Case of foveal hypoplasia with nystagmus

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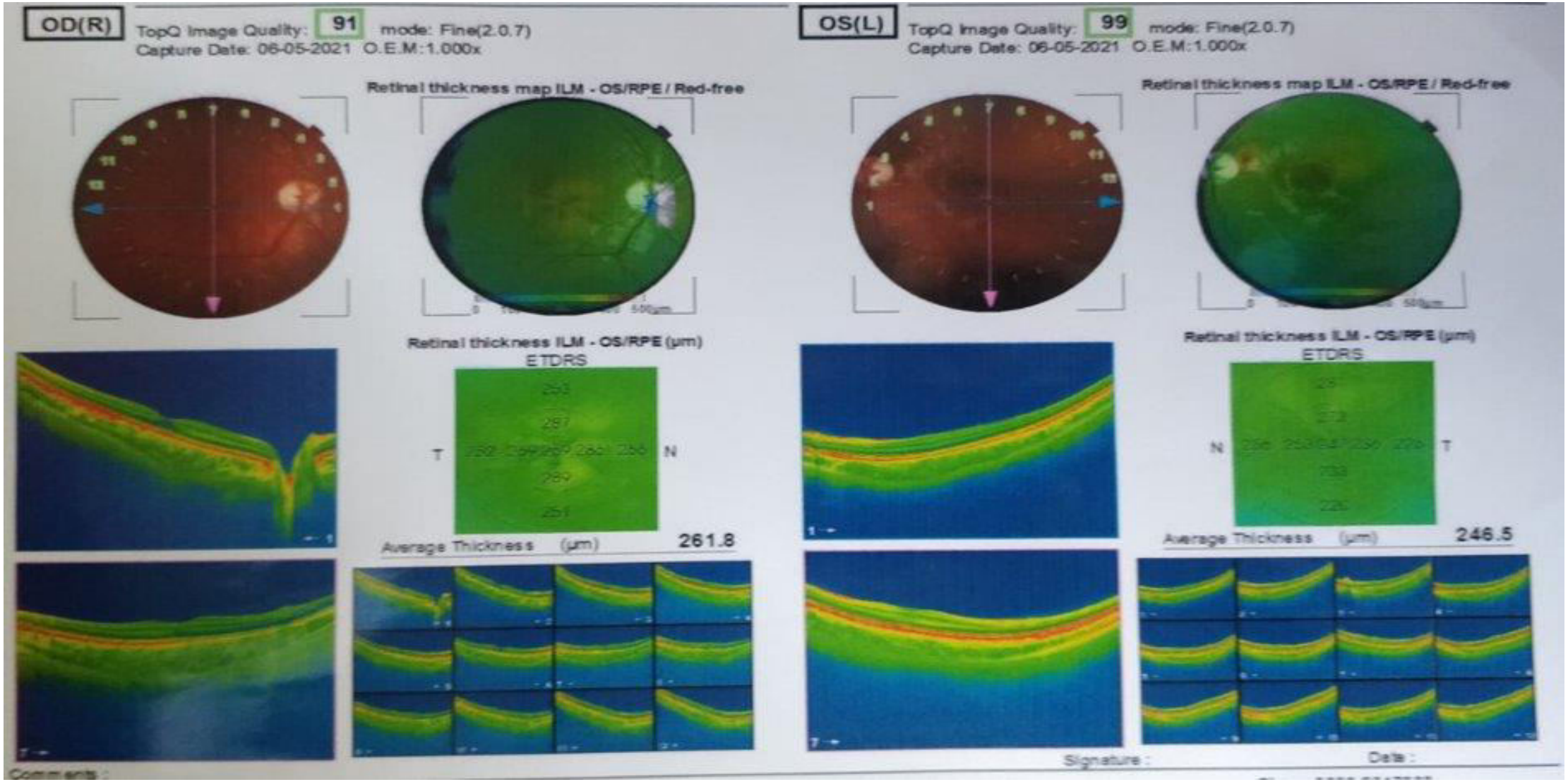
- A 9year old male child was brought to opd by his mother with complaint of defective vision and unable to recognise people since early childhood.
- There is no similar complaint in siblings. On examination there is end gaze nystagmus, bilaterally facial symmetry, full ductions and versions in all gazes .right eye vision – CF 3mts, anterior segment findings are normal, lens- clear. Left eye vision – CF2mts, anterior segment findings are normal, lens – clear. fundus examination of both eyes shows absent foveal reflex.
- On Oct – foveal hypoplasia of both eyes is noted (LE>RE).

OCT Image of both Eyes



DISCUSSION :

- Foveal hypoplasia is a retinal disorder where there is a lack of full development of fovea. Several diseases were known to be associated with foveal hypoplasia which include albinism, aniridia, ROP, incontinentia pigmenti , isolated foveal hypoplasia , stickler syndrome.
- Genetics : Mutations of PAX6 Gene known to cause isolated foveal hypoplasia which has an autosomal dominant inheritance.
- Mutations of GPR143 and SLC38A8 gene were identified to cause autosomal recessive pattern.
- In the above case only poor visual acuity and absent foveal reflex and no other systemic associations are noted.These findings suggest that it is a case of isolated foveal hypoplasia with end gaze nystagmus.