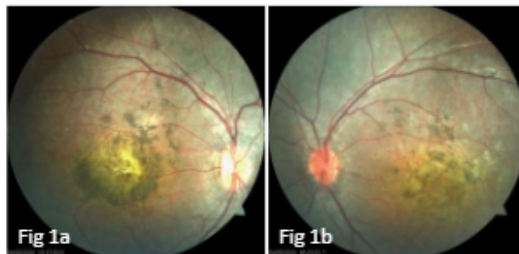
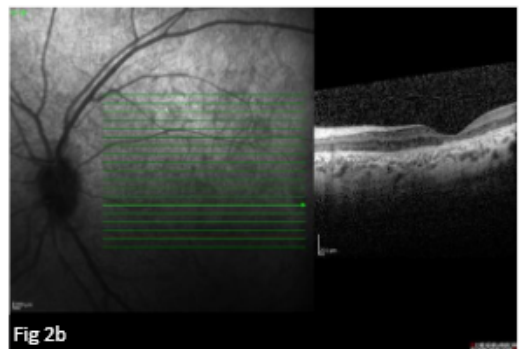
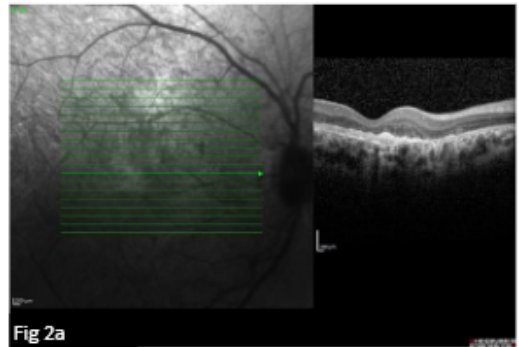


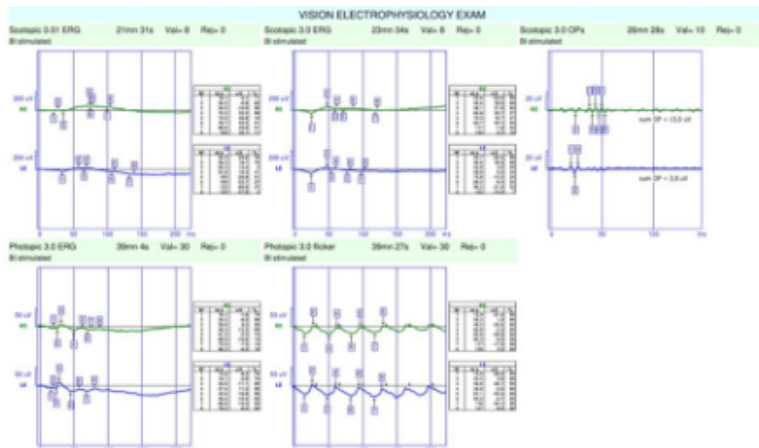
Hypotrichosis with juvenile macular dystrophy (HJMD, OMIM: 601553) has been described as a rare autosomal recessive disease associated with a sparse scalp hair and progressive retinal degeneration caused by *CHD3* (cadherin 3) gene mutation located on 16q22.1. We aimed to report the full-field electroretinographical (fERG) findings of a child with hypotrichosis and retinal dystrophy

A 9-year-old boy with decreased visual acuity, nyctalopia and hypotrichosis underwent complete ophthalmological examination including fundus photography, optical coherence tomography (OCT) and fERG. The patient referred for genetic evaluation.

The best corrected visual acuity was 20/200 on both eyes. Fundus evaluation showed severe atrophy of the central macular region with pigment clumping and retinal atrophy at the posterior pole in particular (Figure 1a: right eye and 1b: left eye). OCT indicated disruption and loss of outer retinal layers with atrophic changes extending along the posterior pole (Figure 2a: right eye and 1b: left eye). fERG revealed severe reduction of light-adapted and dark-adapted amplitudes the latter more prominent which supported the fact that the disease affected the entire retina (Figure 3). A homozygous c.830del mutation was demonstrated on *CDH3* gene.



HJMD should be kept in mind in children with early onset retinal/macular dystrophy and congenital hypotrichosis. ERG is indispensable for the diagnosis and may reveal generalized retinal dysfunction despite predominant macular involvement.



References

1. Hui S, et al. Characterization of *CDH3*-Related Congenital Hypotrichosis With Juvenile Macular Dystrophy. *JAMA Ophthalmol* 2016;134(9):992-1000.
2. Oliveira-Ferreira C, et al. Hypotrichosis with juvenile macular dystrophy. *Ophthalmic Genet* 2016;40(8):574-577.
3. Nassef P, et al. Hypotrichosis with cone-rod dystrophy in a patient with cadherin 3 (*CDH3*) mutation. *Onc Ophthalmol* 2019;13(2):155-160.
4. <http://www.ncbi.nlm.nih.gov/omim/>