CASE REPORT

A RARE CASE REPORT: PHACE SYNDROME

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INTRODUCTION: A syndrome is defined as a recognizable pattern of medical conditions that occur together. Initially described as an association of large cutaneous hemangiomas of the head and anomalies of the cerebral vasculature by Pascual-Castroviejo in 1978. Subsequently been coined the term PHACE association by Frieden et.al. Frieden created the term PHACE, which is an acronym which refers to Posterior fossa anomalies, Hemangioma, Arterial lesions, Cardiac abnormalities/coarctation of the aorta and Eye anomalies.

PHACES SYNDROME is PHACE syndrome plus: Sternal cleft, supraumbilical raphe, or both.

CASE REPORT: A 6 yrs old male child presented with red colour patch over left side of face since birth and progressive increase in size and decrease in vision of the left eye. On examination, the child had an erythematous patch mainly on the left side of face. An ophthalmic examination of the left side revealed microphthalmos and microcornea with corneal opacity. Examination of other systems was unremarkable. Echocardiography was normal, Magnetic Resonance Imaging (MRI) of orbits and brain showed the presence of Hemangioma in left temporal fossa and periorbital soft tissues with intraorbital extension and coloboma of left eye, hypoplastic left cerebellar hemisphere and left unilateral megalencephaly with prominent sulci/cisterns.

INVESTIGATIONS: History and clinical examination, ophthalmic examination, echocardiograph, MRI, magnetic resonance angiogram of the head and neck. Occasionally, a computed tomography angiogram may also be done.

DISCUSSION: Although relatively uncommon, more than 400 cases of PHACE syndrome have been reported in the medical literature. In the PHACE syndrome, apart from facial hemangiomas, structural and vascular anomalies of brain are the most common features (80%), followed by cardiovascular anomalies (33%), ventral developmental defects (25%), and ocular anomalies (20%). It affects girls nine times more than boys. Large segmental infantile hemangiomas are most commonly located on the face. The identification of PHACE syndrome is made by a clinical diagnosis.

Categories of PHACE Syndrome:

Definite PHACE Syndrome.

Facial hemangioma greater than 5cm plus 1 major or 2 minor criteria.

Possible PHACE syndrome.

Facial hemangioma >5cm + 1 minor criteria.

Hemangioma of the neck or upper torso + 1 major or 2 minor criteria.

2 major criteria without hemangioma.

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CONCLUSIONS: PHACE syndrome needs to be managed by a multidisciplinary team of experts. Additional specialties such as cardiology, ophthalmology, neurology, and neurosurgery may need to be involved.

The team of experts should pay close attention to how these children develop throughout the school-age period. As it grows, the hemangioma can break down skin, distort facial features or get in the way of other vital functions, such as breathing, vision, and hearing.

Other complications will depend on what other structures are involved. These could include developmental delay, seizures, headaches, and abnormal muscle tone if the brain is involved.

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