'Pai Syndrome' with anterior alveolar polyp: A variant of a rare clinical entity

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SUMMARY

'Pai syndrome' (PS) is a rare congenital syndrome. Presented here, a new-born baby-girl who exhibited the characteristic features of having a midline nasal (septal) polyp, an anterior alveolar process polyp, and a pericallosal lipoma associated with corpus callosum dysgenesis of the brain. Both polyps were lined with stratified-squamous epithelium. The overall features were largely consistent with those described by Pai et al., in 1987. A midline cleft-lip (with or without cleft-alveolus) is one of the most common features of the syndrome which was however absent in this case. Instead, an anterior alveolar polyp is present, which is relatively rare.

INTRODUCTION

Pai syndrome is a rare syndrome of unknown cause, comprising a particular variety of congenital developmental malformation with variable phenotypical presentations. It was first reported by Pai et al., in 1987 when it was described in a male new-born as an unusual combination of three rare anomalies, i.e., complete median cleft lip, cutaneous polyps, and developmental midline lipomas of the central nervous system.[1] After that, over sixty similar cases have been reported, although their phenotypic presentations were not very similar, ranging from mild-facial dysmorphism to severe frontonasal dysplasia (FND). The perceived impression about the underlying aetiology goes towards a multifactorial origin. No chromosomal abnormalities have been described in patients with PS on karyotype. Diagnosis of the syndrome is based on clinical signs and associated pathology, the common clinical component being midline facial skin masses. Minimum diagnostic criteria as suggested by Castori et al.[2] and Lederer et al.[3], one or more hamartomatous nasal polyps plus median cleft lip (with or without cleft alveolus), and/or alveolar process congenital polyp, and/or peri-callosal lipoma. Pericallosal lipoma is a common finding (85%) in this syndrome which is frequently associated with various degrees of intracerebral malformations such as agenesis or dysgenesis of corpus callosum.[3] Most of the cases of PS have been diagnosed postnatally after birth. Typically, the condition is still not found to have any negative impact on psychoneurological development. In a recent report of an 8-year-long follow-up case, Imai et al., observed some attention-deficit disorder, but its definite relationship with the syndrome is yet to be confirmed.[4]

CASE REPORT

The present case is that of a dysmorphic baby girl, born via lower-segment caesarean section (LSCS) for maternal eclampsia at the gestational age of 38 weeks. This is the first child of non-consanguineous Chinese parents. The mother (in her thirties) developed pregnancy-induced hypertension (PIH). Baby's birth-weight was 3.31kg, length: 49cm, head-circumference: 34cm. She developed hypoxic-ischemic encephalopathy which was successfully managed by immediate intensive care support. Except for PIH in late pregnancy, the mother had no other known obstetric illness. For this case, a prenatal diagnosis was not possible probably because of the non-compliance with regular follow-up.

Family history revealed the presence of Down syndrome from the paternal side. Drug history revealed nothing contributory. The mother also denied any addictive habits such as alcohol, smoking or other harmful substances.

At birth, the baby exhibited two separate soft skin-covered polypoidal masses on the face (Fig. 1), one from the right nasal cavity (nasal septal origin), measuring about 2cm at its visible exterior part, partially obstructing the ipsilateral nasal cavity. Another one arose from the mid-anterior alveolar process, a little smaller and associated with the high-arched palate. No other orofacial deformity was detected. Other systemic examinations were unremarkable. No hypertelorism or other ocular problems were noted. The polyps were surgically removed, and the procedure was uneventful. During the last review when the child was one year of age, she showed good achievement of all milestones.

Chromosomal study result showed a normal female karyotype: 46, XX.

DISCUSSION

Lipoma is the most common soft tissue tumor in adults, but it is uncommon in children and even more rare as an intracranial lesion. However, pericallosal lipoma is a common finding in PS which was present in our patient. It is thought to have originated from over-proliferation of the fat cells present in the leptomeninx, an embryological structure normally differentiated from the meninx primitiva.[5] Based on imaging there are two types of pericallosal lipomas. The tubulo-nodular is the most common type, frequently associated with extensive callosal and often fronto-nasal

This article was accepted: 4 April 2019
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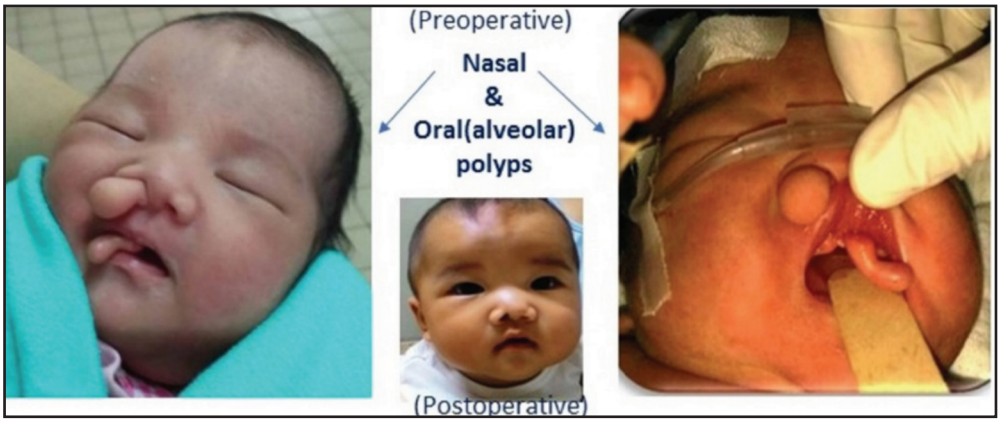

Fig. 1: Images showing the nasal and anterior alveolar polyps of the new born and her postoperative appearance.

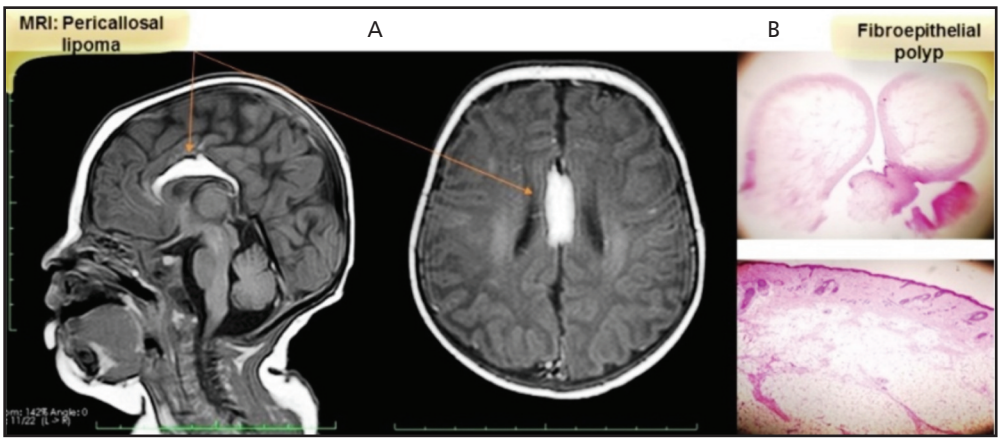

Fig. 2: MRI images of brain showing pericallosal lipoma (A), histopathological view of nasal polyp (B).

anomaly. The second type is the curvilinear lipoma which is less common and probably resulting in a better outcome. This is the latter one, that is involved in our case, in association with dysgenesis of the posterior part of corpus callosum (Fig. 2). Other structures of the magnetic resonance image (MRI) was structurally normal. On MRI, the lipoma showed a hyperintense signal on T1-weighted sequences without fat suppression.

The second feature of this patient was the nasal polyp, which is the hallmark and the most common characteristic of PS (97%).[3] Congenital nasal polyp associated with pericallosal lipoma is only reported in PS. The 3rd most common finding in PS is the midline cleft-lip, which was absent in our case, but this is substituted by the presence of a mid-anterior alveolar congenital polyp, which is a relatively rare finding. Histopathological reports of both polyps are in favour of skin polyps (Fig. 2B). The nasal one is typical fibroepithelial polyp (skin tag), whereas the oral one is nevus lipomatosus superficialis, a rare type of benign hamartomatous skin lesion as defined by the presence of aggregates of mature adipose tissue among the collagen bundles of the dermis.

CONCLUSION

Pai syndrome can have a variable clinical presentation. It may complicate pregnancy but is still manageable with proper resuscitative and surgical measures. Prenatal diagnosis is sometimes possible. In the short-term follow-up, it seems not exhibit any remarkable developmental problem, even in presence of intracranial pathology. Since there is no definite evidence of having any serious impact on the developmental process, the decision to terminate the pregnancy should be carefully examined, especially in cases of minor degree of abnormality. However, long-term follow-up is required in such cases. Further research should be directed towards discovering the aetiology of PS. Any association with Down syndrome may be a point of interest and the presence of such a family history should emphasize on closer prenatal follow-up. Thus far, this is the first reported case of PS in Southeast Asia.

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