Woodhouse - Sakati syndrome
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Section: Neuroradiology
Area of Interest: Cardiac Neuroradiology brain
Neuroradiology spine
Imaging Technique: MR
Imaging Technique: MR-Diffusion/Perfusion
Case Type: Clinical Cases
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Patient: 17 years, male

Clinical History:
A 17-year-old male patient came to our department with a history of abnormal posturing of limbs, abnormal movements of hands and difficulty in speaking for 2 ½ years, which was gradually worsening. Difficulty in swallowing and learning disabilities were also present along with a history of hypogonadism. Ultrasonogram abdomen was normal. Ultrasonogram of scrotum showed bilateral small testis. Complete biochemical workup was done, which was non-contributory.

Imaging Findings:
Imaging Technique: MRI Brain was done in a 3T MRI. Standard Brain Protocol with TIWI, T2WI, FLAIR, DWI and SWI images obtained.

Findings:
Bilateral symmetric areas of gradient blooming noted in susceptibility-weighted images in lentiform nucleus (globus pallidus) (Fig 2). It is hypointense in T2WI (Fig 1).
Relative pituitary hypoplasia also noted (Fig 3).

Discussion:
Woodhouse - Sakati syndrome is caused by a mutation in the DCAF17 gene and inherited in an autosomal recessive manner. Typical clinical features in this disorder include hypogonadism, alopecia, diabetes mellitus, and progressive extrapyramidal signs [1, 3].

The MRI imaging findings described in this disorder include small pituitary gland, basal ganglia iron deposition and white matter changes [1, 2].

It comes under the category of neurodegeneration with brain iron accumulation (NBIA). NBIA represents a group of neurodegenerative diseases featuring extra pyramidal movement disorder, intellectual deterioration and basal ganglia iron deposition.
Written informed patient consent for publication has been obtained.

**Differential Diagnosis List:** Neurodegeneration with brain iron accumulation consistent with Woodhouse - Sakati syndrome, Pantothenate kinase-associated neurodegeneration (PKAN), Aceruloplasminemia, Neuroferritinopathy

**Final Diagnosis:** Neurodegeneration with brain iron accumulation consistent with Woodhouse - Sakati syndrome

**References:**


Description: SWI magnitude image shows bilateral symmetric gradient blooming reflecting iron deposition in globus pallidi. Origin: Barnard Institute of Radiology Madras Medical College, Chennai, India (2019)
Description: T2WI shows bilateral symmetric T2-hypointensities in the globus pallidi. Origin: Barnard Institute of Radiology Madras Medical College, Chennai, India (2019)
Figure 3

Description: Sagittal T1WI shows pituitary hypoplasia Origin: Barnard Institute of Radiology Madras Medical College, Chennai, India (2019)