

Neurocutaneous Melanosis: A Rare Case

Case Report

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Abstract

We report a 15-year-old male patient who presented with sudden onset generalized tonic clonic convulsions and had multiple extensive large pigmented nevi covering entire back. Head CT revealed hyperdense foci in bilateral temporal lobes. MR imaging of the brain revealed multiple T1-Weighted Imaging (T1W) hyperintense foci in bilateral anterior temporal lobes with leptomeningeal enhancement. These findings along with skin lesions are suggestive of neurocutaneous melanosis.

Abbreviations: T1W: T1-Weighted Imaging; SWI: Susceptibility-Weighted Imaging; T2W: T2-Weighted; DWI: Diffusion-Weighted Imaging; FLAIR: Fluid Attenuated Inversion Recovery
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Case Report

15-year-old male patient was brought to the casualty department with a first episode of sudden onset generalized tonic clonic convulsions followed by a fall. There was no history of any other neurological symptoms till date. General examination revealed multiple extensive large pigmented nevi covering entire back (>3) (Figure 1), which were present since birth without any change in size. Family history was non-contributory. CSF revealed normal glucose (63 mg/dl) and protein (460 mg/L) levels with a white cell count of 2 cells/mm³. The CSF findings were thus unremarkable.

Head CT scan revealed small focal hyperdense areas in bilateral temporal lobes (60.5 Hounsfield Unit) (Figure 2) simulating hemorrhagic contusions. MR imaging of the brain revealed multiple T1W hyperintense foci in bilateral anterior temporal lobes corresponding to hyperdense areas noted in CT scan (Figure 3). In addition, a hyperintense lesion was also seen in right frontal

lobe (Figure 4). These lesions were persistently hyperintense on T1W fat saturated (sat) images. These areas were not appreciated on T2-Weighted (T2W) images. No evidence of blooming on Susceptibility-Weighted Imaging (SWI) images was noted thus ruling out hemorrhage. No area of restricted diffusion was noted on Diffusion-Weighted Imaging (DWI) images. No perifocal vasogenic edema was seen. Extensive leptomeningeal enhancement was seen on post-contrast Fluid Attenuated Inversion Recovery (FLAIR) images (Figure 5). MR imaging of spine was unremarkable.

Giant congenital melanotic nevi in the trunk along with classical locations of intraparenchymal melanosis and leptomeningeal melanosis were thus suggestive of neurocutaneous melanosis. The patient was discharged on anti-convulsants with regular follow up.

Discussion

Neurocutaneous melanosis is a rare phakomatosis resulting from congenital dysplasia of the neuroectodermal melanocyte precursor



Figure 1: Multiple extensive hairy pigmented nevi seen over the back.



Figure 2: Axial Plain CT reveals hyperdense areas in bilateral anterior temporal lobes.

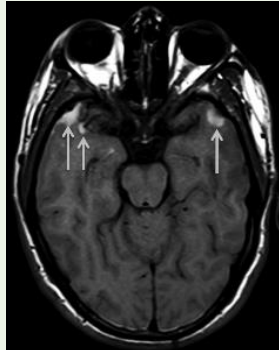


Figure 3: Plain axial T1W images show multiple focal hyperintense lesions seen in bilateral temporal lobes.

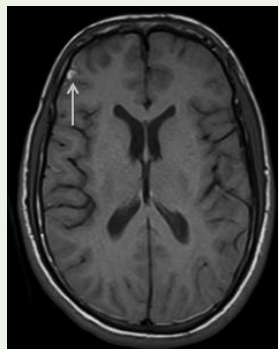


Figure 4: Plain axial T1W images show focal hyperintense lesion in right frontal lobe.

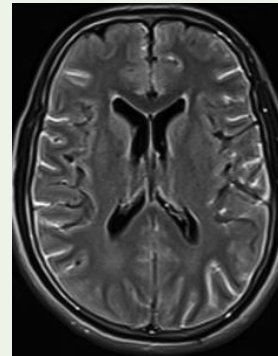


Figure 5: Post-contrast FLAIR images reveal bilateral fronto-parietal leptomeningeal enhancement.

cells [1]. The prevalence of large congenital melanotic nevi in skin is estimated to occur in less than one in 20,000 newborns [2]. The risk of neurocutaneous melanosis in these patients varies from 1 to 12% [2]. Though disease is considered sporadic, Kinsler et al. have reported post-zygotic mutations in codon 61 of NRAS gene to be the cause of this disorder [1,3]. This disease usually presents at 2 years of life, however, a second smaller peak of symptom development occurs during puberty or the young adult years as in our case [4]. Clinical manifestations include focal or generalized seizures, aphasia, and motor or cranial nerve deficits [4].

Though CT is not optimal compared with MR imaging for demonstrating neurocutaneous melanosis, the foci of melanin containing cells may appear as areas of high attenuation. Rebecca YT Ng et al. have reported a case of diffuse leptomeningeal melanocytosis showing the areas of cisternal hyperdensity mimicking subarachnoid hemorrhage; similar to our case in which intraparenchymal melanosis mimics hemorrhagic contusions on CT scan [5]. MR imaging findings of neurocutaneous melanosis include foci (typically 3 cm or less in size) of T1 and T2 shortening within the brain parenchyma and meninges [1]. These imaging characteristics of melanin are thought to be caused by the presence of paramagnetic free radicals (indolesemiquinones and semiquinones) [1]. Most common location of these foci is in the anterior temporal lobe (especially in the region of the amygdala) followed by cerebellar white matter, cerebellar nuclei, and the brainstem [1]. Melanin appears isointense to hypointense on T2W images [6]. Leptomeningeal enhancement has been reported in neurocutaneous melanosis [1].

Substances that appear hyperintense on T1 include methemoglobin, melanin, lipid, protein, and minerals. Findings of absence of blooming on SWI and the persistence of hyperintensity on T1W fat sat images along with the classical location of intraparenchymal lesions and leptomeningeal enhancement in our case is suggestive of melanotic lesions in the brain.

Another differential diagnosis of melanin containing lesion in the brain in a patient with skin lesions includes metastatic malignant melanoma. However multiple foci of metastatic melanoma are predominantly located at the gray-white matter junction and have

surrounding vasogenic edema (similar to other metastasis) with post-contrast enhancement [1].

Follow up is suggested to watch for malignant degeneration into melanoma in the brain by MRI (indicated by progressive growth, surrounding vasogenic edema or mass effect, or development of central necrosis) and in skin by clinical examination (changes in color, size, shape, rapid growth rate, proliferative nodules or ulceration) warranting the need of biopsy.

The treatment is palliative and treatment options include systemic intrathecal chemotherapy, radiotherapy and ventriculoperitoneal shunt in cases of hydrocephaly [4,6]. Regardless of the treatment, the prognosis is bad [6].

Conclusion

Thorough clinical examination and appropriate neuroimaging will help to identify this rare and dreadful phakomatosis.

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