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Neurocutaneous Syndromes: Diagnostic Aspects of Magnetic Resonance Imaging

Antonio G. L. Junior[1]*, Lorena Saraiva de Alencar[1], Nina M. P. Abreu[1], July R.M Machado[1], Hilanne Linhares Andrade de Aquino[1], Juliana Saraiva de Alencar[3], Lucas Olímpio Coimbra[2], Pablo P. A. Coimbra[1].

1.Radiology Unit, Hospital Antonio Prudente, Fortaleza, CE, Brazil.

2.Centro Universitário Christus, Fortaleza, CE, Brazil.

3.Universidade Estadual Do Ceará, Fortaleza, CE, Brazil.

Corresponding Author: Antonio Gomes Lima, Radiology Unit, Hospital Antonio Prudente, Fortaleza, CE, Brazil.

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Abstract

Neurocutaneous syndromes are diseases that generate ocular, cutaneous and neurological changes, being generated by changes in structures that are formed from the embryonic ectoderm and most of them have a genetic origin and can generate tumors in multiple organs. The main examples of this group of diseases are type I Neurofibromatosis, which usually manifests before the age of 10 years, type II neurofibromatosis, which usually affects patients between 20 and 40 years old and other diseases.

For example, Tuberous Sclerosis and the Sturge Weber Syndrome. The specific findings of magnetic resonance imaging and the patient's clinical history are essential to establish the diagnosis of neurocutaneous syndromes. Clinical cases from our institution between 2017 and 2019, which were diagnosed by radiologists specialized in neuroimaging and based on the diagnosis of the clinical history and diagnostic imaging criteria of this syndrome, will be discussed.

***Keywords**: Neurocutaneous Syndromes, Magnectic Resonance Imaging, Neurofibromatosis.*

Introduction

Neurocutaneous syndromes are clinical entities that comprise oculoneurocutaneous disorders characterized by the involvement of structures arising from the embryonic ectoderm. Most are hereditary and can form tumors in various organs, especially in the CNS. The three main diseases in this group are type 1 neurofibromatosis (NF1), type 2 neurofibromatosis (NF2) and von Hippel-Lindau syndrome; also highlighting the tuberous sclerosis complex and some rare syndromes such as Sturge Weber syndrome and neurocutaneous melanosis. NF1 is the most common, manifesting in general up to 10 years of age, mainly in café au lait spots, ephelides, cutaneous neurofibromas, optic gliomas and iris hamartomas, which may be associated with intramedullary astrocytoma. NF2 is less common, affecting patients aged between 20 and 40 years, associated with bilateral vestibular schwannomas, in addition to meningioma, glioma or juvenile/cataract posterior subcapsular lenticular opacities. Tuberous sclerosis complex is characterized by non-malignant hamartomas and neoplastic lesions in the brain, heart, skin, kidney, lung and other organs, in addition to being associated with autism, epilepsy and neurocognitive disabilities. Sturge Weber syndrome is represented by a port-wine stain, usually on the face, and ipsilateral leptomeningeal angioma, seizures, and glaucoma. Neurocutaneous melanosis is characterized by a pigmented skin area and leptomeningeal melanoma. Specific magnetic resonance imaging (MRI) findings added to clinical criteria are capable of establishing the diagnosis of neurocutaneous syndromes. This study is a pictorial essay focused on demonstrating MRI findings and imaging criteria in neurocutaneous syndromes. Clinical cases diagnosed at our institution between 2017 and 2019 by radiologists specialized in neuroimaging will be presented, based on the current diagnostic criteria for neurocutaneous syndromes.

Case Presentation

Clinical Case 1
Neurofibromatosis type I

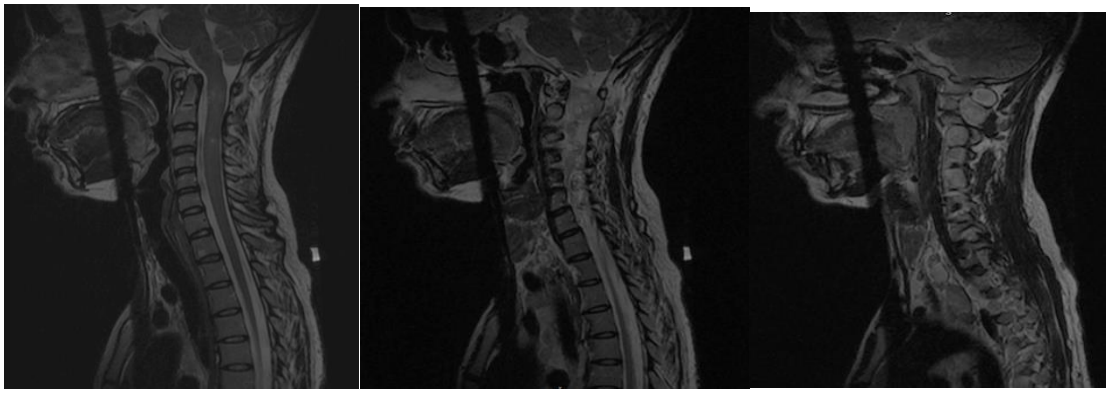


Figure 1.MRI sagittal T2-weighted sections. A and B, cervical neurofibromas (arrows) extending to the foraminal space and to the extra-dural vertebral canal, causing medullary compression with signal change in the spinal cord (C) at the levels of the vertebral bodies of C2 and C3 (○), in a 32-year-old female patient with cutaneous neurofibromas and a diagnosis of NF1.

Clinical Case 2

Neurofibromatosis type II

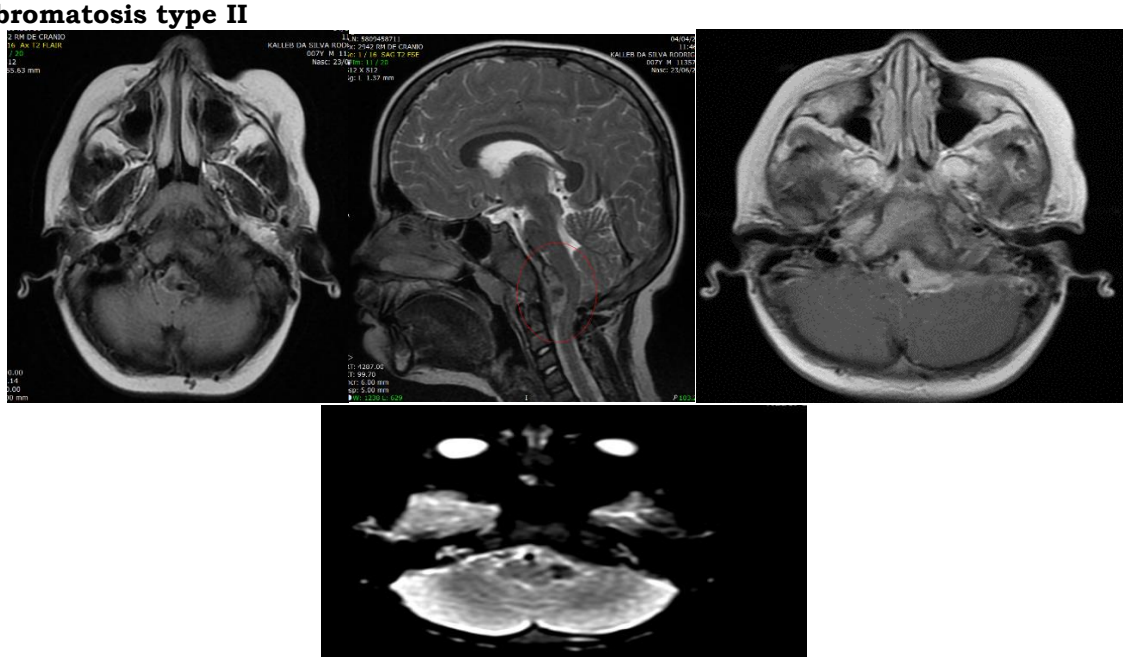


Figure 2. A, axial T2 FLAIR; B, sagittal T2 FSE; C, post-contrast T1; D axial DWI, show solid extra-axial expansive formation (arrows), with diffusion restriction and intense contrast enhancement, located in the craniocervical transition to the left, compressing the ventrolateral face of the medulla and cervical spinal cord , compatible with meningioma. Imaging findings in a 7-year-old male patient with seizures, facial paralysis and skin patches.

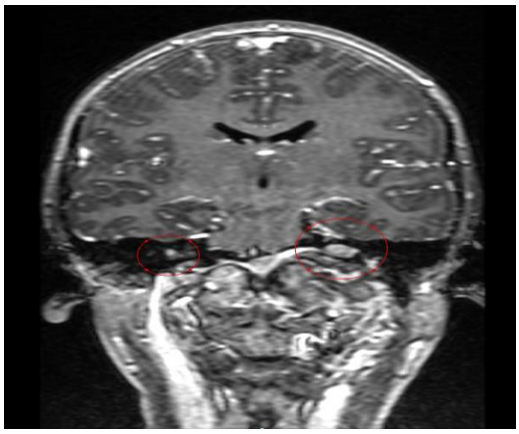


Figure 3. Sagittal T2 FSPGR GD, showing expansile nodular formations located inside the internal auditory meatus (○) bilaterally, showing homogeneous contrast enhancement, compatible with vestibular Schwannomas, in a 7-year-old male patient with seizures, facial paralysis and skin spots. The set of findings is diagnostic of NF2.

Clinical Case 3

Neurofibromatosis Type II

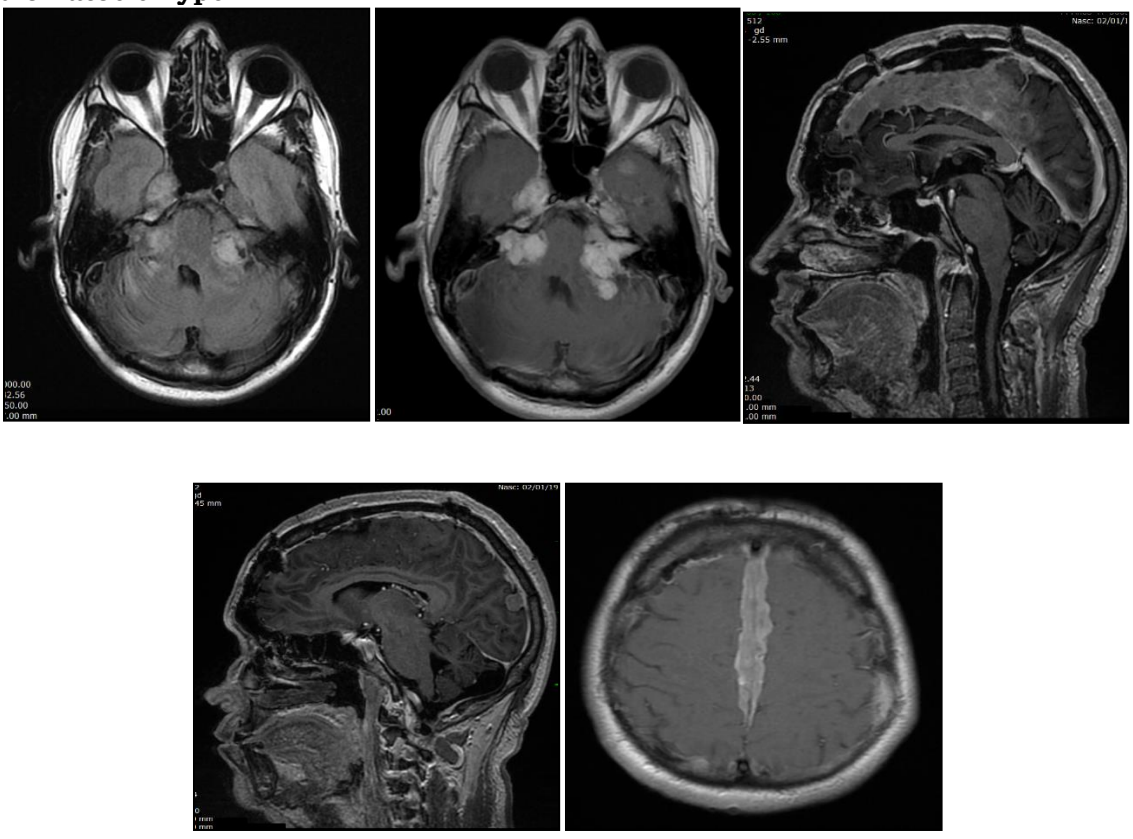


Figure 4. A, axial T2 FLAIR and B, axial T1 post-contrast show expansive formations , with contrast enhancement, located in the cerebellopontine angle bilaterally, determining an expansive effect on the

brainstem, suggesting acoustic schawannomas. C sagittal FSPGR GD, showing diffuse thickening of the sickle, with contrast enhancement, suggestive of plaque meningioma (*); D, sagittal FSPGR and E, axial T1 after contrast, show meningiomas in the sagittal sinuses and high posterior parietal convexity (arrows). Imaging findings in a 44-year-old male patient with diagnostic criteria for NF2.

Clinical Case 4

Tuberous Sclerosis Complex

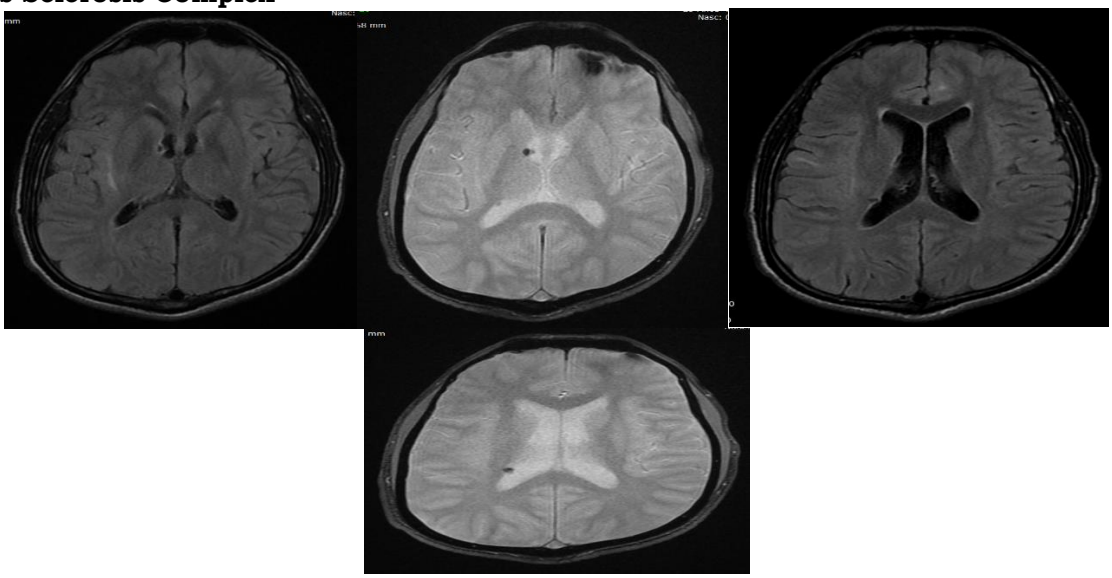
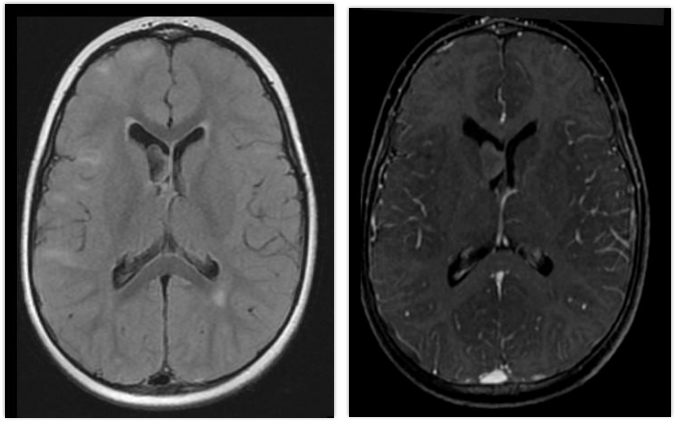


Figure 5. MRI axial sections, A and C T2 FLAIR; B and D T2 GRE show subependymal nodules (○) in the body of the lateral ventricles, some calcified. There are also rare areas of cortical thickening (arrow), which assume a triangular appearance, associated with hypersignal in the white matter, with loss of cortical/subcortical differentiation, suggesting tubercles. Imaging findings in a 23-year-old male patient diagnosed with Tuberous Sclerosis.

Clinical Case 5

Tuberous Sclerosis Complex + Giant Cell Astrocytoma



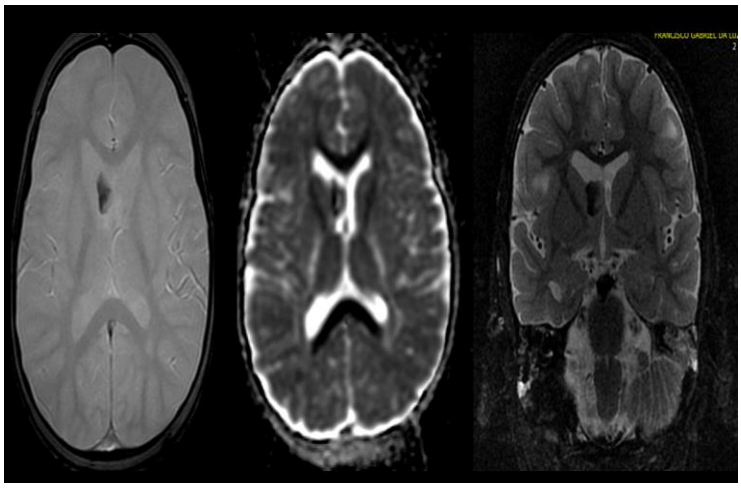


Figure 6.RM axial sections A, T2 FLAIR; B, post-contrast T1; C, T2 GRE; D, DWI; And, coronal STIR section demonstrates an expansive lesion (arrows) located in the anterior horn of the right lateral ventricle, with enhancement after injection of contrast medium, with central calcification, suggestive of subependymal giant cell astrocytoma, in a 2-year-old patient, gender masculine.

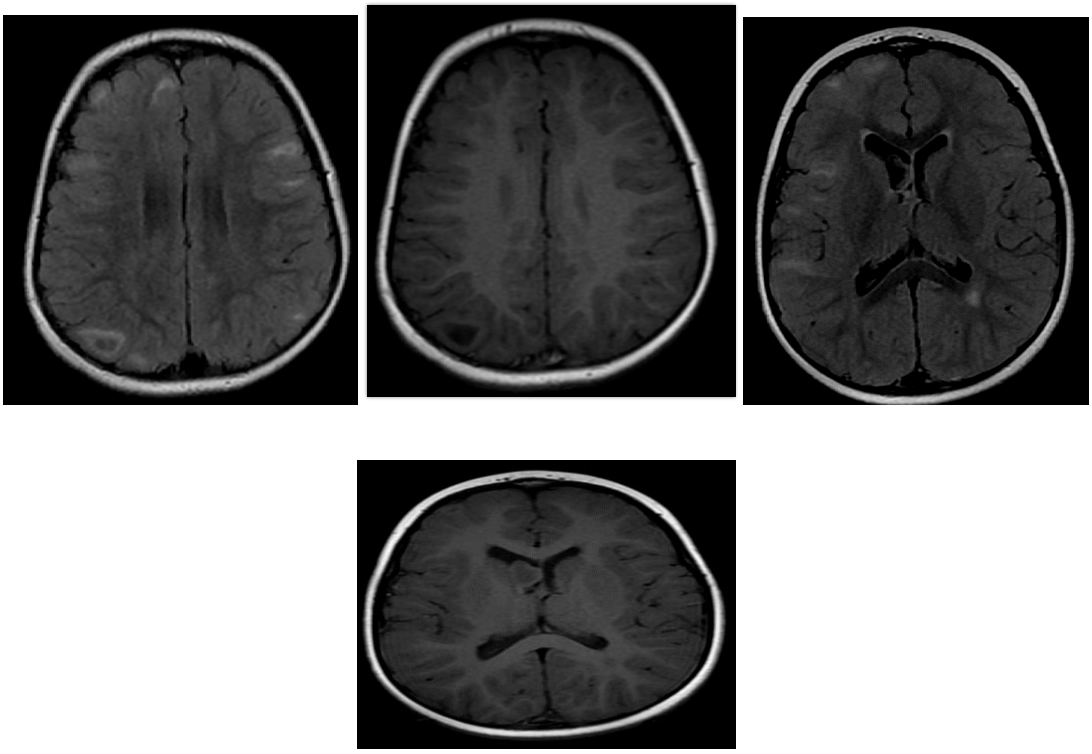


Figure 7. Cortical/subcortical areas of hypersignal in T2 FLAIR (A and C), the largest ones showing low signal correspondence in T1 (B and D) in the cerebral hemispheres, without enhancement after contrast medium injection, without restriction to molecules of water, usually related to cortical tubercles, in a 2-year-old male patient.

Clinical Case 6

Tuberous Sclerosis Complex

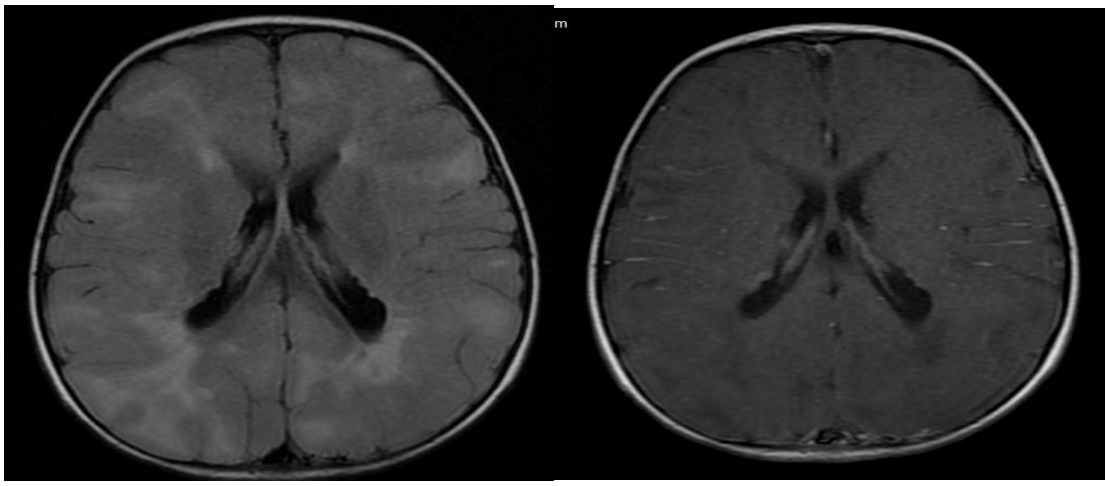


Figure 8. A, axial T2 FLAIR and B, axial T1 post-gadolinium, showing multiple images compatible with cortical tubercles distributed over the cerebral hemispheres and subependymal nodules (arrows) in a 1-year and 4-month-old female patient.

Clinical Case 7

Sturge Weber Syndrome

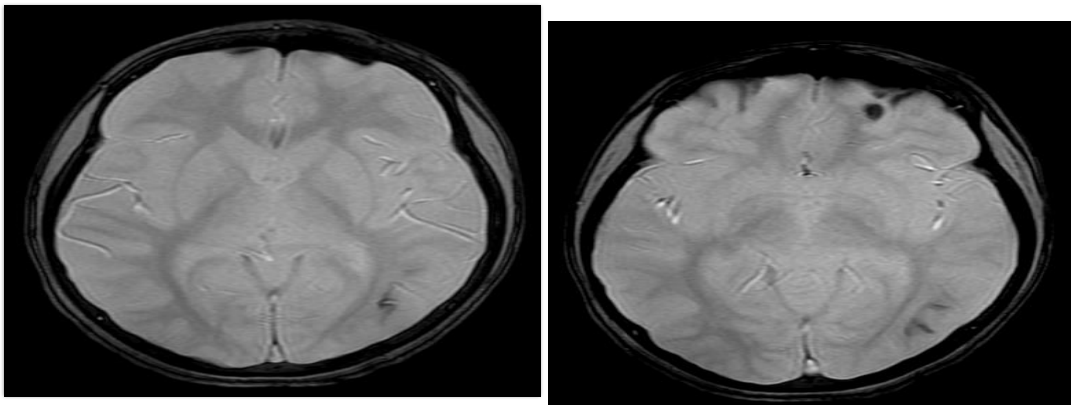


Figure 9. Gyriform T2-weighted GRE (A and B) hyposignal on the surface of the left parieto-occipital transition gyrus (arrows), performed in correlation with an external tomographic study, suggests calcifications associated with the prominence of the regional leptomeningeal vascularization, with no changes in the other sequences of RM, in a nine-year-old male patient with cutaneous-facial angioma, similar in color to port wine.

Clinical Case 8

Neurocutaneous melanosis

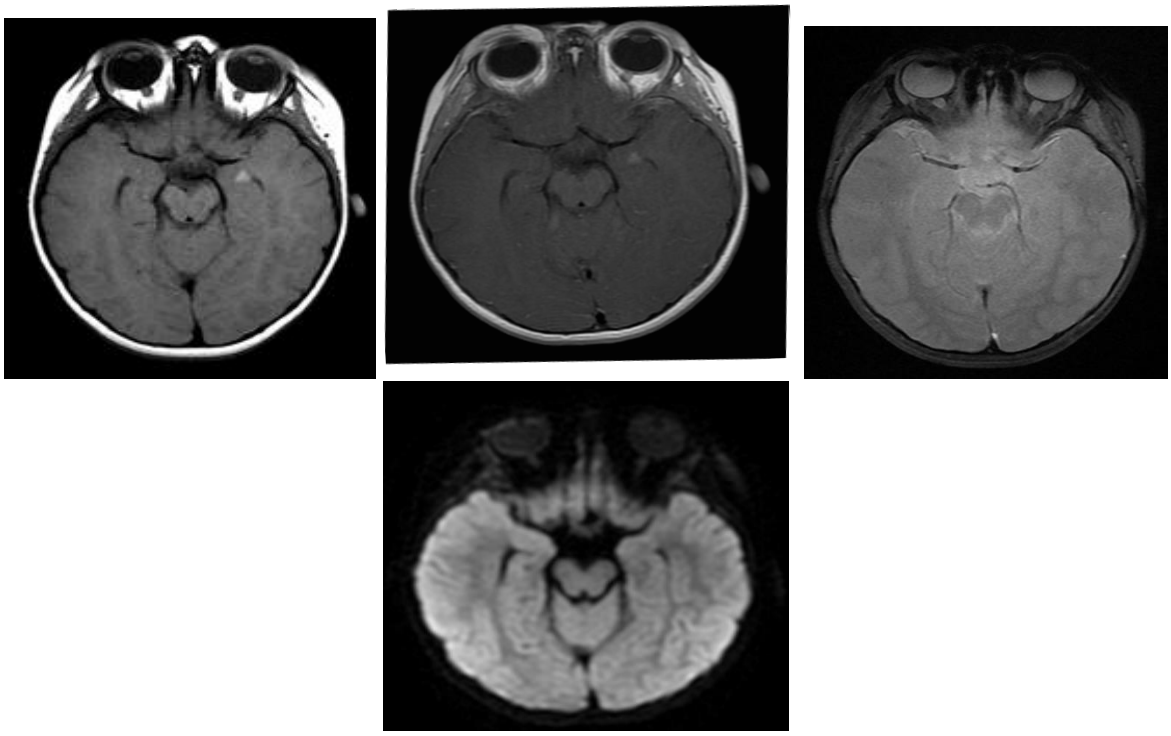


Figure 10. MRI axial sections. A, T1-weighted with evidence of a small area with high signal (arrow) located in the left mesial temporal lobe (tonsilla). B, T1 with paramagnetic contrast, no enhancement. C, T2 GRE; and D, DWI unchanged. Child, female, 1 year and 7 months old with Giant Congenital Neurocutaneous Melanosis.

Discussion

Neurocutaneous syndromes correspond to a set of congenital or hereditary diseases that have many characteristics in common: hereditary transmission, involvement of organs of ectodermal origin (nervous system, eyeball, retina and skin), slow evolution of lesions in childhood and adolescence, and disposition for fatal malignant transformation. Except for Sturge-Weber syndrome, these major neurocutaneous syndromes are genetically determined, although sporadic cases may occur. Our study reviews the clinical features of the most common neurocutaneous syndromes.

Conclusion

In summary, it is essential to carry out an early diagnosis of neurocutaneous syndromes, based on the patient's clinical history and on the specific changes present in the MRI for the proper treatment and for the prevention of neoplasms and other changes present in sick people who have these diseases.

Conflict of Interest Disclosures: The authors declare that they have no conflict of interest.

Ethical Statement: Informed consent was obtained from the patients for publication of this case report.

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