

Pericallosal lipoma associated with Pai syndrome: prenatal imaging findings

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ABSTRACT

Pai syndrome is a rare disorder characterized by the association of a midline pericallosal lipoma, median cleft (palate or lip) and cutaneous polyps of the face. Only seven cases have been reported in the medical literature and we are not aware of any previously reported prenatally detected cases. In this article, we present the prenatal ultrasound and magnetic resonance imaging findings of a syndromic pericallosal lipoma with associated anomalies that led to the prenatal diagnosis of Pai syndrome. We underline the impact on parental counseling following prenatal detection of pericallosal lipoma. Copyright © 2008 ISUOG. Published by John Wiley & Sons, Ltd.

CASE REPORT

A 31-year-old woman, gravida 1 para 0, was referred for evaluation at 26 weeks' gestation because cleft palate and cutaneous nasal and frontal appendages had been detected on routine ultrasound examination. Amniocentesis was performed and the karyotype was found to be normal (46,XX). There was no history of drug ingestion or X-ray exposure during pregnancy. No family history of cleft (palate or lip) or other congenital abnormalities was noted.

Two- and three-dimensional ultrasound examination (Siemens Acuson Antares ultrasound system, Erlangen, Germany) was performed, which showed a bilateral cleft of the upper lip, with a median bud but without significant alveolar or palate extension. We confirmed the presence of three nasal and frontal cutaneous polyps and a slight median indentation of the nose tip suggestive of bifid nose

(Figure 1). Examination of the cerebral structures showed a hyperechogenic pericallosal structure above the anterior and median part of the corpus callosum, highly suggestive of tubulonodular interhemispheric lipoma and possible mild hypoplasia of the splenium (Figure 2a). Fetal magnetic resonance imaging (MRI) confirmed the fatty nature of the lipoma (Figure 2b). Examination of the ventricles, basal ganglia and white matter showed no other abnormalities, and gyration was normal for gestational age. The association of such facial abnormalities with frontonasal polyps, labial cleft and pericallosal lipoma were suggestive of Pai syndrome. The couple were informed of the potentially good cognitive prognosis of interhemispheric lipoma associated with Pai syndrome, as reported in the few published cases.

A 3900-g female infant with a normal Apgar score was delivered vaginally at 40 weeks' gestation after induction of labor. Neonatal physical examination confirmed the facial abnormalities, with good correlation with the three-dimensional ultrasound images (Figure 3a and b). The median cleft of the upper lip was confirmed without any alveolar or palate extension. However, the lip bud seen on ultrasound was actually a fourth pediculated skin appendage. Cerebral ultrasonography and MRI confirmed the prenatal findings, and showed a tubulonodular anterior pericallosal lipoma associated with mild underdevelopment of the distal part of the splenium (Figure 2c).

The median labial appendage was removed at birth to ease feeding, and complete facial surgery was carried out at 3 months of age, with excellent cosmetic results (Figure 3c). Histological examination of the four skin appendages revealed them to be benign fibrofatty epithelial polypoid molluscums. Psychomotor

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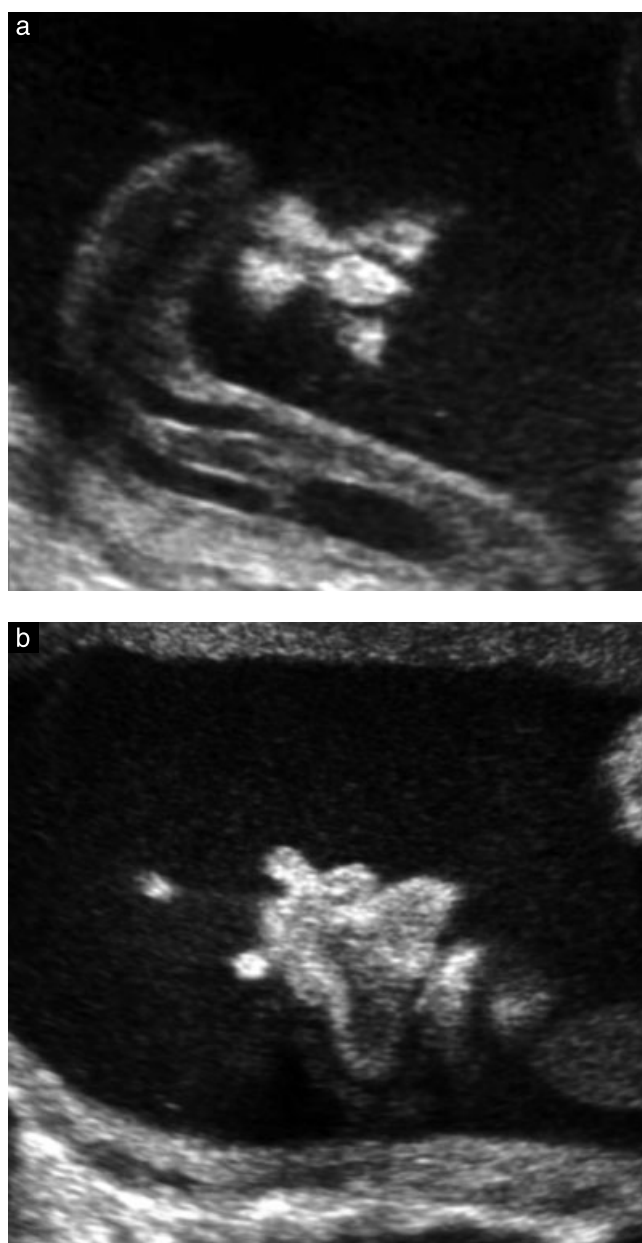


Figure 1 Two-dimensional coronal ultrasound images of the fetal face showing a median bud of the upper lip, a slight median indentation of the nose tip suggestive of bifid nose (a), and nasal and frontal cutaneous polyps (b).

development and neurological status were normal at the 14-month follow-up.

DISCUSSION

Our case illustrates a syndromic pericallosal lipoma, in which the association with specific facial abnormalities was highly suggestive of Pai syndrome.

Intracranial lipomas are rare lesions that represent less than 0.1% of intracranial lesions. Most of these occur at the midline, mostly in the pericallosal cistern^{1–3}. According to Macpherson *et al.*, more than half of midline lipomas are associated with brain malformations of varying degrees, especially midline anomalies⁴. Pericallosal lipomas can interfere with the normal growth of the

corpus callosum between the 11th and 20th weeks, leading to developmental anomalies such as complete/partial callosal agenesis or hypoplasia. The degree of anomaly seems to be in relation to the size and location of the lipoma^{2,5}.

Two morphological types of pericallosal lipoma have been described on the basis of MRI findings^{1,2}. One is a tubulonodular type, appearing round and measuring > 2 cm, and the second type is curvilinear, described as thin, elongated lipoma, measuring < 2 cm in diameter and usually more posterior. Tubulonodular lipoma is much more frequent than curvilinear lipoma. However, Demaerel *et al.* suggested that interhemispheric lipoma can be considered as a unique entity, with variable expression depending on the time and severity of the insult¹. The tubulonodular subtype is most likely the result of a more severe and earlier insult than the curvilinear subtype, and is therefore commonly associated with other cerebral, especially callosal, and frontofacial abnormalities.

With the increasing use of obstetric sonography, a few cases of pericallosal lipoma have been detected *in utero*. Ickowitz *et al.* reported the largest series of pericallosal lipoma (seven cases)⁶. The echogenicity of the lipoma was most often similar to that of parietal bone. The margins were smooth in five patients and irregular in the two patients with the largest lipomas. Extension of the lipoma toward the frontal lobes was visible in two patients and toward the choroid plexuses in another. Ultrasonography was used to correctly establish the extent of the callosal-associated anomaly in all cases except one; fetal MRI confirmed the fatty content of the lipoma in this case and was more effective than sonography in showing its extension and the type of callosal anomaly. The clinical evolution of patients with pericallosal lipoma was normal in all prenatally detected cases, with the limitation of a short follow-up⁶. Indeed, the neurological status seems to be variable and related to the age of the patient, the type of lipoma and the associated anomalies¹. In cases with tubulonodular lipomas, symptoms were encountered mostly in those with the associated dysmorphic face.

In our case, the pericallosal lipoma was an anterior tubulonodular type associated with facial abnormalities, which could have been of concern regarding the postnatal neurological evolution. However, the associated anomalies were suggestive of Pai syndrome, which led to continuation of the pregnancy according to the clinical evolution reported in this syndromic entity.

In its original description, Pai syndrome is characterized by the rare association of median cleft, cutaneous polyps of the face, nasal mucosa and midline pericallosal lipoma⁷. Facial abnormalities in Pai syndrome may be variable, although the median location of cleft palate or lip is a specific feature. Recently, Szeto *et al.* reported a new case of Pai syndrome and reviewed the six previously published cases⁸. All patients showed an interhemispheric lipoma, mainly of the tubulonodular type. A median cleft palate, involving differing stages of primary palate disorder, was encountered in all but one case. Nasofrontal median or paramedian skin appendages were present in all cases.

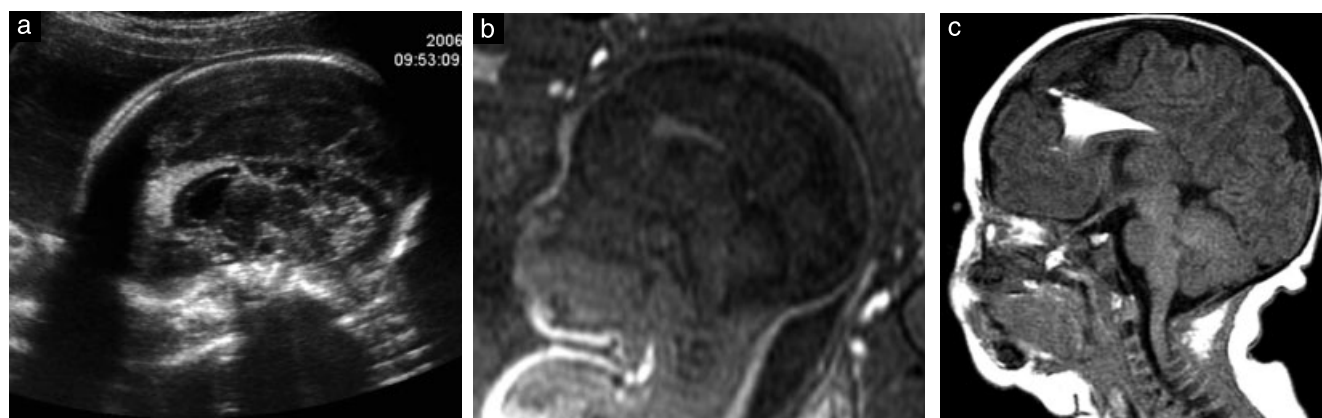


Figure 2 Fetal ultrasound (a) and magnetic resonance (b) mid-sagittal images showing slight splenium hypoplasia and hyperechogenic pericallosal structure with hyperintensity on T1-weighted imaging suggestive of tubulonodular lipoma. (c) Postnatal T1-weighted magnetic resonance image of the midline confirming pericallosal lipoma with splenium hypoplasia.

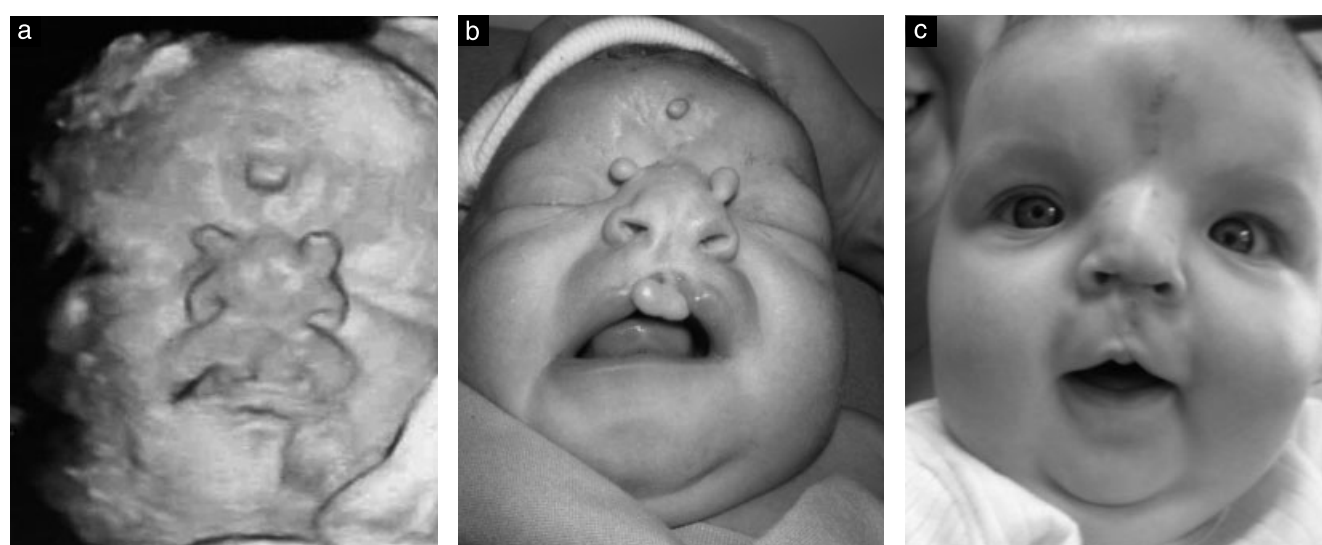


Figure 3 Prenatal three-dimensional ultrasound image (a) and correlation with the clinical aspect at delivery (b). (c) Postoperative result 3 months after delivery.

Our case showed all of the characteristic features of this syndrome including a bifid nose, which was also reported in one case in the review. Interestingly, nasal appendages were also reported as associated findings in one case of the series reported by Ickowitz *et al.* (Case 7), which could also suggest the phenotype of Pai syndrome⁶.

In the series of Szeto *et al.*, no neurological impairment was reported in Pai syndrome, and psychomotor development was normal in all cases⁸. These clinical data were of great help for prenatal counseling in this case of syndromic pericallosal lipoma.

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