e-ISSN: 0975-1556, p-ISSN:2820-2643

Available online on www.ijpcr.com

International Journal of Pharmaceutical and Clinical Research 2024; 16(4); 788-796

Case Series

Case Series on Various Presentations in Common and Uncommon Neurocutaneous Syndromes

Nilufa Ahmed Laskar[1], Bhaskar Jyoti Pegu[2], Tepty Kutum[3], MD Imdadaul Islam[4], Abhishek L[5]

[1,5]3rd Year Resident Trainee, Department of Radiology, Silchar Medical College and Hospital
[3]Assistant Professor, Department of Radiology, Silchar Medical College and Hospital
[4]Assistant Professor, Department of Radiology, Silchar Medical College and Hospital

Received: 25-01-2024 / Revised: 23-02-2024 / Accepted: 26-03-2024
Corresponding Author: Dr. Abhishek L
Conflict of interest: Nil

Abstract:

The phakomatoses are congenital disorders manifesting with central nervous system and cutaneous abnormalities. The structures predominantly affected are those of ectodermal origin, including the skin, nervous system, and eyes. Other organs are also involved in Some syndromes mainly cardiovascular, pulmonary, renal, and musculoskeletal systems. A few rare neurocutaneous syndromes, such as PHACES syndrome and Gomez Lopez Hernandez Syndrome have also been reviewed in this article along with Common Neurocutaneous Syndromes. Neurofibromatosis type 1 and 2, tuberous sclerosis, Sturge Weber's disease or von Hippel Lindau diseases are the four most common types of phakomatoses. In these disorders, imaging of the brain and spine plays an important role in diagnosis and in determining the extent of involvement and the guiding in surgical interventions.

Keywords: Phakomatosis, neurocutaneous, neurofibromatosis, tuberous sclerosis, Sturge- Weber, von Hippel-Lindau, PHACES, Gomez Lopez Hernandez Syndrome, magnetic resonance imaging, computed tomography, brain, spine.

Introduction

The Phakomatoses, also known as neurocutaneous syndromes, consists of a group of disorders that tend to develop hamartomatous malformations and neoplastic growths affecting the skin, the nervous system, and other organs. The more common phakomatoses include neurofibromatosis (NF), Tuberous sclerosis (TS), Sturge-Weber disease (SW), and Von Hippel-Lindau syndrome (VHL). [1]

The uncommon neurocutaneous syndromes include Li Fraumeni syndrome, PHACES syndrome, Basal nevus syndrome, Gomez - Lopez- Hernandez syndrome, Wyburn-Mason syndrome. Neuroimaging plays an integral part of the clinical evaluation by identifying lesions in the central nervous system. It not only aids the initial diagnosis but is also fundamental in delineating the extent of central nervous system involvement, in monitoring disease progression, and in providing an anatomical roadmap for potential surgical intervention. [1].Computed tomography (CT) and ultrasonography (USG) are particularly useful for the study of abdominal manifestations.

Case Series

Case 1- Neurofibromatosis Type I

53year old male with multiple neurofibromas over the body, café au lait spots > 6, presented with headache, confusion, blurring of vision. MRI of the patient showed right sided optic nerve glioma, multifocal glioma, and plexiform neurofibroma. The patient also had history of first degree relative.

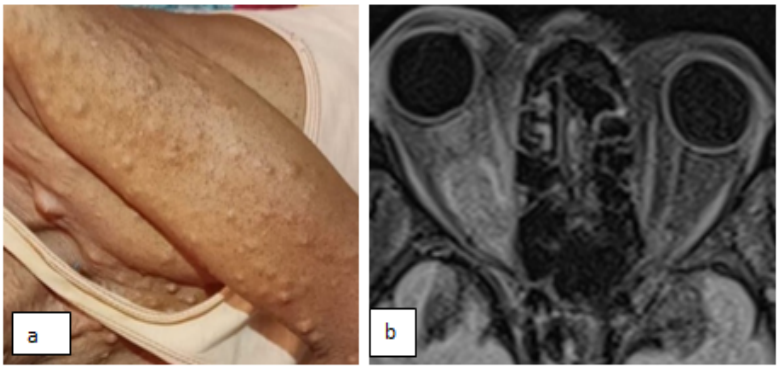


Figure 1: a) shows multiple cutaneous neurofibroma. b) MRI brain FLAIR sequence shows hyperintense fusiform enlargement of the right intraocular segment of the optic nerve suggesting optic nerve glioma

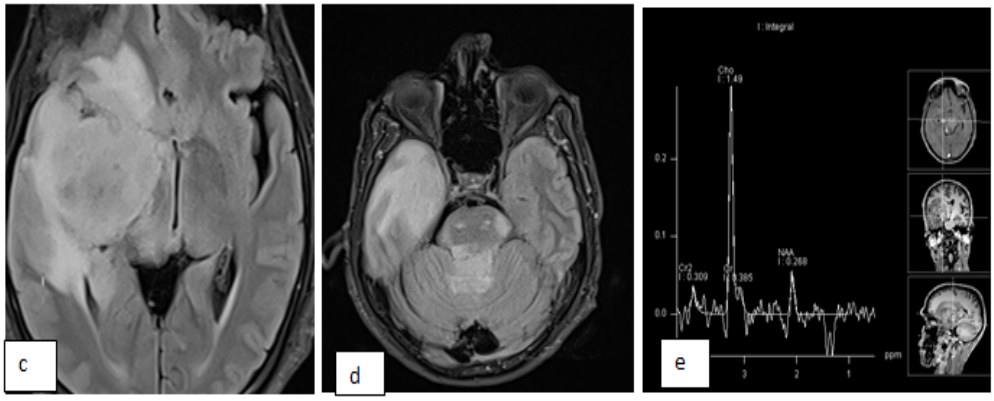


Figure 2: c and d shows MRI BRAIN FLAIR sequence shows multiple variable size and shape intra-axial space occupying lesions in the right temporal , parietal lobe , pons and ,mid brain associated with modeate vasogenic odema

Case 2- Neurofibromatosis Type II

28year old male patient with bilateral sensorineural hearing loss, vertigo, right sided facial weakness along with decreased vision on the left side since 5years.MRI study showed bilateral vestibular schwannoma, left sided parafalcine meningioma, spinal cord ependymoma. On B scan patient had lenticular opacities

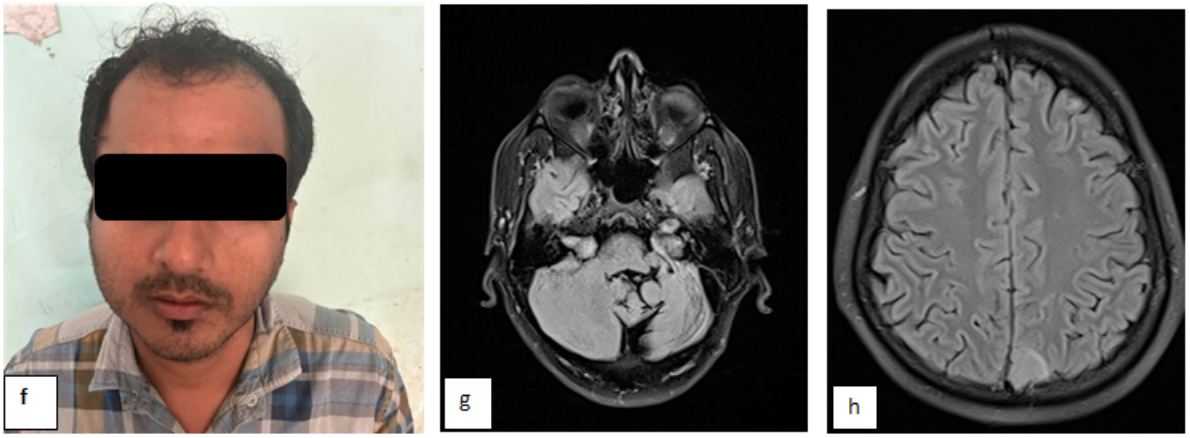


Figure 3: f) shows deviation of face towards the left side suggesting right sided facial nerve palsy associated with premature cataractous changes in the left lens g) MRI BRAIN FLAIR axial image shows hyperintense bilateral cerebellopontine angle masses extending into the internal acoustic meatus showing typical ice cream cone appearance consistent with bilateral vestibular schwannomas. h) MRI FLAIR sequence shows FLAIR hyperintense extra-axial mass in the high parietal lobe along the left parafalcine consistent with left parafalcine meningioma.

Case-3-Tuberous Sclerosis

14 year old female presented with epilepsy, mental retardation and adenoma sebaceum . MRI brain scan showed multiple subependymal nodules along the lateral ventricle and multiple scattered areas of cortical and subcortical hamartomatous lesions noted.

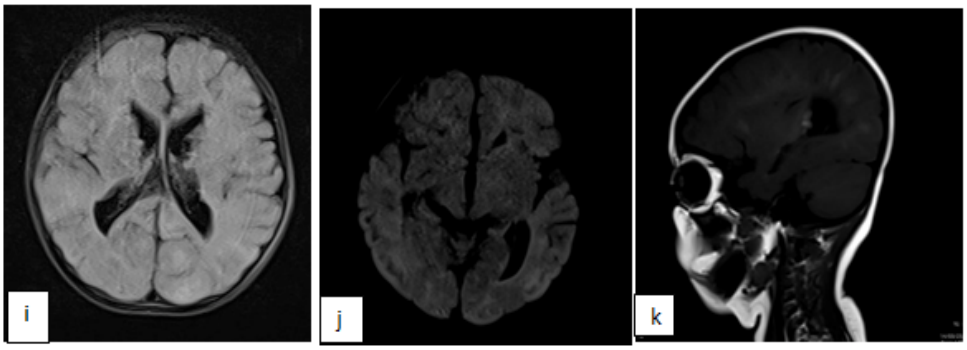


Figure 4: i) MRI BRAIN axial FLAIR image in a patient with tuberous sclerosis shows multiple nodules noted along the ependymal surface of lateral ventricle consistent with subependymal nodules . j and k) MRI BRAIN axial and sagittal FLAIR image shows cortical tubers as well circumscribed areas of high signal intensity with widened gyri.

Case 4- Von Hippel Lindau Disease

23year old female patient with pain abdomen on & off 6-7years. Headache on & off, decreased appetite, nausea, vomiting since 2 months. MRI brain showed cerebellar hemangioblastoma, spinal hemangioblastoma, pancreatic cysts, and multiple cysts in renal parenchyma

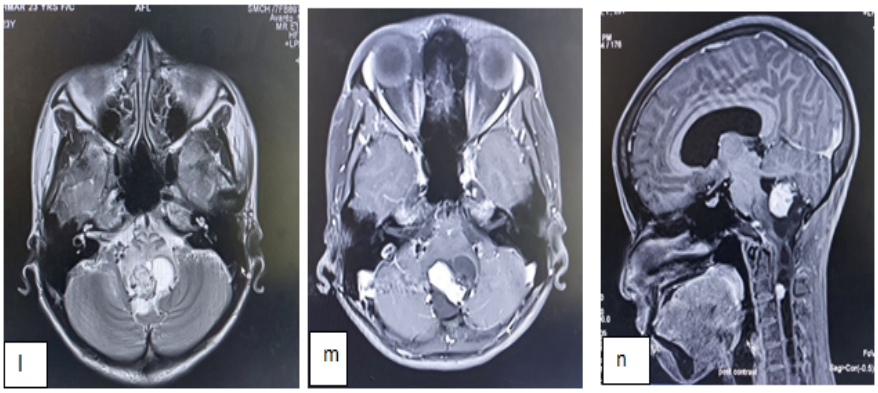


Figure 5: (l-n) T2 axial(l) images of a patient with VHL shows cystic lesion with internal mural nodule in the cerebellar vermis . Post contrast T1 axial and sagittal (m and n) image shows intense enhancement of the mural nodule consistent with cerebellar hemangioblastoma

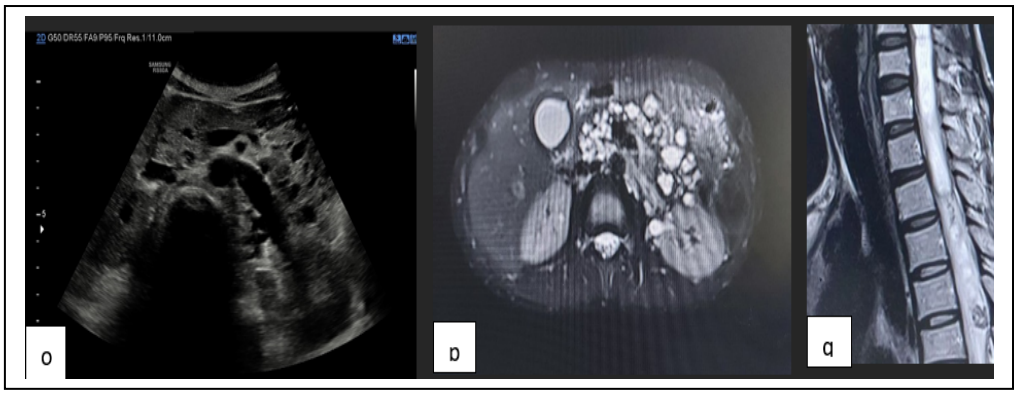


Figure 6: (o and p) USG axial and MR axial T2 FS image of a patient with VHL showing multiple variable sized cysts in the pancreatic parenchyma and left kidney. (image q and n) sagittal T2 (q) and sagittal T1 post contrast fat sat image of the cervicodorsal spine shows intramedullary hemangioblastoma as a solid nodular enhancing mass lesion in the cervical and upper thoracic cervical cord with associated syrinx formation.

Case 5-Sturge Weber Syndrome

A 15 years old male patient who is a known case of afebrile seizures from 7 years of age presented with sudden onset of abnormal movement of left upper limb and left leg for three days, On examination-Port wine stain over right half of the face was seen. His MRI scan showed cortical thinning with features of volume loss and curvilinear hypointensity in grey matter and white matter interface on T2W and Axial FLAIR images of brain. CT brain showed extensive gyral calcification of cortex and subcortical white matter with a tram track appearance in right cerebral hemisphere

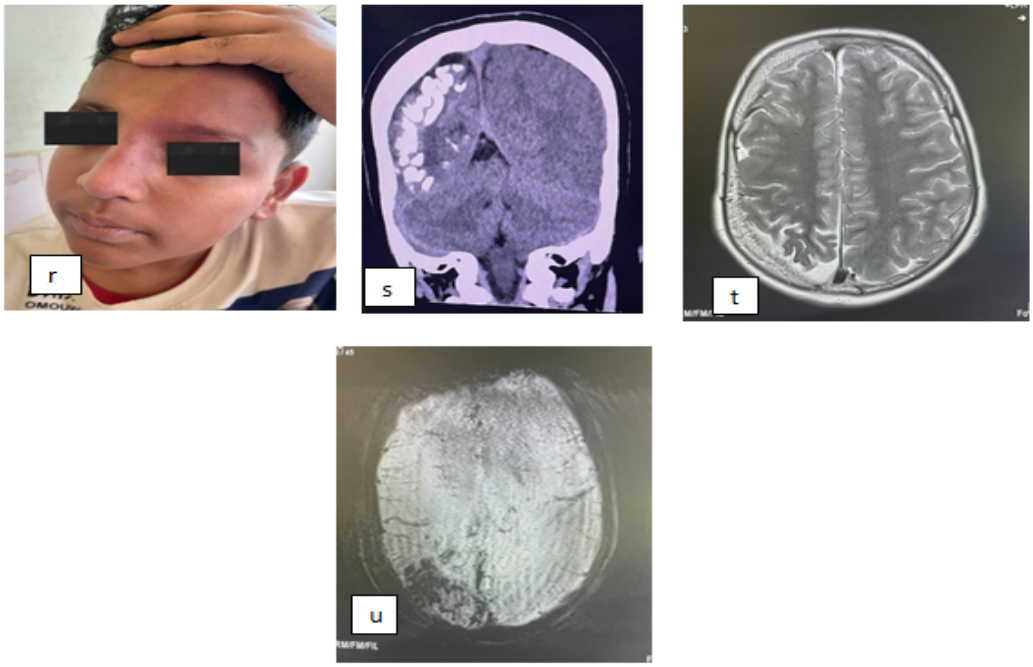


Figure 7: image (r) depicts 15 year old boy with sturge weber syndrome showing port wine stain on the left side. Coronal CT image(s) shows typical curvilinear cortical gyral calcifications in the right cerebral hemisphere with ipsilateral cerebral atrophy .Axial T2WI image (t) shows right sided cerebral hemiatrophy of the same patient .SWI axial image (u) shows curvilinear areas of blooming in the right cerebral hemisphere consistent with calcification

Case 7- Gomez Lopez Hernanadez syndrome

A 28 year old male patient who is a known case of obstructive hydrocephalus with developmental delay. Now presenting with mental retardation, ataxia, scalp alopecia and trigeminal anaesthesia. MRI scan showed Rhombencephalosynapsis, Aqueductal stenosis with partial agenesis of corpus callosum, absent septum pellucidum and Ventriculomegaly.

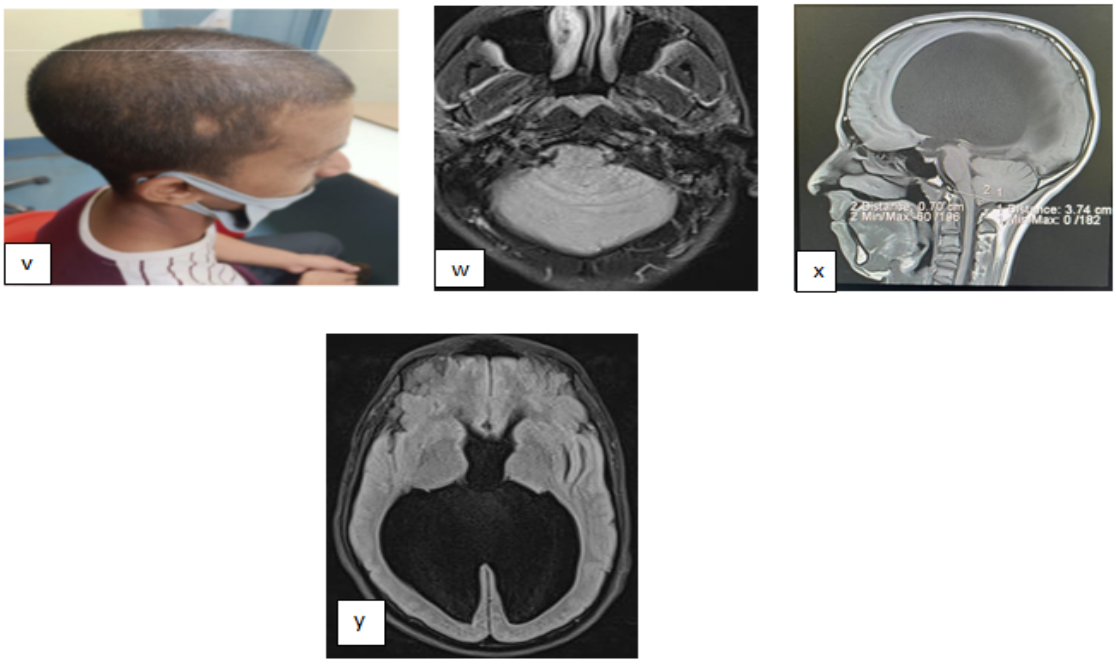
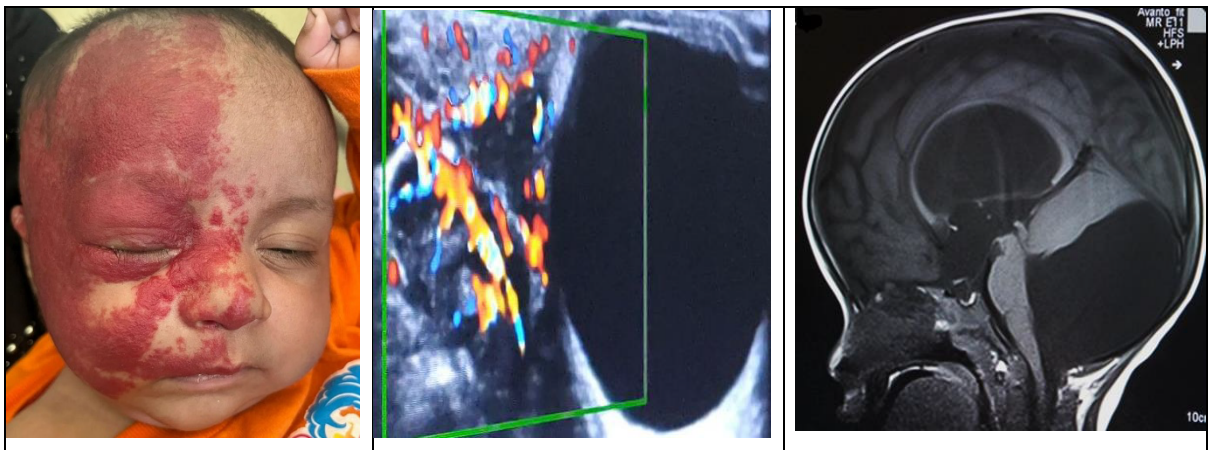


Figure 8: (v) lateral view of the 28 year old male patient with developmental delay with scalp alopecia. AXIAL FLAIR MRI BRAIN Image (w) shows transversely oriented folia with absent cerebellar vermis consistent with Rhombencephalosynapsis. Sagittal T1WI image (x) shows severe obstructive hydrocephalus caused by aqueductal stenosis associated with partial corpus callosal agenesis .Axial FLAIR MRI BRAIN image (y) shows hydrocephalus with partial corpus callosal agenesis.

Case 7-Phaces Syndrome:

4 month old child presented with intractable seizure, mental retardation with presence on angiomatous lesion in frontotemporal distribution.The infant also had respiratory distress along with cyanosis of extremities,. MRI BRAIN showed Dandy walker malformation .USG of right Orbit showed bilateral periorbital Hemangioma.



(i)	(ii)	(iii)

Figure 9: Image (i) shows 4month infant with angiomatous lesion along the trigeminal nerve distribution on the right. USG orbit image (ii) shows echogenic soft tissue lesion in the right periorbital region with prominent vascularity on colour doppler study. Sagittal T1WI MRI image (iii) shows large posterior

fossa cystic lesion associated with cerebellar and vermian hypoplasia elevating tentorium cerebelli and straight sinus communicating with the straight sinus consistent with Dandy walker malformation.

Discussion

NF1

Also known as von Recklinghausen disease is the most common of neurocutaneous and inherited tumor syndromes. The mutation of the NF1 gene, located on chromosome 17q 11.2. It is being inherited in an autosomal dominant manner. The disease affects the brain, skull, orbit, spinal column, muscular system and skin with a variety of clinical manifestations. The earliest and most common clinical findings are café au lait spots. [2]

When two or more of the following are seen, Diagnosis of NF1 is established:

- >/= 6 cafe' au lait spots with size over 5 mm in prepubertal individuals and over 15 mm in post-pubertal individuals .
- >/= 2 neurofibromas of any type or one plexiform neurofibroma ,
- Two or more Lisch nodules (iris hamartomas)
- Axillary or Inguinal Freckles
- Optic nerve glioma
- One or more distinctive osseous lesions like sphenoid dysplasia, pseudoarthrosis
- First degree relative with NF1 by the above criteria

Intracranial manifestations, orbital and skull involvement: CNS lesions are seen in 15%–20% of cases and findings include both neoplastic lesions and hamartomas. Optic pathway gliomas (OPG) are the most common CNS neoplasm. Most are juvenile pilocytic astrocytomas. The most common site of glioma after the optic pathways is mesencephalic tectum.[2–4]

The most common neuroradiologic manifestation observed in approximately 70% of patients with NF1 is nonneoplastic dysplastic white matter lesions, which are commonly seen in children. These are known as foci of abnormal signal (FASI) or unidentified bright objects (UBO) and are seen as multifocal hyperintensities on T2/FLAIR images. These are areas of myelin vacuolization or hamartoma.[2]

Neurofibromas (NFs) are commonly seen in NF1 patients. They may be localized discrete NFs or diffuse plexiform NFs (PNFs). The scalp, orbit, neck, spine, and paraspinal soft tissues are most frequently involved in NFs. The bony dysplasia, which involves a greater sphenoid wing, is another characteristic of NF1. A non-contrast CT scan of the hypoplastic sphenoid wing, which widens orbital fissure and enlarges middle cranial fossa, is demonstrated.[2]

Spinal and other manifestations: Spinal manifestations such as intraspinal and paraspinal neurofibroma, kyphoscoliosis, dural ectasia with posterior vertebral scalloping. Other CNS signs are cutaneous neurofibromas, pseudoarthroses and bowing of the longbones of extremities, ribbon like ribs as well as overgrowth in all or part of a limb.[2]

Neurofibromatosis type 2 (NF2): NF2 is an autosomal dominant disorder with mutation of NF2 gene on chromosome 22q12. In NF2.However, cutaneous manifestations are relatively rare.[2]

Intracranial manifestations: In patients with NF2, CNS lesions are mostly present in all patients. schwannomas of multiple cranial nerves, most commonly vestibular nerves, followed by trigeminal and oculomotor nerves, are characteristic lesions. Bilateral vestibular schwannomas are pathognomonic of NF2. MR scan shows mass in cerebellopontine angle cistern with widening of internal auditory canal (Ice-cream cone appearance). They are characteristically bright on T2W, iso to hypointense on T1W MR Images with strong homogeneous contrast enhancement.

Approximately 50% of patients with NF2 are affected by meningioma. They are multiple and located along falx cerebri and cerebral convexity.[2,3]

Spinal manifestations: Multiple schwannomas occurs along the spinal nerve roots, presents as dumbbell-shaped masses expanding the neural foramina with both intradural and extradural components. Multiple ependymomas can also be seen in spinal cord in NF2 especially in cervical cord and cervicomedullary junction.[2]

In order to diagnose NF2, there are two conditions which must be fulfilled:

I. Bilateral Vestibular schwannomas (VS)

II. Family history of NF2 plus 1 Unilateral VS or 2 any of the following: meningioma, glioma, neurofibroma, schwannoma, or juvenile posterior subcapsular lenticular opacities.

Tuberous sclerosis (TS): Tuberous sclerosis (TS) is a congenital multisystem disorder with prevalence of 1 in 6,000 to 10,000 and inherited in an autosomal dominant pattern.(1–3) Characteristic clinical presentation is triad of skin lesion (facial angiofibroma), mental retardation, and seizures. Hamartomatous growths are seen in multiple organ systems including CNS, eye, skin, kidneys, and lungs.[2]

Intracranial manifestations: The most common intracranial lesions seen are cortical tubers or

hamartomas, which account for 95% of all cases. [4,5]. They are most commonly found in frontal gyri. These lesions are defined by MRI in all age groups. In neonates, the affected gyri appear enlarged and hyperintense relative to surrounding unmyelinated white matter on T1W images and hypointense on T2W MR Images. The lesions appear hypointense on T1W and hyperintense on T2W images due to increasing age and progressive myelination. [2]

Subependymal nodules or hamartomas are seen in 90% of the cases. [5] They are classically found along the ependymal surface of lateral ventricles in striato-thalamic groove between the caudate nucleus and the thalamus, just posterior to the foramen of Monro. The CT and MR imaging appearances of these lesions alter with age, with progressive calcification on CT. Noncalcified lesions appear hyperintense on T1-weighted and isointense to hyperintense on T2- weighted and FLAIR images with variable post contrast enhancement. The calcified lesions typically appear hypointense on T2 and show signal loss on susceptibility weighted MR images. [2]

Subependymal giant cell astrocytoma (SEGA) is seen in approximately 15% of TS patients. [6] They classically present as partially calcified enlarging masses at the foramen of Monro with associated hydrocephalus. On both T1W and T2W images, they show mixed signal intensity and a strong enhancement. [2] In 90% of cases, white matter lesions known as radial migration lines are observed.[6].On T2W MR images, they appear as linear or curvilinear hyperintense area in the white matter, extending from subependymal nodules to cortical tubers. [2]

Other manifestations: Pulmonary lymphangioleiomyomatosis (LAM) is a rare disease, which can be seen in patients with TS and typically affects females. CT chest classically shows multiple well defined thin walled cystic lesions diffusely distributed in lung parenchyma. Almost 70%–80% of patients with TS develop renal angiomyolipoma (AML). [6,7] CT and MR imaging will show intralesional fat . Unlike AML, renal cell carcinoma is relatively rare findings of TS and the imaging findings depend on RCC subtypes. [2]

Sturge weber syndrome (SWS): This is a rare congenital disorder with an incidence of about 1 in 40,000 to 50,000 (1,8) and affects both sexes equally. A rare neurocutaneous, sporadic, congenital, vascular, and somatic condition resulting from somatic mutations in fetal ectodermal tissues that disrupt the establishment of capillary blood vessels. Many of the symptoms and signs of this disorder are caused by angiomas of the choroid, leptomeninges, and face.

Leptomeningeal capillary venous malformation and venous congestion are two pathophysiologic mechanisms that are assumed to be responsible for many of the neurologic consequences. Both mechanisms contribute to hypoxic-ischemic tissue injury.

The clinical findings: The most prevalent type of vascular malformation is port-wine staining of the forehead, upper eyelid, in the areas innervated by the trigeminal nerve (facial capillary malformation); other conditions that are observed include glaucoma, convulsions, focal neurologic deficits, intellectual incapacity, and vision field problems.

Important Diagnostic Characteristics:

Leptomeningeal enhancement from clogged cerebral veins and ensuing parenchymal ischemia on MRI T1/T2 with gadolinium. In an acute ischemia event, reduced diffusion may be one of the early phase alterations. Chronic ischemia and the gliosis that results can be seen with enhanced signal in late phase alterations. Enlarged transmedullary veins and gyriform calcifications.

CT: Prominent choroid plexus, parenchymal volume loss, and subcortical calcification.

X-ray: "Tram-track sign," or gyriform calcification of subcortical white matter. [18]

Von Hippel-Lindau syndrome (VHL): Approximately 1 in 36,000 cases of VHL are inherited through autosomal dominant inheritance [1]. Multiple neoplasms involving the central nervous system and the abdominal visceral organs are characteristic manifestations. The typical central nervous system tumors associated with this condition are hemangioblastomas, which may affect both the brain and spinal cord. The cerebellum accounts for 65 % of intracranial hemangioblastoma, followed by the brain stem of 20 %. [4]. Cyst with a solid enhancing mural nodule is the most common imaging appearance of these lesions. The solid nodule appears to be hyperintense on T2W and isointense to the brain on T1WI. There may also be flow voids inside the nodules. On post contrast study, the mural nodule enhances avidly, whereas the cyst wall rarely enhances. [2]

Patients with VHL show relatively increased incidence of various benign and malignant neoplasms of abdominal organs. The most common findings were pancreatic cysts in 50% to 91% of patients followed by renal cell carcinoma, pheochromocytoma, and neuroendocrine tumor of the pancreas and Serous cystadenoma of the pancreas. [9]

The typical imaging findings of these lesions are shown by the contrast enhanced CT or MR scans in the abdominal region.

PHACES Syndrome: PHACES syndrome is a rare neurocutaneous disorder that usually involves dermatological, neurological, cardiovascular, and ocular abnormalities.

PHACES acronym is based on the association of posterior fossa malformations, large facial hemangiomas, cerebral arterial anomalies, cardiovascular anomalies, and eye anomalies. [9] The hallmark of PHACES is the characteristic infantile facial hemangioma, typically segmental and large. [10] PHACES infantile hemangiomas may not be visible at birth, usually demonstrating a rapid growth phase during the neonatal period, and tend to regress. Facial hemangiomas in PHACES do not follow a dermatomal distribution. [11]

Clinical diagnosis of PHACES syndrome requires the presence of a characteristic segmental hemangioma or hemangioma >5 cm on the head (face or scalp) plus 1 major criterion or 2 minor criteria [12].

Major criteria anomalies:

Arterial:

- anomaly of major cerebral / cervical arteries (ICA, ACA, MCA, PCA, vertebral, basilar)
- dysplasia of the large cerebral arteries (tortuosity, kinking, dolichoectasia)
- arterial stenosis or occlusion with or without moyamoya collaterals
- absence or moderate-severe hypoplasia of the large cerebral arteries
- aberrant origin or course of the large cerebral arteries (except common aortic arch variants such as bovine aortic arch)
- persistent carotid-vertebrobasilar anastomosis (persistent trigeminal, hypoglossal, proatlantal or otic)

Structural Brain:

- posterior fossa anomalies
- Dandy-Walker complex
- midbrain/hindbrain hypoplasia/dysplasia

Ocular:

- posterior segment anomalies
- persistent hyperplastic primary vitreous
- persistent fetal vasculature
- retinal vascular anomalies
- morning glory disc anomaly
- optic nerve hypoplasia
- peripapillary staphyloma

Cardiovascular:

- aortic arch anomalies (except common variants such as bovine arch)
- aortic coarctation
- aberrant origin of subclavian artery
- aortic aneurysm

Ventral or Midline: sternal defects or supraumbilical raphe

Minor criteria:

Arterial:

- Intracranial Aneurysm

Structural Brain:

- midline anomalies
- cortical malformations

Ocular:

- anterior segment anomalies
- sclerocornea
- cataract
- coloboma
- microphthalmia

Cardiovascular:

- ventricular septal defect
- right aortic arch / double aortic arch
- systemic venous anomalies

Ventral or Midline:

- hypopituitarism
- ectopic thyroid
- sternal papule/hamartoma

Gomez Lopez Hernandez Syndrome: Gomez, Lopez Hernndez Syndrome also known as cerebellotrigeminal syndrome dermal dysplasia is a neurocutaneous syndrome with three primary features: partial alopecia of the scalp, rhombencephalosynasis and trigeminal anaesthesia. [14] The presence of rhombencephalosynapsis, with alopecia of the scalp and one of the major craniofacial manifestations, suggests a definitive GLHS. [13] As a result of ectoderm developmental arrest, cerebellar and cranial nerve abnormalities occur in GLHS. [14]

Neuroimaging: Rhombencephalosynapsis is a classic feature in GLHS. Brain MRI shows transversely oriented cerebellar folia with absent cerebellar vermis and fused cerebellar hemispheres, white matter, and dentate nuclei. Other imaging findings include ventriculomegaly/hydrocephalus, cerebellar hypoplasia, and trigeminal ganglia hypoplasia.[17,18]

There are also reports of supratentorial brain anomalies linked to rhombencephalosynapsis, such as abnormal gyri,, corpus callosal anomalies, large massa intermedia, thalamic fusion and lack of septum pellucidum. [17]

Conclusion

Almost 60 or more distinct syndromes are part of the Phakomatoses group. Although genetic testing is

available, the manifestations of these syndromes are in wide range. Detection of abnormalities early and follow up of these lesions for prior diagnosis, imaging plays an essential role. In order to guide the right treatment and prognosis, radiologists must be aware of these syndromes.

Bibliography

1. Lin DDM, Barker PB. Neuroimaging of phakomatoses. Semin Pediatr Neurol. 2006 Mar; 13(1):48–62.
2. Swarup MS, Gupta S, Singh S, Prakash A, Mehndiratta A, Garg A. Phakomatoses: A pictorial review. Indian Journal of Radiology and Imaging. 2020 Apr; 30(02):195–205.
3. Price EB, Moss HE. Osborn's Brain: Imaging, Pathology, and Anatomy. Neuro-Ophthalmology. 2014 Apr; 38(2):96–7.
4. Herron J, Darrah R, Quaghebeur G. Intra-cranial manifestations of the neurocutaneous syndromes. Clin Radiol. 2000 Feb; 55(2):82–98.
5. von Ranke FM, Faria IM, Zanetti G, Hochhegger B, Souza AS, Marchiori E. Imaging of tuberous sclerosis complex: a pictorial review. Radiol Bras. 2017; 50(1):48–54.
6. Manoukian SB, Kowal DJ. Comprehensive imaging manifestations of tuberous sclerosis. AJR Am J Roentgenol. 2015 May; 204(5):933–43.
7. Umeoka S, Koyama T, Miki Y, Akai M, Tsutsui K, Togashi K. Pictorial review of tuberous sclerosis in various organs. Radiographics. 2008; 28(7):e32.
8. Agrawal P, Kharat A, Kuber R, Shewale S. Spectrum of CT and MR findings in Sturge-Weber syndrome: A case report. Med J DY Patil Univ. 2014; 7(4):497.
9. Leung RS, Biswas SV, Duncan M, Rankin S. Imaging features of von Hippel-Lindau disease. Radiographics. 2008; 28(1):65–79; quiz 323.
10. Frieden IJ, Reese V, Cohen D. PHACE syndrome. The association of posterior fossa brain malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities. Arch Dermatol. 1996 Mar; 132(3):307–11.
11. Heyer GL. PHACE(S) syndrome. Handb Clin Neurol. 2015; 132:169–83.
12. Haggstrom AN, Lammer EJ, Schneider RA, Marcucio R, Frieden IJ. Patterns of infantile hemangiomas: new clues to hemangioma pathogenesis and embryonic facial development. Pediatrics. 2006 Mar; 117(3): 698–703.
13. Garzon MC, Epstein LG, Heyer GL, Frommelt PC, Orbach DB, Baylis AL, et al. PHACE Syndrome: Consensus-Derived Diagnosis and Care Recommendations. The Journal of Pediatrics. 2016 Nov; 178:24-33.e2.
14. Choudhary A, Minocha P, Sitaraman S. Gomez-Lopez-Hernández syndrome: First reported case from the Indian subcontinent. Intractable Rare Dis Res. 2017 Feb;6(1):58–60.
15. Rush ET, Adam MP, Clark RD, Curry C, Hartmann JE, Dobyns WB, et al. Four new patients with Gomez-Lopez-Hernandez syndrome and proposed diagnostic criteria. Am J Med Genet A. 2013 Feb; 161A(2):320–6.
16. Gomez MR. Cerebellotrigeminal and focal dermal dysplasia: a newly recognized neurocutaneous syndrome. Brain Dev. 1979; 1(4):253–6.
17. Tan TY, McGillivray G, Goergen SK, White SM. Prenatal magnetic resonance imaging in Gomez-Lopez-Hernandez syndrome and review of the literature. Am J Med Genet A. 2005 Nov 1; 138(4):369–73.
18. https://www.ajnr.org/ajnr-case-collections-diagnosis/sturge-weber-syndrome-0.