Medical Information Document On

Septo-Optic Dysplasia

What is the normal structure of the eye?

The eye is made of three parts:
- A light focusing bit at the front (cornea and lens).
- A light sensitive film at the back of the eye (retina).
- A large collection of communication wires to the brain (optic nerve).

A curved clear window called the cornea first focuses the light.

The light then passes through a hole called the pupil.

A circle of muscle called the iris surrounds the pupil. The iris is the coloured part of the eye.

The light is then focused onto the back of the eye by a lens.

Tiny light sensitive patches (photoreceptors) cover the back of the eye. These photoreceptors collect information about the visual world. The covering of photoreceptors at the back of the eye forms a thin film known as the retina.

Each photoreceptor sends its signals down very fine wires to the brain. The wires joining each eye to the brain are called the optic nerves.

The information then travels to many different special ‘vision’ parts of the brain. All parts of the brain and eye need to be present and working for us to see normally.

Figure 1: The Structure of the Eye
How We See: Beyond the Eyes

There are many different parts of the eye and the brain that need to work together in order for us to see well. The brain gets signals from the eye and sends them to the vision parts of the brain. In order for us to see it is the brain that does most of the work.

Seeing Beyond the Eyes in more detail

When we look, the picture we see travels from each eye along a nerve which connects the eye to the brain called the optic nerve. This is like a cable carrying an internet or TV signal. The signal is carried along each optic nerve and the two optic nerves come together in an area called the optic chiasm where some signals cross over. The signals from the left side of both eyes are usually sent to the right side of the brain, and the signals from the right side of both eyes are sent to the left side of the brain. The signals are sent to two main parts of the brain.

Most of the information is passed to the back of the brain, (the occipital lobes). The left occipital lobe sees the right side of the picture from the eye and the right occipital lobe sees the left side of the picture. To make things more complicated the bottom part of the occipital lobe sees the top part of the picture and the top part of the occipital lobe sees the bottom part.

Figure 2: The Main Pathway for Information Travelling from the Eye to the Brain

Some information is sent to a central part of the brain called the brain stem. This is an important pathway for children with very poor vision as it helps them to pick up movement without thinking. It can help them to automatically move out of the way of objects heading towards them, particularly from the side.

From the occipital lobe the information is then sorted out and sent to three main other parts in the brain. Some of the information goes to the area of the brain called the posterior parietal lobes (these are near the top back of the brain). This is sometimes called the dorsal pathway. Some information goes to the temporal lobes (at the sides of the brain) and this is the where the visual memory is stored. This is sometimes called the ventral pathway. Information is also sent to and from the
frontal lobes (which are at the front of the brain). This is the part that allows us to put information together. The posterior parietal lobes help us to see the whole picture and link up our movement with what we see, the temporal lobes help us to recognise things and the frontal lobes help us to choose what we look at and pay attention to.

Figure 3: The Main Areas of the Brain

The visual brain is a very complex network where information is flowing back and forth all the time. All parts of the brain and eye need to work well. Difficulties with vision can occur if there is damage in or to any of these areas, systems or processes.

**Septo-Optic Dysplasia**

What is Septo-Optic Dysplasia?
Septo-optic dysplasia is a rare condition (1 per 100,000 of the population). In this condition there are at least two of the following:

- Optic nerve hypoplasia
- Midline brain abnormalities
- Hypothalamic – pituitary deficiencies

**Optic nerve hypoplasia** – the optic nerves (the wires joining each eye to the brain), have not fully developed, so they are smaller than usual or don’t work as well.

**Midline brain abnormalities** - structures in the middle part of the brain have not developed fully. These connect the right and left side of the brain. When this happens children can have delays in their development and learning.
Hypothalamic – pituitary deficiencies – the hypothalamic pituitary system controls the production of hormones in the body and temperature control. When the system does not work well, it does not produce enough hormones (deficiency) or control temperature well.

Hormones which can be affected include; growth hormone (growth hormone is needed to help children grow), hydrocortisone (needed to respond to infection, and emotional and physical stress), thyroxine (helps to control energy levels and growth) and anti-diuretic hormone (controls the salt and water levels in the blood).

Septo-optic dysplasia is present at birth, and if the hormone system is affected babies can be very unwell. Sometimes it may not be diagnosed until children are older, even into their teens. The way Septo-optic dysplasia affects children varies a lot from one child to the next.

What is the Cause of Septo-Optic Dysplasia
The cause of Septo-optic dysplasia is not known. There is some evidence to suggest that it is caused by a mutation, or change, affecting one particular gene (a small part of one of your chromosomes). In most cases the gene has not passed down from parent to child, but is a new change that has happened in the child only. Because of this it is very unlikely that future children in the same family will be affected.

Several other causes have been suggested over the years but no good evidence has been found for these. It is likely that Septo-optic dysplasia is caused by many different factors which we don’t fully understand yet and further research is needed.

How does Septo-Optic Dysplasia affect the Child’s Vision?
Vision is only affected in Septo-optic dysplasia if there is optic nerve hypoplasia. The effect on a child’s vision can vary greatly, and some children have a visual impairment. They may have blurring of vision (reduced visual acuity), and difficulty seeing objects of similar colour to the background (reduced contrast sensitivity). The visual field, which is the area of space visible to the eyes when looking straight ahead, may be reduced. Some children may have fast ‘to and fro’ movements of the eyes called nystagmus, which also causes blurring of vision.

What can be done to help this condition?
A child with Septo-optic dysplasia may be supported by a number of specialists depending on how the condition affects them: these may include a paediatrician, paediatric neurologist, endocrinologist, ophthalmologist, teacher of pupils with visual impairment, habilitation specialist.
What Can be Done to Help Children with Visual Impairment?

We use our vision to get around, learn new things and to meet other people and make friends. Children who have visual impairment may need some extra help to do these things.

It is important to know and understand what your child sees so that you can give them the help they need. Young people themselves need to understand how and why they may see differently from others.

If glasses, contact lenses or Low Visual Aids (LVA) have been prescribed, it is important that these are used. These will help your child see more clearly and make sure the vision parts of the brain grow and develop correctly. Even if your child’s vision is very low, try to get them to wear their glasses if prescribed. This will help to give as clear a picture as possible to help get them interested in looking.

Even if a child has very poor vision many useful and practical things can be done to help. All children who have a visual impairment should have an assessment of their needs by a qualified visual impairment teacher and a qualified habilitation specialist. These are the professionals who can give advice and support your child in learning, education and in practical and play activities.

Where Do I find Out More Information on Septo – Optic Dysplasia?

- https://patient.info/septo-optic-dysplasia
- https://www.gosh.nhs.uk/conditions-and-treatments/conditions-we-treat/septo-optic-dysplasia
- https://contact.org.uk/medical-information/conditions/s/septo-optic-dysplasia/

Approved : Month / year

Review Due :

NOTE
This information does not provide a diagnosis and is not a substitute for a consultation with a health professional. It is intended to provide information to support children and families in understanding their condition, how best to manage it and know what treatments and support may be available.