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Reverse Slanting of Split Eyebrows and Palpebral Fissures: A New Dysmorphic Syndrome

By Aamir Jalal Al Mosawi

Abstract- A dysmorphic syndrome is suspected in the presence of more than three minor anomalies which are variations of normal morphological features that are considered of little or no known medical, surgical, or cosmetic significance; more than one major anomaly which is an abnormality that has major medical, surgical or cosmetic significance; and one major anomaly with two or more minor anomalies are also suggestive of congenital syndrome.

Many congenital syndromes are associated with different combinations of hypertelorism (with or without flat mid-face), epicanthic folds, convergent squint, low set ears, upward and downward slanting of the palpebral fissures, and eyebrows abnormalities occurring in association with hypotonia and developmental delay.

The aim of this paper is to describe the occurrence of a new congenital syndrome with the novel association of unique eyebrows abnormalities (splitting with a relatively thick upward slanting medial parts and thin non-slanting lateral parts) with downward slanting palpebral fissures, bilateral convergent squint, hypertelorism with flat mid-face, epicanthic folds, large ears, developmental delay, and infantile hypotonia mostly attributed to congenital myopathy.

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I. Introduction

A dysmorphic syndrome is suspected in the presence of more than three minor anomalies which are variations of normal morphological features that are considered of little or no known medical, surgical, or cosmetic significance; more than one major anomaly which is an abnormality that has major medical, surgical or cosmetic significance; and one major anomaly with two or more minor anomalies are also suggestive of congenital syndrome. Many congenital syndromes are associated with different combinations of hypertelorism (with or without flat mid-face), epicanthic folds, convergent squint, low set ears, upward and downward slanting of the palpebral fissures, and eyebrows abnormalities occurring in association with hypotonia and developmental delay [1,2,3,4].

The aim of this paper is to describe the occurrence of a new congenital syndrome with the novel association of unique eyebrows abnormalities (splitting with a relatively thick upward slanting medial parts and thin non-slanting lateral parts) with downward slanting palpebral fissures, bilateral convergent squint, hypertelorism with flat mid-face, epicanthic folds, large ears, developmental delay, and infantile hypotonia mostly attributed to congenital myopathy.

II. Case Report

A thirteen-month old boy who was the first born child to non-consanguineous parents was first seen at the pediatric neuropsychiatry clinic of the Children Teaching Hospital of Baghdad Medical City because of motor developmental delay. The child had hypotonia during infancy, and was not crawling and was unable to sit without support for long time. He has just started babbling. The boy has distinctive facial features (Figure-1) including:

1. Highly specific unique eyebrows abnormalities consisting of splitting with a relatively thick upward slanting medial parts and thin non-slanting lateral parts.
2. Downward slanting palpebral fissures.
3. Epicanthic folds.
4. Hypertelorism.
5. Depressed nasal bridge.
6. Large ears.
7. Convergent squints of both eyes.

Brain MRI was performed at the age of one month showed normal findings.

Screening for several inborn errors of metabolisms has already revealed no abnormality.

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Figure 1: The boy unique eyebrows abnormalities consisting of splitting with a relatively thick upward slanting medial parts and thin non-slanting lateral parts in association with downward slanting palpebral fissures, bilateral convergent squint, hypertelorism with flat mid-face, epicanthic folds, and large ears.

EMG and nerve conduction studies were performed at the age of seven months (Table-1).

Nerve conduction study (Table-1) was performed by surface and needle electrode on:

Right and left median nerve.
Right ulnar nerve.
Right and left sural nerve.
Right and left common peroneal nerves.

Repetitive nerve stimulation with supra-maximal stimulation of the right ulnar, right facial and right axillary at low rates (3Hz) was performed. The right and right axillary decrement test with 10 pulses (5 trials) showed 1% decrement of motor response.

Needle electromyography (EMG) study was performed on:

Right FDI.
Right deltoid.
Right biceps.
Right and vastus medialis.
Right anterior.

Needle electromyography (EMG) stud showed:

No spontaneous activity.

No myotonic discharges.

The average duration of 20 motor units:

Right deltoid= 5.1 msec (n=8.3 msec).
Right biceps = 4.8 msec (n=8.1 msec).
Right vastus medialis = 4.1 msec (n=8.3 msec).
Right tibialis anterior = 5.3 msec (n= 10.2 msec).
Left tibialis anterior = 5.2 msec (n= 12.5 msec).

30-40% polyphasia of short duration low amplitude was observed.

EMG and nerve conduction studies suggested chronic diffuse non dystrophic myopathic of moderate severity mostly resulting from congenital myopathy.

The proximal lower limb muscles were more severely involved.

Table 1: Summarizes the clinical features of the new syndrome

	Sensory			Motor			
Nerve	Latency msec/cm	Amplitude μV	SNCV m/sec	Muscle	DML Msec /cm	MNCV msec /cm	F-wave Latency
Right median	2.1	26.6	56.2	ABP	3.1	50.2	16.5
Right ulnar	1.9	27.3	56.6	ADM	2.9	51.2	17.2
Right common peroneal				Tibialis Ant. EDB	3.3 4.2	40.2	35.3
Left common peroneal				Tibialis Ant. EDB	3 4.1	40.3	36.3
Left sural	2	15.3	44.6				
Table 4.1: The finding of EMG and nerve conduction studies which were performed at the age of seven months							

Table 2: The clinical features of the new syndrome

Sporadic occurrence
Non consanguineous parents
Splitting of eyebrows with a relatively thick upward slanting medial parts and thin non-slanting lateral parts
Downward slanting palpebral fissures
Epicanthic folds
Hypertelorism
Depressed nasal bridge
Large ears
Convergent squints of both eyes.
Infantile hypotonia attributed to congenital myopathy

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