The Journal of Medical Research

Case Report

JMR 2016; 2(4): 91-93
July- August
ISSN: 2395-7565

www.medicinearticle.com

Pai syndrome without cleft lip. A variation in the expression of the syndrome: A case report

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Abstract

Pai syndrome is a rare disorder that includes midline cleft lip, pericallosal lipoma and cutaneous polyp of the face. It is a rare form of frontonasal dysplasia, first described in 1987. Only few cases have been reported in the literature. We report a case of perinatal diagnosis using cranial ultrasound. The aim of this paper is to describe the clinical and imaging features of a further patient with Pai syndrome.

Keywords: Pai syndrome, Pericallosal lipoma, Corpus callosum agenesis, Congenital nasal polyp, cleft upper lip.

INTRODUCTION

Frontonasal dysplasia (FND) includes a variety of craniofacial defects that affect midline structures of the head and face [1]. FND is etiologically heterogeneous and various subsets are known. Pai syndrome (PS) is one subset [2]. In 1987, Pai described for the first time an unusual form of syndromic FND consisting of three rare developmental anomalies in a male newborn: complete median cleft of upper lip, two cutaneous polyp tags over the nasal septum, and midline lipomas of the central nervous system with normal neuropsychological development [3-7]. Similar findings had been reported earlier by Nakamura in 1984 [8], Ponniah in 1977 [9] and Sharma in 1974 [10]. The cause of Pai syndrome is unknown to date [2] and the phenotype of this rare syndrome is clinically variable [2, 4]. Only few cases have been reported in the literature [6]. The aim of this paper is to describe the clinical and imaging features of a further patient with Pai syndrome.

CASE REPORT

This newborn female patient was born at term by vaginal delivery. Her birth weight was 3.130 kg, length 48.5 cm and head circumference 33 cm. Her parents were unrelated. The Apgar score was 9 at 1 min. She is the first child of nonconsanguineous healthy parents, and there was no family history of neurologic disorders or facial dysmorphism. Mother with history of gestational diabetes mellitus developed in the second half of pregnancy. No other diseases or complications were experienced during pregnancy and no exposure to X-rays or teratogens were described.

The baby was admitted into our hospital with right-side nasal obstruction by an approximately 1 cm bilobulated mass covered with normal skin that protruded from the right nostril (Fig.1), arising from the nasal septum and that was evident immediately after birth.

The vestibular frenum was not involved and the palate appeared normal, just a minimal median alveolar defect (cleft) was seen. Right nasolacrimal duct obstruction also was demonstrated. A superficial plane hemangioma on the forehead midline and left superior eyelid was perceived some weeks later.

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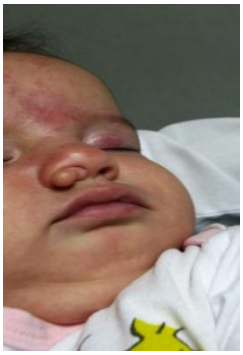

Figure 1: Right nasal polyp

The rest of the physical examination, included neurological exam, was unremarkable and excluded the presence of any further malformation. Fundus oculi and electroencephalogram were normal. Full blood count, erythrocyte sedimentation rate and biochemical parameters were determined as normal in the laboratory tests. Further analysis excluded other visceral abnormalities.

An ultrasound scan of the brain revealed an interhemispheric midline hyperechogenic mass suggesting pericallosal lipoma and agenesis of the corpus callosum.

Computed tomography (CT) scan showed an interhemispheric lipoma-like mass with little calcification (Fig. 2 a) and the point of insertion of the mass on the right anterior section of the nasal septum (Fig. 2 b). The brain CT also revealed colpocephaly and elevation in the third ventricle reflecting corpus callosum agenesis.

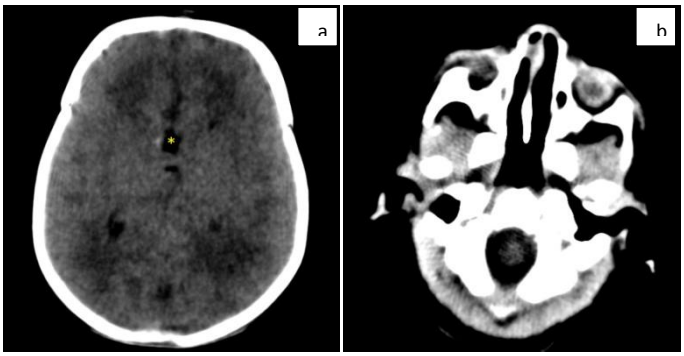

Figure 2: CT. Interhemispheric lipoma-like mass (asterisk) with minimal right peripheral calcification (a). Point of polyp insertion to the right side of the nasal septum (b).

Magnetic resonance imaging (MRI) similarly demonstrated corpus callosum agenesis with an interhemispheric tubulonodular lipoma (high-intensity in T1-weighted and FLAIR images, low intensity in T2-weighted images) and right nasal pedunculated polyp (Fig. 3).

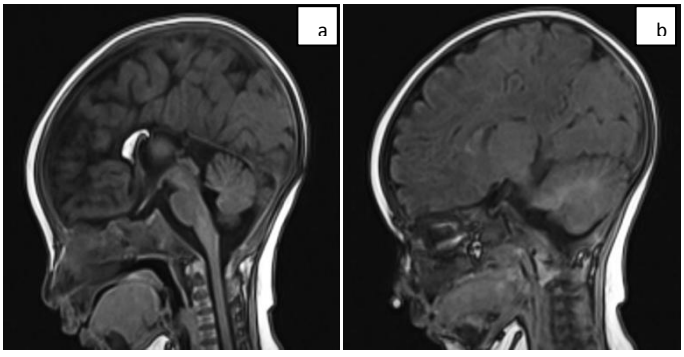

Figure 3: Sagittal T1-weighted MRI shows a pericallosal lipoma, tubulonodular type, appearing as thick hyper-intense band and agenesis of the corpus callosum (a) and a fat-isointense nasal polyp (b).

The baby was monitored for 10 months before surgery showing normal neuropsychological development. At this age, under general anesthesia, the patient underwent a surgical operation to remove the mass. Minimal plasty of the columella and nasal mass excision were performed. There were no intra or postoperative complications. The final aesthetic appearance was excellent with no hypertrophic scarring.

Histopathology of the excised nasal mass revealed mature adipose tissue and normal skin structure.

DISCUSSION

Although there are many other accompanying findings [2,4,6,7], CNS lipomas, median cleft of the upper lip and facial skin polyps form the classic triad to define Pai syndrome [13]. A median cleft lip is a frequent finding in the previously reported Pai syndrome cases in the literature [3,6,13], but, it was not present in our case, Ocak [14], in a similar way, had reported another case of PS without cleft lip. Analyzing the features of all patients with PS, a specific clinical picture and a certain phenotypic variability are revealed [4]. Guion-Almeida et al. [2] reported seven patients with Pai syndrome with phenotype clinically variable. We suggest, as Savasta [7], that malformations seen in our and others' patients support the thought that some variation occur in the expression of this syndrome.

Only few cases have been previously described in literature [3-7,11-13], however, the incidence seems to be underestimated [13]. According to Guion Almeida [2,13] PS is one of the different syndromes described within the FND and suggests that the phenotypic manifestations encompass a continuum, manifested by variable midline anomalies within the FND spectrum and nasal/facial appendages. These phenotypic variability of PS produces diagnostic difficulties and suggests under-reporting [4,6].

Although different CNS and facial skin lesions have been described, it seems that the crucial clinical feature of this syndrome is the midline cleft [4]. In this case there is just a minimal alveolar fissure of scarcely two millimeters. For others authors [11,12], the congenital nasal polyp appears as the main marker of the PS, isolated it is exceptional. It is seemingly a lateral defect, but most originates of the nasal septum that is a structure of the midline or of the columella. Another prominent feature of the PS is the polyp of the oral cavity that originates in the midline of the superior alveolar process, maxillary frenum or gingival surface of the superior lip. According to these authors [11,12] the facial fissure of midline is very common, but not constant (81% of communicated cases) and another frequent discovery is the lipoma of the corpus callosum (85%).

Although a minimum (alveolar) manifestation of the midline cleft was present, our patient did not have a median cleft of the upper lip, which is one of the characteristics of PS, but other criteria were applied (i.e., nasal skin mass, pericallosal lipoma and corpus callosum agenesis) so the authors interpreted the case as a variation in the expression of PS. On the other hand, this baby girl had an associated superficial plane hemangioma on the midline of the forehead and the left superior eyelid. This finding has not been reported among the patients affected by PS.

CNS lipomas are rare [1,4] and account for less than 0.4% of all CNS masses [4]. Patients with CNS lipomas usually present with seizures, but those with PS or FND associated with CNS lipomas do not; this is a constant feature in previous reports [4,7]. In the case presented here, an intracranial tubulonodular lipoma was placed in the area of the genu of corpus callosum.

On ultrasound a lipoma of the corpus callosum appears as a highly echogenic mass in the middle of the interhemispheric fissure. The mass

is hypodense (fat) on CT and minimal calcified outlying components of the interhemispheric lipoma can be observed (Fig. 2). On MRI, intracranial lipomas present as fat-isointense, subarachnoid lesions. They are distinctly hyperintense in T1-weighted sequences (Fig. 3) and typically hypointense in a fat-suppressed serie. On the other hand, in patients with PS the corpus callosum can show variable involvement, ranging from slight anomalies to complete agenesis [15].

In this case, cranial ultrasound led the diagnosis, the association of the ultrasound's findings and the physical exam made suspect the diagnosis.

Long-term prognosis of these patients is excellent with normal neuropsychological development [3,7] We have been following our patient for more than a year and neuropsychological development is normal too.

CONCLUSIONS

Our case seemed to be a variety of Pai syndrome. We recommend imaging of the central nervous system in all the newborn patients with a congenital polyp located in the craniofacial midline to rule out central nervous system involvement. The incidence of PS seems to be underestimated and the diagnostic criteria can be revised. A multidisciplinary approach is the key to successful outcome.

Conflict of interests

The authors have none to declare

Informed consent

Was obtained from the parents.

Author contributions: Case management – I.S.,S.B.,V.F.,C.A.; Patient follow-up – I.S.,S.B.,V.F.; Literature Search – V.F.,L.V., I.O.; Writing – V.F.; Critical Reviews – V.F., L.V. All authors approved the final version of the manuscript.

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