Septo Optic Dysplasia
Also known as Optic Nerve Hypoplasia and/or DeMorsier's Syndrome

A child with Septo Optic Dysplasia (SOD) has underdeveloped nerves from the eye to the brain (optic nerve hypoplasia [ONH]). People with SOD may also have abnormalities of the brain and a poorly functioning pituitary gland.

A child with ONH has optic nerves that are small and poorly developed. Instead of having over 1 million connections (nerve fibers) from each eye to the brain, people with ONH have far fewer connections. The more connections between the eye and the brain, the better the vision. Some people with ONH have near-normal vision in one eye, others have decreased vision in both eyes, and others are severely affected and nearly blind. An eye doctor (Ophthalmologist) can diagnose ONH by looking inside the eye with an ophthalmoscope. The front surface of the optic nerve (optic disc) appears smaller than normal. Most people with ONH have a nystagmus (unusual eye movements). The eyes may seem to move around with no real pattern or purpose. This occurs because the eyes are not able to focus well enough to hold still.

Typically people with SOD have abnormalities of the brain. These abnormalities may include how the brain is formed (brain structure) and how the brain works (brain function). While both usually occur, sometimes a child has a problem only with the structure of the brain and at other times, a child has a problem only with the function of the brain. All problems with the brain can range from mild to very serious. When a child is diagnosed with SOD he or she will undergo a number of evaluations and brain function tests (neurologic tests). A brain specialist (Neurologist) can look at the brain structure through the use of CT (computerized tomography) or MRI (magnetic resonance imaging). After these tests are performed your doctor should be able to tell if there are major problems with the brain structure. However, doctors cannot always predict if a child will or will not have problems with brain function. When a child is less than 3-4 years of age, it is often difficult to predict future brain functions such as speech, intelligence and learning. It is sometimes difficult to assess the brain function and overall development of a child with poor vision. Visually impaired children must be taught and tested in ways that are different than children who are not visually impaired. Be sure that your child is tested and treated by professionals who have experience working with children with poor vision. Testing to determine how you child is developing and to screen for learning problems can be done by specialists such as: pediatricians, psychologists, occupational therapists (OT's), speech therapists (SP's), physical therapists (PT's) and teachers of the visually impaired (VI's). Testing can be done in several different settings including your own home, schools, hospitals, or other clinical settings.

The pituitary gland is found at the base of the brain and serves as the body's "master control gland" because it makes important chemicals (hormones) and directs the making of hormones in glands located in other parts of the body. These hormones are
required for growth, energy control (metabolism) and sexual development. Many people with SOD have problems with their pituitary gland ranging from very minor problems with almost no effect on the child, to problems making one or more very important hormones. When a child has problems making hormones in the pituitary gland it is called hypo-pituitarism. Your child will be tested to see if there are any problems making hormones. A doctor who specializes in gland problems (an Endocrinologist) will order tests to be done at the time of diagnosis. A child who initially does not have hormone problems may develop them at a later time. Therefore, it is very important for your child to be tested regularly as he or she grows up.

Growth hormone (GH) is made in the pituitary gland and controls the growth of the bones. Lack of growth hormone causes a child to grow at a slower than normal rate. Some children who lack growth hormone also have low blood sugar. Without treatment, your child will be shorter than expected when growth is finished. With proper treatment you can expect your child’s growth to be close to what it would have been without a growth hormone problem. About 6 in 10 children with ONH do not make enough growth hormone.

Cortisol is made by the two adrenal glands located on both sides of the middle of the back above the kidneys. A hormone from the pituitary gland (ACTH) signals the adrenal gland to make small amounts of cortisol at various times during the day. Cortisol is also made during times of physical stress (such as during surgery, when suffering from an illness or injury, or when the body is dehydrated). The extra cortisol is necessary to help the body continue to create energy, control the blood sugar and allow for proper heart and lung function. The normal pituitary gland signals the adrenal gland during these periods of stress to make extra cortisol. Your child should have blood tests to see if he or she is able to make enough cortisol. If the level is found to be low, some children will need special tests (called cortisol stimulation tests) to find out if they make some cortisol or if they are not able to make any cortisol. If your child is found not to make enough cortisol, he or she may need to take a cortisol replacement medication by mouth 1 or 2 times each day. Additional cortisol might also be necessary during times of physical stress. If your child has problems with cortisol, you should get additional information about caring for a child with this problem. Thyroid hormone is made by the thyroid gland in the neck. With a poorly functioning pituitary gland, the thyroid gland does not get the proper signal (TSH) to make thyroid hormone when it is needed and the person does not make enough thyroid hormone (hypothyroid). Thyroid hormone helps in normal growth and helps make energy for the body to function (metabolism). Lack of thyroid hormone can lead to poor growth, slowing of mental and muscle function, weight gain, feeling cold, irregular periods (menstrual cycle), hair loss, hoarse voice, brittle nails, and dry coarse skin. Lack of thyroid hormone can be shown with blood tests. The main thyroid hormone measured is called thyroxine (or T-4). Thyroid hormone levels should be checked when the diagnosis of SOD/ONH is made. If your child lacks thyroid hormone, he or she can easily be treated with thyroid pills taken by mouth every day. Your child will need to have T-4 measured regularly to see if the right amount of medicine is being given. With proper treatment, your child's thyroid problem will be corrected and he or she will not suffer ill effects.

The pituitary produces a hormone called Anti-diuretic Hormone (ADH) that is responsible for keeping water in the body by controlling the amount of urine (pee) that is created. Without enough ADH a person cannot control the amount of urine and he or she loses too much water from the body (becomes dehydrated). The person feels very thirsty, has a dry mouth and yet continues to make large amounts of urine. If the child
cannot get enough fluid to replace what is being lost in the urine, this can lead to dehydration and a very high level of sodium (a mineral) in the body. This dehydration can cause serious problems. If this problem is suspected, your child may need to have blood and urine tests. Sometimes a special test called a water deprivation test needs to be done. This involves a test lasting up to 8 hours which is done in the hospital or specially equipped office. Your child will not be able to have anything to drink for the entire test. Blood and urine tests are done to see if the proper amount of ADH is being created. If your child does not make enough ADH (called Diabetes Insipidus [DI] or water diabetes), treatment is available. The most common treatment is a medicine called DDAVP. It can be given in the nose, by mouth, or by injection.

For further information concerning Septo Optic Dysplasia, please contact your endocrinologist.

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