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Beyond the usual: Cortical and subcortical white matter involvement as a presentation of Wilson's disease

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Abstract:
Background*: Wilson's disease or hepatolenticular degeneration is a metabolic disorder with devastating complications. It is also treatable and hence, early diagnosis can tremendously alter the patient's prognosis.*
In this report, we present a 12-year-old boy with behavioral changes including, inappropriate laughter and crying spells due to cortical and subcortical white-matter involvement in a case of Wilson's disease. To the best of our knowledge, cortical and subcortical white matter involvement in a case of Wilson's disease is exceptionally rare. We believe that all clinicians and radiologists when challenged with a case presenting with signs of cortical involvement must be aware of the possibility of this rare presentation of Wilson's disease.
Key Word*:Cortical and Subcortical white matter involvement in Wilson's disease, face of a giant panda, Wilson's, Hepatolenticular degeneration.*

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I. Introduction

Wilson's disease typically presents in childhood. Its neurological manifestations are mostly extrapyramidal. It has numerous potential neuropsychiatric symptoms. [1] Cognitively, this profile is consistent with the disturbance of frontosubcortical networks. [2]The dorsomedial prefrontal cortex (DMPFC) has been proven to be involved in different states, which involve a strong emotional component. [3] Magnetic resonance imaging (MRI) brain typically reveals T2 hyperintense changes in the putamen, lentiform nucleus, thalamus, and brainstem. Cortical and subcortical white matter lesions are unusual but may sometimes be the only signs of Wilson's. [4,5]

II. Case Report

A 12-year-old boy was admitted to our hospital with abnormal posturing of upper and lower limbs and change in behavior for 2 years. The patient had difficulty in performing upper limb movements, abnormal posturing, intermittent contractions, and progressively increasing difficulty in limb movements until finally, the patient became bedridden.

The accompanying changes in the patient's behavior including inappropriate laughter and inappropriate crying. The patient also had bladder incontinence.

On examination: Generalised dystonia was present.

Signs of frontal lobe involvement were present. The frontal lobe is involved in mediating context-specific responses which when affected causes disinhibition including excessive inappropriate laughter or crying. Our patient presented with apathy, indicating the involvement of the frontal lobe.

MRI Findings:

Subcortical T2 hyperintensity was seen in both dorsomedial prefrontal regions [Figure 1]. Subcortical FLAIR hyperintensity was seen in both dorsomedial prefrontal regions and also in both the temporal lobes predominantly on the right side. [Figure 2]. There was a small area of diffusion restriction in the subcortical white matter as well as within an area of the cortex in the right temporal region. The corresponding hypointensity on ADC sequences was also seen. [Figure 3]. There was diffuse tegmental T2 hyperintensity with sparing of the red nucleus [Figure 4].

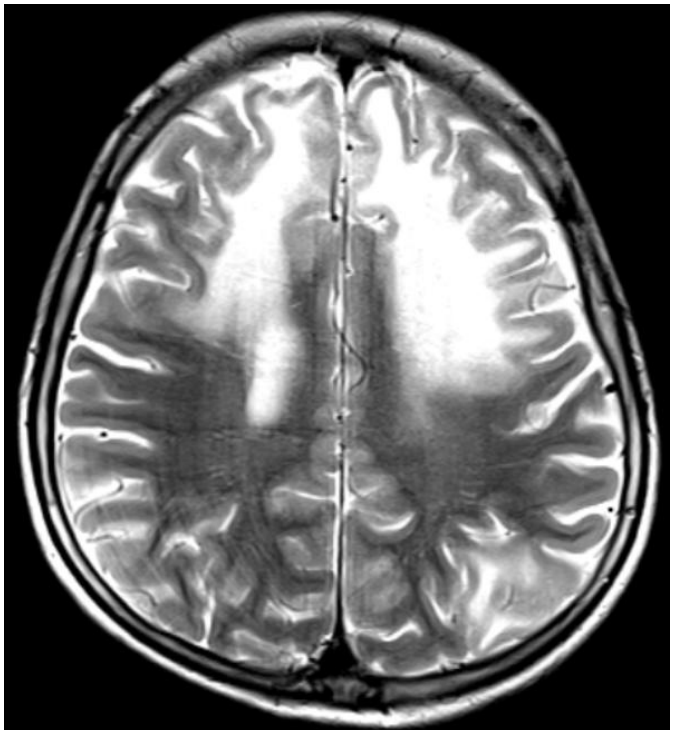
Considering all the neuroimaging findings, a diagnosis of Wilson's disease with involvement of the cortex and subcortical white matter was made.

Serum Ceruloplasmin value was low 0.1 OD [Normal range being 0.2-0.5 OD]

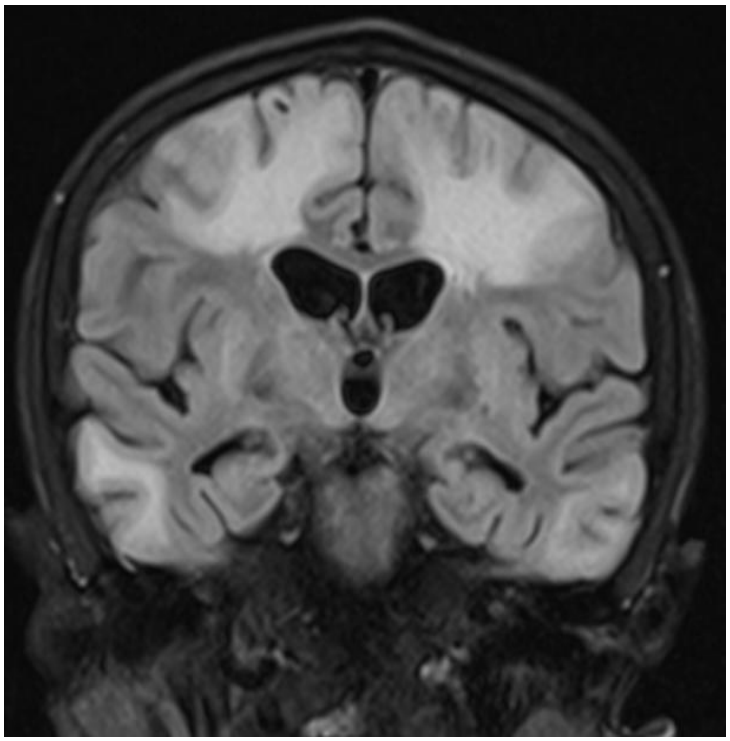
Urine copper value was high 487 microgram/ 24 hours [Normal range being 0-30 microgram/ 24 hours]. This was also confirmed on the slit-lamp examination.

The patient was put on D-penicillamine chelation therapy. The patient has shown progressive improvement.

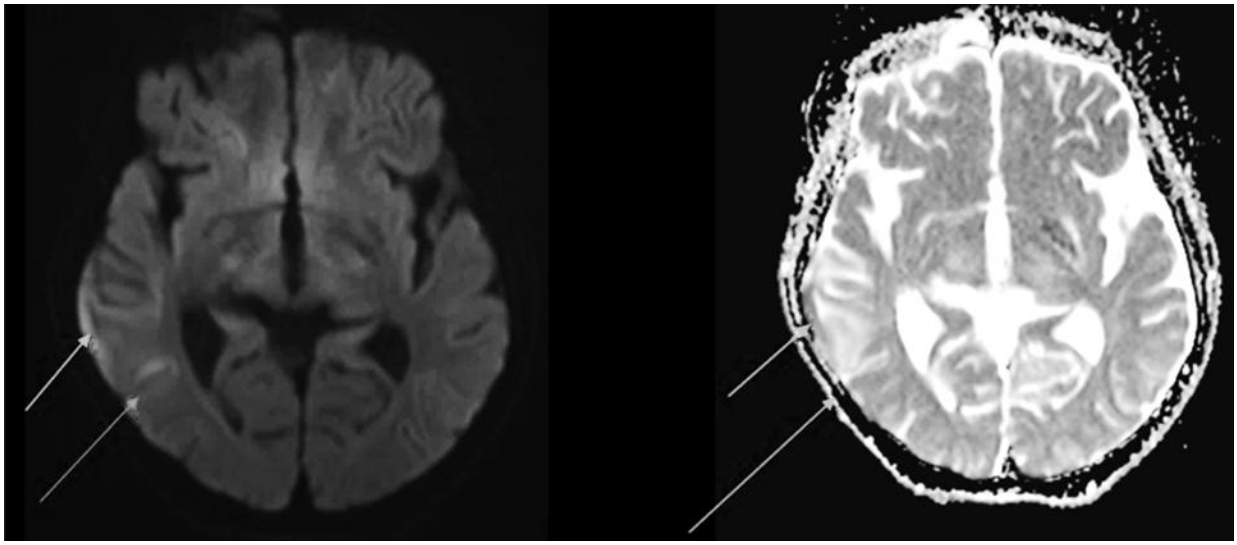
[Figure 1]: Sub cortical T2 hyperintensity in bilateral dorsomedial prefrontal region.



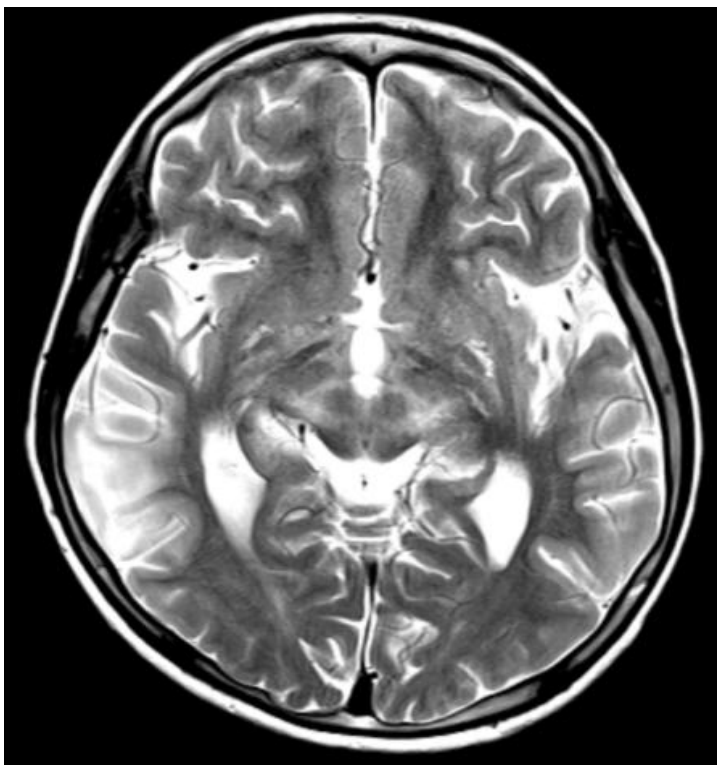
[Figure 2]: Sub cortical FLAIR hyperintensity in bilateral dorsomedial prefrontal region and also in both the temporal lobes on right side more than the left side.



[Figure 3]: There was a small area of diffusion restriction in the subcortical white matter as well as an area of the cortex in the right temporal region. Corresponding hypointensity on ADC sequences is also seen.



[Figure 4]: There is diffuse tegmental T2 hyperintensity with sparing of the red nucleus. There is temporal cortical and subcortical white matter hyperintensity especially on the right side



III. Discussion

In this case report, we presented the case of a young boy who developed frontal cortex involvement as a raremanifestation of Wilson's disease. To the best of our knowledge, cortical and subcortical involvement are very unusual.

The dorsomedial prefrontal cortex is involved in the "sense of self". In this case, the cortex and the subcortical white matter in the region of the dorsomedial prefrontal cortex show T2 hyperintense signal. The apathy that the patient demonstrated was probably a consequence of the same. [3,6,7]

The presence of extrapyramidal symptoms such as cogwheel rigidity and involuntary movements is a result of basal ganglia involvement.[2]

Wilson's disease is caused by an ATP7B gene mutation which results in abnormal ceruloplasmin metabolism, the mutation of which causes abnormal deposition of copper in the liver, brain (especially the basal ganglia), and the cornea.[1]

IV. Conclusion

Cortical and subcortical white matter involvement as the predominant imaging finding in Wilson's disease is very unusual. But occasionally it may be the only imaging finding and this possibility must be kept in mind, to prevent delay in the diagnosis, and misdiagnosis. This case demonstrates the presence of uncommon MRI findings in a relatively common disorder. On treatment, our patient has shown progressive improvement including the return of continence and improved cognitive parameters. This underscores the importance of early detection and management. If not detected sufficiently early, it can lead to severe disability and morbidity.[6] Hence, prompt identification of this rare presentation of Wilson's is a must for its scrupulous management.

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Conflicts of interest There are no conflicts of interest.

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