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Dear Doctor,

Your patient has a rare syndrome, Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS), an autosomal dominant condition caused by a disruption in the NR2F1 gene.

Features present at birth:

- Hypotonia
- Oromotor dysfunction
- Vision problems such as nystagmus and poor tracking

Features which develop over time:

- Development delay/intellectual disability
- Seizures
- Autism Spectrum disorder
- Vision impairment – 90%
    - Due to optic nerve abnormalities +/- brain based vision impairment
    - Optic nerve atrophy or pallor- 82%
    - Optic nerve hypoplasia - 82%
        - Not the classic found in septo-optic dysplasia, we found slightly smaller than normal sized optic nerve heads, no severe hypoplasia
    - Cortical visual impairment - 68%
- Other ocular features:
    - A/hypolacrima (decreased amount of emotional and reflex tearing) - 78%
    - Manifest latent nystagmus (infantile onset of manifest nystagmus, +/- latent component, or latent nystagmus alone - 52%
        - Often improved over time
    - Significant refractive errors
    - Amblyopia

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Suggested evaluation

Targeted History

- History of abnormal visual function?
  - Is it suggestive of CVI?
    - Abnormal vision with normal or mildly abnormal ocular findings
    - As infants, light gazing
    - Difficulty locating objects in a crowded field
    - Variable visual attention, especially in unfamiliar environment
    - Preference for high contrast objects
- History of nystagmus?
- History of abnormal volume of reflex tearing?

Targeted Exam

- Visual acuity (test which is appropriate for patient's age and understanding)
  - Teller acuity cards, Allen pictures, LEA, HOTV, Snellen
  - Color vision (indicator of optic nerve function)
- External exam
  - Manifest nystagmus? Latent component?
- Pupil reactivity
  - Poorly reactive, APD?
- Visual fields
  - Nonspecific abnormalities due to optic nerve abnormalities or CVI
- Intraocular Pressure
  - Glaucoma was not a finding in our pts
- Anterior segment
  - Dry eye findings?
    - Not a feature in our pts
- Optic nerve
  - Atrophy? mild or severe?
  - Mild hypoplasia?
    - Normal optic nerve diameter
      - 3 disc diameters fit in between the center of the optic nerve and the fovea
    - Mild hypoplasia
      - >3 disc diameters fit in between the center of the optic nerve and the fovea

Suggested In-office testing, if able:

- Fundus photography to document optic nerve health
  - Document if pallor exists and the distance between center of the optic nerve border and fovea (rule out mild hypoplasia)
- OCT
  - RNFL to document health of the optic nerve
  - GCL as surrogate for VFs
- Visual fields
  - Confrontation, if able, automated perimetry
    - We were unable to perform VF testing on any patient

Thank you for helping our BBSOAS patients and families,
Jane Edmond, MD

For more information, visit NR2F1 Foundation: nr2f1.org

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