Background: Mutations in MRPS22 cause severe mitochondrial disease with features including cardiomyopathy, lactic acidosis, brain abnormalities, and primary ovarian insufficiency. To date, the MRPS22 gene has not been reported in the fourth decade of life. We report a case of a Turkish patient with MRPS22 mutations.

Result: Neurometabolic disorders can occur in all age groups. We presented a female case diagnosed at an advanced age. Mitochondrial disease should be kept in mind in case of multisystemic involvement.

Case Report:
• 44-year-old female patient.
• Consanguinity in parents was from the same village
• In childhood, her development was normal
• At 33 years, depression occurred.
• Subsequently, tremors in her hands started.
• At 40 years, progressively impaired speech and walking

Physical Examination:
Weight: 43 kg (-2,8 SDS)
Height: 164,5 cm (0,2 SDS)
BMI: 15,9 (-3,5 SDS)
- Dysarthria+
- Dismetria+/
- Dysdiadokinesia+/
- Ataxic gait
- Decreased sense of vibration
- Pes cavus in left foot
- The mitochondrial disease criteria score was 7

Laboratory Findings:
In CSF analysis, the 5-MTHF level was undetectable. (FOLR1 normal)
The Cranial MRI: Bilateral cerebral white matter hyperintensity (Figure 1)
EMG was myopathic.
VEP latency was detected.
ECHO: moderate tricuspid valve regurgitation
The uterine atrophy was detected in MR imaging.
MRPS22 gene: Homozygous c.605G>A (p.R202H) variant
Folinic acid treatment was given because of CSF results. The treatment is still ongoing as it benefits.