Fact Sheet :
Septo-Optic Dysplasia
(De Morsier's Syndrome)

apd
WESTERN CAPE ASSOCIATION FOR PERSONS WITH DISABILITIES
TM: WCAPD

What is Septo-Optic Dysplasia (SOD)?

Septo-Optic Dysplasia is a rare congenital malformation syndrome that features a combination of the underdevelopment of the optic nerve, pituitary gland dysfunction, and absence of the septum pellucidum (a midline part of the brain). Two or more of these features need to be present for a clinical diagnosis — only 30% of patients have all three. French-Swiss doctor Georges de Morsier first recognized the relation of an absent septum pellucidum with hypoplasia of the optic nerves and chiasm in 1956.

Causes of SOD

SOD results from an abnormality in the development of the embryonic forebrain at 4-6 weeks of pregnancy. There is no known single cause of SOD, but it is thought that both genetic (gene mutations) and environmental factors (alcohol or drug use during pregnancy) may be involved or increase the risk. Other factors may include viral infections, certain medications and a blood flow disruption

Typically, people do not have a family history of septo-optic dysplasia, however, there have been a few cases in which multiple family members have been diagnosed

There is no cure for this condition and the treatment is directed toward the specific symptoms in each individual. Children with possible SOD must be kept under careful hormonal follow-up, and, if present, hormone deficiencies should be treated with hormone replacement therapy.

Symptoms of SOD

Typically, the symptoms develop in 3 organs, the brain (which have abnormal formation of midline structures), the eyes (due to optic nerve hypoplasia), and pituitary (due to hypoplasia). About one third of patients present with all of the three main features. However, some symptoms may not appear until childhood or later.

Symptoms may include:

- Blindness in one or both eyes
- Pupil dilation in response to light
- Nystagmus (rapid, involuntary to-&-fro eye movement)
- Inward and outward deviation of the eyes (squint)
- Hypotonia (low muscle tone)
- Seizures

Other common features are:

- Short stature due to lack of growth hormone
- Abnormal thirst, hunger & body temperature
- Low blood sugar
- Sleep difficulties
- Obesity
- Jaundice
- Pituitary hormone insufficiencies
- Intellectual disability or learning disabilities
- Genital abnormalities & problems with sexual development or early puberty
- Developmental delay related to visual or neurological problems

Treatment of SOD

There is no cure for SOD. Pituitary insufficiency can be treated with hormones. Some children can wear spectacles and they should be encouraged to do so. As with most syndromes, please remember the range of symptoms vary from extreme to mild. Each child needs to be evaluated, individually, even with the same diagnosis.

> **"The spectrum of SOD is so diverse and varied, every diagnosis is completely different and no two cases are the same."**
>
> Emily Davison

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Sources :
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