Heimler Syndrome as an differential diagnosis of Usher

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The authors declare no conflicts of interest.

Purpose & Methods

- Highlight possible differential diagnosis of congenital deaf blindness syndromes based on atypical clinical findings.
- A case report of a patient with previous clinical diagnosis of Usher Syndrome.
- The study was approved in the Local Research Ethics Committee.

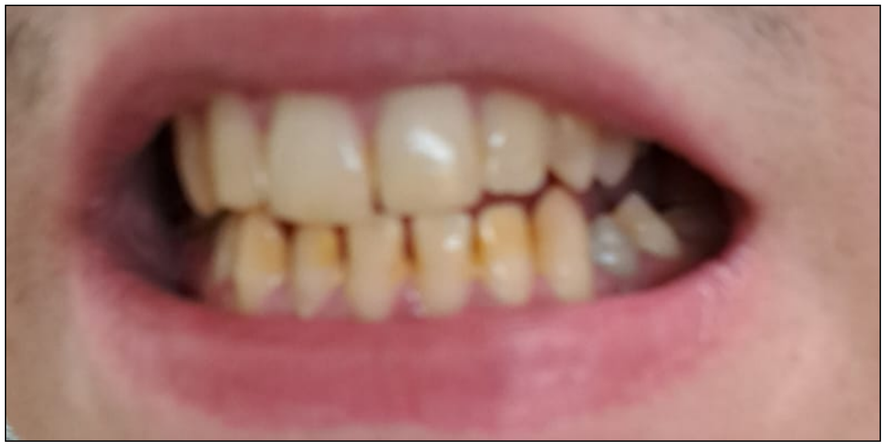
Case report

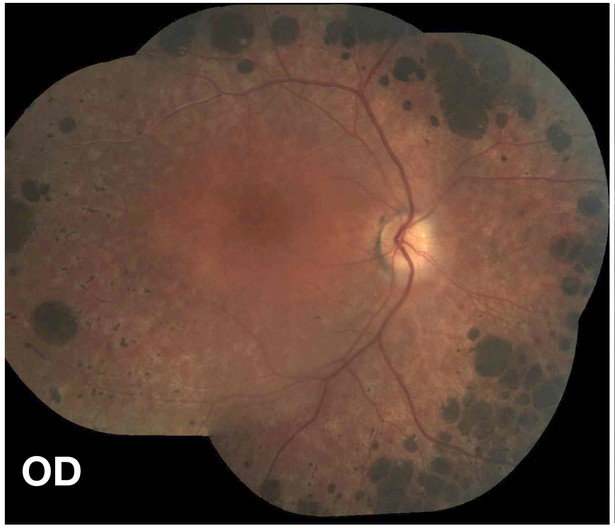
- 18-year-old male diagnosed with Usher Syndrome Type II 9 years ago.
- Audiometry test: severe neurosensorial hearing loss.
- Brainstem Auditory Evoked Potential: absent response.
- No history of prenatal and neonatal infections and complications.
- Hipomatured dentition (Amelogenisis Imperfecta).

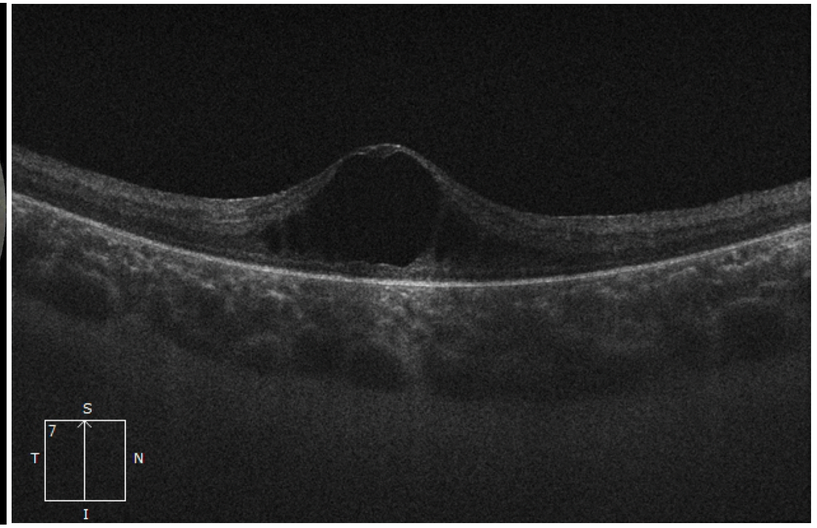
Ophthalmic examination

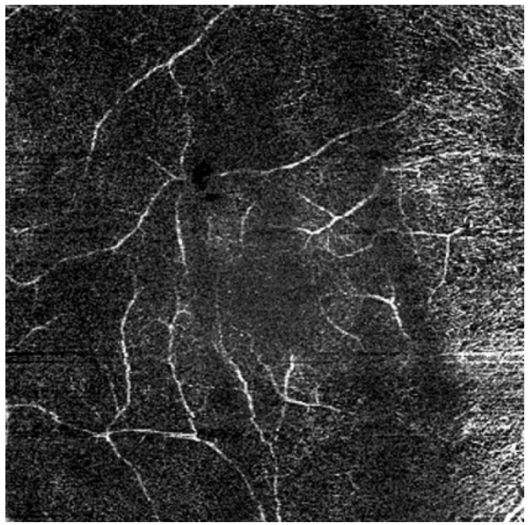
	OD	OS
BCVA	20/100 (+2.00 -3.25 18°)	20/70 +1.75 -2.50 163°
IOP	13mmHg	14mmHg
Humphrey Central Visual Field (10-2)	Foveal response 24dB MD -14.55dB P<1%	Foveal response 24dB MD -13.70db P<1%

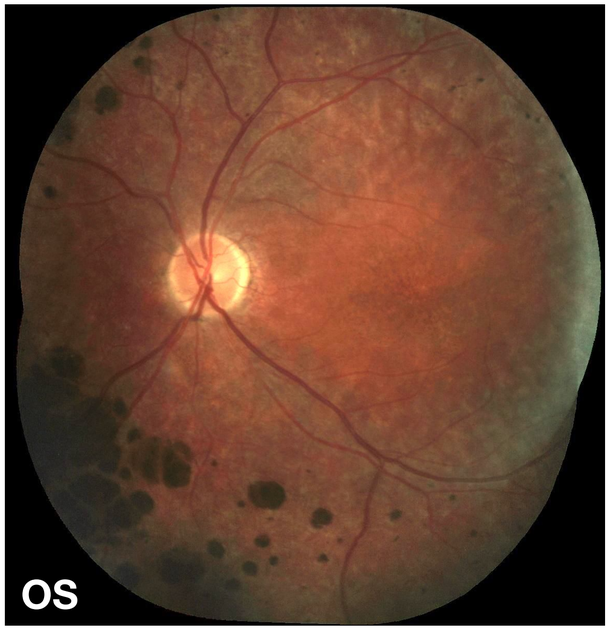
Patient's dentition showing Amelogenesis Imperfecta

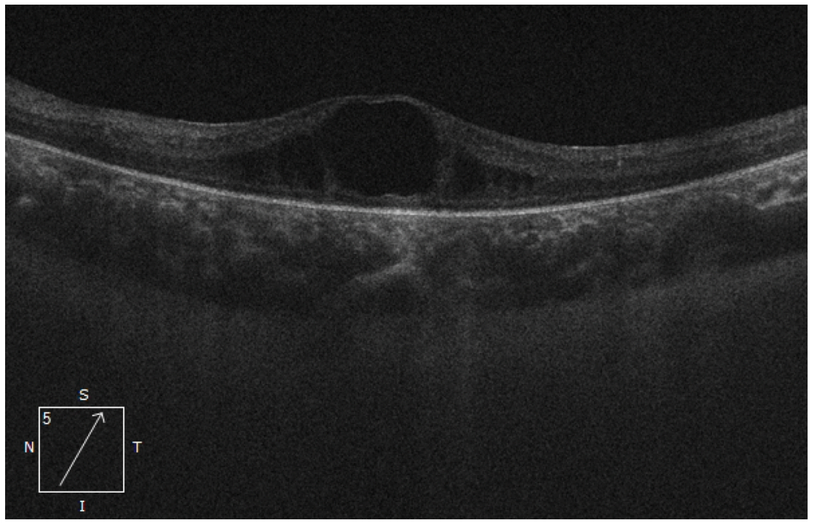


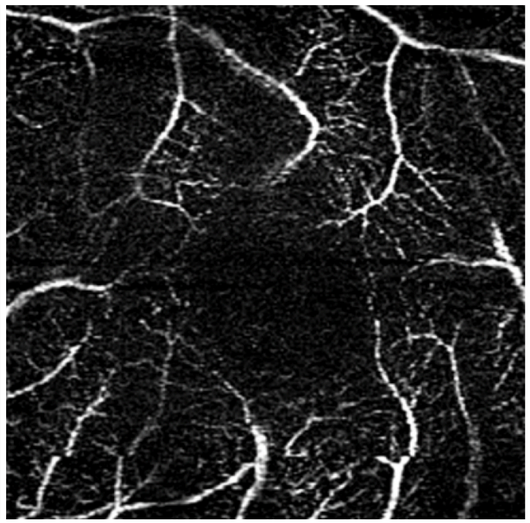












- **Retinography:** diffused bone spicule pigmentation in mid periphery along with retinal atrophy.
- **OCT:** intraretinal macular edema, and photoreceptor loss.
- **OCTA:** in the superficial vasculature, enlargement of foveal avascular zone and areas of edema.

Discussion

- Diagnosis of US is primarily clinical, which is reliable to molecular diagnosis in **97%** of the cases. Nehaus, C. Mol Genet Genomic Med, 2017
    - Retinitis pigmentosa, neurosensorial hearing loss, and, sometimes, vestibular dysfunction. Fuster-Garcia, C. Int J Mol Sci, 2021
- In **3%** of the cases, atypical findings may correlate to a different clinical diagnosis. Nehaus, C. Mol Genet Genomic Med, 2017
- Heimler Syndrome: US + macular edema, and Amelogenesis Imperfecta (non syndromic sensory deficits). Varela, MD. Am J Med Genet, 2020
    - Possible mutations in the genes *PEX1, 6* or *26.* Kin, YJ. Genes, 2021
      Ratbi, I. Am J Hum Genet, 2015