A Hairy Situation

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No relevant financial disclosures exist.

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Case 1

- 21yo woman presents with blurry vision OU.
- Myopia, strabismus
- T1DM, bronchiectasis, anemia, h/o kidney transplant
- BCVA 20/40 OU
- Alt XT, peripheral VF defects OU
- PSC OS















В



Differential Diagnosis

- Retinitis pigmentosa
- Alström Sydrome
- Senior-Loken Syndrome
- Soubert Syndrome
- Stargardt disease
- Bardet-Biedl Syndrome



- The patient's sister is a 22yo woman with a history of myopia, scoliosis, and polydactyly.
- BCVA 20/30 OU













Bardet-Biedl Syndrome

- Bardet-Biedl Syndrome, an AR ciliopathy, was first described in the 1920s.
- ~20 genes (BBS genes) that code for ciliary proteins are associated with BBS.
 BBS1 gene is the most frequently reported, associated with better VA.
- Ciliated cells are widely distributed in the body, many systemic manifestations.
 Obesity, polydactyly/syndactyly, hypogonadism, renal failure, mental and growth retardation, DM



Bardet-Biedl Syndrome

- Photoreceptors are the only cells composed of a modified central cilia that connects the inner and outer segments and are frequently affected as a part of syndromic ciliopathies.
- Abnormal ciliary trafficking of the rhodopsin molecule across the defective cilia and its accumulation in the IS results in eventual PR apoptosis.
- Bardet-Biedl syndrome retinopathy can be widely variable
 - Subtle RPE macular changes bull's eye maculopathy
 - mild peripheral pigmentary retinopathy peripheral atrophy and bone-spicules.
- Multimodal imaging, including OCTA, demonstrates bull's eye maculopathy and peripheral pigmentary retinopathy in the setting of grossly intact retinal and choroidal vasculature.



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