

# Foveal Hypoplasia

- Lack of formation of the foveal depression
  - Foveal plana = absent foveal pit
- associated with poor vision and nystagmus

## Ocular Associations

- Aniridia (PAX6)
- Achromatopsia
- Congenital retinal macrovessel
- Coloboma
- Optic Nerve Hypoplasia
- Retinopathy of Prematurity

## Isolated Foveal Hypoplasia

- Autosomal Dominant from PAX 6 gene variants
- Autosomal Recessive from SLC38A8 gene variants
  - SLC38A8 codes for a glutamine transporter in photoreceptors, mutations in the 6th transmembrane domain cause underdevelopment of the retina
- X-linked from FRMD7 gene variants
  - FRMD7 regulates neuronal outgrowth
  - also associated with familial infantile nystagmus

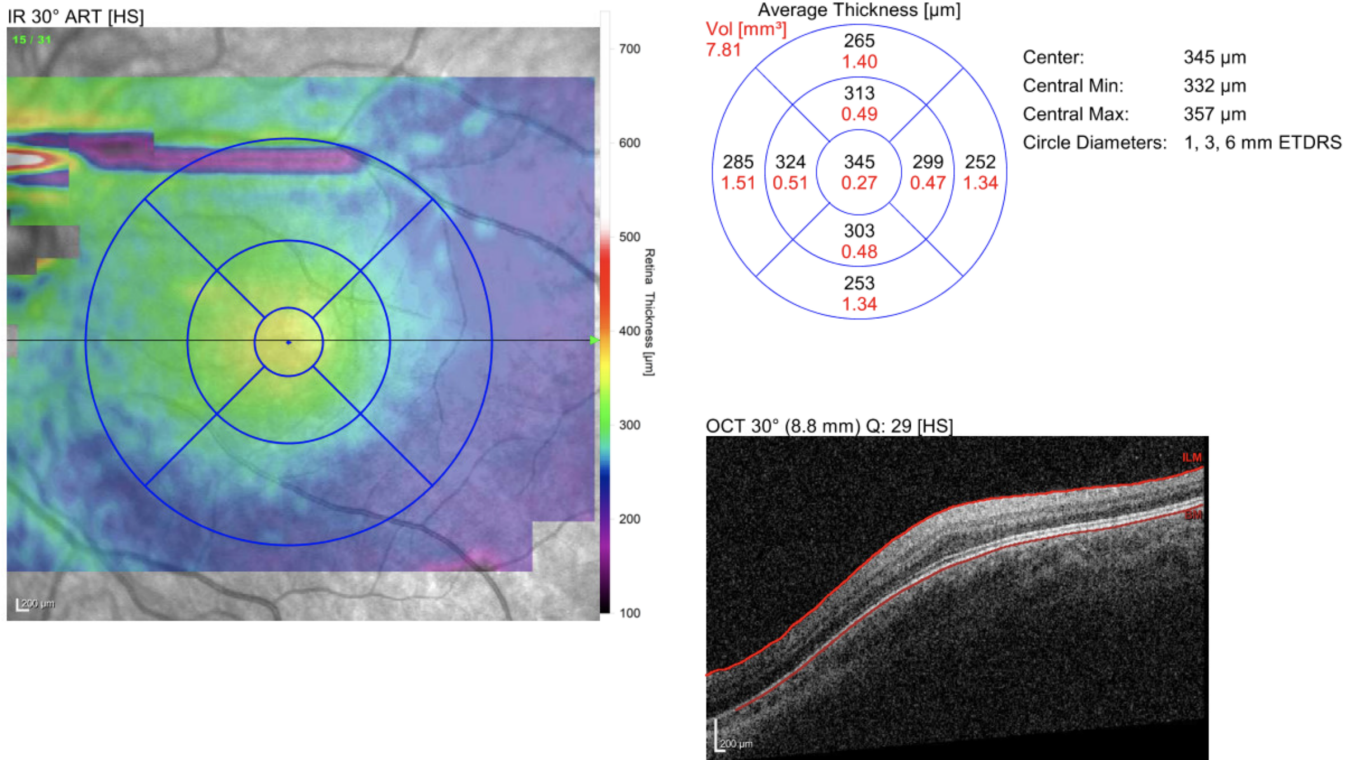
## Systemic Associations

- [Albinism](#)
- [Familial Exudative Vitreoretinopathy](#)
- [Incontinentia pigmenti](#)
- Prematurity
- Stickler Syndrome

## Diagnostic Procedures

- Visual acuity
  - VA is variable but can range from 20/40 to 20/200
- Oculomotor
  - Nystagmus and strabismus
- Ophthalmoscopy
  - absence of foveal reflex and foveal avascular zone and typical macular pigment
- [OCT](#)

- Absence of foveal pit, persistent inner retinal layers through fovea
- Example of an OCT report of a child with foveal hypoplasia



- OCT-A
  - absence of superficial capillary plexus and variable decrease in deep capillary plexus in the fovea

## References

- [Study of the Leicester Grading system, Rufai et al 2020](#)
  - Rufai SR, Thomas MG, Purohit R, Bunce C, Lee H, Proudlock FA, Gottlob I. Can Structural Grading of Foveal Hypoplasia Predict Future Vision in Infantile Nystagmus?: A Longitudinal Study. *Ophthalmology*. 2020 Apr;127(4):492-500. doi: 10.1016/j.ophtha.2019.10.037. Epub 2019 Nov 4. PMID: 31937464; PMCID: PMC7105819
- Hendrickson A, Possin D, Vajzovic L, Toth CA. Histological development of the human fovea from midgestation to maturity. *Am J Ophthalmol* 2012;154:767-778.e2.

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