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A Case of Phacomatosis with Rare Congenital Developmental Malformation of Eye

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Abstract: ***Phakomatoses or neurocutaneous disorders primarily affect the central nervous system; however, skin, viscera, and other connective tissues can also be involved with variable clinical presentation. Imaging plays an important role in diagnosis and classification by early identification of pathognomonic findings in various types of phacomatoses. We present a case of a 1.5 year old male child with complains of blindness since birth. Skin spots and right leg deformity were observed by parents few months after birth. On clinical evaluation and radiological investigations using multimodality approach, diagnosis of phakomatoses i. e Neurofibromatosis type I was established. Atypical Ocular manifestation in the form of persistent hyperplastic primary vitreous with associated vitreous haemorrhage was observed in our case.***

Keywords: Phakomatoses, Neurofibromatosis, B scan, MRI, PHPV

1. Introduction

Neurofibromatosis type 1 (NF1), or von Recklinghausen disease, is a comparatively common hereditary disease in which the skin, nervous system, bones, endocrine glands, and sometimes other organs are the sites of a variety of congenital abnormalities, which often take the form of benign tumors. The typical clinical picture of NF1consists of multiple circumscribed areas of increased skin pigmentation accompanied by dermal and neural tumors of various types. Various ocular findings associated with NF1 include - Lisch nodules, Choroidal freckling, microvascular abnormalities of the retina, Optic pathway gliomas. We present a case of a 1.5 year old male child with complains of blindness since birth. Skin spots and right leg deformity were observed by parents few months after birth. Clinical evaluation revealed pseudoarthrosis involving the leg bone (right Tibia) and few Café - au - lait spots over back and bilateral limbs. X ray lower limbs, ultrasonography (B scan) of both eyes and Magnetic Resonance Imaging of the brain and orbit screening was performed.

2. Case Presentation

A 1.5 year male child brought by parents with complaints of blindness since birth. Parents also noticed skin spots and leg deformity few months after birth. Child was born of a nonconsanguineous marriage. Mother gave history of uneventful antenatal period and a full - term normal vaginal delivery, with no neonatal intensive care unit (NICU) admissions. On clinical evaluation pseudoarthrosis involving the leg bone (right Tibia) was noted. Few Café - au - lait spots were noted over back and bilateral limbs. Xray lower limbs, ultrasonography (B scan) of both eyes and Magnetic Resonance Imaging of the brain were performed in order to establish a syndromic diagnosis.

AP and lateral radiograph of bilateral lower limbs revealed anterolateral bowing of right tibia and fibula at the junction of mid and lower third of tibia. There was cortical thickening of right tibia (as compared to left) with associated sclerosis at the site of bowing.

B scan of bilateral eyes was done. It revealed echogenic membrane/soft tissue showing flow on colour doppler in posterior segment of right eye, most likely suggestive of persistent hyperplastic primary vitreous (PHPV). Thickening of bilateral chorio - retinal layers with subluxation of bilateral lens was also noted.

On Multiplanar MRI of brain, mild cerebral atrophy was noted with J shaped sella. Pituitary gland was small for the age. However, no evidence abnormal signal intensity was noted in brain parenchyma. On bilateral orbital screening, there was thickening of bilateral chorio - retinal layers. A 1.1 x 0.6 x 0.9 m T1/T2 hypointense retrolental soft tissue showing foci of blooming on GRE and not showing restriction of diffusion was seen, extending from the posterior aspect of the lens to the optic nerve head. T1/T2 hyperintense signal was seen within the posterior segment of the right globe with peripheral foci of blooming on GRE. Intraorbital portions of bilateral optic nerves appeared mildly atrophic.

On the basis of clinical and radiological evaluation using multimodality approach i. e. Xray, USG (B - scan), Cross sectional imaging (Multiplanar MRI) diagnosis of Neurofibromatosis - 1 with persistent hyperplastic primary vitreous with associated vitreous haemorrhage was made.

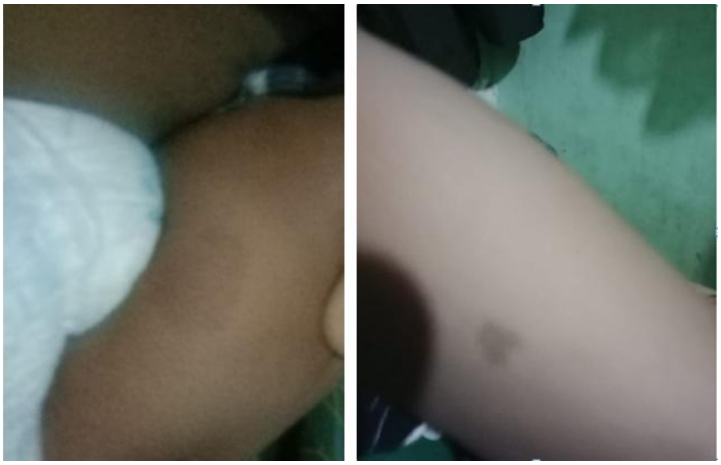

Figure 1 (A) and 1 (B): Light to dark brown well defined pigmented spots over the back and limbs approx.5 - 6 in number, Suggestive of Café - au - laitspots

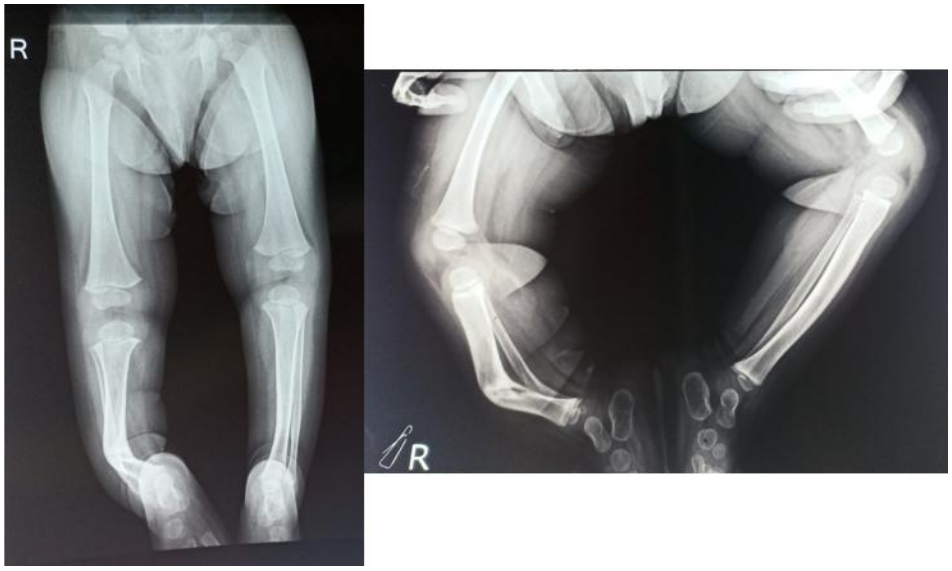

Figure 2 (A) and 2 (B): AP and lateral radiograph of bilateral lower limbs reveal anterolateral bowing of right tibia and fibula at the junction of mid and lower third of tibia. There is thickening right tibia (as compared to left) with associated sclerosis at the site of bowing.

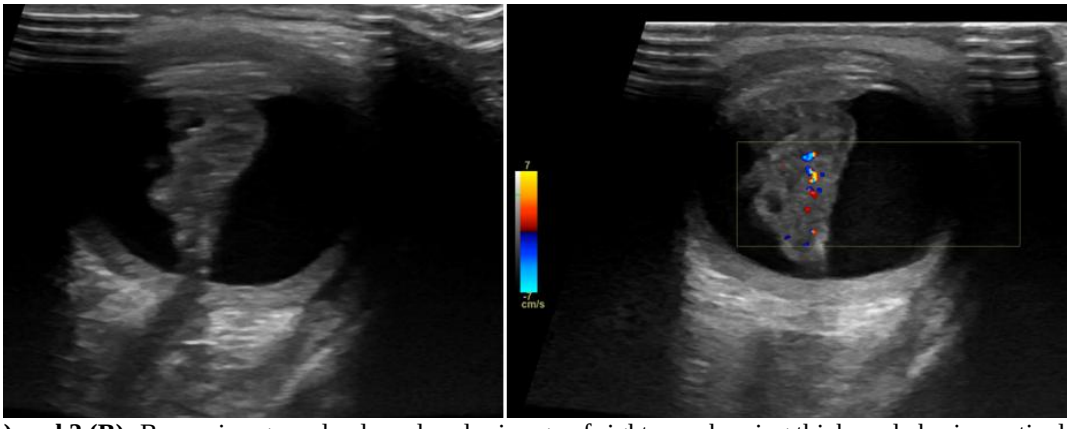

Figure 3 (A) and 3 (B): B scan image and colour doppler image of right eye showing thickened chorio - retinal layers, non appreciation of lens at normal position and Persistent Hyperplastic primary vitreous (PHPV).

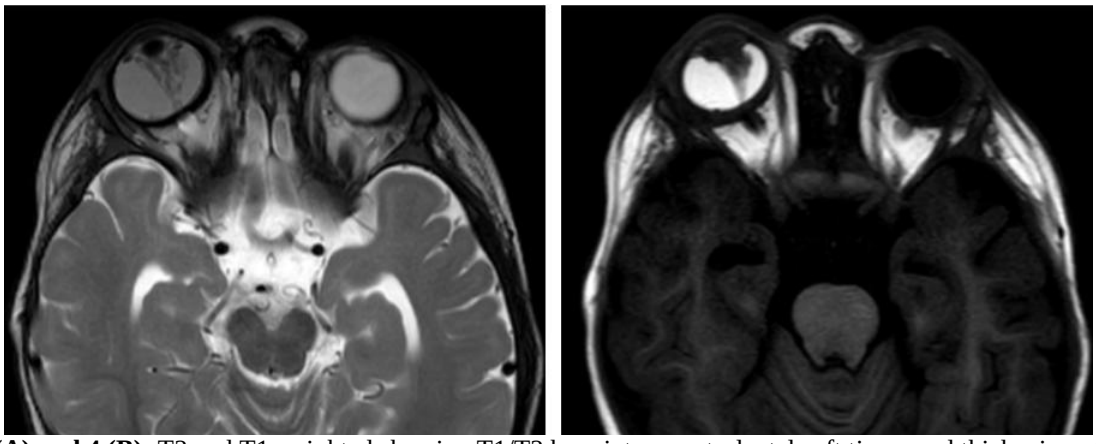

Figure 4 (A) and 4 (B): T2 and T1 weighted showing T1/T2 hypointense retrolental soft tissue and thickening of bilateral chorio - retinal layers

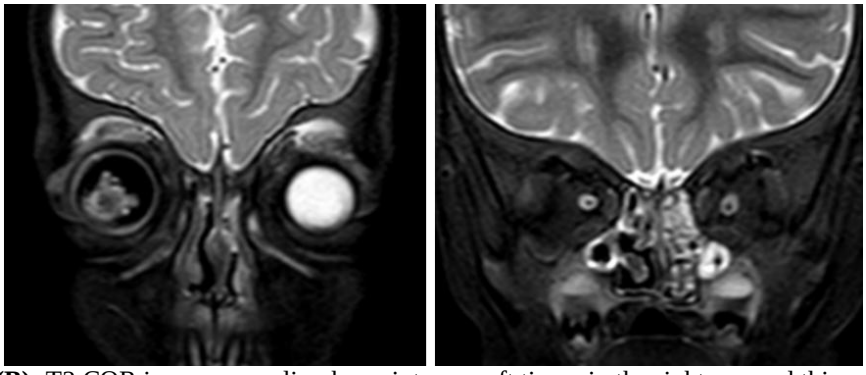

Figure 5 (A) and 5(B): T2 COR images revealing hyperintense soft tissue in the right eye and thinned out bilateral optic nerve

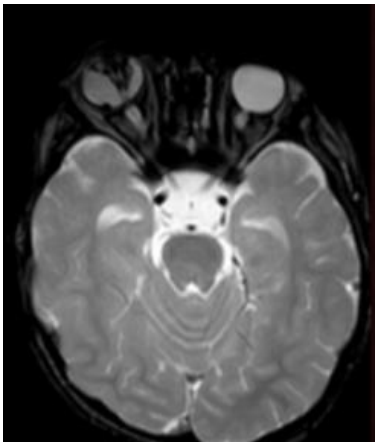

Figure 6: T2 FFE images retrolental soft tissue showing foci of blooming on GRE.

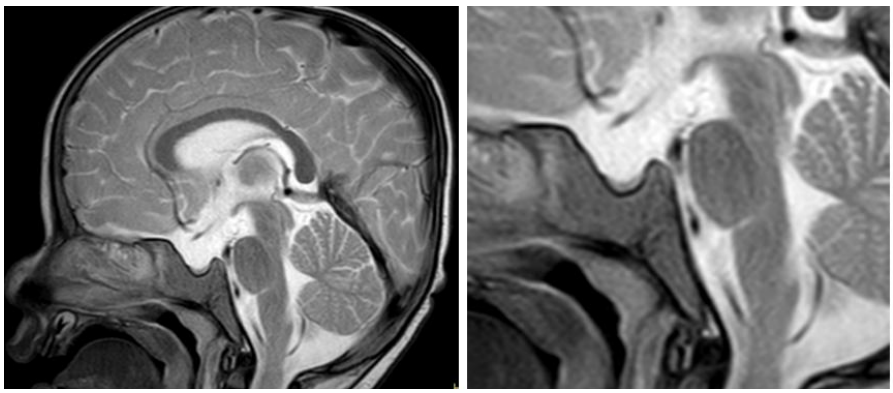

Figure 7 (A) and 7(B): T1w sagittal images of brain showing J shaped sella, pituitary is small in size for age

3. Discussion

Neurofibromatosis type 1 (NF1is autosomal dominant inherited neurocutaneous disorders or phakomatoses that manifest as distinct benign and malignant neoplasms associated with increased morbidity and mortality. Key manifestations of NF1 include café - au - lait macules, axillary or inguinal freckling, neurofibromas or plexiformneurofibromas, optic pathway gliomas, Lisch nodules, and osseous lesions such as sphenoid dysplasia, all of which are considered diagnostic features of NF1. Other manifestations include focal areas of signal intensity in the brain, low - grade gliomas, interstitial lung disease, various abdominopelvic neoplasms, scoliosis, and vascular dysplasia. The various NF1 - associated abdominopelvic neoplasms can be categorized by their cellular origin: neurogenic neoplasms, interstitial cells of Cajal neoplasms, neuroendocrine neoplasms, and embryonal neoplasms. Malignant peripheral nerve sheath tumors and intracranial tumors are the leading contributors to mortality in NF1 (1)

The multiplanar capabilities of MR imaging, particularly with T2 weighting, make this modality helpful in evaluating affected patients and making the diagnosis. The classic peripheral manifestations of NF1 include limb hemihypertrophy, pseudarthrosis, peripheral nerve neurofibromas, and subcutaneous common and plexiformneurofibromas. (2)

Persistent hyperplastic primary vitreous (PHPV) is a rare congenital developmental malformation of the eye that commonly manifests in a full - term infant with leukocoria and microphthalmos.

Persistence of the fetal vasculature is an encompassing term that includes the typical findings of classic PHPV in the retrolental space. Also included are individual legacies of fetal blood vessels that can cause defects throughout the ocular globe, including anterior chamber structures, retinal detachments, and other posterior malformations

Imaging findings depend on the size, thickness, and vascularity of the retrolental fibrovascular mass. At ultrasonography (US), the main finding is an echogenic mass posterior to the lens, with a hyperechoic band extending from the mass to the posterior aspect of the globe. Use of CT will depict microphthalmos and frequently a retrolental focus of increased attenuation. A linear band or septum extending from the posterior aspect of the retrolental mass allows for a confident diagnosis of PHPV. CT also enables the best assessment of the presence or absence of calcifications to help distinguish PHPV from retinoblastoma. At MR imaging, microphthalmos, shallow anterior chamber, enhancing retrolental tissue, persistent Cloquet canal, and retinal detachment all may be evident

PHPV is most often sporadic but may be inherited as an autosomal dominant or recessive trait (3)

In our case diagnosis of NF1 was established, as per 'Revised diagnostic criteria for neurofibromatosis type 1 (NF1) ', 2 major criterias i. e. Café - au - lait spots and pseudoarthrosis of tibia were fulfilled. PHPV with associated vitreous haemorrhage is not a known manifestation of NF1. Although traditionally a sporadic, unilateral, and isolated finding, PHPV also has been reported in trisomy 13, Norrie disease, Warburg syndrome, incontinentiapigmenti, cerebro - oculo - dysplasia - muscular dystrophy, and fetal alcohol syndrome (4).

Our case in unique as PHPV as a manifestation or association is rarely reported in literature with Neurofibromatosis type 1

4. Conclusions

- Phakomatoses or Neurocutanous syndromes need multimodality approach for establishing diagnosis.
- Clinical correlation and syndromic approach is useful to narrow down the investigations required as it is variable with each patient.
- Radiologist play a vital role to identify common and uncommon manifestations of the Phakomatoses.

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