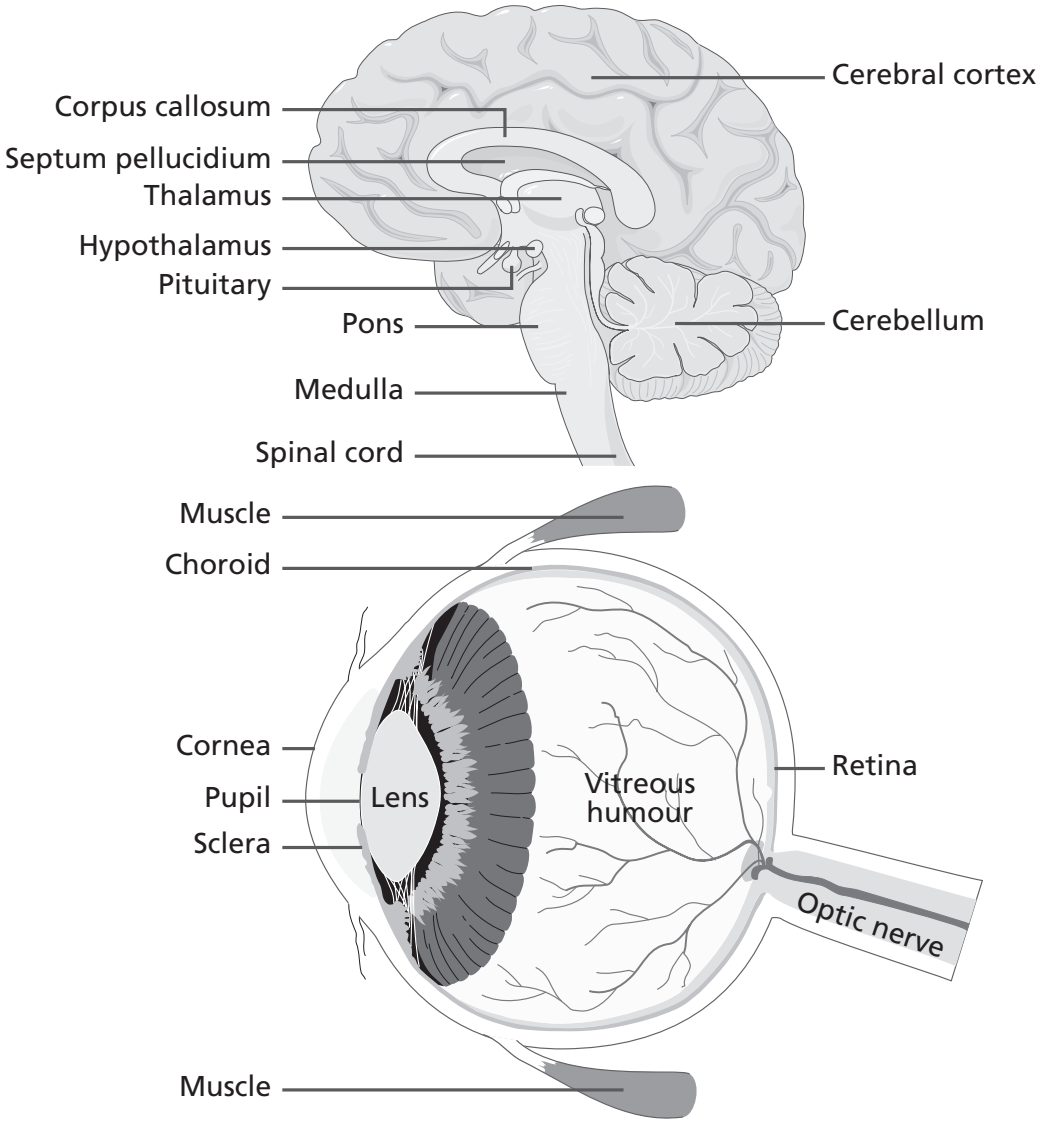
Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Septo-optic dysplasia

This information sheet from Great Ormond Street Hospital (GOSH) aims to explain the medical condition septo-optic dysplasia (SOD) and what to expect when your child comes to GOSH for assessment and treatment.



SOD is the name given to the condition where a child is diagnosed with two or more of the following problems: optic nerve hypoplasia, midline brain abnormalities and pituitary gland abnormalities. It is a rare condition affecting around 1 in every 10,000 births, with boys and girls affected equally. Septo-optic dysplasia is a congenital condition, that is, it is present at birth, although it may not be diagnosed until childhood, or rarely, adolescence. Septo-optic dysplasia was previously known as de Morsier syndrome.

What causes septo-optic dysplasia (SOD)?

There is some evidence to suggest that SOD is caused by a mutation (change) affecting one particular gene (a small part of one of your chromosomes). However, in the majority of cases, it is not thought to be an inherited disorder passed on from parent to child. It is highly unlikely to recur in further children within the family, and this suggests that the cause of the condition is complex. It has been shown to occur more frequently in younger mothers, and environmental factors may play a role. There may have been a particular problem within the pregnancy which is highly unlikely to recur in future pregnancies. More research is needed to confirm or rule out these theories.

What are the signs and symptoms of SOD?

SOD is diagnosed when two or more of the following problems are present: optic nerve hypoplasia, midline brain abnormalities and pituitary gland abnormalities. Only one-third of children diagnosed with septo-optic dysplasia will have all three features. More information about each of these follows:

Optic nerve hypoplasia

This means that the optic nerve has not developed properly during pregnancy and remains small. Normally, the optic nerve contains over a million separate fibres, all of which work together to transmit what the eye sees to the brain to be interpreted. In optic nerve hypoplasia, these fibres have not developed properly. This can affect either one or both eyes and the effect on a child's vision can vary greatly, although most children have a serious visual impairment.

Midline brain abnormalities

Absent or small areas of the middle part of the brain called the septum pellucidum and corpus callosum may be associated with developmental delay; that is, a child will not reach their 'milestones' of development at the expected age, for example, they may walk later or their speech may be late. They may also have movement and coordination difficulties caused by the midline brain abnormality.

Pituitary gland abnormalities

The pituitary gland is located deep in the brain and is the 'master gland' which produces many vital chemicals (called hormones). These hormones pass into the blood stream and control many other glands and parts of the body and make them function normally. Most children with SOD will have an abnormal pituitary gland which will be unable to produce sufficient levels of some of these hormones.

The most commonly affected hormone is called growth hormone (GH), low levels of which cause short stature. Other problems that occur when the pituitary gland is affected include hypoglycaemia (low blood sugar levels due to low GH and cortisol levels), hypernatraemia (high salt levels) and diabetes insidipus (excessive urine production and thirst). It may also affect the release of the stress hormone cortisol from the adrenal glands, low levels of which can be life threatening during severe illness and other stressful events like accidents. It may also be associated with low levels of thyroid hormone as the thyroid gland is controlled by the pituitary gland. In the majority of cases, puberty will be late although in a minority of patients it will occur early. In male patients, the penis may be small and the testes will not have come down into the scrotum.

The severity of symptoms with SOD varies enormously from child to child. Most children have low pituitary hormone levels (most commonly growth hormone deficiency) and visual impairment. Developmental delay is also common, especially if the optic nerve hypoplasia is found to affect both eyes. Many children have sleep disturbances, autistic behaviours and a tendency to weight gain.

How is SOD diagnosed?

Occasionally SOD is diagnosed during routine prenatal ultrasound scanning, but it is most commonly diagnosed during childhood. It is suspected early in childhood if the child has small male genitalia, poor growth, low blood sugar levels and is prone to infections.

If SOD is suspected, various tests and scans will be needed to confirm or rule out the diagnosis. Magnetic resonance imaging (MRI) scans of the brain are used to show the presence and severity of brain abnormalities. Blood tests are used to measure hormone levels. Vision testing is used to measure the severity of the optic nerve hypoplasia. Development assessments are needed to measure developmental delay.

How is SOD treated?

As SOD affects a variety of body systems, a multidisciplinary approach involving different specialists is required to ensure that the best treatments are given. The team may include endocrinologists (hormone specialists), ophthalmologists (eye specialists) and neurologists (brain specialists) as well as input from experts in visual impairment and developmental delay. The basis of treatment is to identify which hormones are absent or not being produced properly and replace these with man-made versions.

What happens next?

Support for visual impairment will enable your child to get the most out of school and social life as can additional help for development delay. Neurodevelopmental services are available providing assessment from physiotherapy, ophthalmologists, occupational therapists, speech therapy and neurologists or neurodevelopmental paediatricians.

Although septo-optic dysplasia is a genetic condition, it is extremely unusual for the affected gene to be identified in an individual at this stage. For this reason, genetic counselling is not usually helpful. However, Professor Dattani is leading a research study currently with our research partners in UCL Institute of Child Health (ICH) to try to establish the genetic basis to this disorder.

Further information and support

At GOSH, talk to our Eye Clinic Liaison Officer who can provide practical information, advice and support on all aspects on visual impairment. You can telephone her on 020 7405 9200 extension 0345, email her at paula.thomas@gosh.nhs.uk or visit her at your next clinic appointment.

Useful numbers

GOSH switchboard – 020 7405 9200

Clinical nurse specialists (Monday to Friday from 9am to 5pm)

Answerphone service for non-urgent queries – 020 7813 8214 – checked at 11am and 3pm. Fax – 020 7829 7958. Email – endocrine.cns@gosh.nhs.uk

Consultant secretaries – 020 7405 9200 – extensions 5813 or 1017 or 8296 to cancel or rearrange appointments and referrals

Out of hours – 020 7405 9200 and ask for the on-call registrar for endocrinology.

Compiled by the Endocrinology team in collaboration with the Child and Family Information Group
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