A 10-year old girl with Ophthalmic Manifestations of Tuberous Sclerosis Complex (TSC)

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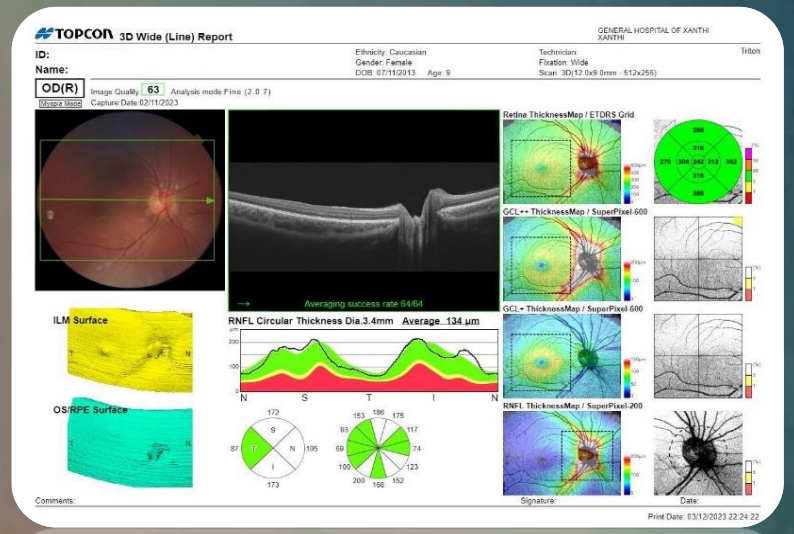
Introduction

- Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder
- Characterized by benign tumors/hamartomas in various organs[1]
- Incidence: 1 in 5,000 to 10,000 births
- Retinal /astrocytic hamartomas are the most common ophthalmological manifestation and satisfy one of the cardinal diagnostic criteria[2] (present in ~50% of cases)

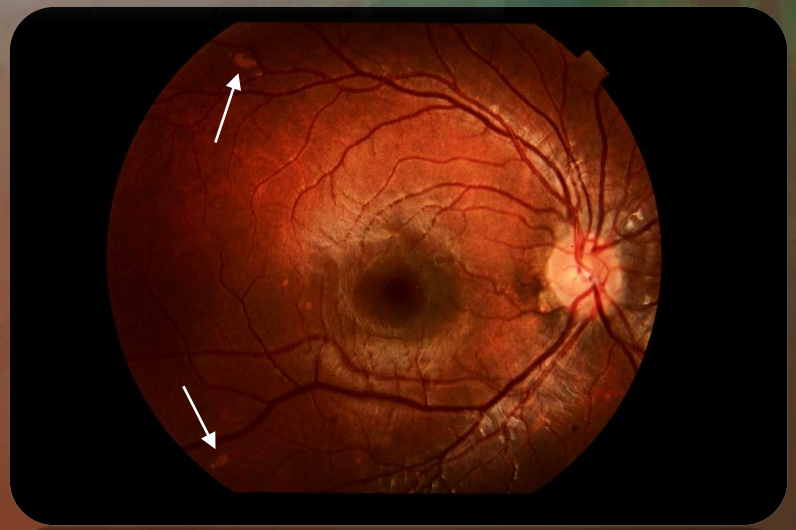


Case Presentation

- 10-year-old female with TSC
- De-novo mutation (not autosomal dominant pattern)
- Previous surgical excision of ovarian neoplasm

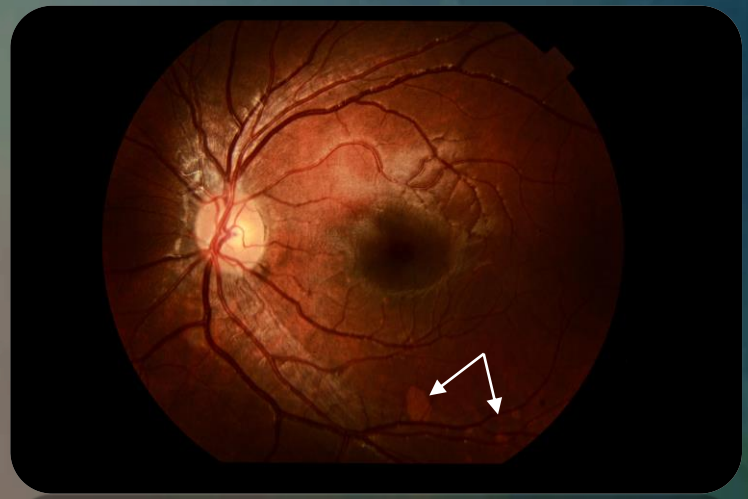
Clinical Findings

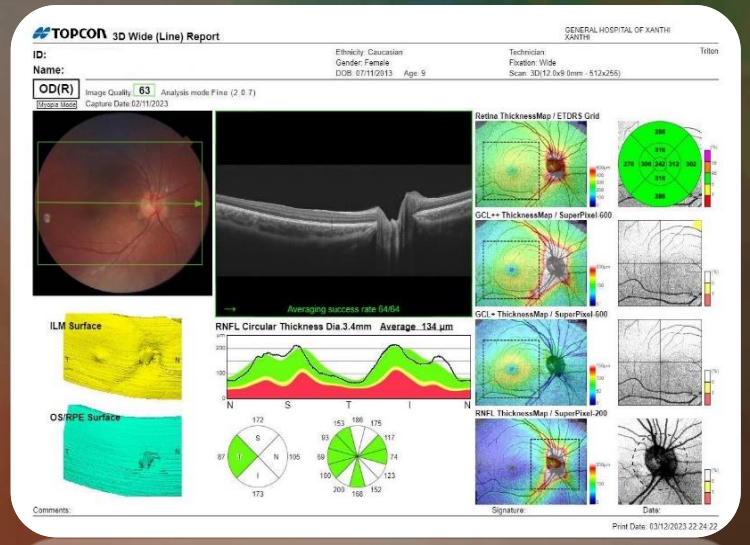
- VA in both eyes: 9/10 with correction (myopia - common in TSC(1))
- Normal extraocular motility and color vision
- Slit lamp exam: deep and quiet AC, normal IOP
- No dermatological stigmata of TSC on eyelids (ash leaf macules or angiofibromas)
- Optic nerves and peripapillary areas grossly normal
- Cup-to-disc ratios: 0.2-0.3 (OD), 0.3-0.4 (OS)
- No papilledema
- Multiple hamartomas present in both fundi
- OCT findings: flat type hamartomas without a hyperreflective projection(3) nor calcifications



Discussion

- ✓ Classic ophthalmological findings of TSC in this pediatric patient[4]
- ✓ Retinal hamartomas(~50% of TSC cases) represent a major diagnostic criterion[2]
- ✓ Lesions appearance consistent with typical TSC-associated hamartomas:
  - *flat, yellowish, translucent*
  - *poorly defined borders*
  - *no hyperreflective projections or calcifications (Grade I by classification)[3]*
  - *peripapillary and arcade distribution[1,5]*
- ✓ Good visual acuity maintained despite multifocal hamartomas
  - *tumors tend to be static[6], rarely impacting vision unless encroaching on fovea or optic nerve*
  - *periodic surveillance with ophthalmoscopy and OCT warranted*
- ✓ De novo TSC mutation is associated with less severe phenotypic expression but multidisciplinary monitoring is still crucial for systemic manifestations[7,8]
- ✓ Retinal findings in TSC are linked to increased risk of renal and cardiac involvement[9]
  - ❖ This underscores the vital value of retinal monitoring in comprehensive patient management
- ✓ Longitudinal observation for tumor growth and visual changes remains essential





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