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CASE REPORT

Aglossia-adactylia sequence and Moebius syndrome involvement.

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ABSTRACT
Adactylia and limb deficiencies are major congenital malformations can be resulted from a long list of etiological factors, in our department the vast majority of these disorders are genetically determined, and only a small fraction of it proved to be sporadic.
We report on a-9-months old male child with Aglossia- Adactylia sequence associated with Moebius syndrome involvement, which in our patient is manifesting itself by left facial nerve and bilateral abducens nerve palsies, total absent of the tongue and absent digits of hands and feet respectively.

Key words: Aglossia-adactylia sequence, Moebius syndrome, Hanhart syndrome, facial nerve palsy, and abducens nerve palsy.

INTRODUCTION

This disorder is characterised by Aglossia-adactylia, and usually the tongue is small but seldom absent. The clinical expression is variable and the limb abnormalities can vary from absence of digits to absence of the distal part of a whole limb. The jaw is small and those who survive usually have normal intelligence. There is a definite association with the Moebius syndrome, in which there are bilateral facial and abducens nerve palsies. A vascular etiology[1] (Robinow et al., 1978), as well as a teratogenic etiology[2,3] (Hall, 1971; Bokesoy et al., 1983) has been implicated. However, most cases are sporadic.

We recently encountered a-9-months old male child presented with the full clinical criteria of "aglossia-adactylia" complex with manifestations of Moebius syndrome, we encountered left facial nerve palsy, and bilateral abducent nerve palsies, the MRI Sagittal section showed the typical picture of absent tongue.

Family search revealed no relevant clinical or radiological features relevant to our patient; therefore, we believe that our patient is a product of a disruptive sequence.

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A-9-months male child was referred to our department because of total absence of digits of the hands and feet

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respectively, he was born full term, a product of uneventful gestation, he was born from vertex presentation. At birth length, OFC, and weight were around the 10th percentile. No associated neonatal complications, apart from the evident absence of digits.
The mother is a-33 –year-old-gravida 1 abortus 0, she experienced 4 years primary sterility, before getting pregnant, married to a –36-year-old-first related man. The child had had remarkable difficulties in sucking and choking was a frequent complaint by the parents, recently he gets used to be bottle fed in small frequent meals to overcome choking.

EXAMINATION:

Developmental examination: Normal motor development, normal hearing, but because of his complex malformations there are a remarkable retardation in coordination and speech development.
Growth: length, OFC, and weight are around the 50th percentile.

Craniofacial: large frontal area, with depressed nasal bridge, though the nose is bulbous, hypertelorism, defective ocular rotation and strabismus. Long philtrum, very thin upper lip, and inward depressed lower lip, because of the absent support of the tongue, micrognathia, and low set ears.

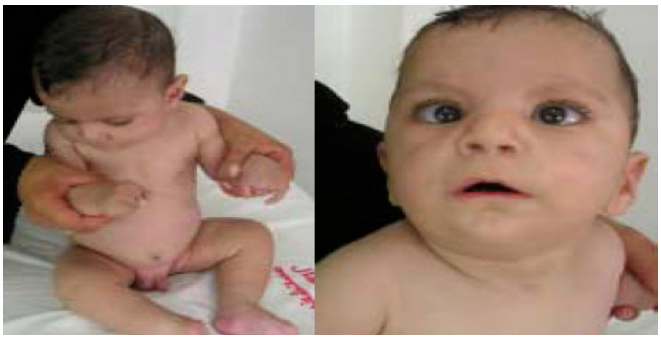
Skeletal abnormalities:

Chest: No Poland anomaly.
Hands: (Figure 1) bilaterally affected, total absent of the terminal phalanges of right hand with preservation of the thumb only, whereas the left hand total absence of digits and sparing only the thumb, and the middle phalange of the 5th finger.
Feet: bilateral total aplasia of the digits.
Pelvic bones: normal.
Spinal column: normal.
Genetalia: normal
Abdominal ultrasound: normal.

Investigations: Chromosomal study, metabolic screening, and all basic hematological tests revealed normal results.

Family history: nothing of significance apart from the primary sterility of the mother, no history of similar condition, in both maternal and paternal sides.

Figure 1



A **B**

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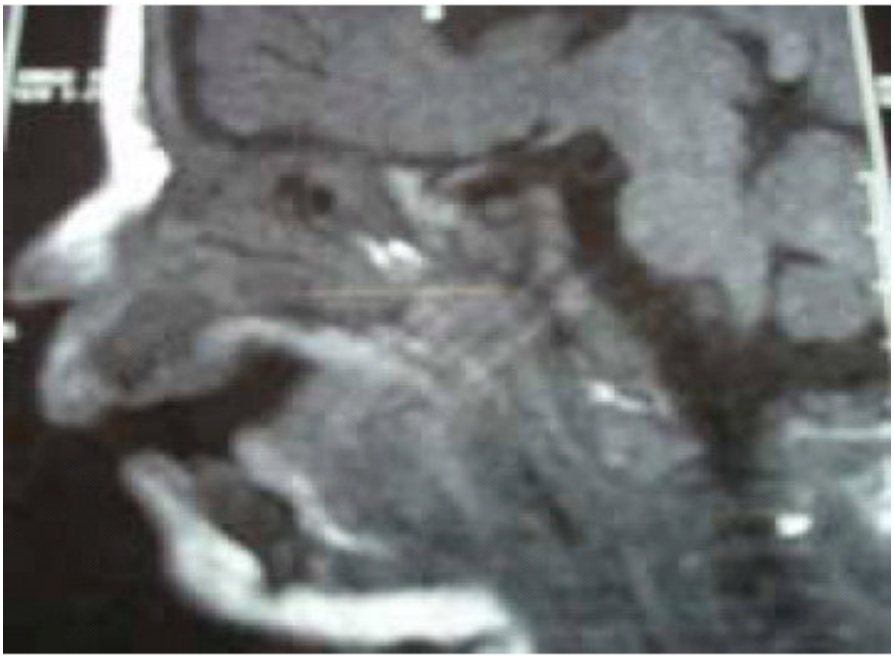
A. Male child with the bilateral hands and foot adactylia, left facial nerve palsy and bilateral abducent nerve palsies (defective ocular rotation), note the depressed inferior lower lip because of the absent tongue and oligodontia.

B. Note expressionless face, the unilateral facial nerve palsy (the deviation angle is on the right side), and the inability to execute horizontal eye movements.

Imaging Findings

Figure 2: MRI Saggittal section showed the absent tongue, with only minute trace of it.



DISCUSSION

Hall[3] called attention to the number of ambiguous and the significant overlapping entities that exist in the literature on this subject. He proposed a classification of syndromes with Oromandibular limb hypogenesis (OMLH) based on the presence of hypoglossia. It should be noted that mild degrees of hypoglossia are difficult to detect and may, in fact go unnoticed.

There is an overlap and similarities between different syndromic entities among the similarities with OHLH,

including long list of syndromes; Moebius syndrome, hypoglossiahypodactylia syndrome, Hanhart syndrome, glossopalatine ankylosis syndrome, limb deficiency-splenogonadal fusion syndrome, and Charlie M syndrome[4]. All are very uncommon except the Moebius syndrome.

Kaplan et al., (1976) considered this group of disorders as a syndrome community in which the overlapping phenotypes of the define syndromes reflect underlying developmental relationships.
Cohen et al., (1971)[6] reported a patient with minute tongue, limb deficiency, and Moebius involvement, they suggested that hypoglossia-hypodactylia syndrome, Hanhart syndrome, and glossopalatine ankylosis syndrome might represent the same recurrent pattern syndrome.

Hermann et al., (1976)[7] analysed 7 personally studied cases and 62 previously reported cases of Oromandibular –limb hypogenesis. They found that severity of upper limb involvement and especially malformation of the feet, but not the presence of cranial nerve palsies, was significant in differentiating cases, and the group of patients with cranial nerve palsies included some with limb defects similar to those observed in Hanhart syndrome and others with Poland anomaly, finally, cases with cranial nerve palsies without limb involvement were documented. Most reported cases of aglossia – adactylia "ankylosing superior", and Moebius syndrome represented instances of Hanhart syndrome, besides most cases of Moebius syndrome combining a chest defect and /or symbrachydactyly represented Poland/Moebius syndrome[7]. And the cranial nerve palsies obviously occurred in several etiologically distinct conditions.

Temtamy and McKusick (1978)[9] recognised three clinical entities within the Moebius syndrome category: 1) Sixth and seventh cranial nerve palsy alone. 2) sixth and seventh nerve palsy with limb anomalies, and 3) sixth and seventh nerve palsy with arthrogryposis.

Gillerot et al., (1991)[10] reported a case with hydrocephalus secondary to aqueduct stenosis. There was an "intense fibroblastic proliferation of meningeal tissue around the brainstem" with "small vessels penetrating deeply into the parenchyma inducing an astroglial reactional proliferation and ending in a necrotic calcified zone, in our patient no associated hydrocephaly was noted.

Dunham and Austin (1990)[11] reported a black male infant presented with Aglossia-situs inversus, and discussed two further cases from the literature, in our patient the abdominal ultrasound revealed normal visceral components.

Our impression that our patient's malformation complex which is composed of, "Aglossiaadactylia", and the sixth and seventh cranial nerve palsies, all these constellation of manifestation are strongly suggesting the second categorisation of Temtamy and McKusick (1978)[7], we are focussing in this study on the importance of Imaging studies to further clarify the disorder, however, children with small tongues can escape the diagnosis, moreover, our hypothesis that the malformational complex in this child probably resulted from a disruptive sequence, in addition the family were under two risks, firstly the consanguinity, and secondly the primary sterility of the mother, and with the birth of this child the factor risk became higher.

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