



Mahidol University
Faculty of Medicine
Siriraj Hospital

Spot diagnosis 2





Septo-Optic Dysplasia (SOD)

- Developmental forebrain disorder at 4–6 weeks gestation
- Rare, heterogeneous, phenotypically variable

Classical triad (30% of patients)

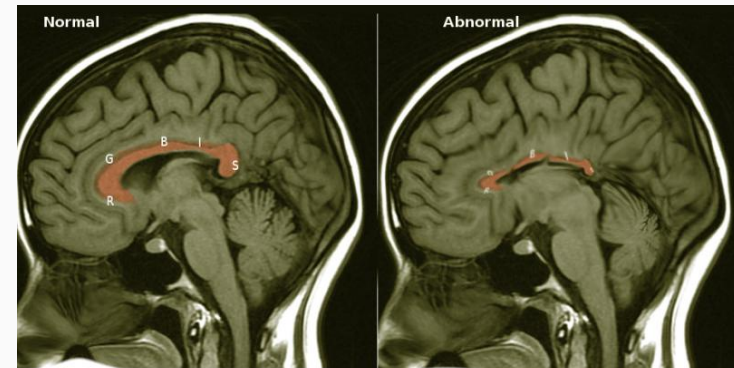
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1. Optic nerve hypoplasia: strabismus, nystagmus, visual impairment
2. Hypopituitarism: 62–80%
 - GH deficiency: most common 50-80%
 - TSH 15-40%, ACTH 10-30% , gonadotropin deficiencies 10-20%
 - Dynamic progression over time
3. Midline brain defects (septum pellucidum / corpus callosum)

Septo-Optic Dysplasia (SOD)

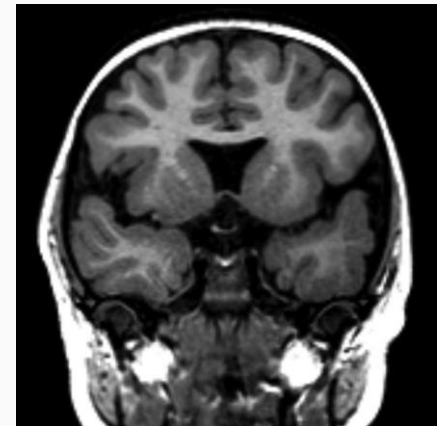
Other manifestation

- Developmental delay, autism, behavioral problem
- Seizure
- Cerebral palsy



MRI Brain Findings

- Absence of the septum pellucidum
- Optic nerve hypoplasia
- Abnormalities of the corpus callosum (such as hypoplasia or agenesis)
- Pituitary hypoplasia (small anterior pituitary gland)/ ectopic posterior pituitary



Septo-Optic Dysplasia (SOD)



Molecular genetic

1. *HESX1* – transcriptional repressor
 - Autosomal recessive & dominant forms
2. *SOX2* – severe bilateral eye defects + pituitary hypoplasia
 - De novo mutation

Genetic mutations identified in <1% overall

Evaluation

- Clinical assessment
- Ophthalmologic examination
- Pituitary hormone testing
- MRI brain to assess optic nerves, pituitary gland, and midline brain structures



Management

- Replace deficient hormones
- Developmental & visual support
- Regular growth assessment
- Surveillance for ACTH deficiency
- Pubertal evaluation in adolescence
- Monitor obesity & autism spectrum features
- Genetic counselling: most cases are sporadic