Pediatric Ophthalmology Special Syndromes (Downs, Goldenhar, etc.) and Craniosynostosis

Learning objectives:

1. Understand the differences between plagiocephaly, brachycephaly, scaphocephaly, trigonocephaly, acrocephaly in terms of head shape and sutures involved.
2. Be able to list the ocular complications of craniosynostosis syndromes.
3. Be able to list the ocular findings, major systemic findings, genetic mutations, and inheritance patterns of anophthalmia, branchial arch syndromes, Goldenhar syndrome, Treacher Collins syndrome, Pierre Robin sequence, fetal alcohol syndrome, infantile malignant osteopetrosis, Wilson's disease, Turner syndrome, Stickler syndrome, ocular albinism, Alagille syndrome.

Pre-work:

1. **Read BCSC Pediatric Ophthalmology sections below**
   - Orbital Disorders specifically Craniofacial malformations, Craniosynostosis, Nonsynostotic craniofacial conditions
   - Albinism subsection

2. **Skim through** the articles below and note ocular findings, major systemic findings, genetic mutations, and inheritance patterns of each disease:
   - Article on ocular findings in Down Syndrome https://www.mdpi.com/2227-9067/10/2/341
   - Article on ocular findings in Turner Syndrome (by Dr. Moshirfar) https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9692343/
   - Article on Alagille syndrome https://disorders.eyes.arizona.edu/disorders/alagille-syndrome
   - Article on Wilson's disease https://encyclopedia.pub/entry/26244
   - Attached article on Stickler syndrome

Lecture Outline:

1. Group Quiz on various findings in craniosynostosis syndromes ~15 minutes
2. Lecture on types of craniosynostoses ~ 15 minutes
3. Socratic teaching with one craniosynostosis case and various ocular findings in craniosynostosis ~30 minutes
   a. Discussion of evaluation and multidisciplinary management of patients with craniosynostosis
4. Pictionary on special syndromes listed above

a. Each resident picks a condition from a hat and has to draw out its associated findings and convey the inheritance pattern. Then each resident presents their condition to the group who has to identify the disease. The group discusses the diagnosis and management of the condition. ~45 minutes