

## Nystagmus, Congenital optic nerve disorders, and Phakomatoses

### Learning Objectives:

1. Understand the definition of nystagmus and how to distinguish it from other types of eye movements such as [convergence-retraction nystagmus](#), [opsoclonus](#), saccadic intrusions, [square wave jerks](#), [superior oblique myokymia](#), [ocular flutter](#), [volitional nystagmus](#) (AKA voluntary flutter), and [oculomasticatory myorhythmia](#).
2. [Congenital Nystagmus](#): Be able define and distinguish between congenital motor nystagmus, congenital sensory nystagmus, periodic alternating nystagmus, and latent nystagmus (AKA fusion maldevelopment nystagmus).
  - a. Be able to list ocular pathologies associated with nystagmus in childhood.
3. Acquired Nystagmus: Be able to diagnose and localize the lesion in [spasmus nutans](#), [see-saw nystagmus](#), [upbeat nystagmus](#), [downbeat nystagmus](#), [monocular nystagmus](#) (Heimann-Bielschowsky phenomenon), [gaze-evoked nystagmus](#), [peripheral vestibular nystagmus](#), [Bruns nystagmus](#), [periodic alternating nystagmus](#), [torsional nystagmus](#), and [acquired pendular nystagmus](#) (including association with oculopalatal myoclonus or tremor).
4. List and explain various treatments of nystagmus including medications, prisms, and surgeries.
5. Understand the pathophysiology of congenital optic nerve pathologies, their systemic and ocular associations, and their expected visual potentials. Conditions to know include optic nerve hypoplasia, optic nerve coloboma, optic nerve pit/hole (topless/bottomless disc), myelinated nerve fiber layer, tilted disc, Bergmeister papilla, megalopapilla, peripapillary staphyloma, optic nerve aplasia, and melanocytoma.
6. Phakomatoses: Know ocular and systemic manifestations, inheritance pattern, various names, and long-term monitoring needed for neurofibromatosis type I and II, tuberous sclerosis, von Hippel Lindau, Sturge Weber syndrome, Wyburn Mason syndrome, Klippel-Trenaunay syndrome, ataxia telangiectasia, and incontinentia pigmenti.

### Pre-Work:

1. Watch
  - The associated hyperlinked videos with each of the diagnoses above.
  - Maybe this will be helpful for you! Baylor ophtho [resident's mnemonics for phakomatoses](#) video in "Sketchy Micro" format.
2. Read
  - [BSCS Pediatric Ophthalmology and Strabismus](#):
  - "Nystagmus and esotropia" under chapter "Esodeviations"
  - Chapter "Childhood Nystagmus"
  - Chapter "Optic Disc Abnormalities"



- “Phakomatoses” under chapter “Ocular manifestations of systemic diseases”
- BCSC Neuro-ophthalmology:
- Chapter “The patient with nystagmus or spontaneous eye movement disorders”-- Skip the optic atrophy section, as it was already covered in a prior lecture.

3. Prepare

- Come prepared to share some of your favorite mnemonics for types of nystagmus and phakomatoses.

**Lecture outline:**

1. Brief overview lecture on approach to nystagmus (10 minutes)
2. 3 oral boards-style nystagmus cases (20 minutes)
3. Congenital optic nerve disorders worksheet regarding identifying the condition based on photos/imaging and knowing associated findings. To be completed in pairs in "speed dating" style. I will walk around and answer questions. (45 minutes)
4. Phakomatoses Jeopardy game. Residents break into three groups. (45 minutes)
  - Residents share their favorite mnemonics