PHACE SYNDROME A RARE PAEDIATRIC CASE

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ABSTRACT
PHACE syndrome is a very uncommon syndrome that involves multiple systems of the body. We are presenting our experience of PHACE syndrome in an infant which was admitted to Department of Pediatrics, Lahore General Hospital at the age of two months. This syndrome is treated symptomatically as there is no standardized treatment option for the affected individuals.

Key Words: PHACE syndrome, Hemangioma, Neurocutaneous syndrome, Coarctation of Aorta.

How to cite this article: Liaqat I, Zahra H, Rashid A. Phace Syndrome a Rare Paediatric Case. Pak Postgrad Med J 2024;35(2): 86-88

DOI: https://doi.org/10.51642/ppmj.v35i02.596

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INTRODUCTION
PHACE syndrome is a neurocutaneous syndrome found in 2% of infantile hemangioma. It is rarely seen congenital cervicofacial hemangioma. [1] Exact cause of this syndrome is not known and is considered as a hereditary condition. Mostly sporadic but its correlation with mutations in X-linked genes has been found. Cases have been seen more commonly in females.[2] Clinically it presents in the form of posterior fossa malformation, hemangioma of cervicofacial region, arterial anomalies, cardiac anomalies and eye anomalies. First time, PHACE term was described by Frieden and colleagues in 1996. According to a case series of 29 children with PHACE syndrome, 69% have abnormal neurodevelopment including 44% language delay, 36% with gross motor delay, 8% with fine motor delay.[3] Due to multiorgan involvement we have to perform many investigations to see hemangioma in different organs, including CBC, chest Xray, CT brain, CT angiography, doppler studies, echocardiography, ultrasound abdomen, PTA, BERA and fundoscopy. As there is also involvement of endocrine glands so thyroid profile, HbA1c, serum electrolytes, parathyroid level are also done. As the syndrome involves multiorgans, clinical management require multidisciplinary approach. The hemangioma is treated successfully with oral beta blockers (propranolol), it should be used with caution because of the risk of cerebrovascular accident in patients with heart or blood vessels problems. We can also use systemic steroids, surgery or laser therapy as a treatment option. Aspirin is given to prevent ischemic accidents; surgical intervention may require for cardiovascular abnormalities and hormonal replacement therapy for endocrinopathies. Patients having major organ involvement like vessels of brain and heart have bad prognosis.

CASE REPORT
A two months old male baby presented in the emergency with complaint of respiratory distress and cough for one week. In addition, he had history of gross motor delay, as well as complaint of sweating and tachypnea while feeding. On examination, there were red raised lesions all over body, respiratory rate was 48/min, pulse rate 142/min, temperature 100 F, BP 80/55, SPO2 96% with oxygen. On systemic examination there was subcostal recession, normal vesicular breathing, bilateral crepitations, apex beat

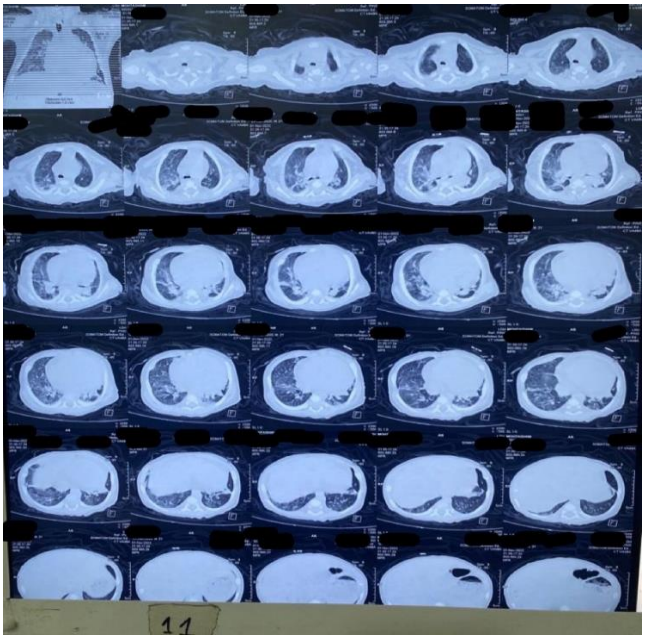
was in the 4th intercostal space medial to midclavicular line, both heart sounds were audible with no added sound. On abdominal examination there was only hepatomegaly and rest of examination was unremarkable. We suspected it clinically as PHACE syndrome. CBC was unremarkable, radiological investigations favor our clinical diagnosis as Chest Xray showed cardiomegaly. CT chest and abdomen revealed many infantile hemangiomas. HRCT showed enlarged heart, bilateral ground glass appearance and prominent hilar vasculature, few ataractic bands. Echocardiography revealed, severe pulmonary hypertension. Hearing and visual assessment was normal. Doppler studies show coarctation of abdominal aorta beyond the takeoff of celiac axis. On the basis of history and clinical examination, we suspected it clinically as patient has multiple haemangiomas on face, neck and torso, and upper arm with one major criteria in the form of cardiovascular involvement.

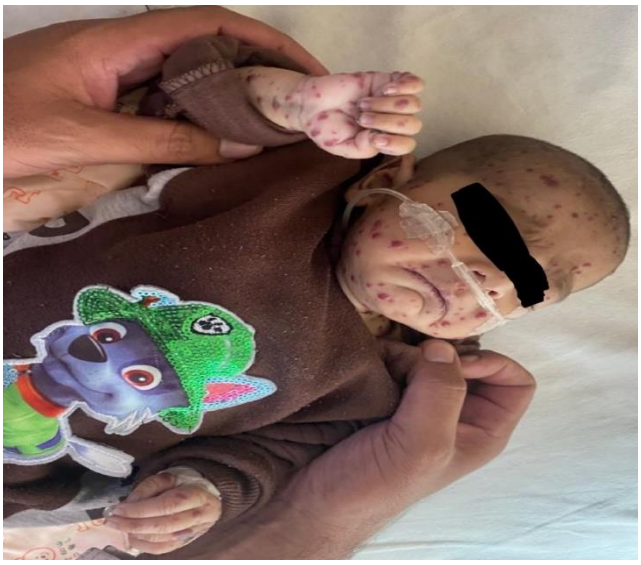
CRITERIA OF PHACE SYNDROME:

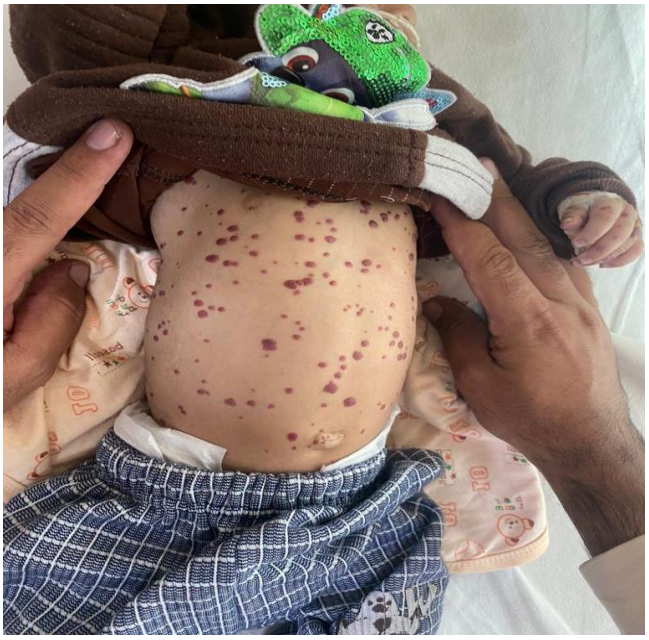
Definite=Facial hemangiom of>5cm plus 1 major or 2 minor criteria

Possible=1) Facial hemangioma greater than 5 cm in diameter plus 1 minor criterion; 2) hemangioma of the neck, upper torso, or torso and upper arm plus 1 major criterion or 2 minor criteria; or 3) no hemangioma plus 2 major criteria.

After establishing provisional diagnosis,treatment was offered to reduce the preload and after load with use if spiromide and digoxin.Beta blocker was given for hemangioma and consulted with cardioogist,cardiac surgeons and interventional radiologist for future management plan.







DISCUSSION

PHACE syndrome is associated with dandy walker malformation, vascular anomalies such as co-arctation of aorta, aneurysmal carotid dilations, eye findings include glaucoma, cataract, microphthalmia optic nerve hypoplasia, ventral defects. Endocrinopathies such as hypothyroidism, hypopituitarism, growth hormone deficiency and diabetes insipidus may be seen.[4] All finding may not be present in a single patient. It is an under diagnosed syndrome, facial hemangiomas are the primary criteria considered in the diagnosis of this syndrome. In our case, multiple facial hemangiomas are noted involving both cheeks, nose, eyelids, forehead, ears, neck and whole body.

Cutaneous hemangiomas are common but extracutaneous hemangiomas have been reported in

the brain, lungs, bowels, liver.[5]In our case multiple hemangiomas were present in the lungs and liver. Hemangiomas are benign but if they involve eyelids, ears, airways they can interfere with vision, hearing and breathing which was seen in our case as patient had severe respiratory distress and was oxygen dependent but hearing and vision was not affected. Studies have highlighted that some patient have brain vessels malformations which was not seen in our case PHACE syndrome is very rare, so most pediatrician will probably see few cases in their lifetime. Patient reporting with cutaneous haemangioma should be looked for extra cutaneous haemangiomas in major organs. Management plan may be decided by multidisciplinary team. In order to guidelines for pediatrician we can say that it can be treated symptomatically involving multidisciplinary team.

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