Video and Ophthalmic examination: Clues in Sialidosis diagnosis

Teaching Neuro*Images*

Neurology

Resident and Fellow Section

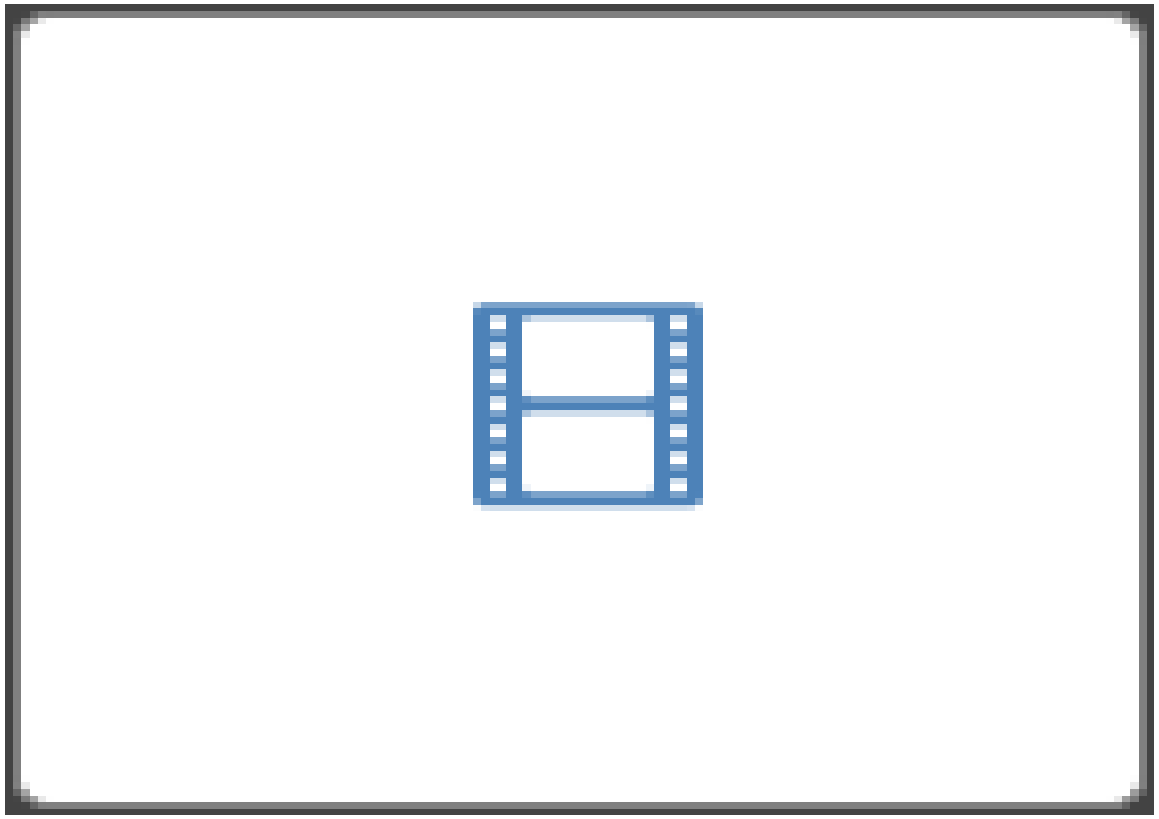
Vélez Gómez et al.

Vignette

- A 53-year-old woman was presented for evaluation of visual disturbances, generalized and multifocal myoclonus, and progressive ataxia that began at age 30.
- Bilateral cherry-red spots in the macula was observed in the ophthalmic examination.
- Reduced neuraminidase activity in fibroblasts and the homozygous mutation c.403G>A in the NEU1 gene confirmed the diagnosis of sialidosis type I.

Imaging – cherry-red spot

Video – Cortical Myoclonus



Conclusions

- Cherry-red spot is a red zone at the centre of the macula surrounded by retinal opacification. It is due to the accumulation of different lipid, sphingolipid, or oligosaccharide material in the ganglion cells of the retina.
- Generalized and multifocal myoclonus at rest, which worse during a sustained posture or with tactile sensory stimulation are characteristics of cortical myoclonus.
- Cherry-red spots on the retina, cortical myoclonus and progressive ataxia are essential clues to suspect Sialidosis, also called cherry red spot-myoclonus syndrome.