TITLE : Homozygous sequestosome 1 (SQSTM1) mutation: a rare cause for childhood-onset mixed movement disorder ,cerebellar ataxia with vertical gaze palsy

Aims and Background

- Mutations in SQSTM1 gene have been recently identified as a rare cause of progressive childhood neurodegenerative disorder. They were earlier associated with frontotemporal dementia and ALS.
- This case reports a progressive childhood-onset mixed movement disorder with cerebellar ataxia and vertical gaze palsy with normal MRI and homozygous SQSTM1 mutation.

Materials and Methods

- Study Type: Case Study
- Patient: 13-year-old male, born of third-degree consanguinity, with normal developmental milestones.
- History: Progressive neurological illness of 8 years with poor scholastic performance, involuntary movements, unsteadiness, and recent onset headache.
- Examination: Impaired frontal and parietal lobe functions, bilateral EOM restriction, dystonia, decreased tone, normal motor power, absent reflexes, no sensory or autonomic dysfunction.
- Investigations: Normal serum ceruloplasmin, lactate, ammonia, vitamin B12, vitamin E, thyroid tests, MRI brain normal, WES showed homozygous nonsense variant in exon 2 of SQSTM1 gene.

Results and Conclusion

- Result: Homozygous nonsense mutation in exon 2 of SQSTM1 gene detected.

- Discussion: Childhood-onset neurodegeneration with ataxia, dystonia, and gaze palsy is caused by homozygous SQSTM1 mutations, characterized by onset in the first or second decade, with features such as gait ataxia, cognitive decline, dysarthria, and dystonia.

- Conclusion: SQSTM1 mutation should be considered in differential diagnosis of cerebellar ataxia, dyskinesias, and ophthalmological manifestations.