KREP

2025

KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE PRESENTATION

Case of Manganese transport disorder

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MANGANESE TRANSPORT DISORDER

INTRODUCTION

Manganese Transport Disorder (MTD) is a rare inherited neurodegenerative condition caused by mutations in SLC30A10 or SLC39A14, leading to excessive manganese accumulation in the brain. Manganese is paramagnetic and thus has a strong impact on MRI signals, especially T1-weighted imaging.

PATHOPHYSIOLOGY

SLC30A10 mutation results in impaired biliary excretion of manganese, while SLC39A14 mutation causes impaired cellular transport. Both lead to manganese accumulation in the brain, particularly in metabolically active areas.

CLINICAL CORRELATION

- Adult-onset dystonia, parkinsonism, gait disturbances, and hepatic dysfunction. MRI aids in early diagnosis and differentiation from other metabolic or toxic encephalopathies.

RADIOLOGICAL INVESTIGATIONS -Modality of Choice: MRI Brain

- **T1-Weighted Imaging (Mainstay)**
- Characteristic Findings:
  - Bilateral symmetric hyperintensities (bright signal) due to Mn deposition.

  Key Regions Involved:
  - Globus pallidus (most consistent)
  - Putamen & caudate nuclei
  - Substantia nigra
  - Dentate nuclei of cerebellum
  - Midbrain (red nucleus spared)
  - Dorsal pons
  - Superior cerebellar peduncles
  - Anterior pituitary

Mnemonic: "BGP-DAMPS" – Basal ganglia, Globus, Pituitary, Dentate, Anterior brainstem, Midbrain, Pons, SCP

T2-weighted & FLAIR / DWI-ADC

- T2-weighted: Typically normal or hypointense.
- DWI/ADC: No diffusion restriction (helps rule out infarcts).

CT Brain

- Not sensitive.
- May show faint hyperdensity in basal ganglia.
- Cannot detect early parenchymal Mn deposition.

Diagnostic pearl

- Bright T1 signal in globus pallidus + normal DWI/T2 = Think manganese.
- Differentiates from Wilson's disease, which shows:
  - T2 hyperintensity
  - "Face of giant panda" sign (midbrain)
  - ATP7B mutation + low ceruloplasmin

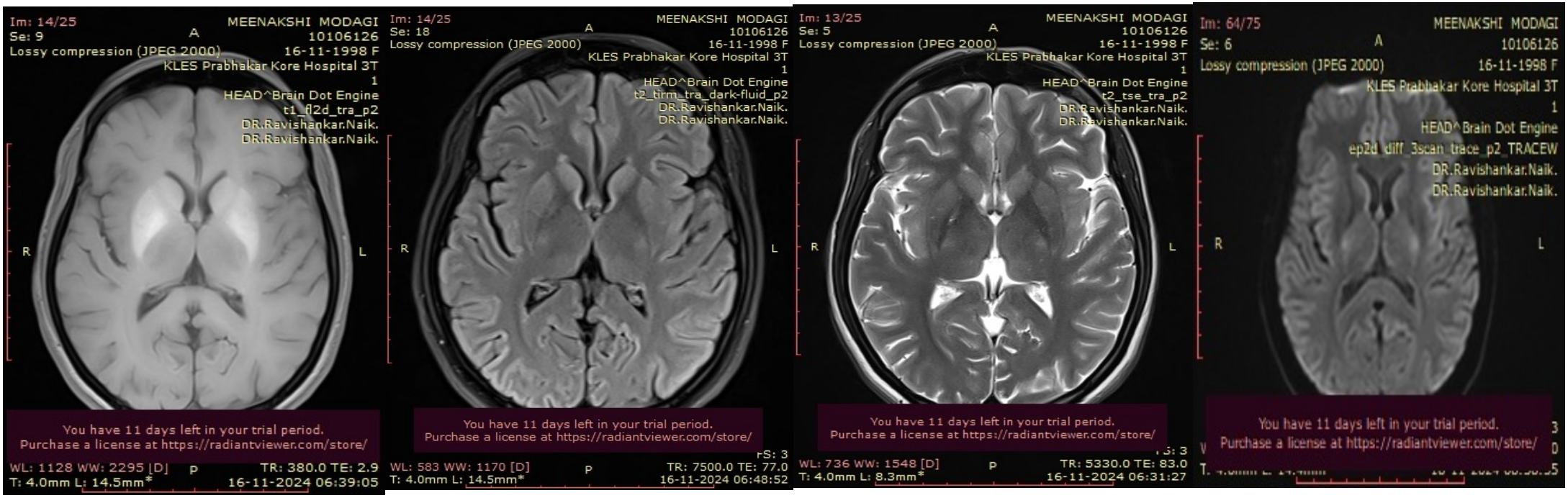
Confirmatory Diagnosis

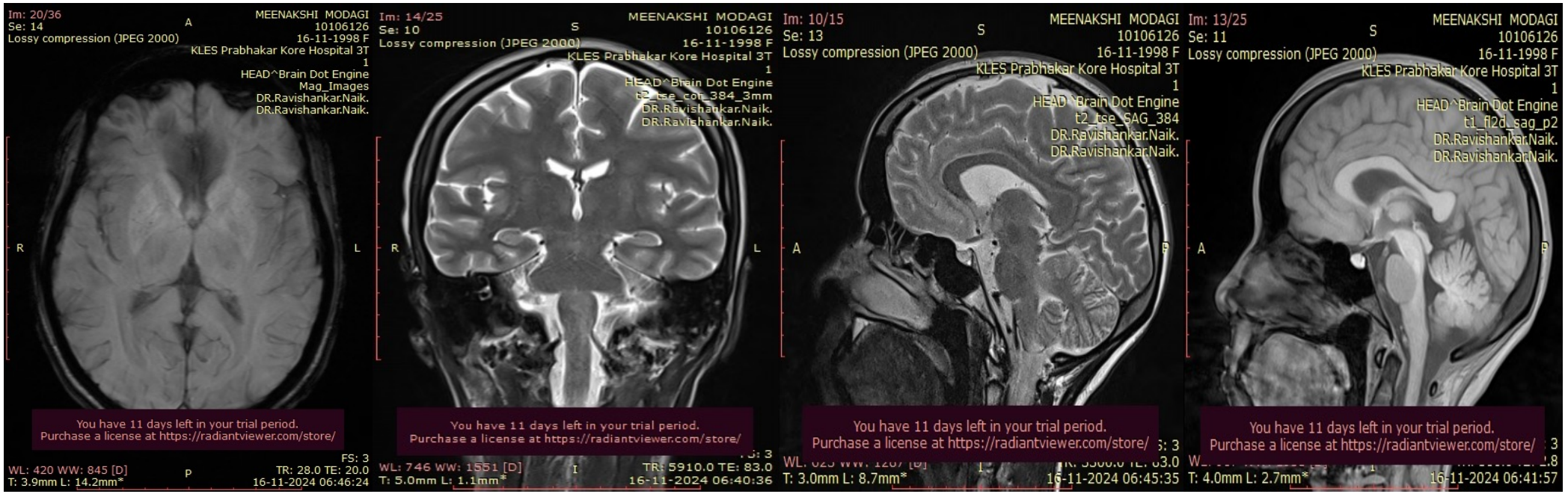
- Serum manganese levels: Elevated
- Genetic testing: SLC30A10 / SLC39A14 mutations

Differential Diagnoses

Condition	T1 Signal	T2 Signal	Other Imaging Features
Manganese Disorder	↑	±Normal	No DWI restriction, pituitary hyperintensity
Wilson's Disease	↓	↑	T2 basal ganglia, "Face of panda" sign
Chronic liver disease	↑	Normal	Pallidal T1 hyperintensity
Leigh syndrome	Normal	↑	Brainstem/basal ganglia necrosis

MANGANESE TRANSPORT DISORDER

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IMAGING FINDINGS

- Bilateral symmetrical T1 hyperintensities noted involving the bilateral basal ganglia, dentate nucleus, cerebral peduncles, midbrain (sparing the red nucleus), dorsal pons, superior cerebellar peduncles and anterior pituitary.
- No evidence of diffusion restriction noted on DWI sequence.

SUGGEST: GENETIC WORK UP FOR SLC30A10 & SLC39A14 TO RULE OUT MANGANESE TRANSORT DISORDERS

LABORATORY REPORT

Redcliffe labs

Patient NAME	: **Mrs MEENAKSHI**		
DOB/Age/Gender	: 26 Y/Female	Report STATUS	: Final Report
Patient ID / UHID	: 10523721/OF10523721	Barcode NO	: ZF408342
Referred BY	: Dr. Ramakrisha Neurologist	Sample Type	: Whole blood EDTA
Sample Collected	: Nov 21, 2024, 07:16 PM	Report Date	: Nov 26, 2024, 06:15 PM.

Test Description	Value(s)	Unit(s)	Reference Range

Manganese, Blood

Manganese ICPMS *(Blood)*	**154.24**	µg/L	4 - 15

Medical Remarks: Kindly correlate clinically.

Interpretation:

1. Manganese is an essential element used as a co-factor for a number of enzymatic reactions. Main intake is derived from food, vegetables, germinal portions of grains, fruits, nuts, tea and some spices.
2. It is used in industries dealing with steel alloys, dry cell batteries, electrical coils, ceramics, matches, glass tiles, welding rods, animal food additives & fertilizers. Individuals working in industries with high concentrations of manganese dust show a 30 times greater incidence of respiratory disease than normal individuals.
3. Exposure to Mn in work place primarily from inhalation of dust and fumes. Smaller particles get deposited in the lower respiratory tract and mainly absorbed into the blood and lymph nodes. Manganese exposure leads to abnormal electrocardiogram and inhibits myocardial contraction.
4. Human exhibits toxicity to Mn when exposed to large quantities of dusts containing the metal, in mining, ore crushing, machining of Mn alloys, construction and destruction of brick. Mn toxicity is also a concern in newborns and children receiving long term parenteral nutrition.

Increased Levels : Acute hepatitis, Industrial exposure, Myocardial infarction

Decreased Levels : Seizures, Phenylketonuria

*** End Of Report ***

ROLE OF IMAGING

Purpose	Utility
Early detection	Bright T1 signal in basal ganglia
Monitoring progression	Assessing signal intensity changes
Response to therapy	Reduced T1 hyperintensity over time
Screening of siblings	In familial cases

THANKYOU