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Papilledema in Children (Idiopathic Intracranial Hypertension)

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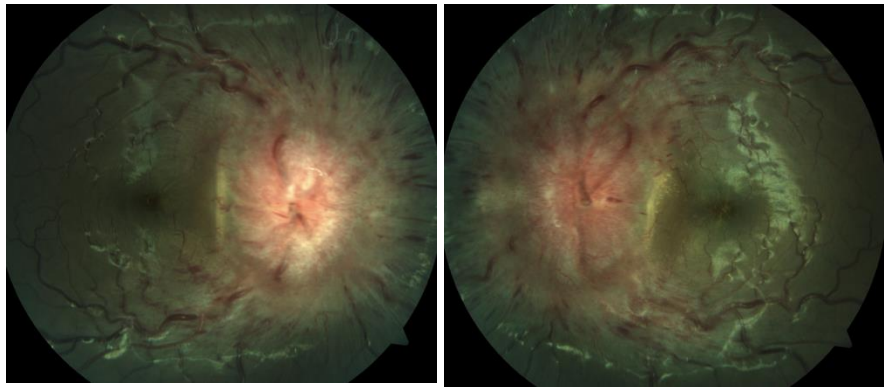
AAPOS 2026 Workshop

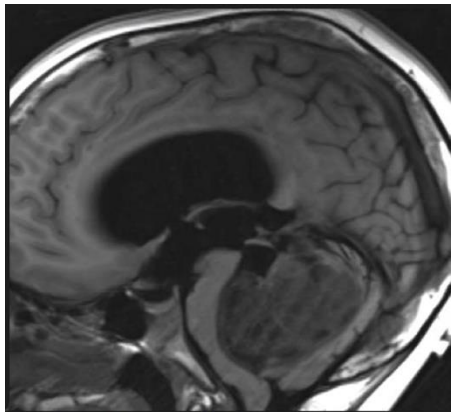
Financial Disclosures

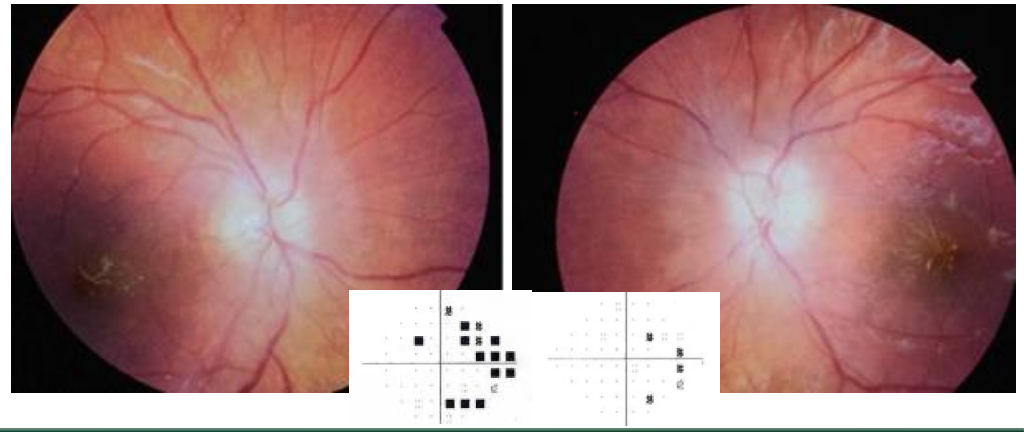
- None

Papilledema

- Acquired optic disc swelling caused by elevated intracranial pressure (ICP).
- Associated with serious medical diagnoses and can result in permanent visual loss.
- A standardized approach to evaluate the patient with papilledema is recommended.



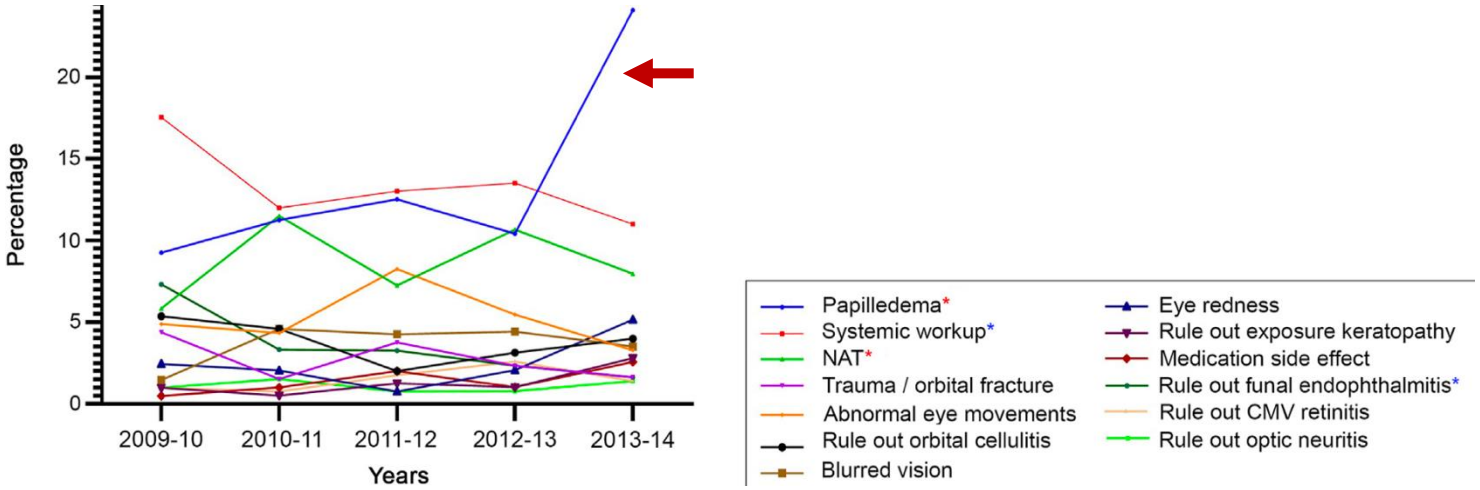




Pediatric papilledema

- Screening for papilledema ~ **14.2%** of inpatient & ED ophthalmology consultations.
  - Papilledema in 3.72-3.75%
  - Idiopathic intracranial hypertension (IIH) in **(51%)**
- "The incidence of true papilledema among children referred for suspected papilledema based on fundus examination is very low." Kovarik, KK, et al. J AAPOS;19:344-348, 2015

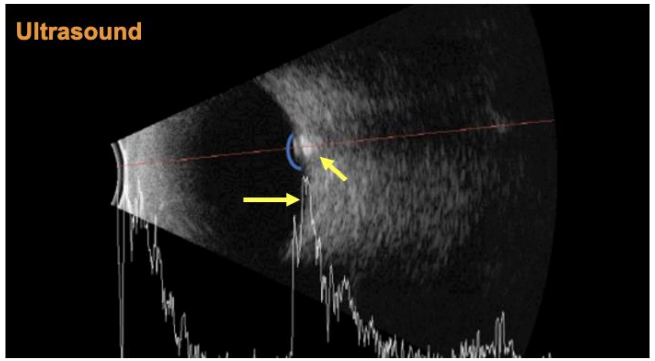
There may be an over-referral for children with suspected papilledema who do not have raised ICP.

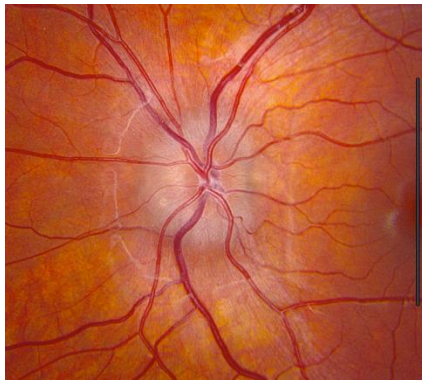


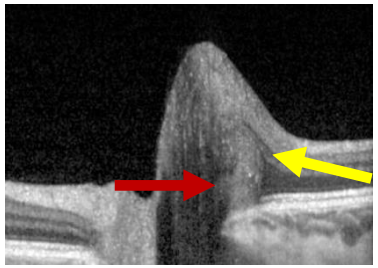
Gautam, N. et al. J AAPOS27:75.e1-5, 2023
Fischer AF, et. al. J AAPOS Apr;29(2):104158, 2025

Pediatric pseudoapilledema

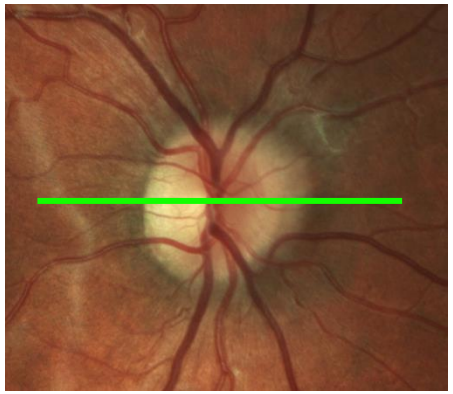
- The presence or absence of papilledema is a diagnostic dilemma
- Mimickers of papilledema
  - drusen, PHOMS, hyperopia, congenital anomaly, myopic tilted disc



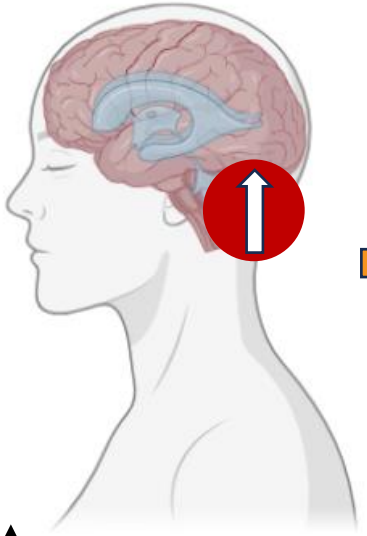




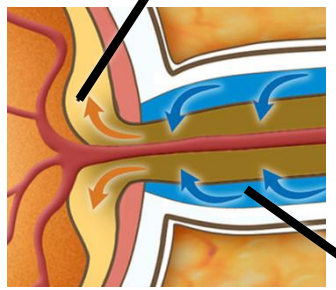
Eshun, E, et al. JAAPOS, 2022.

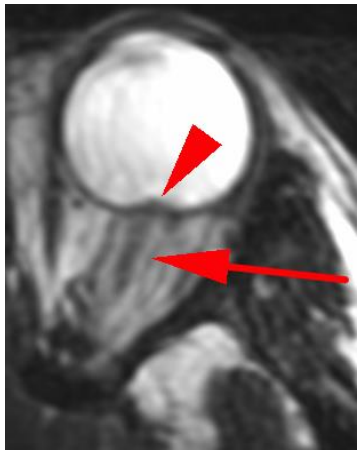


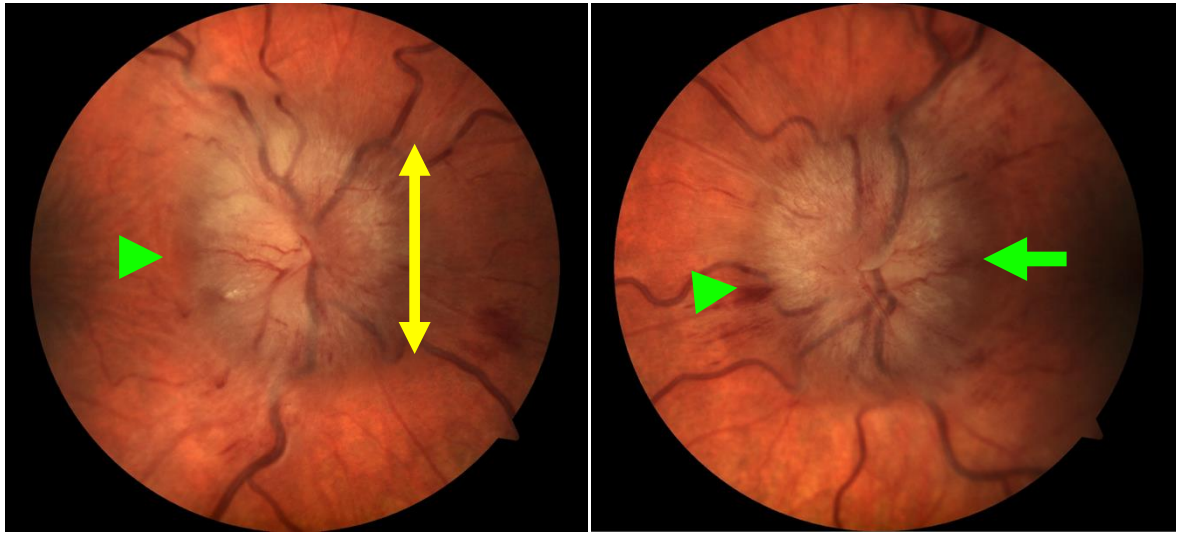
Pathophysiology



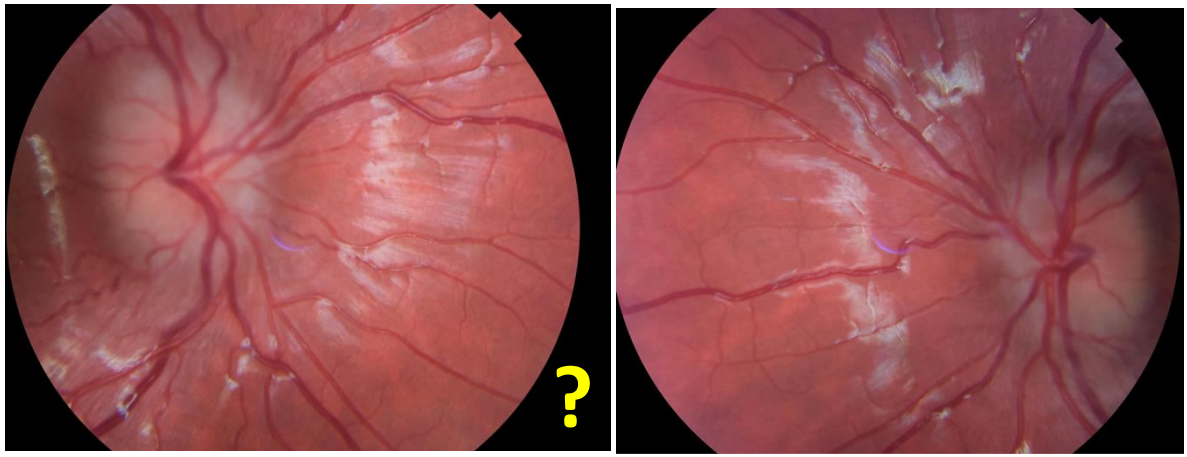
- ↑ in brain or cerebral blood volume
- Mass effect
- ↑ in CSF production
- ↓ Decrease in CSF drainage
- Cranial abnormalities





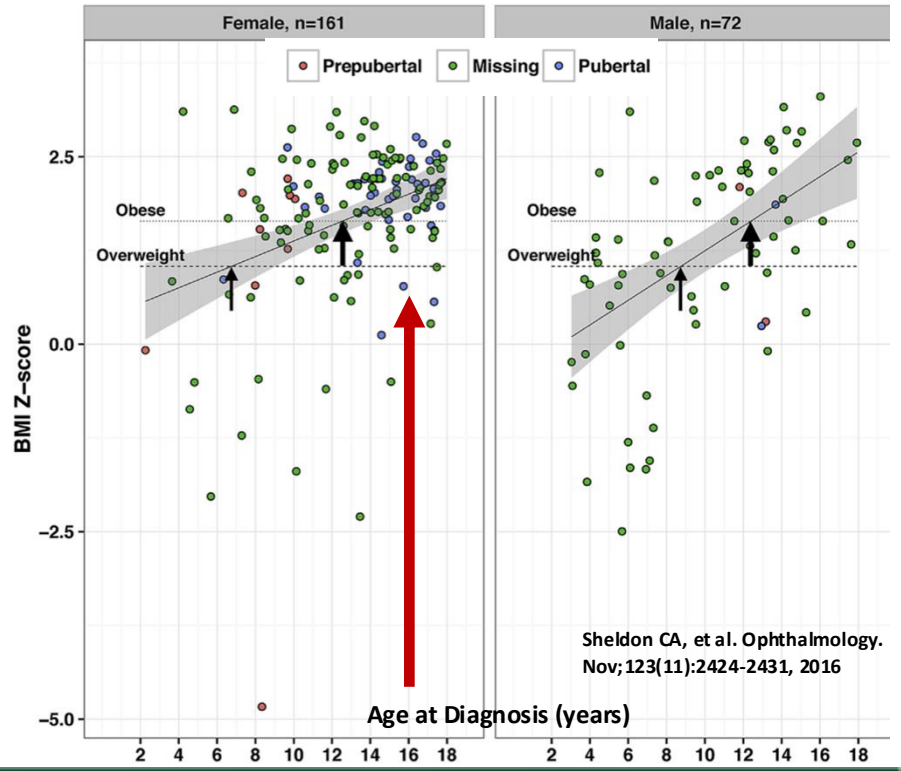


Acute, high-grade papilledema



Idiopathic Intracranial Hypertension (IIH)

- Intracranial hypertension with normal brain parenchyma, CSF constituents
- **Primary = (IIH)** vs. Secondary PTCS= (systemic & drug induced)
- IIH incidence: 0.63 - 0.90 in 100,000
  - Most common cause of PTCS in adults
  - **50% of pediatric papilledema**
- Prepubertal vs. pubertal pediatric IIH
  - Prepubertal: no gender or body habitus predilection
  - Pubertal: overweight females
- Metabolic and hormonal signals regulate CSF dynamics



Pseudotumor Cerebri Syndrome

Primary (Idiopathic Intracranial Hypertension)

Pre-pubertal
- male or female
- thin

Post-pubertal
- female
- obese
- polycystic ovarian syndrome

Secondary

Meds/Toxic:
- Cyclines (tetra, mino, doxy)
- Sulfa drugs
- Retinoic acid
- Steroid withdrawal
- Hypervitaminosis A
- Growth hormone
- Thyroxine
- Lithium

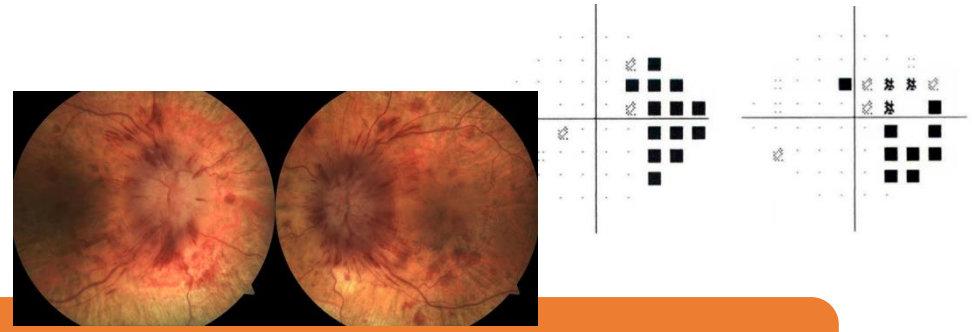
Vascular:
- CVST
- Ear infection with mastoiditis
- SVC syndrome
- Arteriovenous fistula
- Prior intraventricular infection; SAH

Associated Conditions:
- Addison disease
- Hypoparathyroidism
- OSA
- Pickwickian syndrome
- Anemia
- Renal failure
- Turner syndrome
- Trisomy 21
- Craniosynostosis
- Post infectious

Rangwala LM, Surv Ophthalmol. 52(6):597-617, 2007
Beres SJ. Semin Neurol. 40(3):286-293, 2020

IIH Treatment

- Conservative
  - Weight loss & healthy lifestyles



Medical (majority of patients)

- Acetazolamide (10-25 mg/kg/day)
- Topiramate (25 mg qday, ↑weekly)
- Lasix
- **Glucagon-like peptide (GLP)-1 Receptor Agonists (Liraglutide, Semaglutide)**
  - **reduce CSF production**

Surgical (sight threatening papilledema)

- Optic nerve sheath fenestration
- CSF diversion
  - Lumbar drain (temporary)
  - Shunt (permanent)
- Endovascular cerebral venous sinus stenting
- *Max medical therapy: { acetazolamide, steroids

Mitchell, JL.et al. Brain;146(5):1821-1830, 2023.
Subramanian PS.. Curr Neurol Neurosci Rep. Aug;24(8):265-272. 2024
Ploof J, et al. J Child Neurol. 2021 Oct;36(11):1047-1053.

Clinical Diagnostics

Clinical Diagnostics

- Perimetry
- Fundus photography
- Fundus autofluorescence
- Optical Coherence Tomography
  - Enhanced Depth Imaging
  - Retinal nerve fiber layer thickness
  - Ganglion cell–inner plexiform layer (GC-IPL)
  - En face reconstruction
- Ultrasound
- Fluorescein angiography
  - Oral
  - Intravenous

Congenital optic neuropathies with systemic associations

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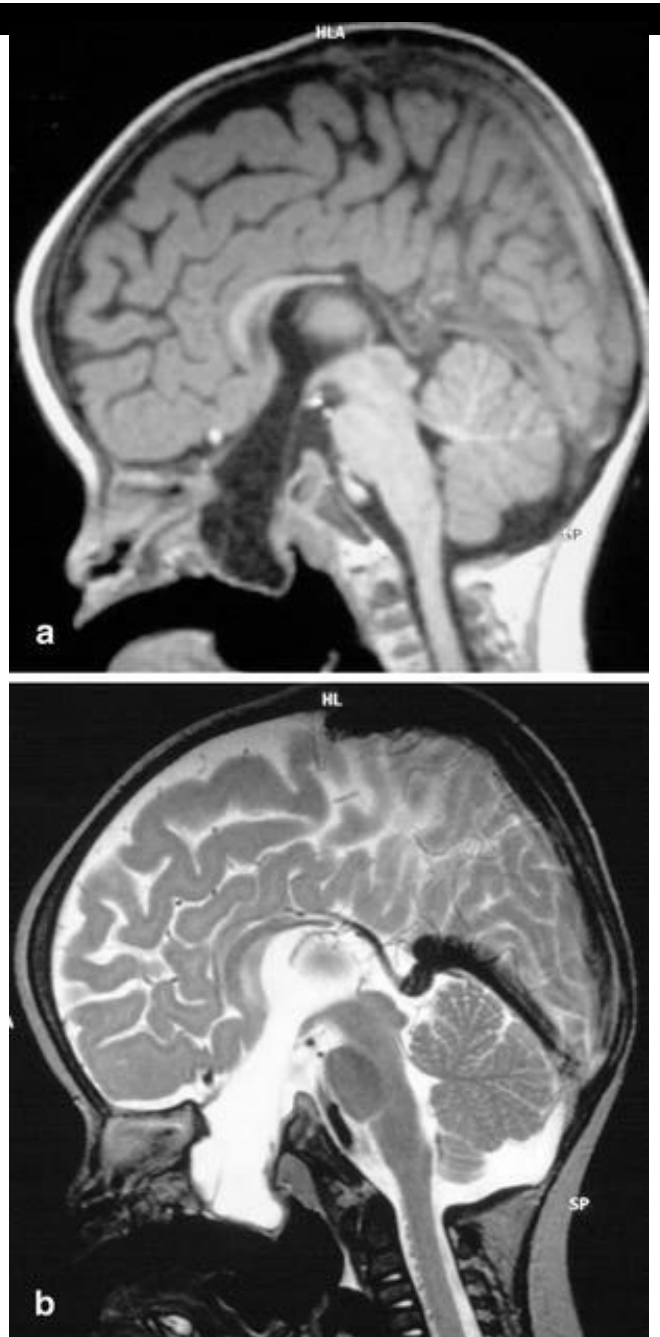
EMORY EYE CENTER

Financial disclosures

- I have no relevant financial relationships to disclose

Pre-test question 1

- What congenital optic neuropathy is most commonly seen associated with the finding depicted in the image?
  - A) Morning glory disc anomaly
  - B) Optic nerve hypoplasia
  - C) Peripapillary staphyloma
  - D) Optic disc coloboma

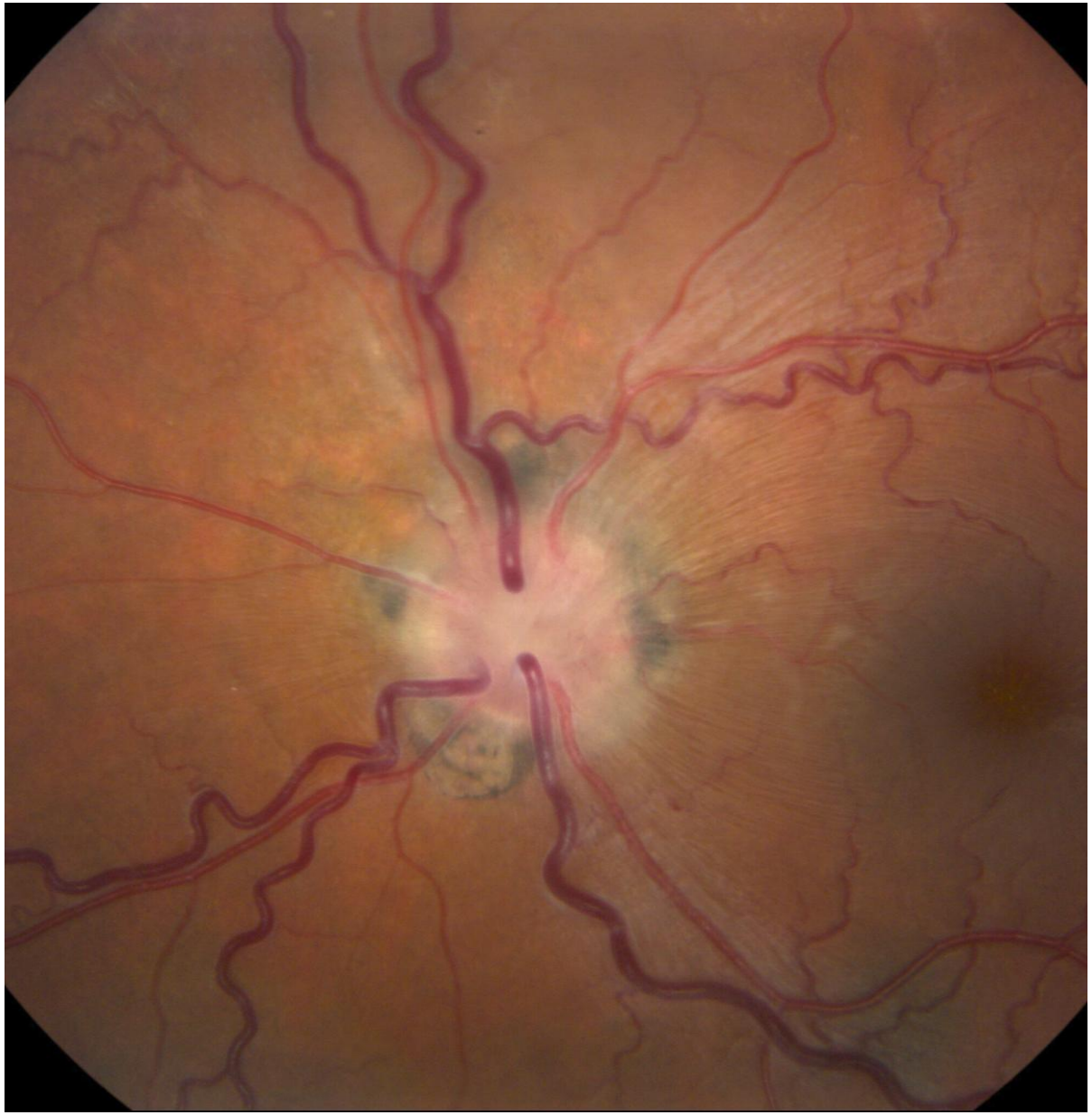


Chen CS, David D, Haniah A. *Childs Nerv Syst* 2004;20:87-90

ARS Question 1

Pre-test question 2

- A patient presents with the optic nerve appearance depicted in the image. What systemic problem does this patient need to be screened for?
  - A) Intracranial vascular anomalies
  - B) Hormonal deficiencies
  - C) Seizure disorder
  - D) Renal impairment



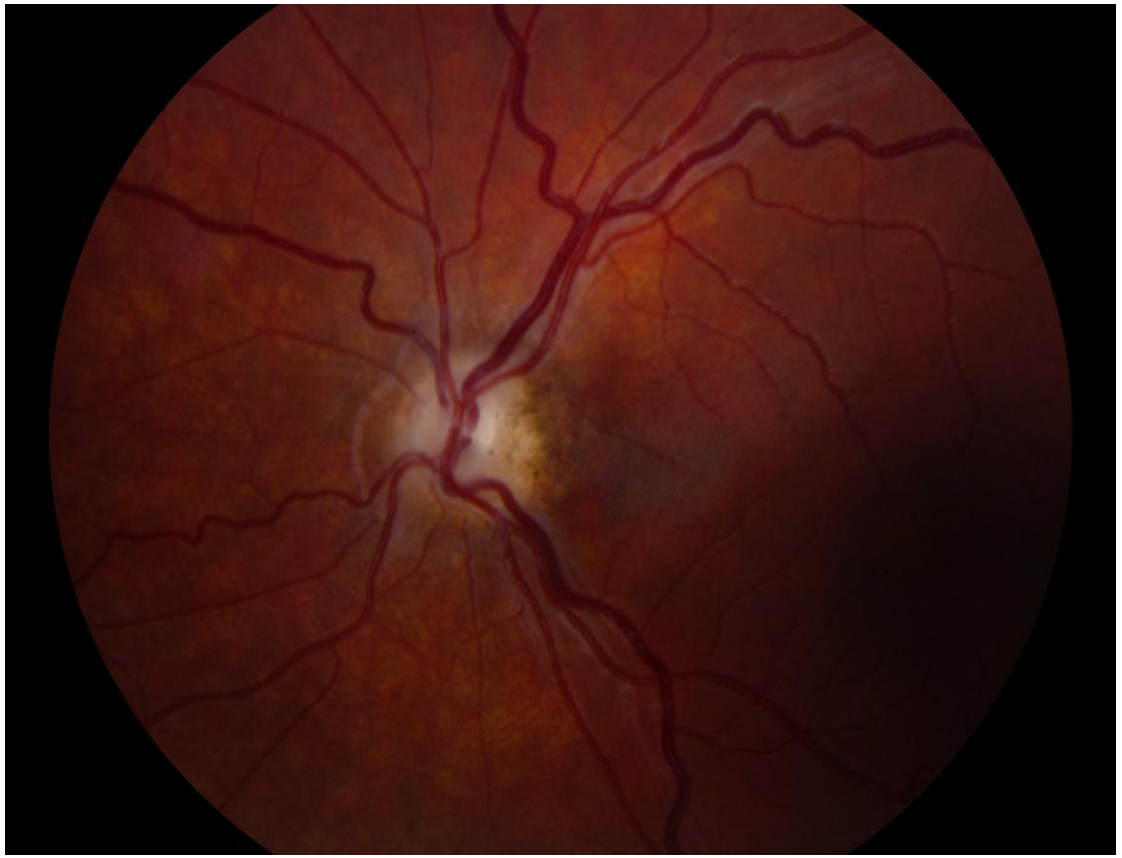
ARS Question 2

Congenital optic nerve abnormalities

- Optic nerve hypoplasia
- Optic nerve coloboma
- Morning glory disc anomaly
- Aicardi syndrome
- Papillorenal syndrome
- Congenital tilted disc syndrome
- Optic nerve aplasia
- Optic pit
- Peripapillary staphyloma
- Megalopapilla
- Myelinated nerve fibers
- Bergmeister’s papilla

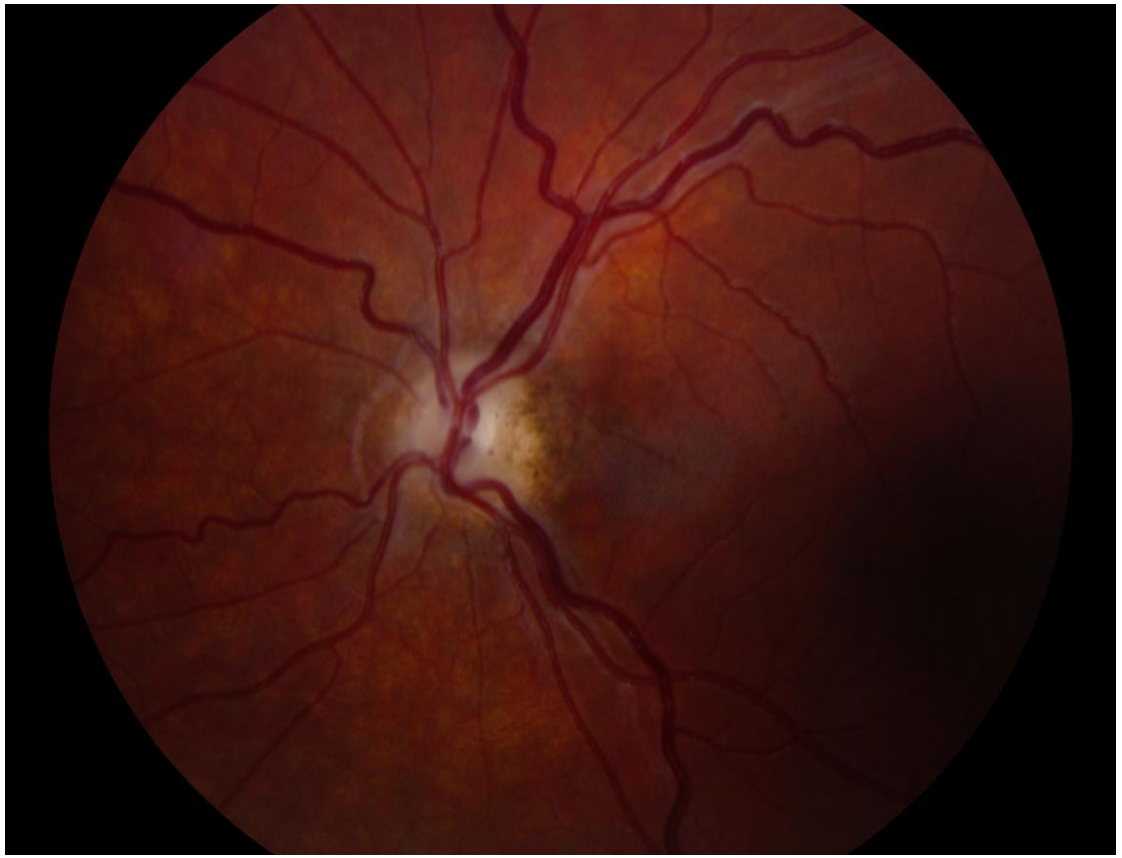
Optic nerve hypoplasia

- One of the most common congenital optic disc anomalies encountered
- Small nerve
  - Decreased number of axons
  - Normal support tissue
- “Double ring” sign
  - Normal junction of sclera and lamina cribrosa (outer ring)
  - Abnormal extension of retina and RPE over outer portion of lamina cribrosa (inner ring)
- Tortuous vessels

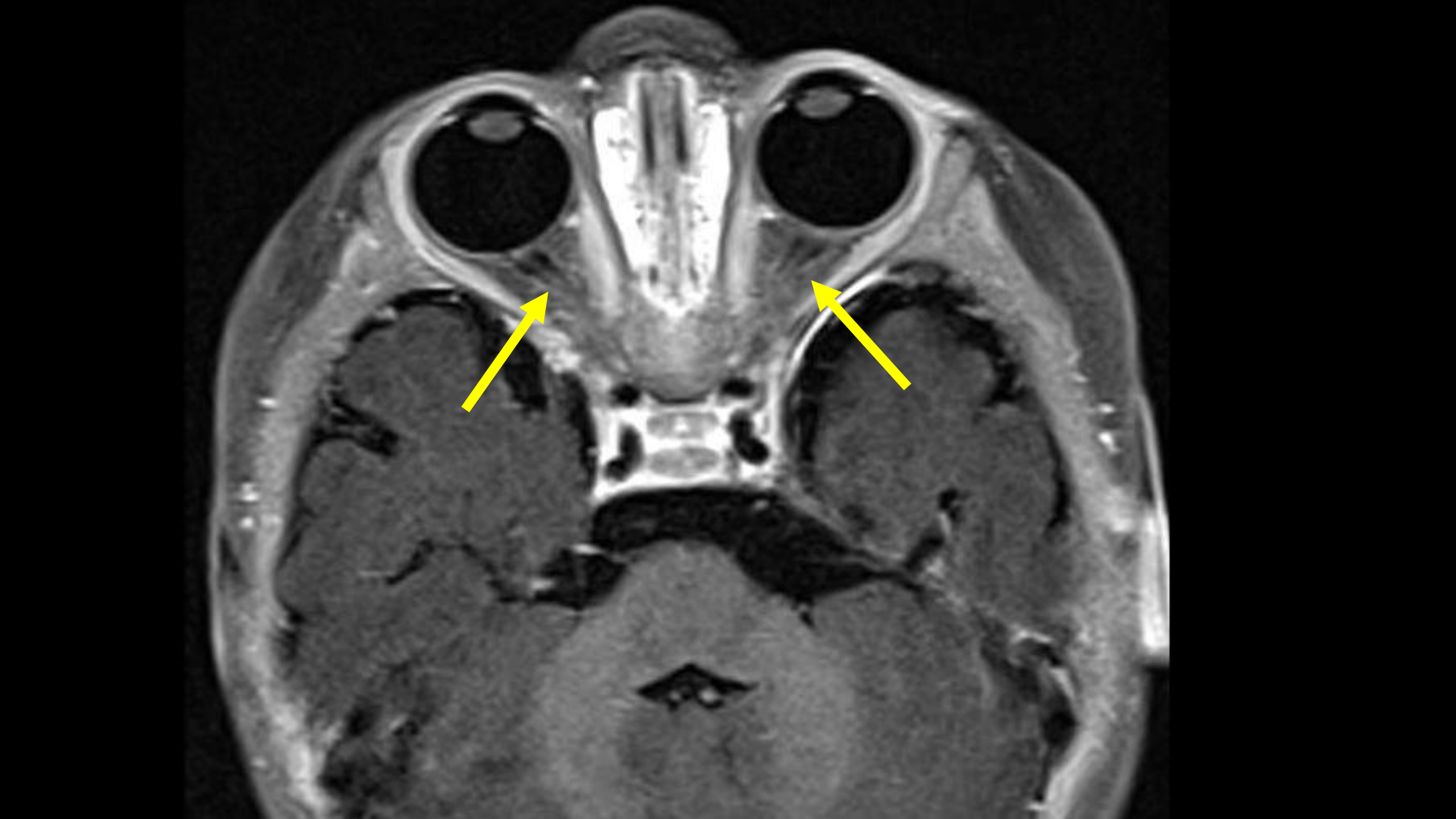


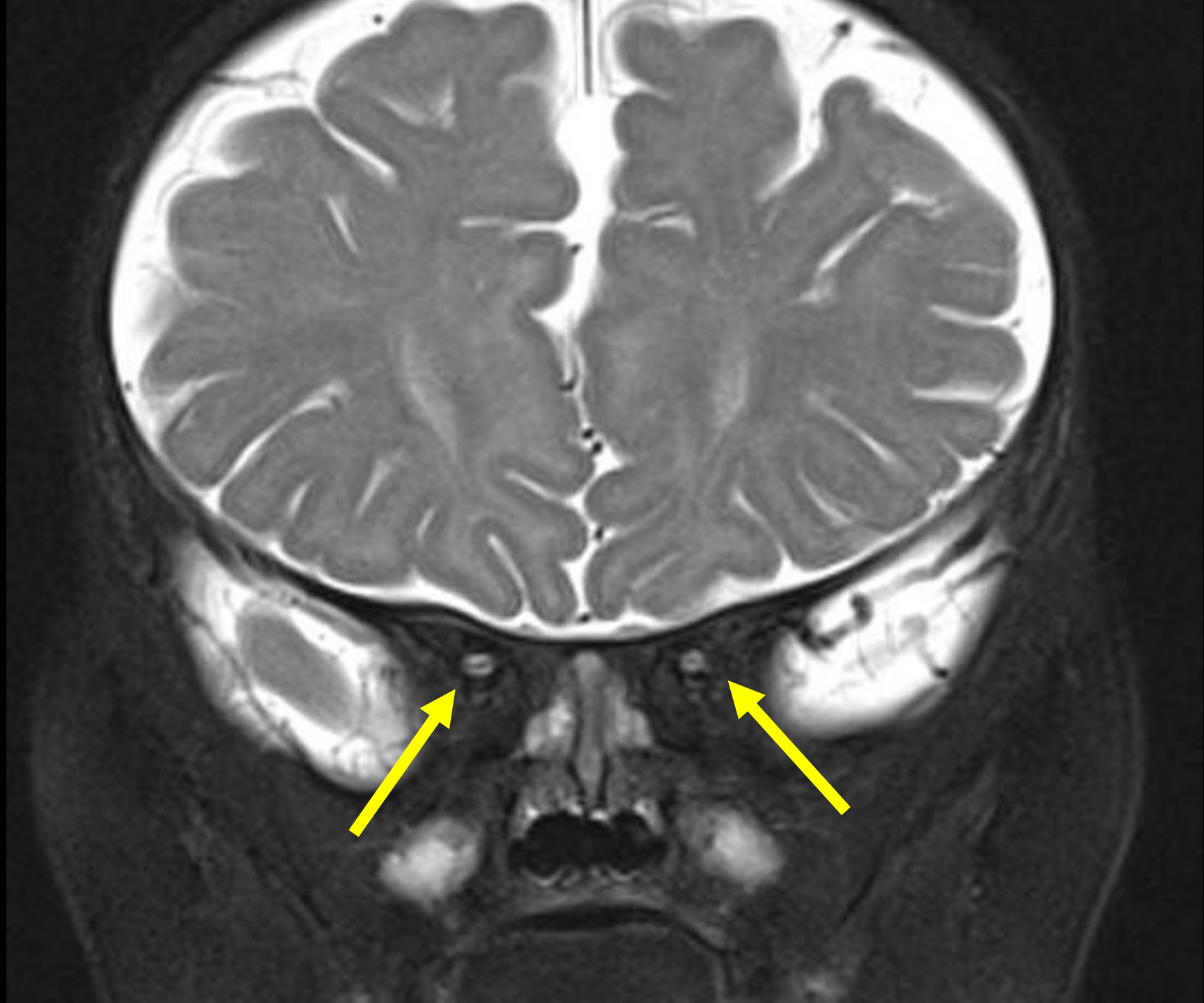
Optic nerve hypoplasia

- Normal optic nerve development:
  - 3.7million axons at 16-17 weeks GA
  - Apoptosis to 1.1million axons by 31 weeks GA
- Optic nerve hypoplasia
  - Failure of development of axons OR
  - Intrauterine degenerative process
- Visual acuity
  - 20/20 to NLP
- Genetic associations
  - Rare - <1% have a *HESX1* mutation



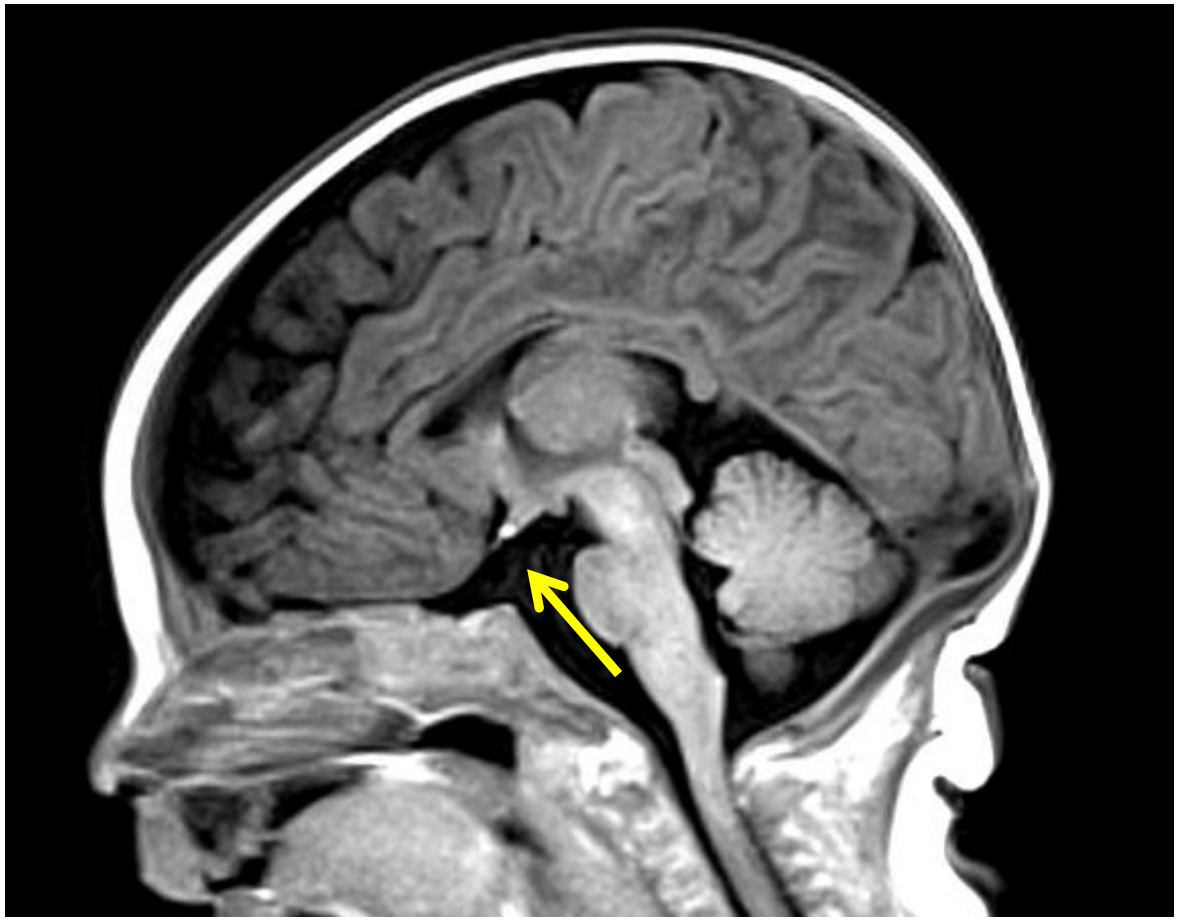
McNay DEG, Turton JP, Kelberman D, et al. *J Clin Endocrinol* Metab 2007;92:691-7





Optic nerve hypoplasia – systemic concerns

- Agensis of corpus callosum
- Hormonal abnormalities - pituitary dysfunction
  - Present in up to 75% of ONH patients
  - Endocrinopathies can be: growth hormone deficiency, hypothyroid, ACTH deficiency, diabetes insipidus
- If untreated sudden death can occur in ACTH deficiency due to inability to regulate blood sugar and blood pressure during febrile illness



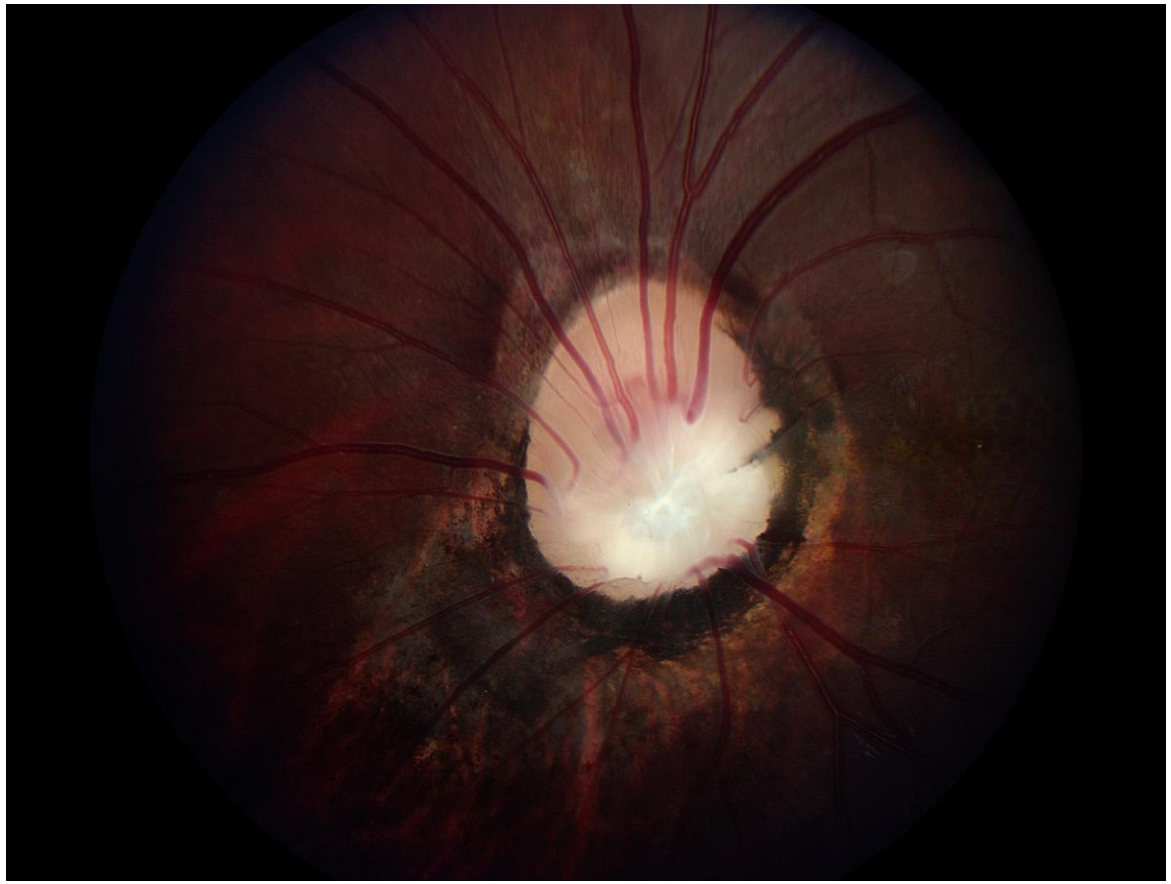
Ahmad T, Garcia-Filion P, Borchert M. et al. *J Pediatr* 2006;148:78-84

Optic nerve hypoplasia - recommendation

- All children with ONH get:
  - MRI brain
  - Endocrine evaluation

Optic nerve coloboma

- Abnormal closure of the embryonic fissure
  - Defect occurs b/w 5^{th}-7^{th} week of gestation
- Can be associated with colobomatous defects of other parts of the eye
- Can have decreased visual acuity – papillomacular bundle involvement
- Can have serous RD
- Can have large orbital cysts, microphthalmia

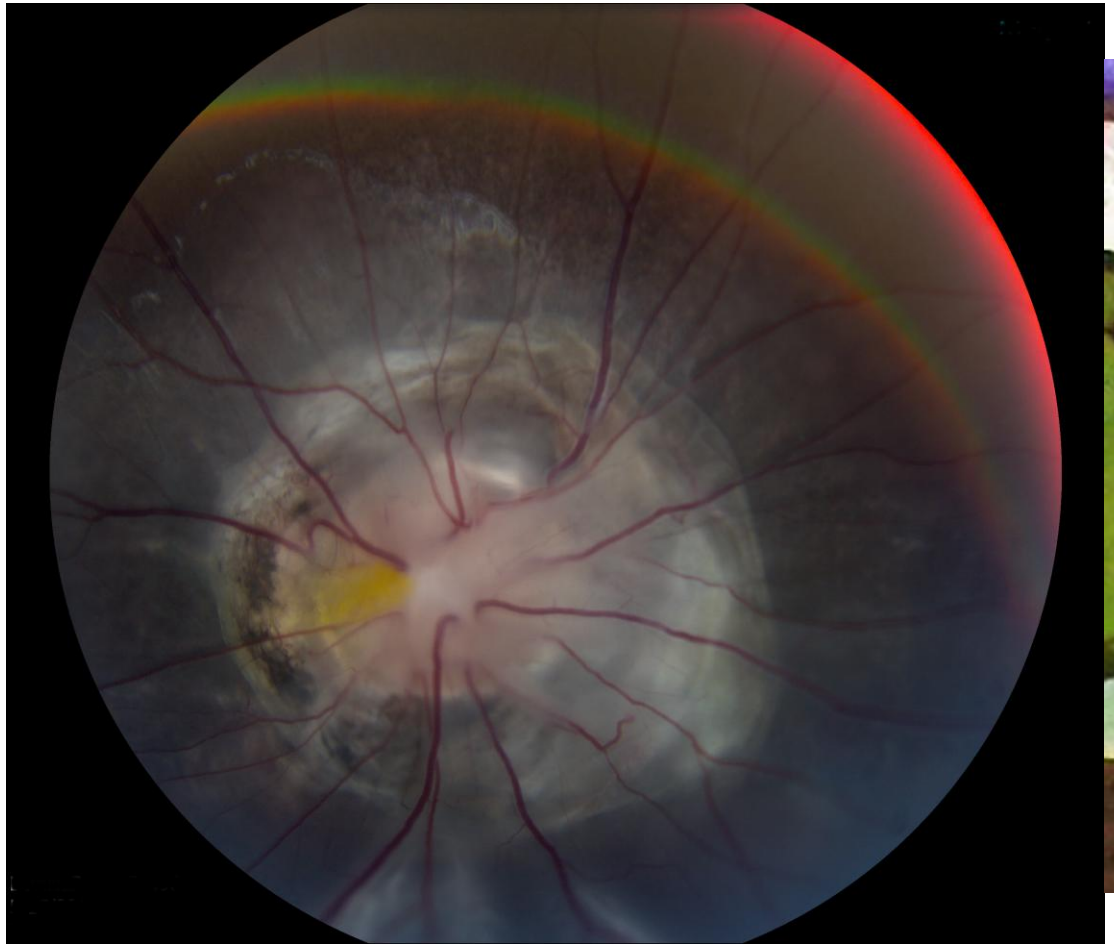


Optic nerve coloboma – systemic concerns

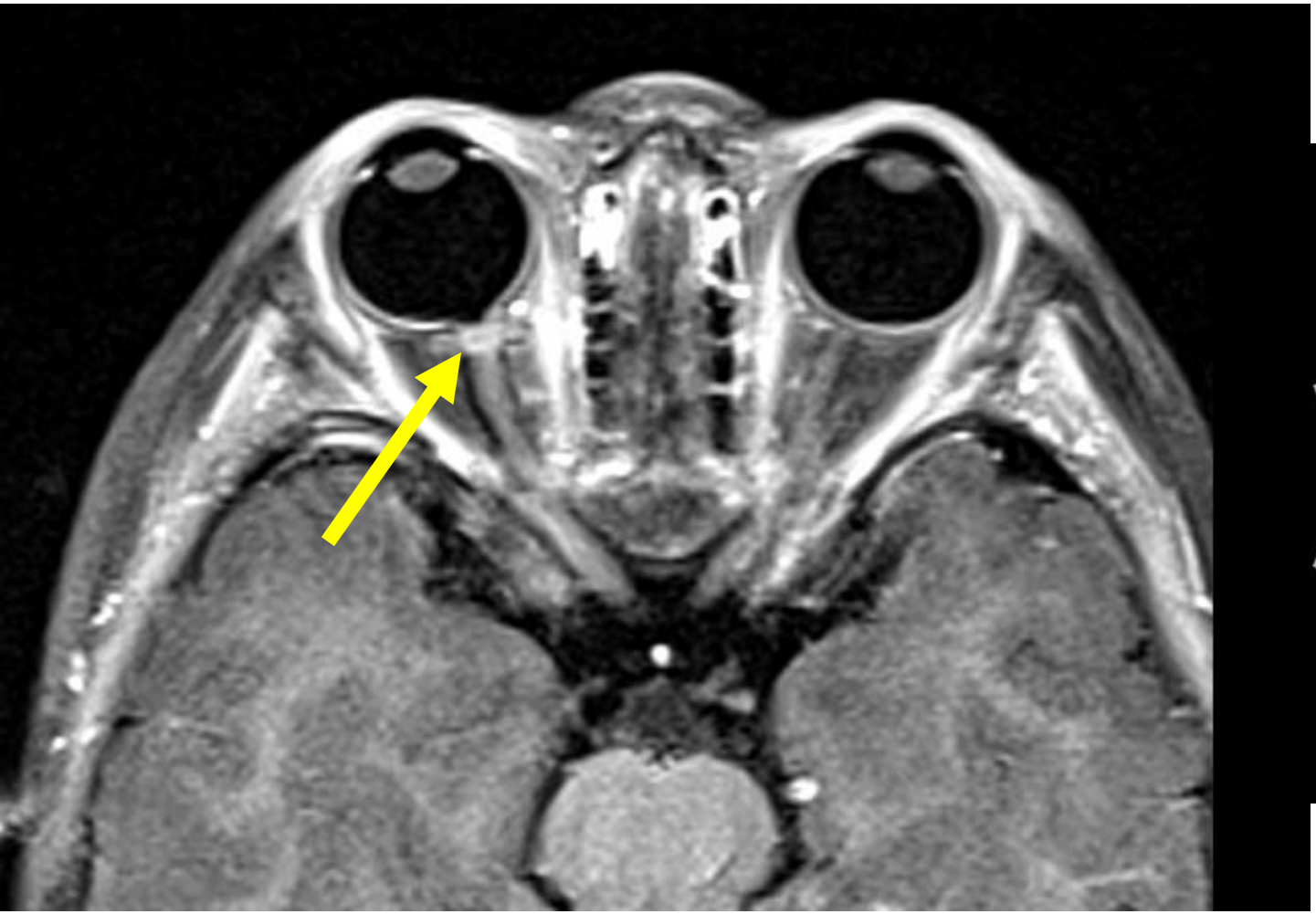
- Can be sporadic or AD
- Associated with CHARGE syndrome (Coloboma, Heart defect, Atresia choanae, Retarded growth and development, Genital hypoplasia, Ear anomalies/deafness
- *CHD7* mutations are most frequently associated with CHARGE syndrome
    - Genetic testing and counseling is recommended

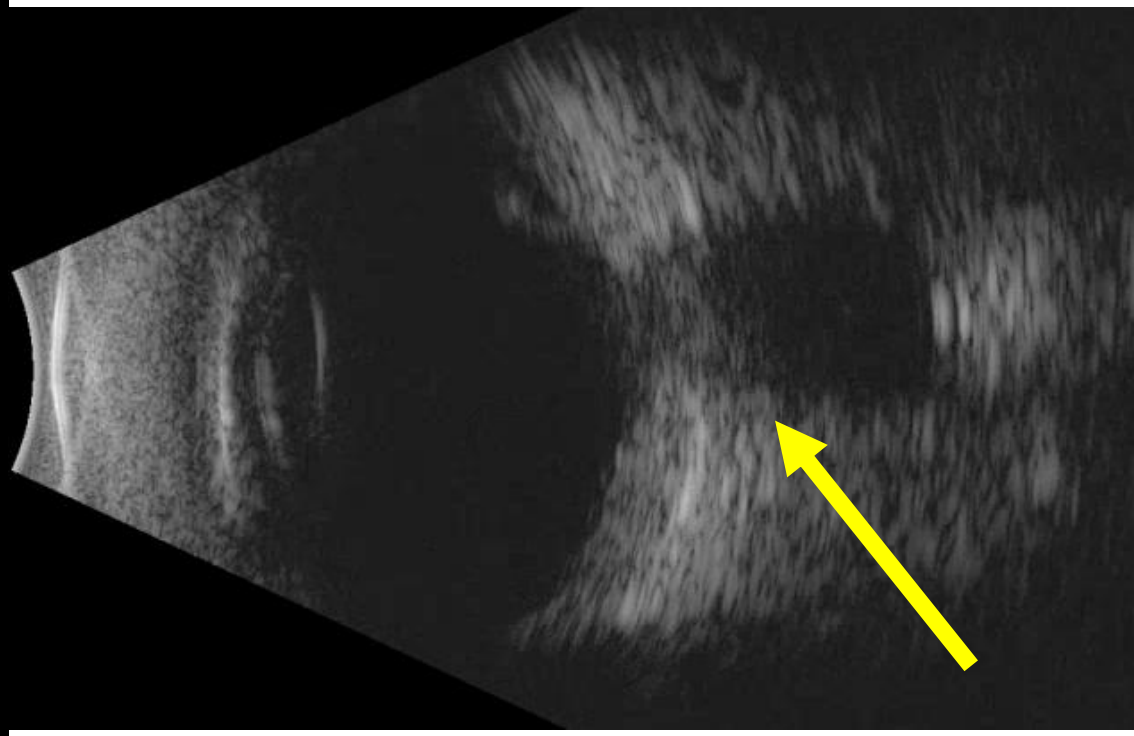
Lingam G, et al. Ocular coloboma – a comprehensive review for the clinician. *Eye* 2021;35:2086-2109.
George A, et al. Genetics of syndromic ocular coloboma: CHARGE and COACH syndromes. *Exp Eye Res* 2020;193:107940

Morning glory disc anomaly



Morning glory disc anomaly



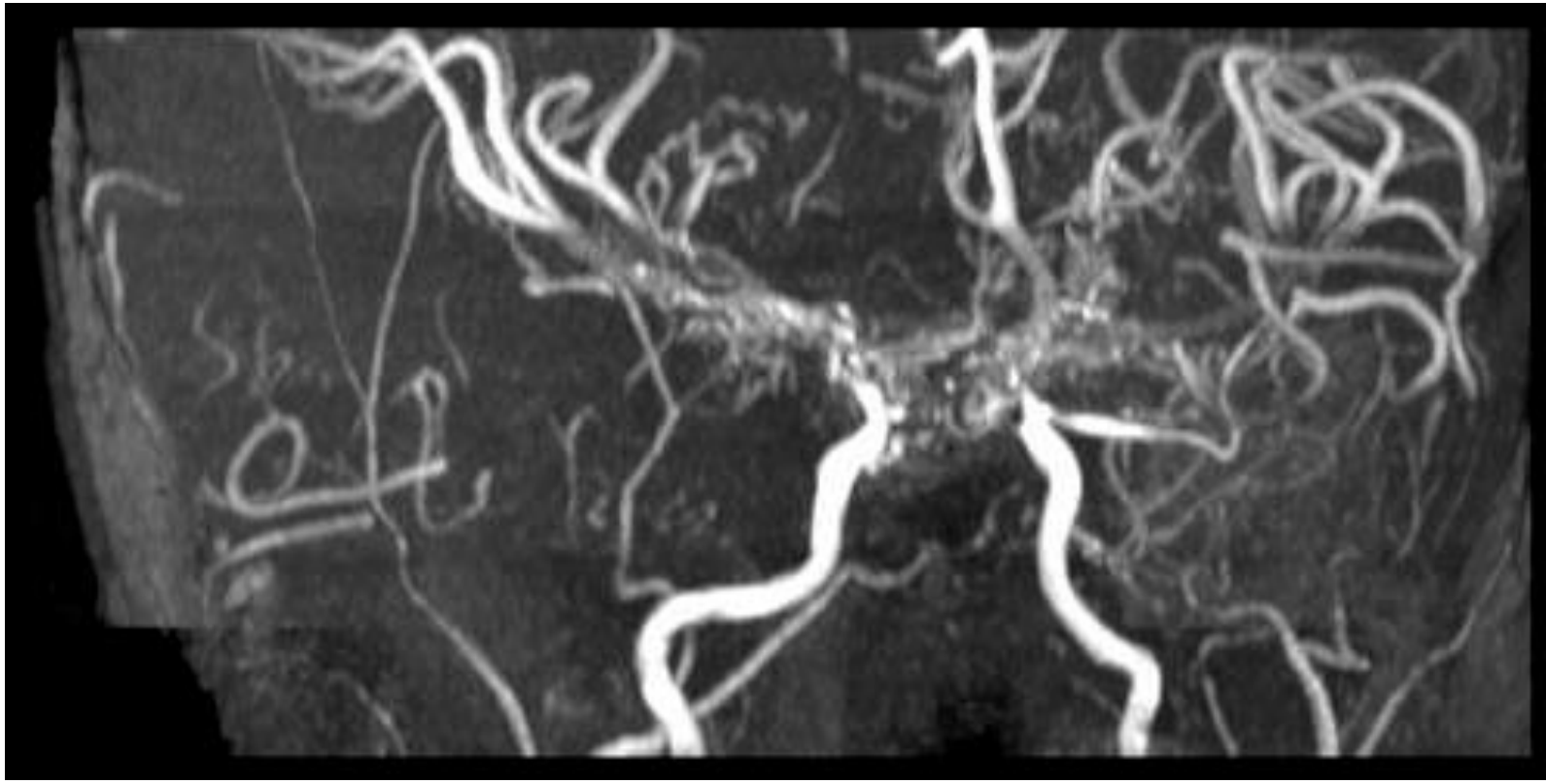


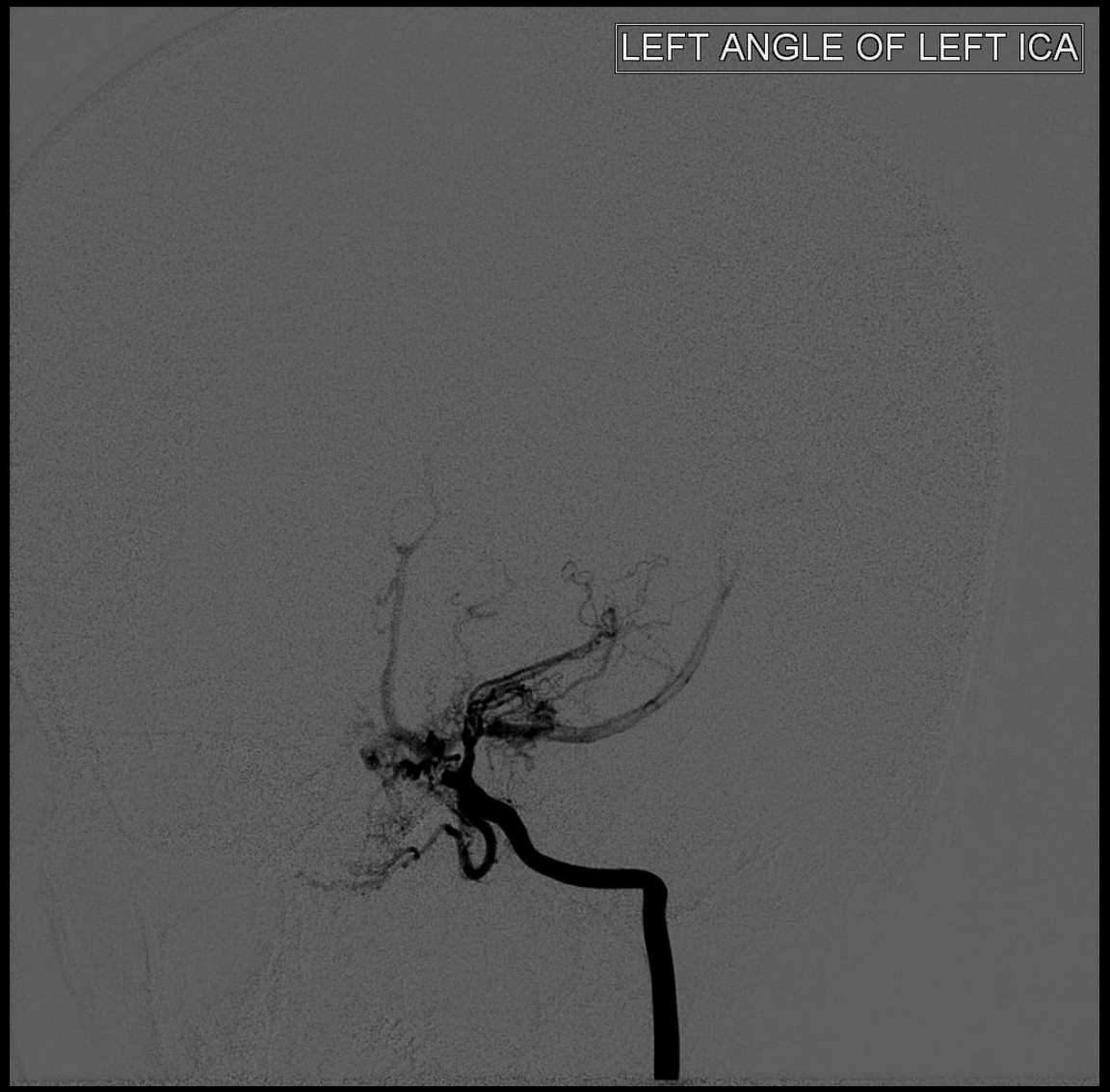
Morning glory disc anomaly

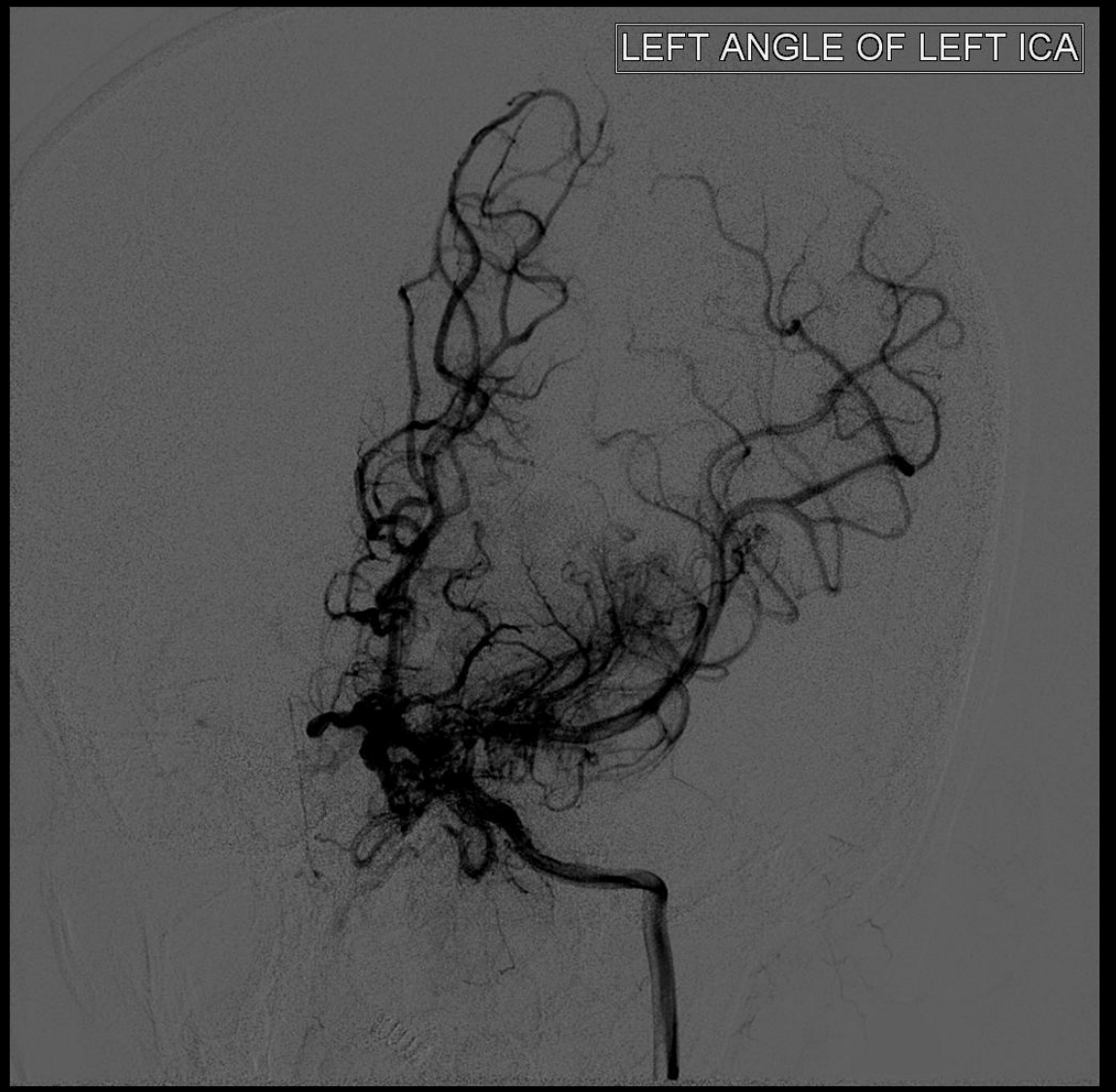
- Sporadic defect – possible primary mesenchymal abnormality (partial development of lamina cribrosa and/or sclera)
- Usually unilateral
- Genetic associations: *PAX6* and *MMP19* mutations
- Vision: usually between 20/200-CF
- Can develop serous RD

Morning glory disc anomaly: Systemic implications

- Ipsilateral intracranial vascular dysgenesis
  - Hypoplasia of carotid arteries
  - Moyamoya disase – “puff of smoke”
    - Associated with developmental delay, seizure, TIA, stroke
    - Can be treated with antiplatelet regimen
    - Treatment may include revascularization surgery
  - Lenhart et al: 9/20 MGDA patient had cerebrovascular anomalies

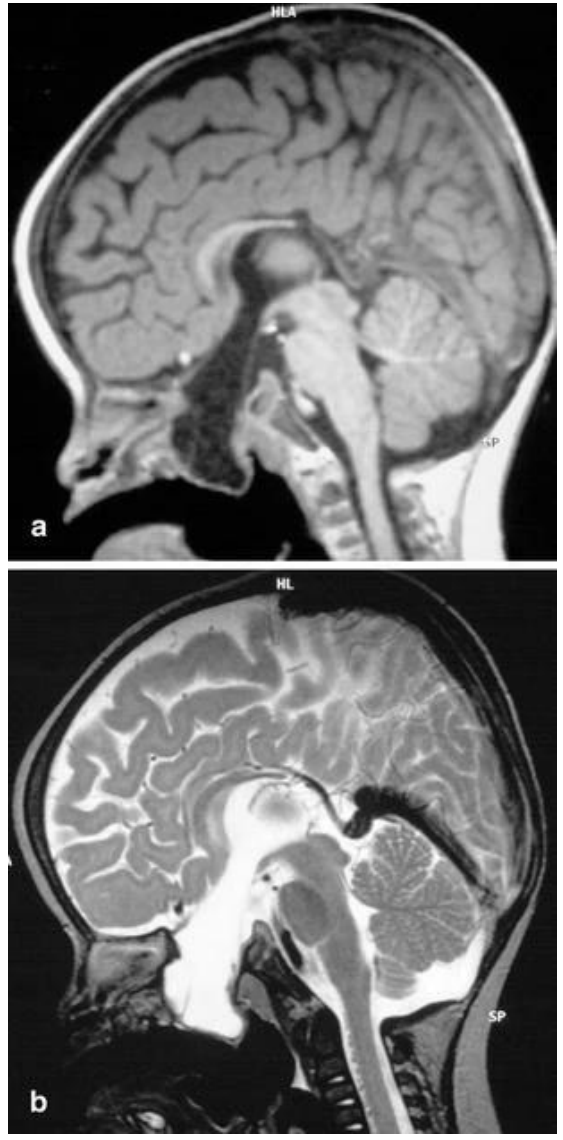



LEFT ANGLE OF LEFT ICA


LEFT ANGLE OF LEFT ICA

Morning glory disc anomaly: Systemic implications

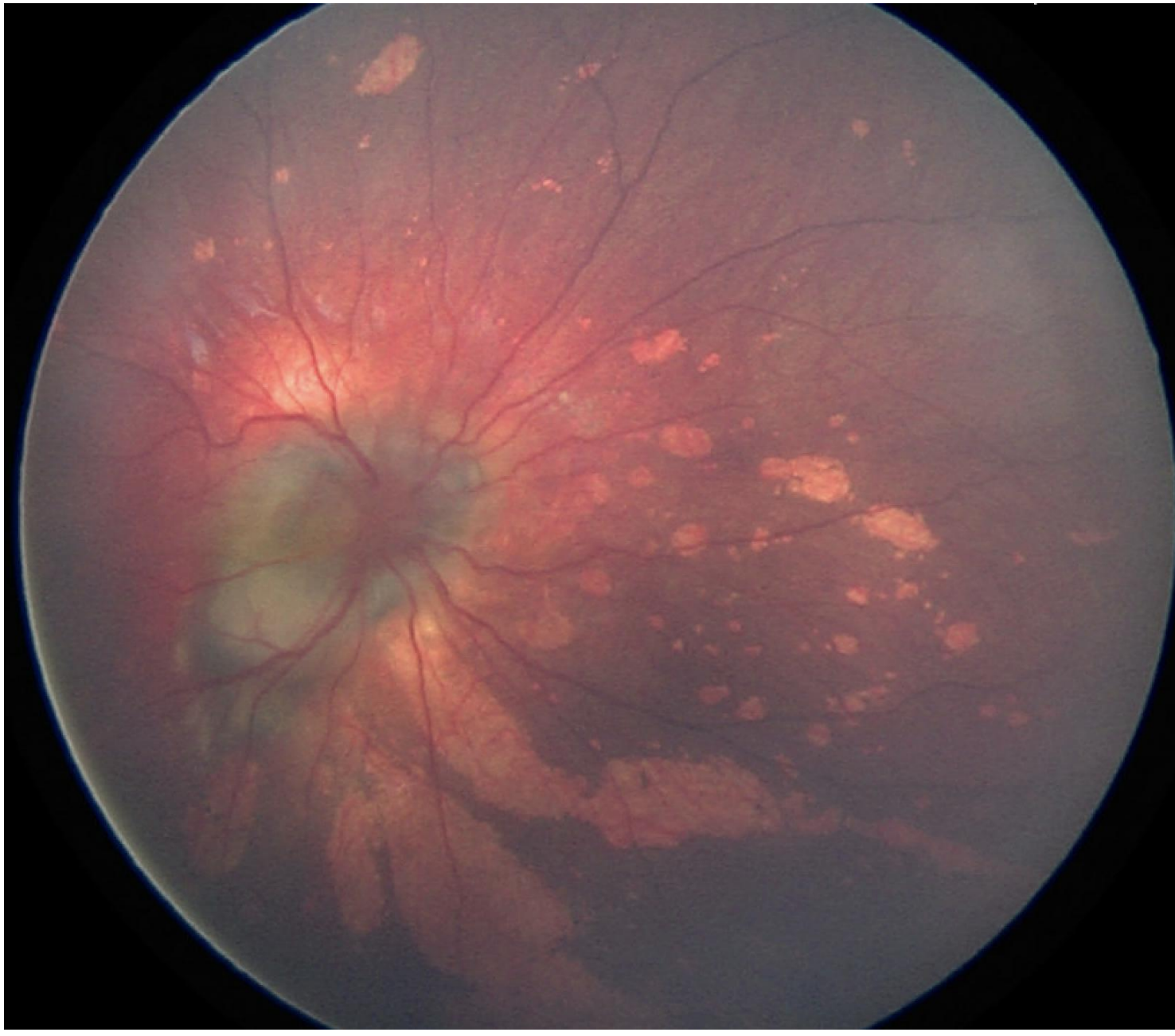
- Other structural anomalies:
  - Cleft lip/palate
  - Hypertelorism
  - Agenesis of corpus callosum
  - Chiari I malformation
  - Basal encephalocele



Chen CS, David D, Haniah A. *Childs Nerv Syst* 2004;20:87-90

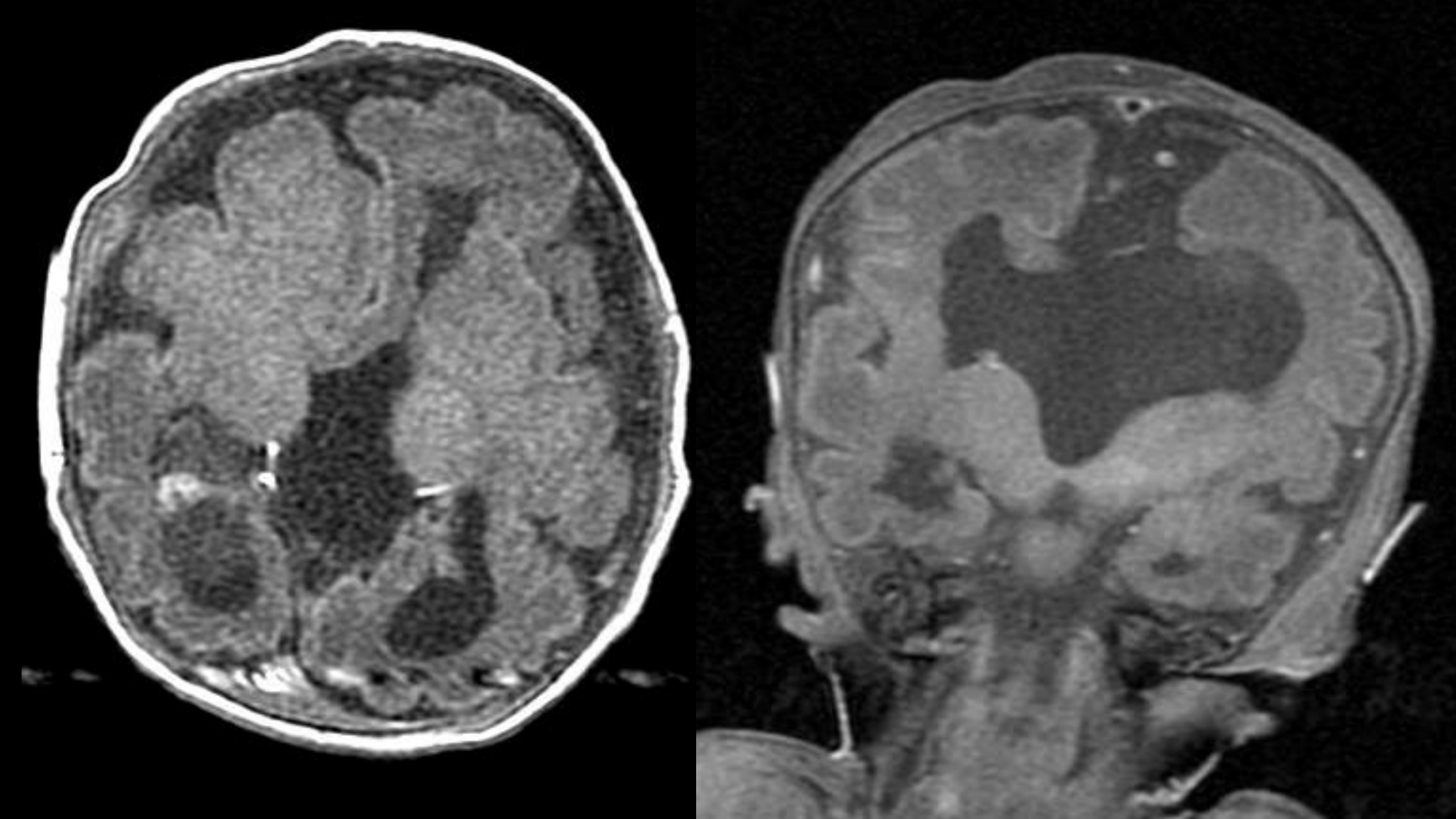
Aicardi syndrome

- Cerebroretinal disorder
- Dysplastic/colobomatous/hypoplastic optic nerve
- Retinal lacunae

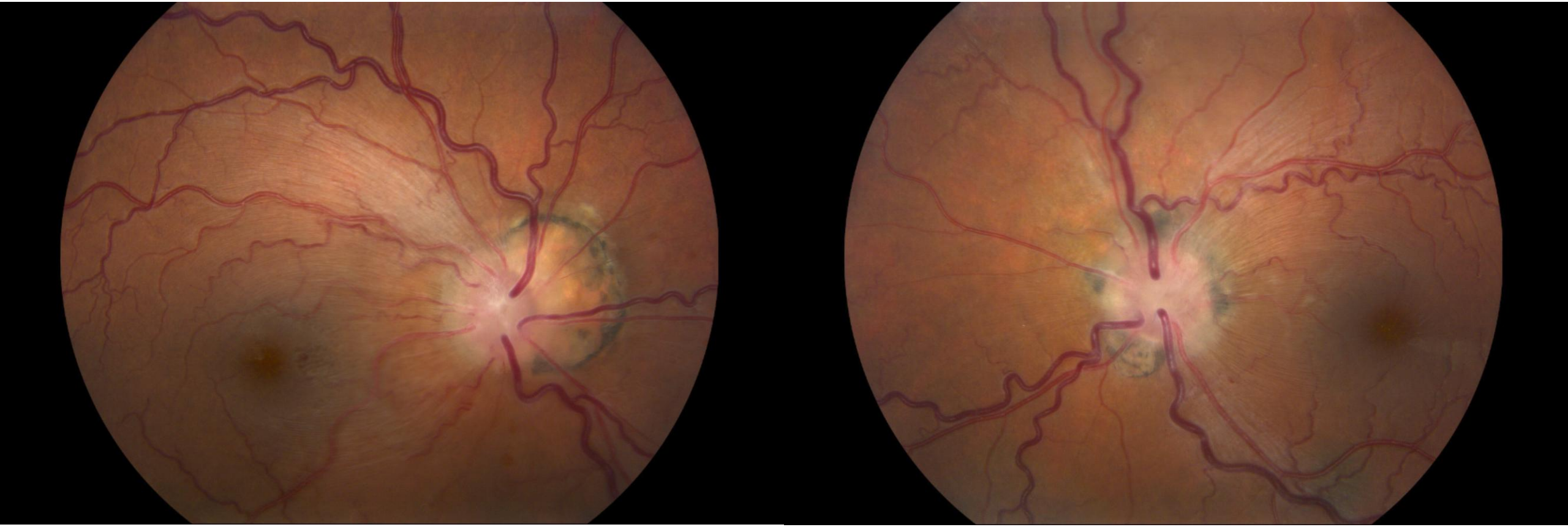


Aicardi syndrome – systemic implications

- X-linked disorder – lethal in males
- Associated with seizure disorders
- Severe developmental delay (91% do not progress beyond 1yr milestones)
- Costal malformations
- MRI: agenesis of the corpus callosum, cortical migration abnormalities/malformations

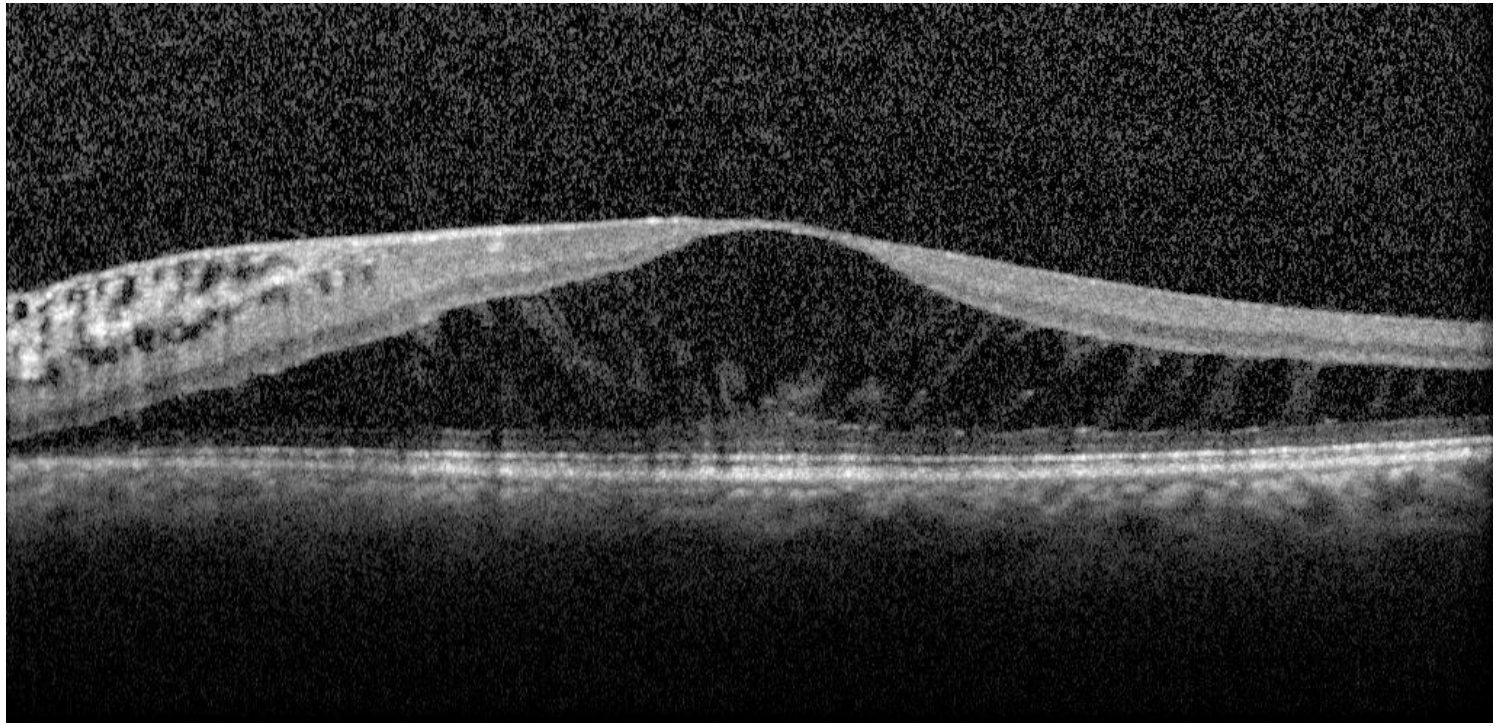


Papillorenal syndrome



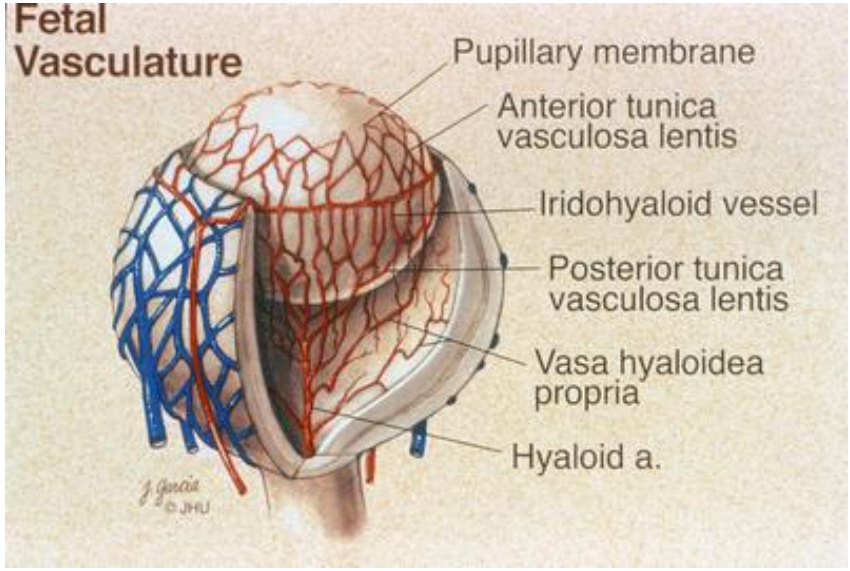
Papillorenal syndrome

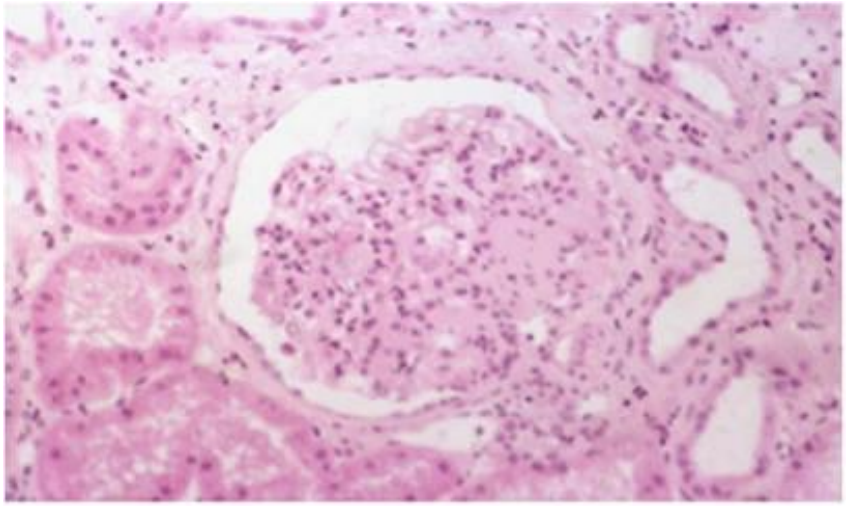
- Retinal vessels emanate from periphery of nerve
- Vision is frequently normal until retina is involved
- May have retinal and choroidal hypoplasia
- Patients develop retinoschisis
- May develop serous RD



Papillorenal syndrome Systemic associations

- May be sporadic (65%) or familial – frequently due to mutations in *PAX2* gene – AD inheritance (99% penetrance)
- Primary failure of angiogenesis: failure of hyaloid system to develop -> absent central retinal vessels
- Dysfunctional angiogenesis -> dysplastic kidneys, glomerulosclerosis
- 10% have high frequency hearing loss
- Average age to ESRD: 19.5 years





Bower M, Salomon R, Allanson j, et al. *Hum Mutat*. 2012;33:457-466

Thank you!

Congenital and Acquired Optic Neuropathies – What Not to Miss

Surveillance and Management of Optic Pathway Gliomas in the Setting of NF1

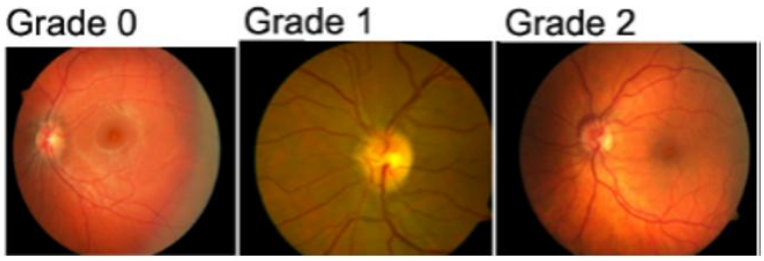
Gena Heidary, MD, PhD

I. General Comment on NF1 OPG versus sporadic and that the focus will be on NF1 OPG
II. General Guidelines for Surveillance
    A. In the setting of café au lait spots
        1. Vision function
        2. Lisch nodules
        3. Choroidal abnormalities (new diagnostic criterion)
        4. Evidence of optic nerve findings

    B. When diagnosis of NF1 is known
        1. In children younger than 8 years old
        2. In children 8 years and older
        3. What elements would prompt neuroimaging?
            a. Change in visual function
            b. Change in optic nerve appearance
III. Management of NF1 OPGs
    A. Case examples contrasting presentation of optic nerve edema early in NF1-OPG and disease course of these case examples
    B. Protocol for evaluation
    C. Protocol for surveillance
    D. Indications for treatment
        1. Reduction in visual function (VA and VF)
        2. How to incorporate OCT
        3. Protocol for surveillance once treatment is initiated
    E. Clinical course of the two case examples
    F. Summary

Optic nerve head drusen and pseudopapilledema handout

Frisén scale: easiest is to look for vessel obscuration



Could be papilledema or pseudopapilledema

Should never be confused for pseudopapilledema (vessel obscuration, mushroom shaped nerve)

Key paper

This is the paper that standardized OCT-based diagnosis of optic disc drusen (ODD) and helped reduce confusion between:

- true ODD
- papilledema
- and later, PHOMS-like structures being mislabeled as drusen
- It's a consensus recommendations paper (not a treatment trial)

The Optic Disc Drusen Studies Consortium Recommendations for Diagnosis of Optic Disc Drusen Using Optical Coherence Tomography

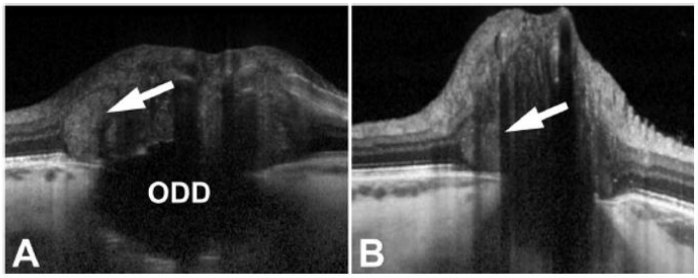
Lasse Malmqvist, MD, Lulu Bursztyn, Msc, MD, Fiona Costello, MD, PhD, Kathleen Digre, MD, J. Alexander Fraser, MD, Clare Fraser, MMed, Bradley Katz, MD, PhD, Mitchell Lawlor, FRANZCO, PhD, Axel Petzold, MD, PhD, Patrick Sibony, MD, Judith Warner, MD, Marianne Wegener, MD, Sui Wong, MD, Steffen Hamann, MD, PhD

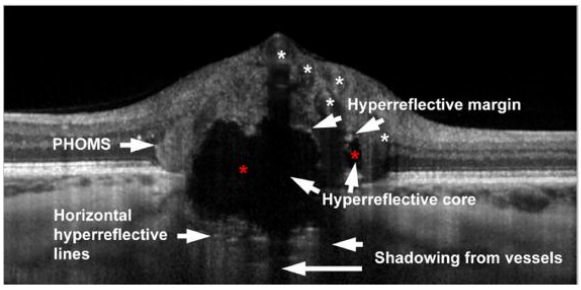
Background: Making an accurate diagnosis of optic disc drusen (ODD) is important as part of the work-up for possible life-threatening optic disc edema. It also is important to follow the slowly progressive visual field defects many patients with ODD experience. The introduction of enhanced depth imaging optical coherence tomography (EDI-OCT) has improved the visualization of more deeply buried ODD. There is, however, no consensus regarding the diagnosis of ODD using OCT. The purpose of this study was to develop a consensus recommendation for diagnosing ODD using OCT.
Methods: The members of the Optic Disc Drusen Studies (ODDS) Consortium are either fellowship trained neuro-ophthalmologists with an interest in ODD, or researchers with an interest in ODD. Four standardization steps were performed by the consortium members with a focus on both image acquisition and diagnosis of ODD.
Results: Based on prior knowledge and experiences from the standardization steps, the ODDS Consortium reached a consensus regarding OCT acquisition and diagnosis of ODD. The recommendations from the ODDS Consortium include scanning protocol, data selection, data analysis, and nomenclature.
Conclusions: The ODDS Consortium recommendations are important in the process of establishing a reliable and consistent diagnosis of ODD using OCT for both clinicians and researchers.

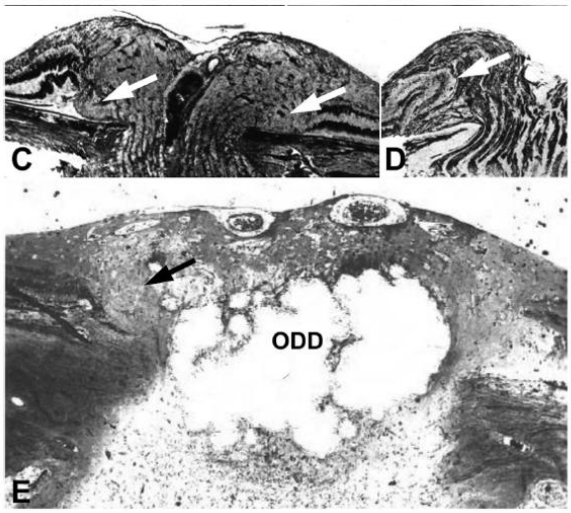
Journal of Neuro-Ophthalmology 2018;38:299–307
doi: 10.1097/WNO.0000000000000585

Optic disc drusen (ODD) are predominantly calcified deposits in the optic nerve head with an estimated prevalence of 2.4% (1). The condition is generally benign, with rare complications such as anterior ischemic optic neuropathy (2,3) and retinal vascular occlusions (4,5). Confusion can arise because the frequently observed protrusion of the optic disc and blurring of the optic disc margin can be mistaken for optic disc swelling (6–8). Separating ODD from other causes of optic disc elevation such as papillede-

Images from the paper

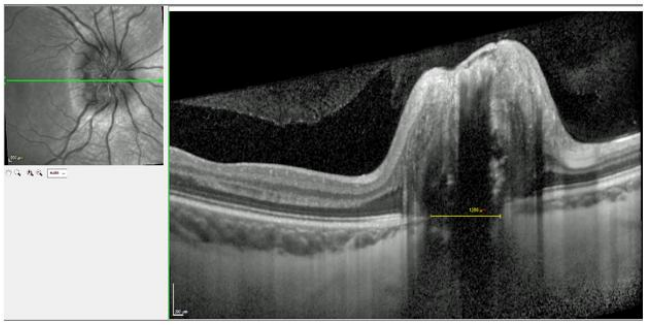




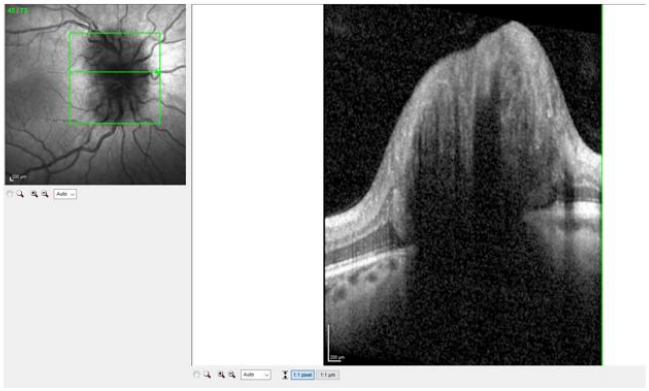


2018 consensus paper

Drusen: hyporeflective core: partial hyperreflective margin



PHOMS: peripapillary hyperreflective ovoid mass-like structures



Multirater Validation of Peripapillary Hyperreflective Ovoid Mass-like Structures (PHOMS)

Axel Petzold[a,b], Valerie Biousse[c], Lulu Bursztyn[d], Fiona Costello[e], Alison Crum[f], Kathleen Digre[g], Clare Fraser[h], J. Alex Fraser[i], Bradley Katz[f], Neringa Jurkute[j], Nancy Newman[k], Jette Lautrup-Battistini[l], Mitchell Lawlor[h], Petra Liskova[m], Birgit Lorenz[n], Lasse Malmqvist[o], Jason Peragallo[c], Patrick Sibony[p], Prem Subramanian[q], Robert Rejdak[r], Katarzyna Nowomiejska[r], Valerie Touitou[s], Judith Warner[t], Marianne Wegener[o], Sui Wong[u], Patrick Yu-Wai-Man[v,w], and Steffen Hamann[x]

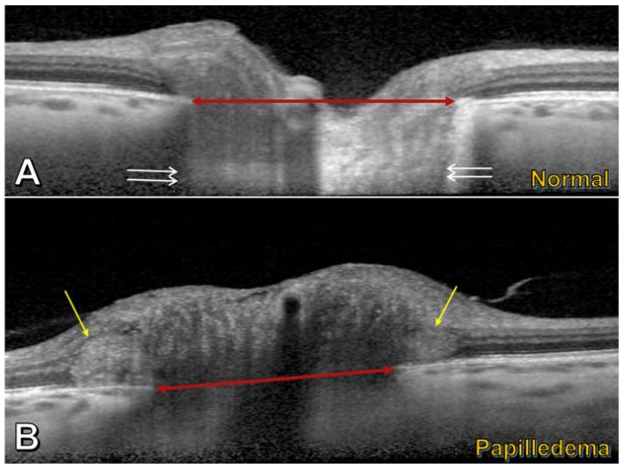
- Refined PHOMS definition:
- Re-defined PHOMS by:
- Location Strictly peripapillary Sitting on top of Bruch's membrane on OCT B-scan
- Causes an upward deflection ("ski slope") of at least two retinal layers
- Signal appearance/Reflectivity similar to RNFL-- Consistent with the idea that PHOMS may reflect axonal stasis
- Helps distinguish PHOMS from normal axon exit anatomy (which can mimic it in tilted discs, elevated disc borders, and myopia)

ABSTRACT

Peripapillary hyperreflective ovoid mass-like structures (PHOMS) are a new retinal optical coherence tomography (OCT) finding. The *Optic Disc Drusen Studies Consortium* had made recommendations to distinguish PHOMS from true optic disc drusen (ODD) in 2018. While publications on PHOMS have increased since then, the accuracy of the definition of PHOMS and reliability of detection is unknown. In this multi-rater study, we demonstrate that the 2018 definition of PHOMS resulted in a poor multi-rater kappa of 0.356. We performed a Delphi consensus process to develop a consistent and refined definition of PHOMS with clear principles around the nature of PHOMS and how they differ from normal anatomy. Fifty explanatory teaching slides, provided as supplementary material, allowed our expert group of raters to achieve a good level of agreement (kappa 0.701, 50 OCT scans, 21 raters). We recommend adopting the refined definition for PHOMS.

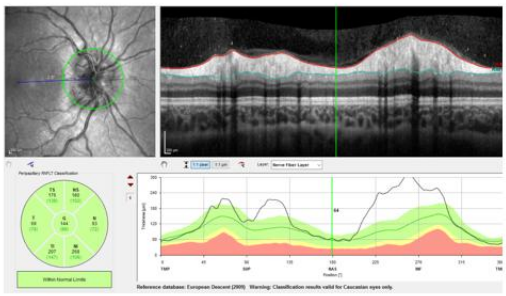
PHOMS: an OCT marker of axoplasmic stasis in the optic nerve head

J. Alexander Fraser, MD[1], Patrick A. Sibony, MD[2], Axel Petzold, MD, PhD[3], Caroline Thaung, MB, ChB, DPhil[4], Steffen Hamann, MD, PhD[5], ODDS Consortium[6]

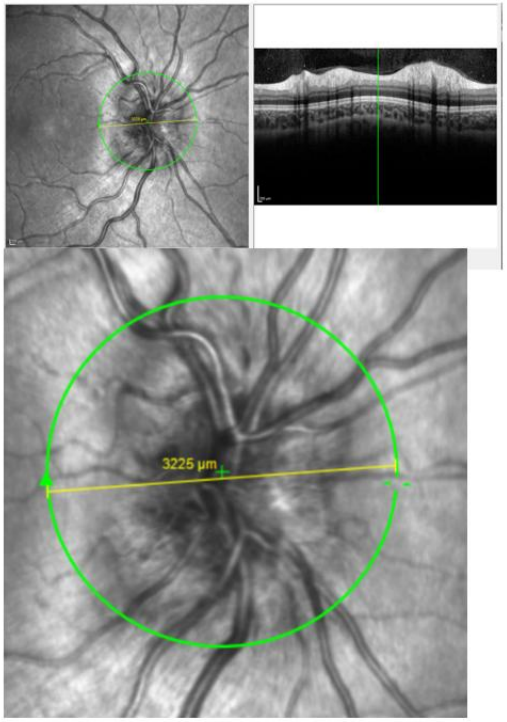


- PHOMS are a common but non-specific OCT marker of axoplasmic stasis in the optic nerve head.
- They are ODD or ODD precursors, though they can be seen in association with ODD as well as a other conditions.
- They do not exclude papilledema and often accompany it.

Sometimes, in a hyperopic child, the RNFL appears thick, should we worry about superimposed papilledema?

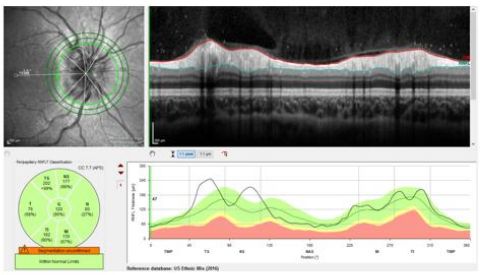


RNFL 144 µm
BMO 1.5 mm²

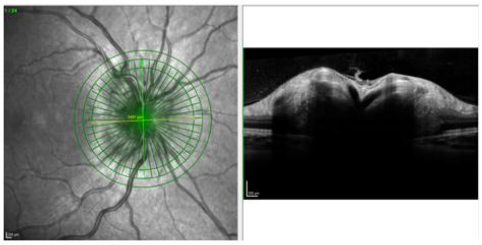


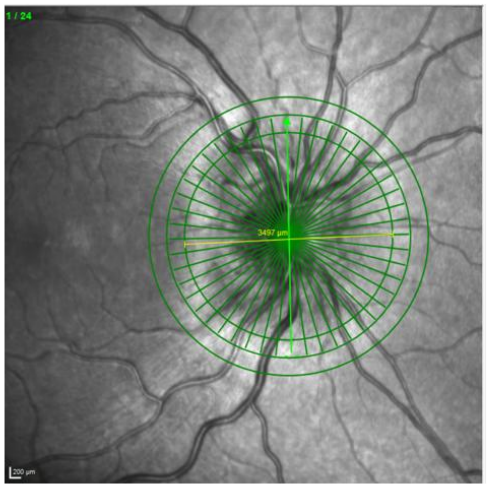
Size of projected circle 3225 µm (should fall at 3500 µm in a 23 mm eye

GMPE protocol: same child as above, same day

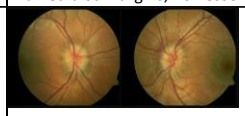
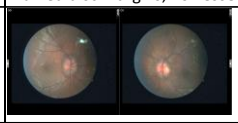
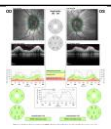
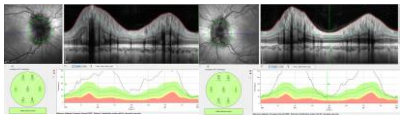
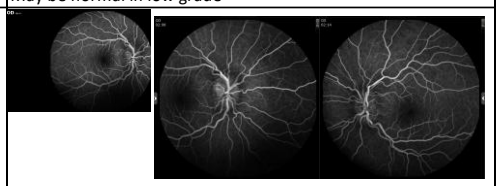
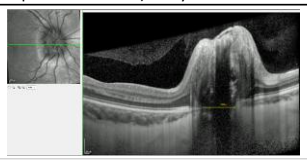
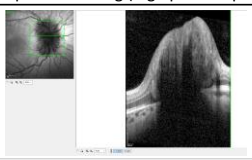


RNFL 120





Size of projected circle
3489

Visual function (vision, color, pupils, visual fields)	Normal	Normal
Fundus photos	Blurred disc margins, no vessel obscuration	Blurred disc margins, no vessel obscuration
		
Retinal Nerve Fiber Layer	Should be normal but it can be thickened due to optical artifact of hyperopic eye. May need to use GMPE or measure how far the RNFL projects away from nerve	Thickened. Usually>130 but could see RNFL low if baseline is lower
		
Ganglion cell layer	Normal	Normal
Fundus Autofluorescence	May show optic nerve drusen (40% sensitivity)	Normal
Fluorescein angiogram	Late nodular staining. May leak if there is a peripapillary choroidal neovascular membrane.	May be normal in low grade
		
OCT through the optic nerve head	Usually normal to small Bruch Membrane opening <1450 microns, no upward bowing of Bruch Membrane (low negative predictive value). May see drusen.	May show upward bowing (high positive predictive value)
		
Variability over time	Less than 5 micrometers variability over time	More than 5 mircometer variability