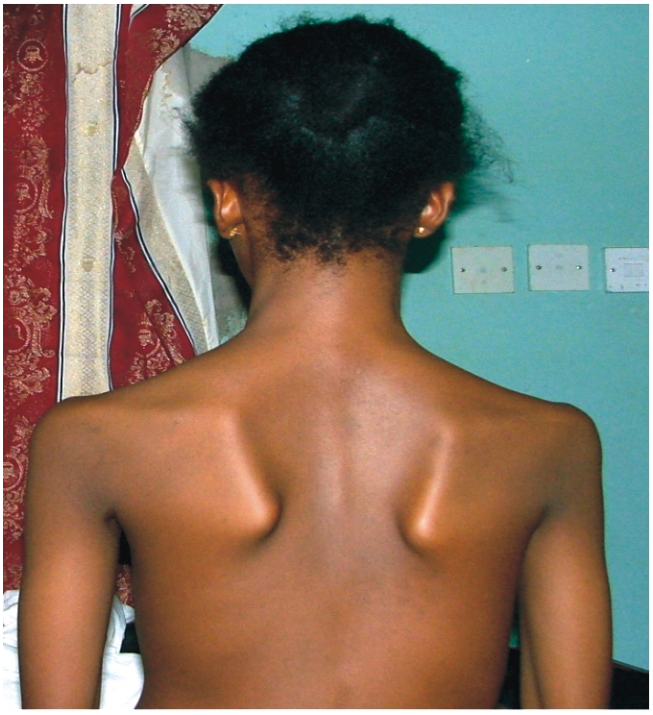

Fig. 2. Back view of the patient showing 'winged' scapulae and wasting of the shoulder muscles.

DISCUSSION

The facioscapulohumeral muscular dystrophy (FSH) was described by Landouzy and Dejerine in 1884[2]. It has an incidence of 1 in 20,000 in the USA[3]. There is no report of its incidence or prevalence in Nigeria.

Transmission is usually autosomal dominant although both autosomal recessive and X linked cases have been described[4]. Males are more affected than females. About 30% of those affected are said not be

aware of the disease presence[3]. Recent advances in molecular genetics have elucidated genetic heterogeneity in FSH. In majority of cases deletions of telemetric heterochromatin at chromosome 4q35 are responsible but the particular gene and its products remain unknown whereas in some families the disease is linked to chromosome 10[3,5-,9]. There is a significant correlation between disease severity and the size of the deletion[7-,9]. Carrier detection and prenatal diagnosis are thus possible.

There is usually a positive family history but it is worth remembering that family histories can be unreliable or impossible to obtain. More than 50% of sporadic cases represent new mutations[3].

Onset is usually insidious and often in late childhood or adolescence although diagnosis is usually made some time after this. Weakness in most cases begins in the face and progresses to the shoulder girdle with winging of the scapulae. Weakness may subsequently develop in the lower limbs with foot drop in approximately 50% of patients[4]. Pelvic girdle weakness occurs later in 20% of patients producing a waddling gait as demonstrable in our patient. Severe functional impairment can result thereof. About 15%- 20% will become wheelchair bound[8]. Reflexes are only impaired at the biceps and triceps. Pseudohypertrophy and contractures are rare. There is usually a normal life span and a normal intelligence though mental retardation complicates about 40% of cases.

Creatinine kinase is usually normal or mildly elevated. Electromyography shows a myopathic pattern. A prominent inflammatory exudates with multifocal distribution is present in some muscle biopsy samples. Others may show typical myopathic features with increased variation in fibre diameters, occasional necrosis and regeneration and an increase in connective tissue and fat. Unlike in the Duchenne muscular dystrophy where there is commonly cardiac involvement, other organ systems including the cardiovascular system in FSH are rarely affected. Nevertheless labile hypertension has been reported to be common in FSH and there is an increased incidence of nerve deafness[3]. Cardiac manifestations of tachycardia, cardiomegaly and arrhythmias (ventricular and atrial extrasystoles) may also be seen[10]. Coat's (eye) disease may also occur in some cases. Coat's syndrome consists of FSH plus sensorineural deafness, mental retardation, retinal telangiectasia, exudation and detachment[11].

Our patient's clinical features are very much typical for FSH (see Figs. 1 and 2). However she did present with some unique features. These include the slow mentation, the high arched palate and the rather significant abnormalities of cardiac function as detected by ECG and echocardiography. These cardiac findings though rare may be encountered[10]. It may thus be easily concluded that these findings are the result of serendipity but we contend that it once more highlights the need for thoroughly evaluating every case of muscular dystrophy on its own merit as unexpected details may be present and contribute to the morbidity and mortality of the patient.

It is further intriguing that this patient was the proband in the family. Whether this represents the result of a new mutation or simply a lack of proper historical documentation (vide supra) remains a poser. EMG was not available in our center, nevertheless the totality of her clinical features and investigative results were sufficiently diagnostic for facioscapulohumeral muscular dystrophy.

It was difficult to determine between the muscular and the cardiac abnormalities, which was the greater factor in contributing to the functional impairment in this patient.

Though no specific treatment is available for FSH, albuterol given as a slow release formulation at a dose of 16mg/day orally for 3 months has been shown to improve muscle strength in 12% of patients[12]. Ankle foot orthoses are helpful for foot drop. Scapular stabilization programmes improve scapular winging but may not improve function. Other supportive measures may be necessary depending on specific entities encountered. Our patient needed stabilization of her cardiac functions.

The concepts of genetic counseling and prenatal diagnosis are still evolving in Nigeria. The traditional and conservative natures of the Nigerian society make these options sensitive in nature.

In conclusion, it must be noted that the muscular dystrophies are here with us in Nigeria. There may be distinct differences in the mode of presentation of these disorders locally as here documented. Bearing this possibility in mind meticulous attention should be given to the clinical evaluation of these patients when they present in order to pick up hitherto unrecognized features.

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