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Case Report

Orodental manifestations in cases with partial agenesis of corpus callosum-rare phenomena

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ARTICLE INFO

Article history:
Received 22 April 2014
Accepted 18 April 2015
Available online 6 June 2015

Keywords:
Corpus callosum malformation
Mental retardation
Dysmorphic facial features
Multiple structural abnormalities

ABSTRACT

This article focuses on the associated signs and symptoms of patients with partial agenesis of the corpus callosum. The orodental manifestations of such cases have been given special weightage which will prove to be of great help to oral physician when encountered with such cases.

Case details: Two siblings, aged 14 and 16 years, reported with a chief complaint of severe crowding of teeth with mouth breathing habit. They were low birth-weight babies and had been born to non-consanguinous parents. The distinguishing features of these children were craniofacial abnormalities, delayed developmental milestones, mild mental retardation and abnormal gait. The nosological features and the clinical manifestations of this syndrome and the plausible autosomal recessive inheritance of this rare syndrome have been elicited. The diagnosis was based on characteristic phenotype, in particular striking craniofacial and skeletal abnormalities and neuroimaging.

Conclusion: It is a challenge for healthcare professionals to help these youths to maximize their potential as human beings and help them achieve a meaningful adulthood. On the other hand, diagnosing such cases can be a challenge to dentistry. A systematic protocol, if adhered, can lead to a more appropriate diagnosis. Managing such cases in a clinical setup involves a multispeciality and interdisciplinary approach.

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http://dx.doi.org/10.1016/j.jobcr.2015.04.003

1. Article focus:
   - This article focuses on the associated signs and symptoms of patients with partial agenesis of the corpus callosum.
   - This syndrome involves a multidisciplinary approach to managing such cases.
   - The orodental manifestations of such cases have been given special weightage in this article which will prove to be of great help to oral physician when encountered with such cases.
2. Key messages:
   - Diagnosing such cases can be a challenge to dentistry.
   - A systematic protocol, if adhered, can lead to a more appropriate diagnosis.
   - Managing such cases in a clinical setup involves a multispeciality and interdisciplinary approach.
3. Strengths and Limitations:

A detailed workup had been done for this article including FISH and chromosomal analysis despite financial constraints.

1. Introduction

Acrocallosal syndrome is an atypical congenital disorder which was first described by Albert Schinzel in 1979 and may also be referred to as 'Schinzel Acrocallosal syndrome'.[1] The siblings were rare cases of physical and behavioral phenotypes where chromosomal analysis showed normal karyotyping and no interstitial deletions. However, the anatomical and radiological findings closely supported the diagnosis of: "ACROCALLOSAL SYNDROME".

2. Case report

Two siblings, aged 16 and 14 years respectively, had reported to the clinic with a chief complaint of mouth breathing habit and severe crowding of the upper and lower front teeth. They were born at term by normal delivery and history regarding any maternal illness, drug intake or exposure to radiation during antenatal and prenatal period was negative. Both were low birth weight babies; the boy weighed 2200 gm and girl, 2000 gm respectively. After 3 months of age, the mother noticed that both the children had abnormal facies, with delayed developmental milestones like walking and talking, hypophonia and difficulty in keeping up with their peers in school. She also felt pulsations on the top of the head of the male child even after 2 years of age. The respondent also reported extraction of the upper front teeth in the male child by a local dentist in an attempt to relieve crowding of teeth.

Clinical appraisal of both the siblings revealed the following: (Fig. 1)

- Physical and mental growth retardation with delayed developmental and skeletal milestones. Parameters like height- 139 cm (normal: 160 cm), weight- 23.49 gm (normal: 51.28 kg) were falling below the 3rd percentile.
- Neurological examination revealed borderline mental retardation, moody and irritable behavior. There was scissoring gait due to hypotonicity of the adductor muscles of the lower limbs.
- Urinary bladder and bowel control not achieved.
- No abnormalities were detected in the other systems.
- SMR (Tanner's sexual maturity rate) was 3 with micropenis.
- Anthropometric measurements showed short stature, dolichocephaly (head circumference- 47 cm (normal: 52 cm), frontal bossing and triple hair whorls.
- Limb abnormalities: Dermatoglyphics revealed simian crease in both the cases and clinodactyly of the fifth finger in the girl.
- Pedigree chart analysis revealed that their first cousin had similar features and had died at 6 years of age. (Fig. 2)

Fig. 1 – Anthropometry measurements (NCHC).

The patients exhibited characteristic dysmorphic features like hypertelorism, strabismus, upward slanting and narrow palpebral fissures (with mongoloid slant in the girl) of the eyes. They had broad nasal bridge, short philtrum, incompetency of lips, posteriorly angulated malformed ears and prominent occiput. Malar prominences and mandible were hypoplastic with convex profile. Intra oral examination revealed normal mouth opening, crowding of the upper and lower anterior teeth, anterior open bite, high arched palate and missing 11 and 21. (Fig. 3) Differential diagnosis thought of were Aicardi, Andermann, Shapiro, Greig's