Johanson-Blizzard syndrome

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Keywords: congenital abnormalities, craniofacial abnormalities, otitis media with effusion, hearing loss.

INTRODUCTION AND LITERATURE REVIEW

The Johanson-Blizzard Syndrome (JBS) is hereditary autosomal recessive. It bears ectodermal dysplasia, endocrine and exocrine failure, and there can be mental and development failure. It is marked by nasal wing hypoplasia or aplasia and dental abnormalities1. There may be prenatal growth failure; sensorineural hearing loss; hypotonia; microcephaly; skull mid-line defects; less hair or frontal toupee; nasolacrimal fistula; cleft palate; hypothyroidism; pancrætic failure; malabsorption. First described in 1971 by Johanson & Blizzard2. There were 51 cases reported in the literature by 2004.

CASE DESCRIPTION

A three-year old Caucasian female, with development retardation. Born at the 8th month of gestation, out of a C-section, she was discharged from the hospital after three days. At four months of age she could not support her own head, when she was submitted to surgery to correct craniostenosis. She managed her own head, when she was submitted to surgery to correct craniostenosis. She managed her own head. She was unable to hear. She could stay standing with support but could not walk. She had undergone an uneventful gestation. PHYSICAL EXAM: hypertelorism; thin nose; micrognathia; global hypotonia and ligament laxity; level V muscle strength in the four limbs; myotactic hyporeflexia; primary hypothyroidism; pancreatic failure with malabsorption. First described in 1971 by Johanson & Blizzard2. There were 51 cases reported in the literature by 2004.

DISCUSSION

The cases considered JBS, have a variety of clinical and anatomical findings differing a little from the original description. In this patient there is no hypothyroidism - present in 1/3 of the cases3,4 and pancreatic failure was not investigated because she did not have symptoms which would justify it. There are many cases in which pancreatic failure was not mentioned because the diagnosis was intrauterine, or because the focus was the radiological study of the temporal bone4. There are cases of siblings with JBS and consanguinity is considered a risk factor, and there are doubts concerning the recessive autosomal inheritance. In most of the cases there was no genetic investigation and there are well defined cases with normal caryotype5.

EEC malformation in this syndrome had not been described. It was only in one case that there was a CT scan in which there was bilateral cystic dilatation of the cochlea and vestibule. There are no reports of secretory otitis media, which was possible upon CT scan. The malformation did not allow for otoscopy and tympanometry.

FINAL REMARKS

Early diagnosis is important because of pancreatic failure and hypothyroidism, which can be the main risk factors for mental retardation5. In the prenatal ultrasound scan, the sigmoid was dilated and there was nasal wing aplasia (beak-shaped nose), which may represent the earlier diagnosis in JBS, especially in gestations of consanguineous couples1. In relation to sensorineural hearing loss, one must consider rehabilitation with a personal hearing amplification device and cochlear implant. As to conduction hearing loss, clinical treatment must be considered.

REFERENCES


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Paper submitted to the BJORL-SGP (Publishing Management System – Brazilian Journal of Otorhinolaryngology) on July 24, 2006; and accepted on May 15, 2010. cod. 3295