PHACES syndrome with cataract and Horner's syndrome: a case report

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PHACES syndrome (Posterior Fossa Malformations, Hemangioma, Arterial Anomalies, Cardiac Defects and Coarctation of the Aorta, Eye Abnormalities, and Sternal Abnormalities or Ventral Developmental Defects) is a rare neurocutaneous syndrome, which characteristic feature is large segmental hemangioma. Extracutaneous involvement is an important cause of morbidity in this syndrome.

Described below is an infant with large hemangiomatous lesions on the right side of the face, suprasternal notch, midline sternal defect and supraumbilical raphe. Based on the new consensus on the diagnostic criteria of PHACES syndrome, a definitive diagnosis of PHACES syndrome has been corroborated. Accordingly, our patient was analyzed with regards to other clinical features through magnetic resonance imaging of the head, neck and abdomen, all of which were normal. In echocardiography, the patient showed atrial septal defect, in addition to ipsilateral conductive hearing loss, contralateral cataract and Horner's syndrome, which was reported only in very few cases. The clinical presentation of the present case was different from most previous reported ones, as segmental hemangioma in PHACES syndrome was, for the most part, located on the left side of the face and ocular involvement was predominantly reported ipsilateral to hemangiomatous lesion.

Key words: PHACES syndrome, cataract, Horner's syndrome, conductive hearing loss

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INTRODUCTION

Hemangioma is the most common benign tumor in infancy with an approximate prevalence of 10% in the first year of life. It is commenced with a period of rapid growth in the first few months and a spontaneous regression over the following 5-7 years. Such type of tumor is asymptomatic without any need for treatment intervention, except close observation. Hemangioma, based on its extent of involvement, is categorized into localized, segmental, indeterminate and multifocal categories. Areas of involvement in face are classified into frontotemporal (S1), maxillary (S2), mandibular (S3), frontal and central (S4) [1].

Segmental hemangioma entails higher risks of complications such as ulceration disfigurement, functional impairment of vital organs and associated structural anomalies such as PHACES syndrome. With ratio 3 to 1, conventional hemangioma is more prevalent in females; however, in PHACES syndrome (posterior fossa malformations, hemangioma, arterial anomalies, cardiac defects and coarctation of the aorta, eye abnormalities, and sternal abnormalities or ventral developmental defects), this ratio is as high as 9/1 [3]. In this article, a patient with PHACES syndrome is described [2].

CASE PRESENTATION

A 5-month-old female infant was referred to surgery clinic in Afzalipour Hospital with

an atrophic skin in sternal region and multiple hemangiomatous lesions on face and chest.

She was a product of a normal vaginal delivery with 9 months gestational age and no perinatal complications. Her parents were not consanguineous and there was no history of hemangioma or other serious diseases running in the family.

Physical examination revealed a fully circumscribed segmental dark red plaque, covering the right side of the face with extension to periauricular, tragus, neck and lower lip. Further observed was a 3× 4 centimeter mass of hemangioma on sternal area with depigmented and atrophic skin, which was retracted during respiration. There was an atrophic area on supraumbilical which was consistent with supraumbilical raphe (Figure 1).

Both eyes were of a bluish-gray color. Ophthalmic examination revealed anterior cataract in her left pupil which, in contrast to the right pupil, had no normal light reflex. Consulting with an otolaryngologist, a hemangioma lesion on gingiva near the lower incision teeth and the involvement of periuricular and tragus, which led to auditory obstruction and conductive hearing loss, were reported. Airway examination (by laryngoscope), the neurologic development, and magnetic resonance imaging (MRI) of the brain were all normal.

The last echocardiography revealed mild atrium septal defect (ASD), but pulmonary hypertension, ASD and tricuspid regurgitation were reported in pervious echocardiography. Chest X-ray, magnetic resonance imaging with contrast and ultrasonography of abdomen were all normal.

Laboratory tests including complete blood cell count, urine analysis, stool examination and thyroid function test were within normal limit. A written informed consent to report the case was obtained from the parents of the patient.

Following consultation with pediatric neurologist and cardiologist, it was decided that propranolol be primarily employed for the treatment of hemangioma. We further consulted with thorax surgeon and ophthalmologist for a surgical reconstruction of sternal defect and cataract treatment. Unfortunately, the parents did not consent to any treatment modalities, hence the fact that no follow-up on the patient was obtained.

DISCUSSION

According to the 2016 consensus on diagnostic criteria for PHACES syndrome, definite diagnosis of PHACES syndrome requires a large segmental hemangioma of face or scalp with a diameter size of more than 5 centimeters or a surface area involvement of 22 cm^2 and one major or two minor criteria. Previous case reports showed that 31% of patients with such characteristic features of hemangioma had other diagnostic criteria of PHACES syndrome [2]. Our case had a segmental hemangioma covering a large surface area on the right side of her face with a diameter larger than 5 centimeter and one major criterion as sternal defect and supraumbilical raphe, corroborated via the diagnosis of PHACES syndrome.

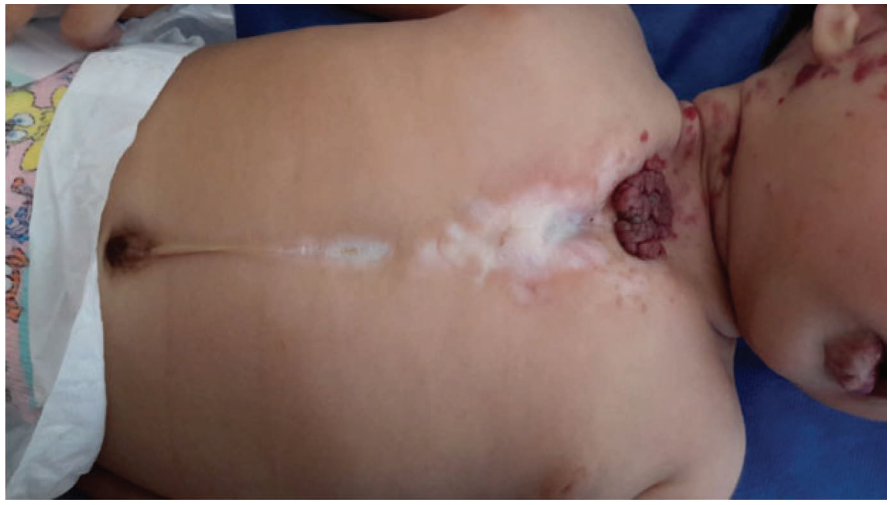

Figure 1. A large segmental plaque which was covered the right side of face with a hemangioma on depigmented and atrophic skin of sternal area and supraumblical raphe.

Most hemangiomatous lesions developed on the left side, yet, in our case, hemangioma was located on the right side, which is relatively rare [3]. Visceral hemangioma is observed in 22% of PHACES syndrome cases. The present case showed no extracutaneous hemangioma [2].

A very common extracutaneous manifestation of PHACES syndrome is cerebrovascular defect with posterior fossa anomalies, including Dandy-Walker's syndrome, with a prevalence ranging from 30.4% to 81% in previous case reports; our patient, on the other hand, had no neurologic abnormalities [4].

In previous literatures, the prevalence of cardiovascular anomalies was from 41% to 67%. The most common cardiac involvement is coarctation of the aorta (19-30%). Other cardiac abnormalities, descending by frequency, are patent ductus arteriosus (PDA), ventricular septal defects (VSD), atrial septal defects (ASD), and pulmonary stenosis [5]. The present case had a mild ASD in her last echocardiography, which is only rarely reported [2].

Ocular manifestations are most frequently located on the same side of hemangiomatous lesion, and have been observed in 30% of the cases with a dominant involvement of posterior segment of ophthalmic area. The most prevalent ophthalmologic abnormalities are microphtalmia, optic atrophy, iris and optic nerve hypoplasia, coloboma and Horner's syndrome. Ocular findings in the present case report were anterior cataract and Horner's syndrome, which were surprisingly located on the contralateral side relative to hemangiomatous lesion. Anterior segment involvement of the eye such as anterior cataract was observed very rarely, making it a minor criteria for the diagnosis of PHACES syndrome [2,6].

In line with the present research, sternal defect has been observed in 30% of the cases. S3 segment involvement, also found in our case, entailed a higher risk of sternal defect and supraumblical raphe [3,6].

Our patient had a conductive hearing loss due to the pressure of hemangioma exerted on the auditory canal. There are a few reports on conductive or sensory neural impairment in patients with PHACES syndrome, which is always ipsilateral to hemangiomatous lesion and mostly observed with S1 or S3 segment involvement [7].

There exist myriad treatment modalities for hemangioma, including corticosteroid (topical, intralesional and systemic), vincristin, interferon, imiquimod, beta blockers, cryosurgery and laser. The first line of treatment for PHACES syndrome is oral corticosteroids, although there are a few case reports on the use of propranolol with good efficacy. Brain imaging and consultation with cardiologist prior to treatment with propranolol in PHACES syndrome are necessary, because the adverse effects of propranolol can be high-output cardiac failure and stroke, particularly in infants with cerebrovascular anomalies [1,8].

CONCLUSION

In an infant with large segmental hemangioma, it is indispensable to evaluate the involvement of the cardiovascular system, central nerves system and eye by a thorough physical examination, echocardiography, magnetic resonance imaging of the head and neck, abdominal ultrasonography and laboratory tests for thyroid function.

Conflict of Interest: None declared.

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