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"NEUROIMAGING SPECTRUM OF PHACOMATOSIS: INSIGHTS FROM RADIOLOGY"

Radio-Diagnosis

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ABSTRACT

"Phacomatoses" denote a diverse array of neurocutaneous disorders that impact the central nervous system (CNS), eyes and skin. This abstract provides an exploration of the neuroimaging characteristics of phacomatoses, including Sturge-Weber syndrome, tuberous sclerosis, neurocutaneous melanosis and neurofibromatosis 2. Each disorder exhibits unique radiological features crucial for diagnosis and management. For instance, tuberous sclerosis manifests cortical tubers, while Sturge-Weber syndrome presents with leptomeningeal angiomatosis. Neurofibromatosis 2 is characterized by vestibular schwannomas and meningiomas, and neurocutaneous melanosis shows melanocytic deposits. A comprehensive understanding of these radiological findings aids clinicians in formulating effective management strategies tailored to individual patient needs. Thus, this abstract underscore the significance of neuroimaging in the evaluation and treatment of phacomatoses, emphasizing the role of regular surveillance in enhancing patient care and prognosis.

KEYWORDS

Phacomatoses, Tuberous Sclerosis, Sturge-Weber syndrome, Neurofibromatosis 2, Neurocutaneous melanosis.

INTRODUCTION

The term "phacomatoses" was coined by the ophthalmologist Van der Hoeve in the 20th century when he described the retinal hamartomas associated with neurofibromatosis. [1]. Phacomatosis are group of neurocutaneous disorders mostly involves structures arising from embryonic ectoderm. Most commonly affected organs are central nervous system, skin and eyes. These are also known as neurocutaneous disorders. These consists about 60 different diseases, of which most common are tuberous sclerosis, neurofibromatosis 2, neurofibromatosis 1, Von hippel Lindau syndrome and Sturge weber syndrome. As most of these disorders have cranial manifestation, imaging is an important tool for diagnosis.

We report a few cases of phacomatosis that presented in our department.

Case 1

A one-year-old boy presented with complaints of seizures and mental disability. Clinical examination shows hypomelanic papules.

On MRI brain, there were areas of extensive T2/FLAIR hyperintensities in cortical and subcortical white matter of bilateral fronto- temporo-parietal lobes suggestive of cortical and subcortical tubers. Multiple hyperintense radial bands were noted radiating from periventricular region to cortical surface. Multiple subependymal nodules were noted along bilateral ventricular margins and few of them showing enhancement after contrast administration. (Figure 1)

These findings were consistent with diagnosis of tuberous sclerosis.

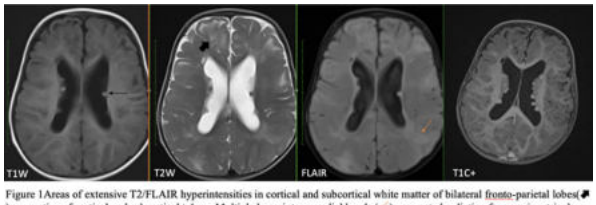

Figure 1Areas of extensive T2/FLAIR hyperintensities in cortical and subcortical white matter of bilateral fronto-parietal lobes( ) suggestive of cortical and subcortical tubers. Multiple hyperintense radial bands ( )were noted radiating from periventricular region to cortical surface. Multiple subependymal nodules(←)were noted along bilateral ventricular margins and few of them showing enhancement after contrast administration.

Case 2

A five years old female came with complaints of seizures and mental disability.

MRI brain showed multiple subependymal nodules along bilateral lateral ventricles appearing hyperintense on T2W, FLAIR and T1W images. Few of these nodules showed enhancement on contrast administration. Some nodules showed blooming on SWI- likely calcified subependymal hamartomas. (Figure 2)

These findings were consistent with diagnosis of tuberous sclerosis complex.

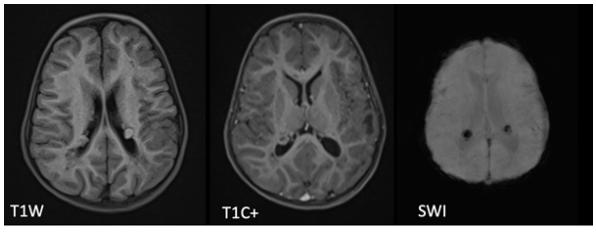

Figure 2 MRI brain showed multiple subependymal nodules along bilateral lateral ventricles appearing hyperintense on T1W images. Few of these nodules showed enhancement on contrast administration.
Some nodules showed blooming on SWI- calcified subependymal hamartomas.

Tuberous sclerosis

Tuberous sclerosis complex (TSC) is a multisystem neurocutaneous disorder with a genetic origin, distinguished by the formation of non-cancerous growths known as hamartomas in diverse organs including the brain, skin, kidney, lung, retina, and heart.The classic triad of symptoms associated with TSC includes facial adenoma sebaceum, epilepsy, and cognitive impairment.

Tuberous sclerosis complex (TSC) is marked by distinct radiological features in the brain, reflecting the presence of hamartomas, cortical tubers, subependymal nodules (SENs), and other anomalies crucial for diagnosis and monitoring. Cortical tubers, focal areas of abnormal cortical development, are prominent on neuroimaging, showing as regions of hyperintensity on T2-weighted MRI and hypointensity on T1-weighted images. SENs, small benign growths along the lateral ventricles, are pathognomonic for TSC, appearing as round lesions adjacent to the ventricular lining. Subependymal giant cell astrocytomas (SEGAs), specific tumors linked with TSC, are typically seen as contrast-enhancing masses near the foramen of Monro causing obstructive hydrocephalus. White matter abnormalities, including dysmyelination and gliosis, are common. Angiomyolipomas (AMLs), often renal, may sporadically occur in brain parenchyma, displaying fat density on CT and hyperintense on T1-weighted MRI. Additional findings encompass intracranial calcifications, ventriculomegaly, and corpus callosum abnormalities. These radiological hallmarks are pivotal in understanding and managing the neurological aspects of TSC. [2,3]

Case 3

A 56 years old female presented with complaints of seizures and

difficulty in vision from right eye. During the clinical examination, on the left side of the face, a port wine stain was observed.

On NCCT brain, gyriform calcifications are noted in right parieto-occipital lobes (figure 3A) as well as choroidal calcifications in right globe (figure 3B). A diagnosis of Sturge-weber syndrome was made.

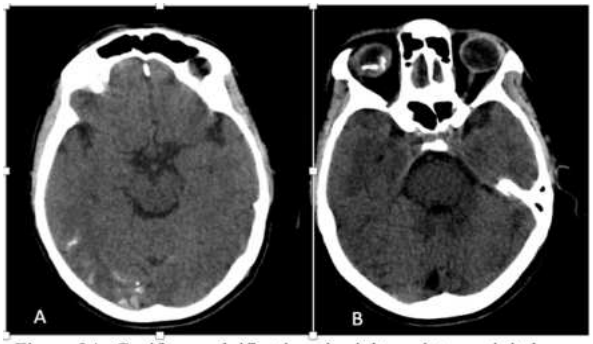

Figure 3A. Gyriform calcifications in right parieto-occipital lobes. 3B Choroidal calcification in right globe

Sturge weber syndrome

Sturge-Weber syndrome (SWS), alternatively termed encephalotrigeminal angiomatosis, is a rare type of neurocutaneous disorder consisting of vascular malformations affecting the skin, eyes, and brain. Cranial radiological imaging assumes a pivotal role in the diagnostic process and management of SWS, revealing distinct findings associated with the disorder. One of the hallmark radiological features of SWS is leptomeningeal angiomatosis, characterized by abnormal vascular proliferation involving the leptomeninges, particularly the leptomeninges overlying the cerebral cortex. This can lead to gyriform calcifications and cortical atrophy, often referred to as "tram track" calcifications due to their linear appearance on imaging studies. Other cranial radiological manifestations in Sturge-Weber syndrome encompass, cerebral atrophy seen as cortical thinning and volume loss, which can contribute to neurological symptoms and cognitive impairment. unilateral cerebral hemiatrophy, typically ipsilateral to the side of the facial cutaneous vascular malformation known as a port-wine stain is also seen. This hemiatrophy may be evident on imaging as asymmetrical enlargement of the lateral ventricle and sulcal prominence on the affected side. skull changes including thickening of the skull and bony overgrowth and less common, intracranial hemorrhage is also evident. These findings are essential for diagnosis, prognostication, and guiding management strategies in individuals with SWS.[4]

Case 4

A 26 years old female came with complains of pain and numbness in lower extremities and blurring of vision.

On MRI brain, nodular avidly enhancing masses were noted in bilateral cerebellopontine angle suggestive of vestibular schwannomas. There was also an enhancing lesion in cisternal segment of left occulumotor nerve – left oculomotor schwannoma(Figure 4). Multiple meningiomas with dural tail were noted along frontal parafalcine, left frontal convexity , right parietal convexity, right petroclival region, right cerebellar hemisphere.(Figure 5)

On MRI Spine, enhancing intradural extra medullary mass with dural tail at C2-C4 causing compression and displacement of cervical cord was seen. Also, multiple enhancing intradural extra medullary mass at D5-6, D6-7, D7-8 and D11 level causing compression and displacement – multiple schwannomas(Figure 6).The case was diagnosed to be of MISME syndrome (neurofibromatosis 2).

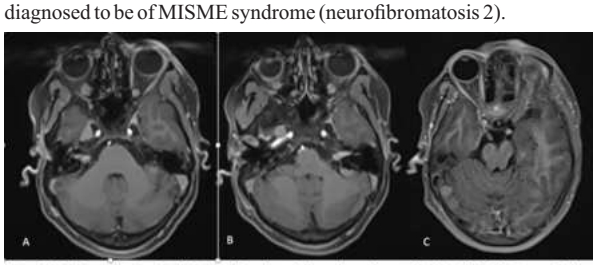

Figure 4 Nodular avidly enhancing lesions in bilateral cerebellopontine angles on right(A) and left side(B) – vestibular schwannomas. Another enhancing lesion in cisternal segment of left oculomoter nerve – schwannoma (C)

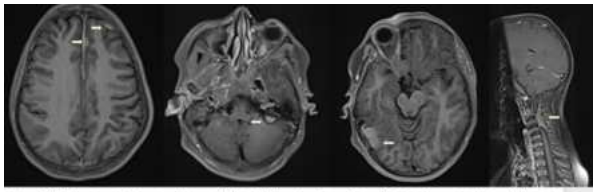

Figure 5 Multiple extra axial enhancing lesions with dural tail along frontal parafalcine, left frontal convexity, left cerebellopontine angle and right parietal convexity and in spine at C2-4 level – multiple meningiomas

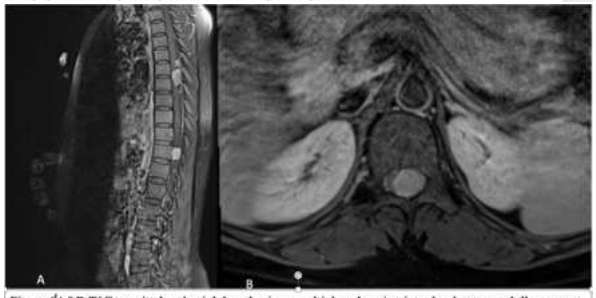

Figure 6 A&B T1C+ sagittal and axial dorsal spine - multiple enhancing intradural extra medullary mass at D5-6, D6-7, D7-8 and D11 level causing compression and displacement of thoracic cord to the right side – multiple schwannomas.

Neurofibromatosis 2

Neurofibromatosis type 2 (NF2) is indeed a rare genetic disorder and follows an autosomal dominant inheritance pattern. NF2 is chiefly attributed to loss-of-function mutations in the NF2 gene, situated on chromosome 22q11.2. This gene encodes the merlin protein, which plays a crucial role in regulating cell growth, particularly in Schwann cells.

Neurofibromatosis type 2 (NF2) is marked by the formation of nervous system tumors, primarily schwannomas and meningiomas.

Schwannomas are the hallmark tumors of NF2 and commonly involve the cranial nerves, particularly the vestibulocochlear nerve (CN VIII). Bilateral vestibular schwannomas are pathognomonic for NF2 and are typically visualized as enhancing masses along the internal auditory canal and cerebellopontine angle on MRI scans. These tumors can cause hearing loss, vestibular dysfunction, and other neurological deficits.

Meningiomas are another common intracranial tumor in NF2, arising from the meninges surrounding the brain and spinal cord. These tumors can occur at multiple locations within the intracranial compartment, including the convexities, falx cerebri, and skull base. On imaging studies, meningiomas typically appear as well-defined, contrast-enhancing masses with dural attachment in addition to cranial tumors, individuals with NF2 may develop spinal cord schwannomas and meningiomas. These tumors can cause spinal cord compression, leading to sensory and motor deficits, as well as bowel and bladder dysfunction.

Ependymomas are also seen which are typically benign tumors found within the spinal cord, presenting in as many as 53% of individuals with NF2. Although intracranial occurrences are rare, they have been reported. Notably, NF2 often presents with multiple ependymomas along the spinal cord, a characteristic feature of the condition.[5]

Cranial radiological imaging plays a crucial role in the diagnosis, monitoring, and management of NF2, revealing specific findings associated with the condition.

Case 5

An 18-year-old male came with complain of headache and projectile vomiting. On examination multiple melanotic nevi were seen on arm which were present since birth.

On NCCT brain moderate communicating hydrocephalus and multiple nodular and gyriform hyperdensity with foci of calcification in left insular cortex, left frontal lobe, lower pons and medulla was noted. (Figure 7)

On MRI Brain, nodular as well as gyriform lesion in left insula showing intrinsic T1 hyperintensity, appearing T2 and showing enhancement on post contrast T1 images and showing blooming SWI (Figure 8). On MRI spine, multiple nodular as well as plaque like

lesions in thecal sac of dorsal spine in intradural extramedullary space appearing T1 hyperintense, T2 hypointense and show avid enhancement on post contrast were seen (Figure 9). The case was diagnosed as neurocutaneous melanosis.

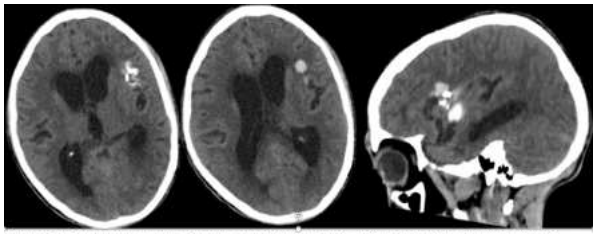

Figure 7 Multiple nodular and gyriform hyperdensity with foci of calcification in left insular cortex and left frontal lobe with moderate communicating hydrocephalus.

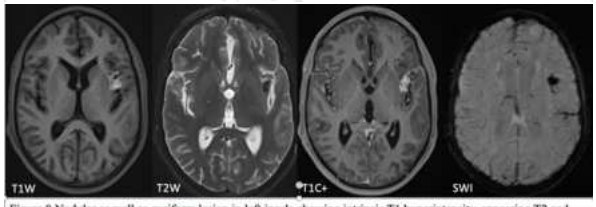

Figure 8 Nodular as well as gyriform lesion in left insula showing intrinsic T1 hyperintensity, appearing T2 and showing enhancement on post contrast T1 images and showing blooming SWI.

Figure 9 Multiple nodular as well as plaque like lesions in thecal sac of dorsal spine in intradural extramedullary space appearing T1 hyperintense , T2 hypointense and show avid enhancement on post contrast

Neurocutaneous melanosis

Neurocutaneous melanosis (NCM) is a rare disorder defined by the presence of extensive or multiple pigmented nevi along with the occurrence of leptomeningeal melanosis or melanoma. Importantly, this condition manifests without malignancy in the skin lesions and without affecting other bodily organs. The first documentation of NCM dates back to 1861, credited to Rokitanski. It's believed that the pathogenesis of Neurocutaneous melanosis (NCM) entails the dysplasia of neuroectodermal melanocyte precursor cells. This dysplasia triggers the proliferation of melanin-producing cells within both the skin and the leptomeninges. Nonetheless, the exact mechanism driving this process remains elusive. People with NCM also face the potential risk of malignant transformation into leptomeningeal melanoma and cutaneous melanoma

On CT scans, melanocyte accumulations in neurocutaneous melanosis (NCM) appear as subtle areas of hyperdensity. MRI is the favored imaging technique for NCM, wherein melanocytic deposits usually appear hyperintense to isointensie on precontrast T1-weighted images because of the paramagnetic characteristics of melanin. The anterior temporal lobe, notably the amygdala, represents the primary site of melanocytic deposits in NCM, with other common locations encompassing the inferior surface of the cerebellum, the ventral aspect of the medulla, the pons, the cerebral peduncles, and the upper cervical spinal cord. Typically, the dura remains unaffected, and primary involvement of the cerebral parenchyma may happen through melanin-containing macrophages and melanocytes. Secondary involvement occurs via spread through the Virchow-Robin spaces, sparing the deep cerebral parenchyma. Malignant progression to leptomeningeal melanoma and cutaneous melanoma poses a significant risk for individuals with NCM, manifesting in approximately 40 to 60% of symptomatic cases. Distinguishing between benign and malignant parenchymal melanocytosis on MRI presents challenges, yet certain features such as the presence of mass lesions, nodular or thick plaque-like contrast enhancement, edema, growth, necrosis, and hemorrhage may indicate malignant transformation. [6,7]

CONCLUSION

In conclusion, neuroimaging, particularly CT and MRI, is indispensable in diagnosing and managing cranial phacomatoses like neurofibromatosis 2, tuberous sclerosis, neurocutaneous melanosis and Sturge-Weber syndrome. By discerning distinct radiological features, clinicians can tailor treatment and prognostic assessments accurately. Regular surveillance with imaging detects disease progression and complications early, optimizing patient outcomes.

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