http://jmscr.igmpublication.org/home/
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v9i1.24

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

PHACE Syndrome: A Rare Case Study

Authors
Dr Hari Sankar T[1], Dr S K Pradhan[2], Dr C S Patra[3], Dr S S Mishra[4]
[1]Junior Resident, Department of Pediatrics, MKCG Medical College
[2]Assistant Professor, Department of Pediatrics, MKCG Medical College
[3]Senior Resident, Department of Pediatrics, MKCG Medical College
[4]Associate Professor, Department of Pediatrics, MKCG Medical College

Abstract
PHACE is a clinic-radiological syndrome first described by Friedon in 1996 as an infantile segmental hemangioma with other features like posterior fossa abnormalities, arteriopathy, cardiac defects and eye anomalies.
Long term outcome varies according to the severity of associated defects.
Keywords: *PHACE, Hemangiomas, VSD, CoA, LASER.*

Introduction

PHACE syndrome stands for P:-Posterior fossa brain defects like dandy walker malformation or other brain defects, H:- facial infantile Hemangiomas, A:- Arterial cardiovascular anomalies like aneurysms and stroke ,C:-co-arctation of aorta and E:-Eye anomalies. Overall there is a female preponderance. Almost 88% of PHACE infants are females[1]. Early detection and specialized care by an experienced multidisciplinary team is essential.

Case Study

5 month old female child , product of 3rd degree consanguineous marriage ,from lower socioeconomic status family presented with chief complaints of cough and cold , and respiratory distress for 8 days and mild fever for 3 days. child was neurodevelopmentally normal and received only birth dose of vaccination.

Child was delivered at term with a birth weight of 2.8 Kg. No history of birth asphyxia.

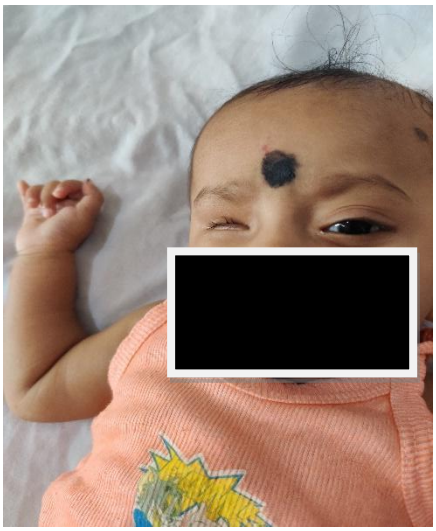
At 2 months of age, child had history of excessive cry with difficulty in breathing with bluish discoloration of body noticed by mother. There is a history of multiple episodes of uprolling of eyes with fisting of both hands for 4-5 days. Admitted to MKCG MCH Berhampur, diagnosed to have congenital acyanotic heart disease ,VSD. 4 days after discharge child had a similar attack with left sided focal seizures –MRI done ,shows left frontal, temporal and parietal lobes shows relatively smaller size/atrophy ,with mild prominence of left lateral ventricle and prominent extra cerebral spaces in vicinity. White matter in these regions show low volume as compared to right and minimal altered signal intensity. Child was started on levetiracetam syrup and no further seizure occurred.

O/E:- Child is conscious, alert, HR:-150/mt, Spo_2 :-96% in RA., RR:-65/mt. Anthropometric

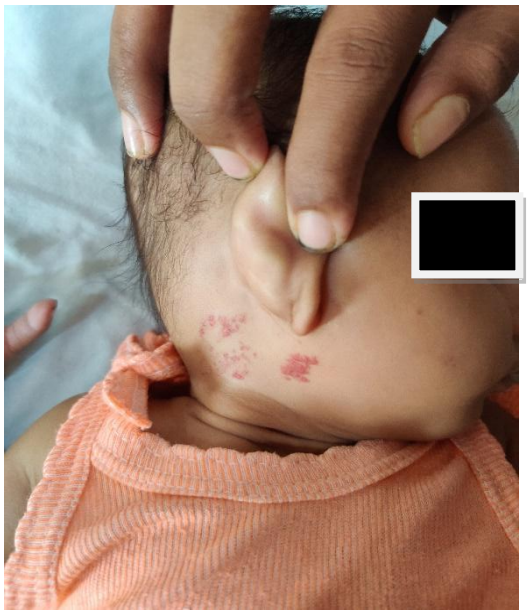
measurements shows HC :-38cms (between -2 to -3 SD), Wt :- 4.8 KG. Length:-61cms. (Wt/length between -2 to -3 SD). No pallor, icterus , cyanosis, clubbing, lymphadenopathy or edema. Head to foot examination reveals right sided anophthalmia. Capillary hemangiomas are seen all over the face and right side of neck. There is reduced movements noticed in right side.

On systemic examination:- in CVS ,a pan systolic murmur is heard in LLSB, soft blowing with radiation to right sternal border .Respiratory system shows conducted sounds in all lung fields. Per abdomen shows liver 2cm below right costal margin and spleen just palpable .AF at level.

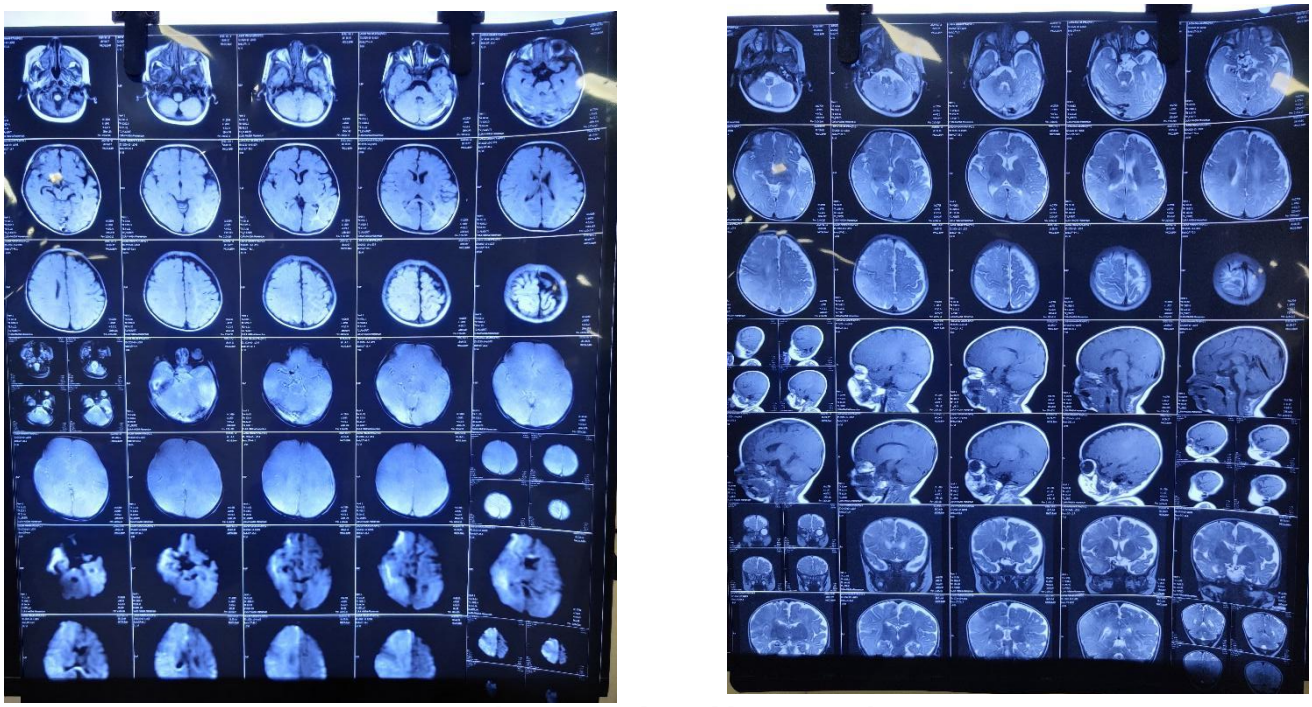
Investigations: CBC shows microcytic hypochromic anemia with hemoglobin of 8 gm% and other parameters are within normal limits. CRP was positive and echocardiography was done showing ACHD, single large perimembraneous VSD, mild PR, discreet post ductal co–arctation of aorta, severe PAH.



Anophthalmia in right eye



hemangiomas on neck and face



MRI showing unilateral brain atrophy

Discussion

PHACE/S is a syndromic form of infantile segmental hemangioma described by Friedon et al in 1996[2]. Other features of PHACE syndrome include posterior fossa brain malformations like Dandy walker malformations, hypoplasia and agenesis of cerebellum, cerebrum, corpus callosum, septum pellucidum. There can be arterial anomalies like coarctation of aorta, cardiac anomalies like VSD, PDA, arterial aneurysms and eye anomalies. Overall there is a female preponderance.

In our case we have got a female child with MRI findings suggestive of atrophy of left frontal temporal and parietal lobes with mild prominence of left lateral ventricle and low volume white matter in left side. Echo study shows Coarctation of aorta with VSD, PR and PAH. Patient is having right sided anophthalmia with capillary hemangioma in face and right side of neck.

The differential diagnosis of PHACE syndrome includes other neurocutaneous syndromes like Wyburn mason syndrome and Sturge weber syndrome[3]. Port wine stain in case of Sturge weber syndrome may mimic facial hemangiomas. But absence of glaucoma on eye examinations and MRI findings in our patient excludes Sturge weber syndrome.

Structural brain anomalies are the most common extracutaneous manifestation in PHACE syndrome. The commonest posterior fossa abnormality is the Dandy walker syndrome[4]. Cardiac investigations may reveal complex coarctation of aorta, the commonest cardiac lesion[5]. Structural heart defects are seen in 15% of the cases[6], 40% are known to have vascular abnormalities. Approximately 1/3 rd cases are known to have eye involvement[7].Children with vascular abnormalities are at risk of aneurysms and cerebral infarction.

According to a case series of 29 children with PHACE syndrome 69% had abnormal neurodevelopment, including 44% with language delay, 36% with gross motor delay and 8% with fine motor delay.

The underlying pathogenesis of PHACE syndrome remains unknown though evidence that infantile hemangiomas may result from abnormal growth and differentiation of hemogenic endothelium highlights some avenues for further investigations. Due to the involvement of multiple organ systems in PHACE syndrome clinical care should require multidisciplinary input. The beta blocker Propranolol is emerging as a treatment for infantile hemangiomas associated with PHACE syndrome.

Conclusion

PHACE syndrome is a rare syndrome in children. A diagnosis is generally made from the physical examination, along with imaging of head and chest, and an eye examination. Mostly diagnosed among female infants. Long term quality of life varies. Propranolol is emerging as a treatment of infantile hemangiomas associated with this syndrome. LASER and other surgeries can make a substantial positive impact on appearance.

References

1. Metry DW, Siegel DH, Cordisco MR, Pope E, Prendiville J, Drolet BA et al .A comparison of disease severity among affected male versus female patients PHACE syndrome. J Am Acad Dermatol 2008;58:81-7
2. Frieden IJ, Reese V, Cphen D. PHACE syndrome: The association of posterior fossa brain malformations, hemangiomas ,arterial anomalies, coarctation of aorta and cardiac defects, and eye abnormalities. Arch Dermatol 1996
3. Enjolras O, Riche MC, Merland JJ.Facial port wine stains and Sturge –Weber syndrome.Pediatrics.1985;76:48-51.
4. Denzer F, Denzer C, Lenners BS, Bode H, Wabitsch M. A case of PHACE syndrome and acquired hypopituitarism. Int J Pediatr Endocrinol. 2012;1:20-1.
5. Sripornsawan P, Chotsampansharoen T, Kritsaneepaiboon S. Successful treatment

of PHACE syndrome with oral propranolol. J Hematol Transfus Med. 2015;25:55-60.

6. Kehkshan Tahir, Aysha Umar Khan, Shahbaz Aman. PHACE syndrome: A Case report. Journal of Pakisthan Association of Dermatologists 2017.
7. Hartemink DA, Chiu YE, Drolet BA, Kerschner JE. PHACES syndrome: A review. Int J Pediatr Otorhiolaryngol 2009;73:181-7.