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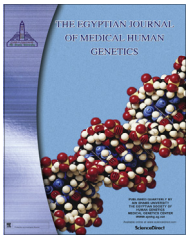

CASE REPORT

Moebius syndrome with macular hyperpigmentation, skeletal anomalies, and hypoplasia of pectoralis major muscle in an Egyptian child

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KEYWORDS

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Cranial nerves;
Ptosis;
Facial nerve palsy

Abstract We report a 4 month old female infant, 3rd in order of birth of the first cousin consanguineous parents. The patient has congenital right facial nerve palsy, with asymmetry of facial expression during crying and difficulty in swallowing. Associated anomalies include abnormal facial features, bilateral finger anomalies, bilateral talipes equinovarus, kyphoscoliosis, hypotonia, high frequency hearing loss. Bilateral macular hyperpigmentation was detected in our patient on fundus examination which was not reported previously in Moebius syndrome cases. In addition there is hypoplasia of the right pectoralis major muscle.

1. Introduction

Moebius syndrome is a congenital, nonprogressive complete or partial facial nerve palsy, with limited abduction of one or both eyes [1]. Other associated features may include other cranial nerve dysfunction with deafness as well as orofacial, musculo-skeletal and neurodevelopmental problems [2–5], but they are not necessary for diagnosis, making the syndrome extremely variable in its clinical manifestations.

Although neither the etiology nor the pathogenesis of the syndrome have yet been elucidated, there are two theories: a developmental rhombomeric defect involving predominantly motor nuclei and axons as well as traversing long tracts due to a genetic cause [6], or an interruption in the vascular supply of the brainstem resulting in ischemia in the region of the facial cranial nerve nuclei owing to an environmental, mechanical or a genetic cause [7,8]. Moebius syndrome can also be considered as a complex regional developmental disorder of the brainstem [9].

We report a case with the typical features of Moebius syndrome who has in addition some unreported features after taking consent of the parents.

2. Case report

A 4 month old female, 3rd in the order of birth of 1st cousin consanguineous marriage. The patient was delivered at full

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term by cesarean delivery after uncomplicated pregnancy with no history of fever, drug intake or smoking by the mother. Her birth weight was 1.250 kg (< 5th percentile). The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of developmental delay and poor weight gain. Since birth the patient was admitted to neonatal intensive-care unit for 12 days due to poor suckling, difficulty in swallowing and low birth weight. There was no improvement of weight gain in spite of nutritional management. The mother noticed that her baby did not close her right eye completely during sleep with no blinking. The patient suffered drooling of saliva from the right side of the mouth. She also had developmental delay as she cannot support the neck. Family history was unremarkable. She had two healthy sibs. Both parents were normal.

On examination, her weight was 3.100 kg (below 5th percentile), her length was 53 cm (below 5th percentile), and her skull circumference was 35 cm (below 5th percentile). The patient had mask face, facial asymmetry, epicanthal folds, broad protruding flat low set ears more marked on the left side which had flat auricle and rudimentary ear lobule, microretrognathia, high arched palate, and tongue tie (Figs. 1 and 2). The patient had clenched fists, adducted thumbs, bilateral overlapping 2nd finger over thumb, bilateral flexion deformity of 4th and 5th proximal interphalangeal joints, left simian crease, dystrophic nails, and bilateral talipes equinovarus deformity (Figs. 3 and 4). There is kyphoscoliosis of the back more marked on thoracolumbar vertebrae. Abdominal, genital, and cardiac examinations were normal. Neurologic examination demonstrated mild hypotonia, normal pupil size and reactivity, convergent squint, limited abduction of the right and the left eye (6th nerve palsy), unilateral right facial palsy with no blinking, and the right eye was opened during sleep and crying (Fig. 5). She also had decreased response to loud voice. She had central uvula with normal tongue movement. Right pectoralis muscle is hypoplastic.

Abdomino-pelvic ultrasonography and ECHO cardiography were normal. Extended metabolic screen, serum lactate,

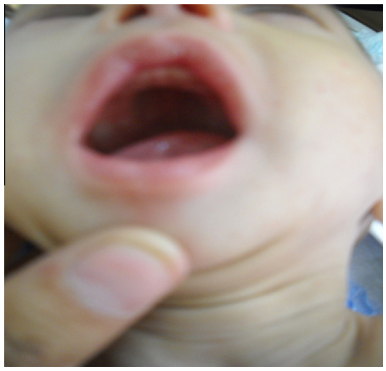

Figure 2 High arched palate.

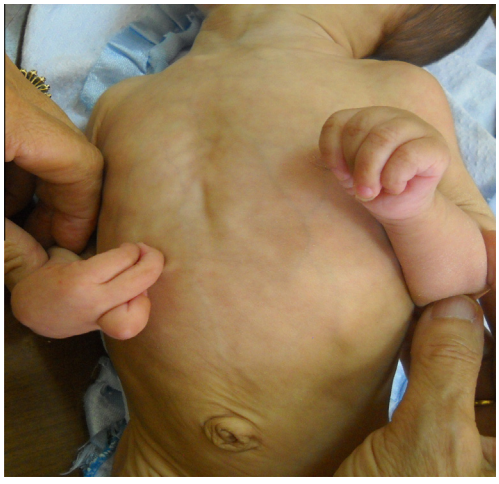

Figure 3 Clenched fists, adducted thumbs, bilateral overlapping 2nd finger over thumbs, bilateral flexion deformity of 4th and 5th fingers.

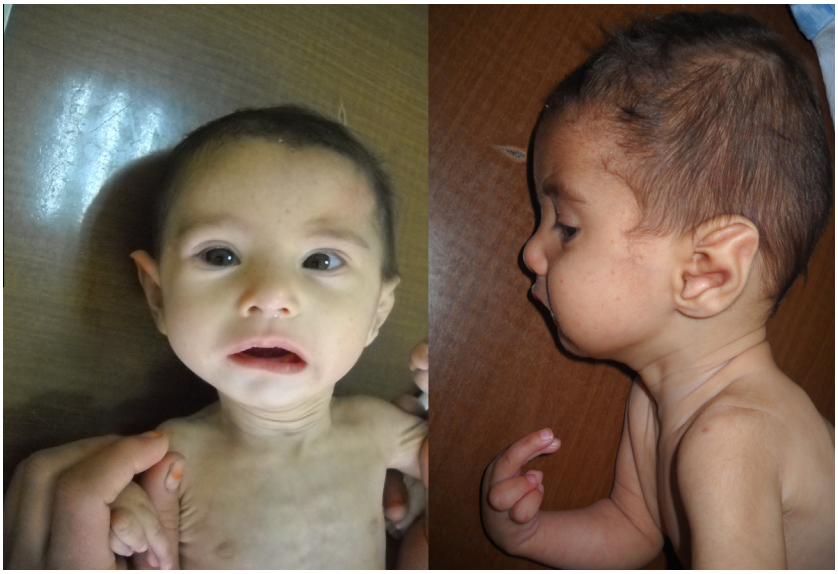

Figure 1 Photo of the patient showing dysmorphic features, asymmetry of the masked face with loss of subcutaneous fat in neck and whole body.