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Acrocephalosyndactyly, Apert type, in a newborn: Cerebral sonography

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Apert syndrome;
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Abstract We describe the clinical and cerebral ultrasonographic features of a rare case of type 1 acrocephalosyndactyly (Apert syndrome). The patient was a newborn male whose twin had died *in utero*. Most cases of Apert syndrome are sporadic, although autosomal dominant inheritance has also been reported. Diagnosis is based on physical examination together with imaging data. Since Apert syndrome can give rise to numerous CNS abnormalities, affected newborns should undergo echoencephalography for more complete characterization of their malformations.

Sommario Vengono descritti il caso clinico e le caratteristiche ecografiche cerebrali di un gemello (in gravidanza con gemello premorto) affetto da acrocefalosindattilia, Tipo Apert, quadro malformativo complesso e raro con implicazioni genetiche, appartenente al gruppo delle disostosi cranio-facciali. La diagnosi si basa principalmente sull'esame clinico e sulle indagini per imaging. Poiché la sindrome di Apert è accompagnata da diverse anomalie del Sistema Nervoso Centrale, è necessario e utile prevedere una indagine ecografica transfontanellare in epoca neonatale.

Introduction

The first case of Apert syndrome was reported exactly 100 years ago in the Bulletin of the Medical Society of Paris. The author was the French physician afterwhom the syndrome is named [1]. Apert syndrome, which is also known as type 1 acrocephalosyndactyly (ACS1 — OMIM 101200), is a rare genetic disorder characterized by multiple malformations. It affects 15.5 out of every 1,000,000 live births [2]. Although many cases are sporadic, others appear to result from the inheritance of an autosomal dominant trait linked to advanced paternal age. The molecular bases of the syndrome are still obscure.

Clinically speaking, the syndrome is characterized by brachycephaly or turricephaly, hypoplasia of the facial mass, exophthalmos, hypertelorism, canthal dystopia, hypoplasia of the upper airway cavities and ethmoid, cleft palate, cutaneous and/or bony syndactyly of the hands and feet. The cerebral anomalies described in the literature include agenesis of the corpus callosum (CC), progressive

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and nonprogressive hydrocephalus, and hippocampal abnormalities. Most of the patients are mentally retarded [2,3].

In this report, we describe the sonographic features of the cerebral structures in a newborn with Apert syndrome. The syndrome can be diagnosed prenatally, especially if 3D sonography is used, but there are very few descriptions of findings obtained with transfontanellar sonography [4].

Case 1

A male infant was transferred to our unit shortly after normal vaginal delivery in another hospital. The gestational age was 40 weeks, and the birth weight was 4300 g. The mother was 32 years old, and the father was 29. A previous pregnancy had ended in abortion, and the present pregnancy had been complicated by the death *in utero* of the patient's twin. The 1- and 5-min APGAR scores were 9 and 8. Multiple malformations were evident at birth, including turricephaly, frontal bossing, wide anterior fontanel, flattened face, hypertelorism with down-slanting palpebral fissures, bulging eyes, micrognathia (Fig. 1), high arched palate, cleft palate, cutaneous syndactyly of the hands and feet with deformity of the great toes (Fig. 2), and hypotonic extremities. On transfontanellar sonography all components of the ventricular system were present. The midline structures were characterized by extreme craniocaudal elongation, and the lateral ventricles appeared mildly enlarged and square-shaped on anterior coronal scans. The anterior and middle portions of the CC appeared normal whereas the posterior CC was underdeveloped with sulci arranged in a radial pattern (Figs. 3 and 4). A small cyst was noted at the level of the right anterior plexus. The 2D US study was supplemented with color Doppler studies of the major cerebral arteries (anterior cerebral, middle cerebral, internal carotid) to identify any signs of increased intracranial pressure. Qualitative and quantitative analysis of the flow curve revealed anterograde systodiastolic flow and a normal Resistance Index (0.70) for the anterior cerebral artery.

Skeletal films revealed craniofacial dysostosis characterized by a tower-shaped skull, synostosis of multiple sutures, signs of intracranial hypertension, and hypoplasia of the facial mass. The metacarpal bones of the feet were shortened, and the first metatarsal was larger than normal and bifid. When present, the first ray consisted exclusively in a large, stubby bone element. The bones of the hands did not appear to be fused, but the phalanges were markedly shortened. Computed tomography of the brain with 3D-VR reconstruction confirmed the presence of brachy/turricephaly caused by premature fusion of the coronary sutures and partial lambdoid synostosis. The sagittal and metopic sutures appeared to be widened (Fig. 5). The orbital cavities were smaller than normal, and the roofs and lateral walls were elevated due to anterior displacement and ''frontalization'' of the greater wing of the sphenoid. The result was bilateral proptosis, hypertelorism, and partial stenosis of both optic canals. Cerebral MRI without contrast enhancement was performed along with MRI angiographic studies of the venous structures. These examinations confirmed the presence of cranioencephalic dysmorphism with decreased anteroposterior and interparietal diameters. They also revealed: mild cerebellar hypoplasia with signs of lamina quadrigemina hypertrophy; elongation of both optic nerves and of the hypophyseal peduncle caused by deformity of the base of the skull (reduction of the angle formed by the ethmoid—sphenoid plane and the posterior surface of the *clivus*), hypoplasia of the CC and absence of the splenium in the posterior portion, dysmorphism of the dural sinuses and signs of hypertrophy and

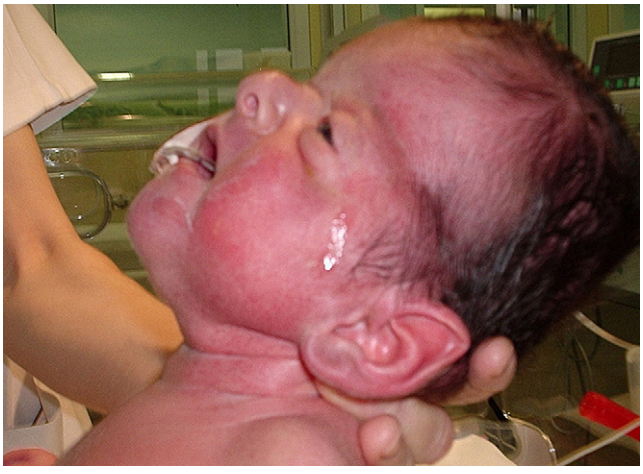

Fig. 1 Typical Apert syndrome facies: high, broad, flat forehead; hypertelorism; depressed nasal bridge; down-slanting palpebral fissures; bulging eyes; open mouth; low-set ears.

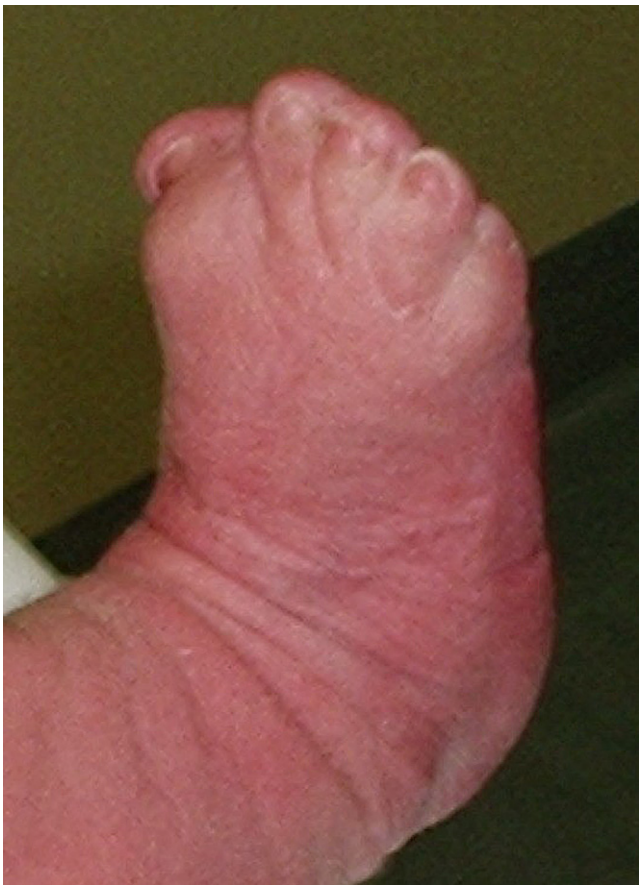

Fig. 2 Complete syndactyly involving all five digits of the right foot; the great toe is also enlarged.