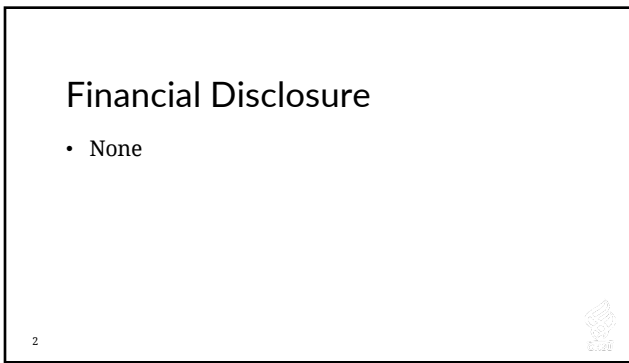
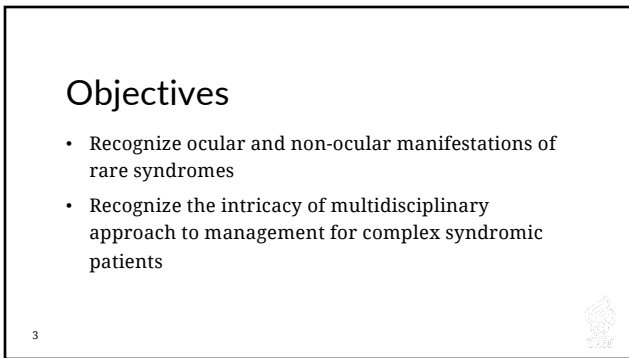


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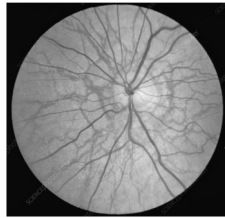
Ehlers Danlos Syndrome

- Group of genetic connective tissue disorders characterized by joint hypermobility, skin hyperextensibility, and tissue fragility.
- Ocular involvement: keratoconus, myopia, blue sclera, epicanthal fold, retinal detachment, glaucoma, angioid streak
- Non-ocular involvement: easy bruising, chronic joint pain, mitral valve prolapse, hernias, gastrointestinal disorders, dysautonomia
- Management: treat ocular manifestations, multi-disciplinary/specialty approach (pain specialist, GI, neurology, cardiology)

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Angioid streak



Keratoconus with Munson sign

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Charles Bonnet Syndrome

- Presence of vivid, complex visual hallucinations in patients with significant visual loss
- Management: reassurance, education, and address any underlying visual impairment if possible
- Characteristics:
 - Hallucination: vary widely in complexity and content, can involve multiple sensory modalities
 - Not associated with any psychiatric disorders
 - Usually maintain insight into the fact that their hallucinations are factitious
 - Triggers: significant periods of low visual stimulation (dimly lit environment or visual fatigue)
- 6• Management: reassurance, education, and address any underlying visual impairment if possible

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Steven Johnson Syndrome (SJS)

- Severe hypersensitivity reaction that affects the skin and mucous membranes, including the eyes.
- A medical emergency and can be life-threatening!
- Triggers: Many but common ones include: NSAIDs, anticonvulsants, antibiotics (sulfonamides like Bactrim), allopurinol (for gout) and infections such as herpes and mycoplasma
- Begins as flu-like symptoms such as fever, sore throat, and fatigue. Within a few days, a painful rash develops, which progresses to blistering and widespread skin detachment. In severe cases, involvement of the mucous membranes of the eyes, mouth, throat, and genitals can lead to significant complications, including impaired vision, difficulty eating and breathing, and increased susceptibility to infections.

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Steven Johnson Syndrome (SJS)

- Ocular manifestation:
 - Conjunctivitis
 - Corneal involvement: erosion, ulcer, and perforation
 - Symblepharon formation
- Management: frequent ocular lubrication, antibiotic ointment and/or drops, amniotic membrane placement. Involve our dermatology colleague.

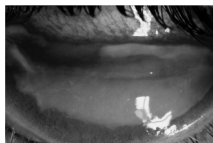
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SJS with a corneal epithelial defect and diffuse conjunctival injection



Membranous conjunctivitis is characterized by the formation of a layer of tissue consisting of fibroblasts, blood vessels, fibrin and inflammatory cells

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Sturge-Weber syndrome

- Also known as encephalotrigeminal angiomatosis, is a rare congenital disorder characterized by abnormal blood vessel development in the skin, brain, and eyes
- Typically due to somatic mutation of the GNAQ gene leading to vascular malformation of the capillaries

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Sturge-Weber syndrome

- Cutaneous: facial port-wine stain since birth caused by dilated blood vessels
- Neurologic : seizures, developmental delays, intellectual disability, hemiparesis, and cognitive impairment.
- Ocular: Glaucoma (elevated IOP from malformation of trabecular meshwork) and choroidal hemangioma

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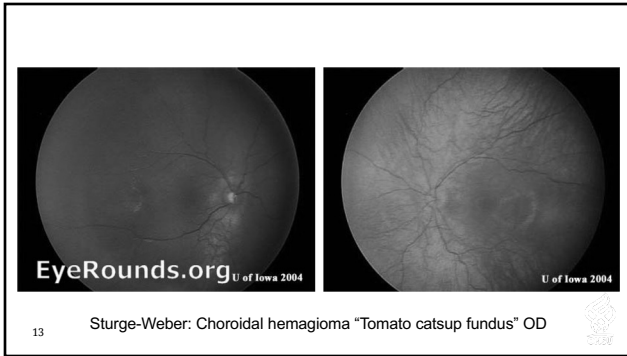
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Neurofibromatosis Type 1 (NF1)

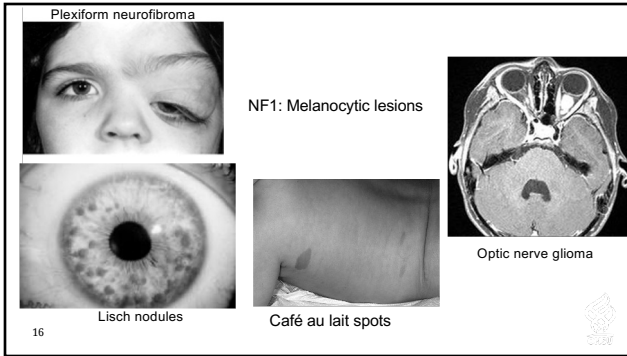
- Genetic disorder (NF1 gene encodes neurofibromin) causing development of neurofibromas

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Neurofibromatosis Type 1 (NF1)

- Ocular: lisch nodules (tiny, pigmented nodules on iris), optic nerve gliomas (sight threatening), sphenoid wing dysplasia (lead to proptosis)
- Others: café-au-lait spots, plexiform neurofibromas (S-shaped eyelid causing ptosis and deprivation amblyopia), brain tumors (gliomas), scoliosis, learning disabilities, cardiac defects
- Managements: monitor for development of optic nerve glioma, consider fixing plexiform neurofibromas, supportive care

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Waardenburg Syndrome

- Genetic disorder (PAX3, MITF, SOX10, EDNRB, EDN3) with facial features, hearing loss, and pigmentary abnormalities to the skin, hair, and eye

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Waardenburg Syndrome

- Face: high, broad forehead; a wide nasal bridge; a prominent nasal root; a broad nasal tip; and a prominent, wide-set eyes (hypertelorism)
- Ears: hearing loss
- Additional: developmental delay, intellectual disability, musculoskeletal anomalies, or gastrointestinal abnormalities
- Pigmentary changes
 - Hair: distinctive pattern of hair pigmentation, characterized by a white forelock (a patch of white hair near the forehead) or premature graying of the hair.
 - Eyes: heterochromia iridis, ptosis, strabismus, synophrys, telecanthus, dystopia canthorum
- Management: multidisciplinary approach with the involving organ systems

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