Congenital Cataract

Case Description
parents of a child have noted white pupils in his both eyes

Image Description

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external photograph shows dense white cataract involving the nucleus of the lens

Data acquisition

History
- onset & course
- TORCHS → maternal illness during pregnancy
- maternal drug ingestion during pregnancy
- ROP → premature birth
- systemic disease → growth & development delay
- ocular/systemic diseases in patient
- steroid use
- radiation
- family hx of eye disease
  - congenital cataract
  - retinoblastoma

Physical Exam
- VA
  - grating cards
  - Teller cards
  - tumbling E's
  - pictures
- IOP
- RAPD
  - cataract should not cause RAPD
- corneal diameter
  - PFV
  - microcornea
- slit lamp exam
  - cataract
    - location
    - size: <3 mm vs >3 mm
    - density: retina visible with direct ophthalmoscope through undilated pupil?
  - persistent fetal vasculature
  - examine family members
- dilated fundus exam
  - stalk between optic disc and lens
  - persistent fetal vasculature
  - medulloepithelioma
  - retinoblastoma
  - ROP
  - IP, Norrie disease
- cycloplegic refraction

Additional Testing
lab tests are indicated in patients with bilateral cataract

lab tests not indicated if:
- no known systemic diseases
- positive family history of isolated congenital/childhood cataract
- examination of parents shows lens opacities

- if no fundus view → B-scan ultrasound
- urine testing
  - galactosemia → reducing substances
  - Lowe syndrome → amino acids
  - Alport syndrome → amino acids
- galactosemia → RBC galactokinase
- diabetes → serum glucose
- hypoparathyroidism → serum calcium (low) & phosphorus (high)
- serology for TORCH

Assessment
congenital cataract (nuclear)

Treatment
- observation
- temporizing vs. definite rx for small cataracts
  - dilating drops
  - treat amblyopia
  - <3 mm → if visually not significant
  - small unilateral cataracts may still result in amblyopia
- cataract surgery
  - within days to weeks
    - +primary posterior capsulotomy
    - +anterior vitrectomy
  - >3 mm → if visually significant
- refractive rehabilitation
  - no IOL for children <6 mo of age
    - spectacles
    - aphakic contact lens
    - IOL later in life
      - pediatric IOL power —> aim for hyperopia; develop myopic shift later
      - no IOL for cataract associated with JRA

Referrals
refer patients with bilateral cataract to pediatrician for systemic evaluation

Patient Education/ Counselling
- Prognosis: good final visual acuity if removed early
- Complications: amblyopia treatment may be necessary
  - risk of amblyopia
- Follow-up
  - monitor for cataract progression and amblyopia
  - monitor for postoperative complications
  - opacification of posterior capsule
- Don'ts: children with rubella must be isolated from pregnant women

Differential Diagnosis

congential/childhood cataract

bilateral
- ***familial (hereditary)***
  - AD → most common
  - X-linked
  - AR → rare
- ***chromosome abnormality*** → trisomy 13, 18, 21
- ***craniofacial syndromes***
- ***renal syndromes***
  - Lowe syndrome → ± posterior lenticonus
  - Alport syndrome → anterior lenticonus, anterior subcapsular cataract
- ***musculoskeletal diseases*** → myotonic dystrophy → polychromatic lenticular deposits ("Christmas tree" cataract)

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- ***metabolic diseases***
  - diabetes mellitus → within the first few weeks of life, may be reversible with treatment
  - galactosemia → oil droplet cataract

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- ***intrauterine infections***
- ***ocular anomalies***
  - aniridia
  - anterior segment dysgenesis
  - retinitis pigmentosa
- ***iatrogenic***
  - corticosteroids
  - radiation
- ***idiopathic***

unilateral
- persistent fetal vasculature
- posterior lenticonus/ lentiglobus
- posterior segment tumor
- retinal detachment
- trauma (child abuse)
- radiation
- idiopathic

retinoblastoma
ROP
Norrie disease — XLR — boys only
incontinentia pigmenti — XLD — girls only
PFV
medulloepithelioma
unilateral

Ophthalmology Mind Maps/Arman Mashayekhi, MD