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CRANIOSYNOSTOSIS: ACROCEPHALOSYNDACTYLY (APERT SYNDROME) DIAGNOSED IN A NEWBORN

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ABSTRACT

We report a 10 days old newborn with brachycephaly, midfacial hypoplasia, syndactyly and broad distal phalanx of thumb and big toe. At the 20th gestational weeks an enlargement of the left cerebral ventricle and malformation of the fingers of the hands and toes were noticed on a regular ultrasound examination. The aforementioned malformations were observed at birth and at the age of 11 months. The large fontal was closed; the small one was palpable at the tip of the finger. Brachycephaly was evident with high full forehead, flat occiput, and irregular craniosynostosis especially at the coronal suture. Cutaneous syndactyly was present at both hands (fingers II-V), with almost complete fusion of the second, third and fourth fingers. Distal phalanges of the thumbs were broad as well as distal hallux. There was cutaneous syndactyly of the feet. Mental development at the age of 11 months was normal.
Apert syndrome is a sporadic disorder. Rarely, inheritance is autosomal dominant. Appropriate management includes surgical treatment of the syndactylies, follow up of the eventual airway compromise and hearing difficulties. This is a report of a patient identified as a newborn.

Keywords: cranyosynostosis, syndroma Apert, newborn

INTRODUCTION

Apert syndrome is characterized primarily by craniosynostosis, midface hypoplasia, and syndactyly of the hands and feet. There is also a tendency to fusion of bony structures.

First described by Apert in 1906 [1] who compared the hand to spoon, or when thumbs were free to obstetric hand. Most of the patients are sporadic. Nevertheless an autosomal dominant inheritance was also reported.

We describe a patient recognized as Apert syndrome in his newborn period.

CASE REPORT

We report a 10 days old newborn with brachycephaly, midfacial hypoplasia, syndactyly and broad distal phalanx of thumb and big toe. The girl is the second child of young and healthy parents with uneventful pregnancy and delivery. At the 20th gestational weeks an enlargement of the left cerebral ventricle and malformation of the fingers of the hands and toes were noticed on a regular ultrasound examination. The pregnancy continued until vaginal delivery at 40th gestational week. At birth and at the age of 11 months the

aforementioned malformations were observed. The large fontal was closed; the small one was palpable at the tip of the finger. Brachycephaly was evident with high full forehead, flat occiput, and irregular craniosynostosis especially at the coronal suture. There was a supraorbital horizontal groove, hypertelorism with shallow orbites, down slanting palpebral fissures, small nose, and maxillary hypoplasia. Cutaneous syndactyly was present at both hands (fingers II-V), with almost complete fusion of the second, third and fourth fingers. Distal phalanges of the thumbs were broad as well as the distal hallux. There was cutaneous syndactyly of the feet. Publication of the images was declined. Mental development at the age of 11 months was normal. Height and weight were normal. No heart or kidney malformations were detected. X-rays of the skull was not done. MRI is pending, as well as the genetic analysis. Appropriate surgical and clinical follow-up is going on.

DISCUSSION

This is a rare case in which the diagnosis was done in the newborn period. Further follow-up only confirmed the diagnosis. The fusion of the cervical vertebrae was found in 68% of patients [2] with Apert syndrome. The c2-c3 fusions in Crouzon syndrome and C5-6 fusions in Apert syndrome could therefore be used to separate those entities. The severity of the syndactyly was also scored: type 1, the thumb and part of the fifth finger are separate from the syndactylous mass; in type 2, where the little finger is not separate; and type 3, where the thumb and all fingers are affected [3].

Mental retardation has rarely been reported [4]. Malformations of the corpus callosum, the limbic structures, have also been observed [5]. Early scull decompression improved the IQ [6]. Esophageal stenosis was also observed [7]. Several ocular abnormalities were also reported: 14% amblyopia, 60% strabismus, 19% anisometropia, and 34% of eyes had ametropia [8].

Mostly sporadic, Apert is rarely reported in families. Mother daughter [9, 10], mother and son with Apert syndrome were reported [11].

FGFR2 gene caused Apert syndrome [3, 12]. 64% of patients were found to have S252W P253R mutation, while 33% had P253R [13]. Postsurgical facial appearance was better in patients with P253R mutation [14]. Birth prevalence was estimated to be 12.4 per million live births [15].

In brief, this is a rare description of Apert syndrome diagnosed at birth.

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Резиме

CRANYOSINOSTOSIS: ACROCEPHALOSYNDACTILY (APERT SYNDROME) ДИЈАГНОСТИЦИРАН КАЈ НОВОРОДЕНЧЕ

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Прикажано е новороденче со карактеристики на Апертов синдром: брахицефалија, хипоплазија на средишниот дел на лицето, синдактилија и широки дистални фаланги на палецот на рацете и нозете. Ултрасонографски преглед во дваесеттата гестациска недела откри проширени летерални вентрикули. По раѓањето големата фонтанела беше затворена, а малата имаше дијаметар на мал прст. Краниосиностозата беше особено изразена на коронарната сутура. Кутаната синдактилија се наоѓаше меѓу вториот и четвртиот прст, речиси со полна фузија на вториот, третиот и четвртиот прст. Дисталните фалангуи на палците беа широки, а постоеше и кутана синдактилија на стапалата. На 11 месеци менталниот развој беше нормален.

Апертовиот синдром е спорадичен, иако ретко се опишани фамилијарни случаи. Хируршки третман на синдактилијата, поретко краниотомија, и следење на нарушувањата на слухот и дишните патишта се задолжителни. Ова е редок приказ на дијагноза на новороденче на Апертов синдром.

Клучни зборови: краниосиностоза, Апертов синдром, новороденче