Co-management Guide

Pediatric Endocrinology	Septo-optic dysplasia

Guidelines Referenced	Eur J Hum Genet. 2010 Apr; 18(4): 393–397. OMIM Entry - # 182230 - SEPTOOPTIC DYSPLASIA *https://www.omim.org/entry/182230*
Background	Septo-optic dysplasia is a clinical diagnosis that is made when 2 or more of the following features are present: 1. Optic nerve hypoplasia 2. Midline brain defects (agenesis of septum pellucidum or corpus callosum) 3. Pituitary dysfunction (MRI may or may not be abnormal) a. GH deficiency b. Central hypothyroidism c. Secondary adrenal insufficiency d. Diabetes insipidus e. Abnormalities in pubertal development (can be precocious, delayed or absent) Three genes (HESX1, OTX2, SOX2) have been associated with SOD but these are rare causes of the disorder. In most cases, the cause is unknown. It is felt to likely be due to a combination of genetic and environmental causes. It is equally present in males and female infants. It is more common in infants born to younger mothers (<21 years of age). Estimated epidemiology: • Incidence is ~ 1:10,000 live births • 30% have complete manifestations • 50-60% have pituitary dysfunction with GH deficiency being most common • 25% have visual impairment • 60 % with developmental delay (more common in bilateral optic nerve hypoplasia versus unilateral optic nerve hypoplasia) Pituitary dysfunction may evolve over time, making on going follow up necessary. The earlier the diagnosis, the better the long term outcome as underlying hormonal abnormalities can be addressed such as hypoglycemia, risk of adrenal crisis, growth issues.
Initial history	These children can present at birth or later with growth failure or visual issues. Consideration in a newborn or infant: • strabismus or nystagmus • hypoglycemia • jaundice • microphallus • midline defects (ex, cleft palate)

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Initial Management	Baseline labs CMP IGF-1 or GH if child is < 1 year TSH and FT4 LH, FSH starting at 12 years ACTH and cortisol MRI with pituitary protocol Referral to Endocrinology and Ophthalmology	
When to Refer	When an infant or child is identified as having SOD or optic nerve hypoplasia	
Pre-Visit Work Up	Labs as above, MRI Accurate growth points on a growth curve	
Co-management Strategy (as appropriate)	**Specialist scope of care** • Assessing growth • Monitoring for signs and symptoms of pituitary dysfunction, by history and exam as well as monitoring labs • Treating pituitary dysfunction as indicated	**Primary care scope of care** • Assessing growth • Monitoring for signs and symptoms of pituitary dysfunction, by history and exam
Return to Primary Care Endpoint	Point at which care can be transferred back to primary care… Pediatric Endocrinology will typically follow the child for several years to monitor for pituitary dysfunction. If we have established there is no pituitary dysfunction and depending on child's age and course, we will typically discharge from our clinic. We then ask the primary care provider to monitor growth and for any future concerns for pituitary dysfunction.	