

OPTIC NERVE HYPOPLASIA

A GUIDE FOR PARENTS

This pamphlet contains general information about ONH. It is not meant to be substitute for any information given to you by your health care providers. You are encouraged to have regular health care visits and to talk with your health care provider about your child's condition and his or her needs.

Words printed in blue are explained in the Glossary

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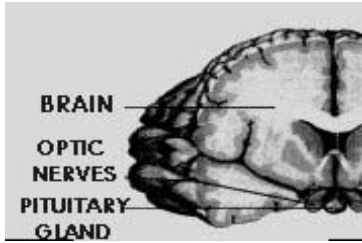
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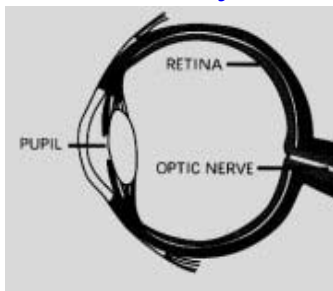
About the Authors and Sponsors

Feel free to print all of the booklet out to share with family, friends, teachers, doctors ect.



A person with **Optic Nerve Hypoplasia (ONH)** has small eye nerves (optic nerves) from the eye to the brain. Some people with ONH also have an abnormal brain and a poorly functioning **pituitary gland**. This brochure explains the problems that can occur in children with ONH. Your child may have none, any or all of these problems in a mild or more serious form. Depending on the person's problem sometimes the disease is called Optic nerve Hypoplasia (ONH), **septo-optic dysplasia**, or **De Morsier's syndrome**

How The Eye Works



The eyes receive light from the outside world and send these pictures to the brain along the optic nerves.

The Vision Of People With Optic Nerve Hypoplasia

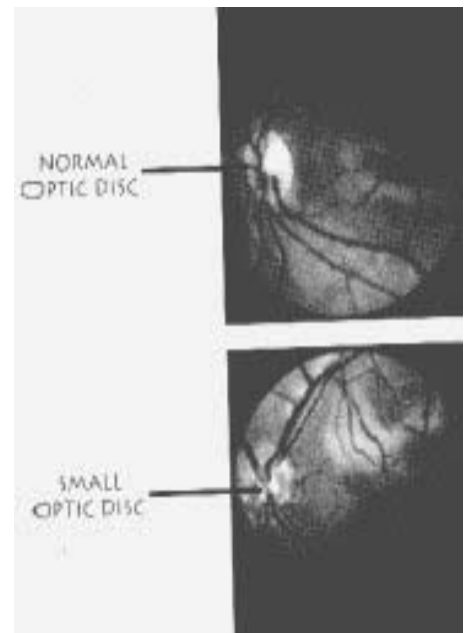
A person with ONH has optic nerves which are small and poorly developed. Instead of having over 1 million connections (**nerve fiber**) from each eye to the brain, people with ONH have far fewer connections. The more connections between the eye and brain the better vision. Some people with ONH have near normal vision in one eye, others have decreased vision in both eyes, and others are severely affected and nearly blind.

The Eye Examination

An eye doctor (**ophthalmologist**) can diagnose optic nerve hypoplasia by looking inside the eye with an ophthalmoscope. The front of the optic nerve (**optic disc**) appears smaller than normal.

Your child will undergo a number of eye examinations to determine his or her vision. The younger the child the more difficult it is to tell the amount of vision present. Depending on your child's ability to cooperate with the examination, an experienced ophthalmologist can usually tell your child's ability to see. However this usually can not be done with children under the age of 3-4 years. It takes a number of years to be able to tell what a child's vision will be like. The vision does not usually worsen over time from ONH. It may, however, improve over time.

Most children with ONH have unusual eye movements (**nystagmus**). They may have eyes that 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; this pattern is often seen with children who have poor vision.



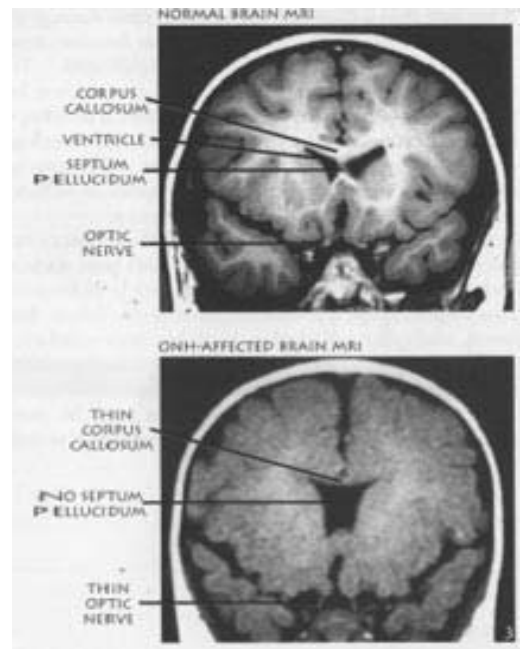
The Brain: Formation and Function

Many people with ONH 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 the problems with the brain can range from minor to very serious.

The normal brain is made up of two equal parts ([hemispheres](#)), which are connected by nerve fibers ([corpus callosum](#)) and are separated by fluid-filled spaces ([ventricles](#)). The ventricles have dividers between them ([septum pellucidum](#)).

Some people with ONH have a problem with the formation of the septum pellucidum; this is called septo-optic dysplasia or DeMorsier's syndrome. Others have an abnormal corpus callosum. Some people also have other parts of the brain which are abnormally formed. These can be seen with the [CT \(computerized tomography\)](#) or [MRI \(magnetic resonance imaging\)](#). Problems with the formation of the brain can be quite varied often involving the hemispheres and the ventricles. These can lead to small brains, excessively large ventricles, and fluid filled sacs ([cysts](#)) in the ventricles.

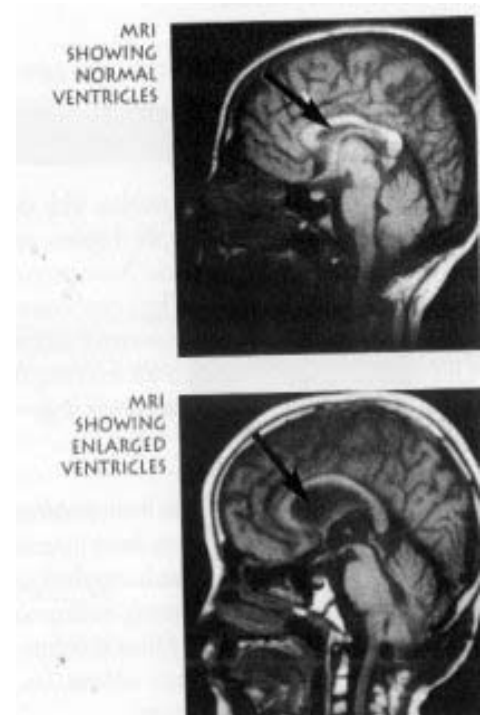
In some people with ONH, these brain problems are minor and do not cause abnormal brain function. In others, brain function is affected, ranging from mild to severe. The major areas which can be involved are the use of large muscle ([gross motor](#)) and small muscle ([fine motor](#)), intelligence and learning, speech, and interacting with people. Many of these problems can be helped with therapy. Some children with ONH also have [seizures](#) (fits, [convulsions](#)) and may need to take medicines to control the seizures.



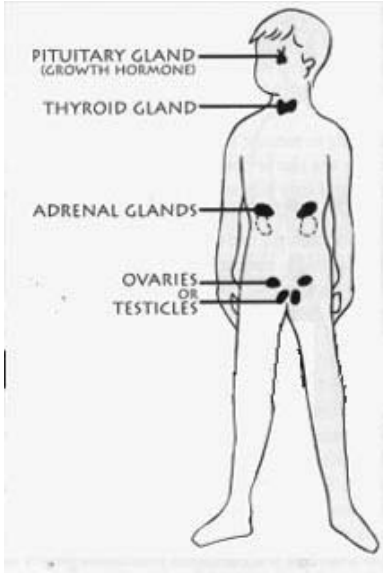
Brain Evaluation and Tests

When your child is diagnosed, and at regular times during your child's life, he or she will undergo a number of evaluations and brain function tests ([neurologic tests](#)). These are usually done by a brain specialist ([neurologist](#)). To show the structure of the brain, radiologic tests such as CT or MRI can be done by an imaging specialist ([radiologist](#)). Testing to determine how your child is developing and to screen for learning problems can be done by specialists in child development (pediatricians, [psychologists](#), occupational/ physical therapists, and or teachers). Testing can be done in several settings including schools, hospitals, or other clinical settings.

At the time your child is diagnosed with ONH your doctors should be able to tell you if there are major problems with your child's brain structure. 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. All of these brain problems may worsen if your child has poor vision ([visually impaired](#)). 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 different ways. Be sure that your child is tested and treated by professionals who have experience working with children with poor vision.



The Pituitary Gland



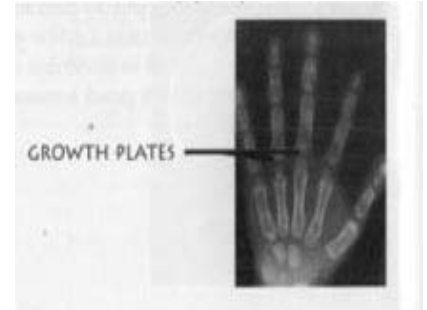
The **pituitary gland** is found at the base of the brain and serves as the body's "master control gland" because it makes important chemicals (**hormones**) and directs the making of hormones in glands located in other parts of the body. These hormones are required for growth, energy control (**metabolism**), and sexual development.

Many children with ONH 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 person has problems making hormones in the pituitary gland it is called **hypopituitarism**.

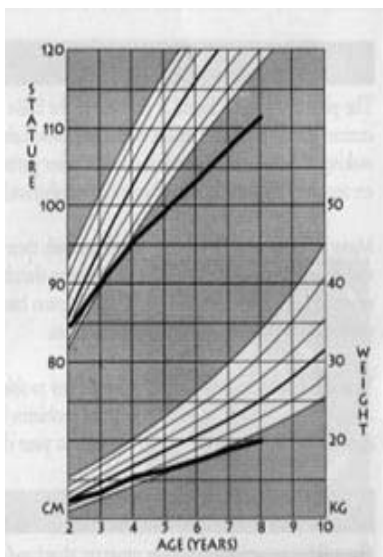
Your child will be tested to see if there are any problems making hormones. This is done by a doctor who specializes in gland problems (endocrinologist). Tests are done at the time of diagnosis and regularly as your child grows up. A child who initially does not have hormone problems may develop them at a later time. Therefore, it is very important for your child to be tested regularly as he or she grows up.

Growth Hormone

The child's small size and slow growth can be seen by plotting the child's height on a growth chart and comparing the child's growth over time with the growth of other children of the same age. Children without enough growth hormone have bones that are less developed than expected for the child's age. Your doctor may order a bone age X-ray of the hand and wrist (or if younger than 2 years an X-ray of half of the body) as part of the evaluation of your child's growth.



Blood tests to measure hormones important for growth and controlled by growth hormone can also be done. These are called **IGF-1 (somatomedin C)** and **IGFBPs**. These blood tests help tell if one has the ability to make enough **growth hormone**.



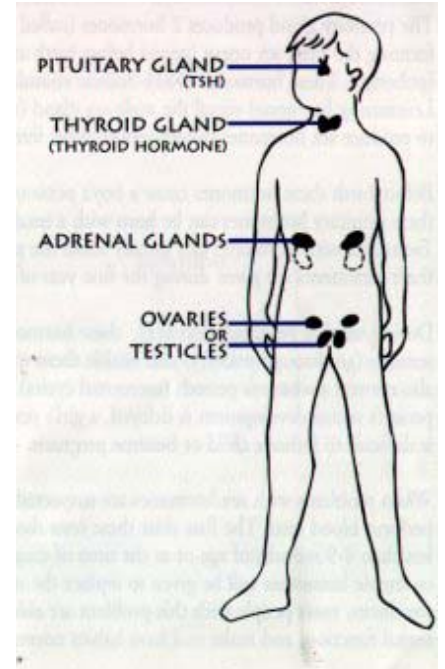
The endocrinologist can do a special test call a **growth hormone stimulation test** to see if the child is making enough growth hormone. This is done by giving the child special medicines that make the pituitary gland release growth hormone. Small amounts of blood will be taken frequently over a period of about 2-3 hours to measure the amount of growth hormone made by the pituitary gland.

About 6 in 10 children with ONH do not make enough growth hormone. If your child is found to not make enough growth hormone to grow normally (called growth hormone deficiency), he or she may need to get growth hormone shots 6 nights a week at least until the child has finished growing and probably throughout his or her adult life. In adulthood, growth hormone treatment is important to maintain strong bones and normal metabolism. These shots are easy to give and almost all parents are able to give them to their children after receiving instruction. With proper treatment you can expect your child's growth to be close to what it would have been without a growth hormone problem

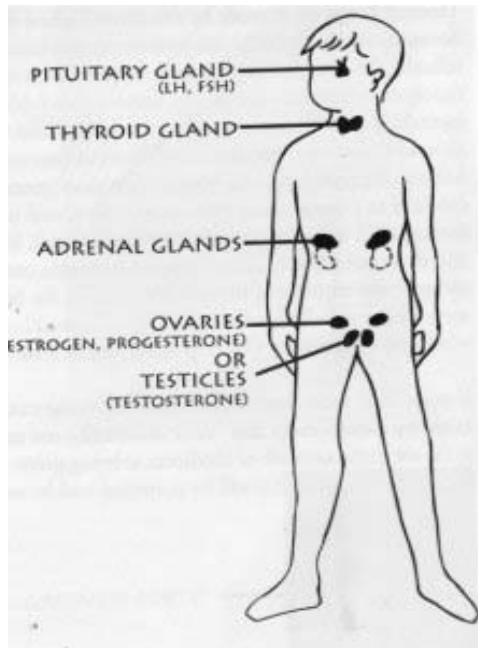
Thyroid Gland and Thyroid Hormone

Thyroid hormone is made by the **thyroid gland** in the neck. The thyroid gland normally makes the right amount of thyroid hormone because the pituitary gland tells the thyroid gland how much thyroid hormone it needs to make. 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 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 which measure the amount of thyroid hormone in the body. The main thyroid hormone measured is called **thyroxine** (or **T-4**). Thyroid hormone levels should be checked when the diagnosis of ONH is made and at least every year after the diagnosis.

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.



Sex Hormones



The pituitary gland produces 2 hormones (called **gonadotropins**) involved with forming the male sex organ (penis) before birth and sexual development (**puberty**). These hormones (**FSH-follicle stimulating hormone**; and **LH-Leutinizing hormone**) signal the male sex gland (**testicle**) or female sex gland (**ovary**) to produce the sex hormones (male-**testosterone**; female-**estrogen** and **progesterone**).

Before birth these hormones cause a boy's penis to form normally. Boys missing these pituitary hormones can be born with a smaller than normal sized penis. Treatment with hormones can usually make the penis grow to normal size. Often these treatments are given during the first year of life.

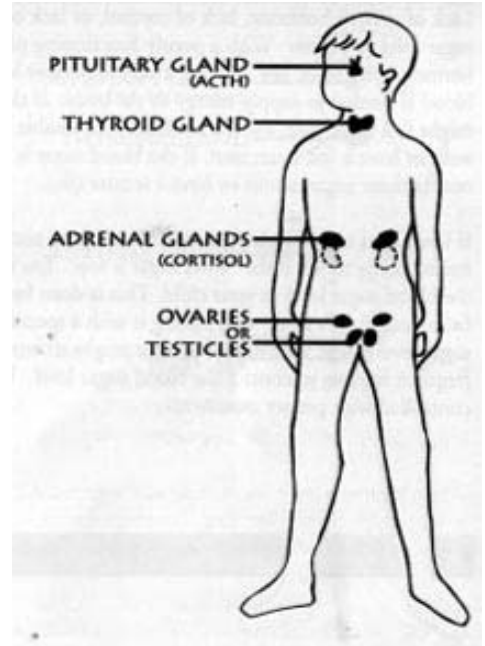
During teenage years (**adolescence**), these hormones cause a boy or girl to mature sexually (go through **puberty**) and enable them to make babies (**reproduce**). They also control a woman's periods (**menstrual cycles**). Without these hormones, a person's sexual development is delayed, a girl's periods are absent or irregular, and it is difficult to father a child or become pregnant.

When problems with sex hormones are suspected, the endocrinologist will perform blood tests. The first time these tests should be done is when your child is less than 8-9 months of age or at the time of diagnosis. If a problem is found, male or female hormones will be given to replace the missing hormones. With proper treatment most people with this problem are able to develop sexually, have normal sexual function, and make and have babies normally.

Adrenal Glands and Cortisol

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 to not make enough cortisol, he or she may need to take cortisol 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.



Low Blood Sugar (Hypoglycemia)

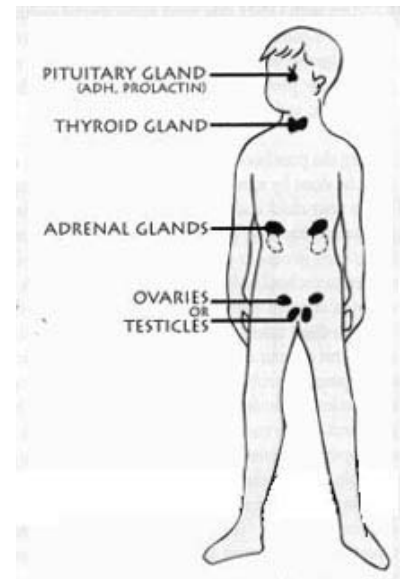
Lack of growth hormone, lack of cortisol, or lack of both can cause a body's blood sugar level to be low. With a poorly functioning pituitary gland, these two hormones might be low, causing a person to have low blood sugar. The sugar in the blood is needed to supply energy to the body. If sugar level is low a person might feel tired, pale, sweaty, confused, be unable to think, to use their muscles well, or have a very fast heart beat. If the blood sugar is very low the person might pass out, become unconscious or have a seizure (fit).

If low blood sugar levels are suspected, a blood test can be done when the person is feeling badly to tell if the blood sugar is low. You might be taught how to measure the blood sugar level in your child. This is done by getting a small amount of blood from your child's finger and testing it with a special blood sugar meter. If the blood sugar level is low, the endocrinologist might recommend hormone treatments and/or frequent feedings to control the blood sugar level. Low blood sugar can usually be controlled with proper treatment.

Anti-Diuretic (ADH)

The pituitary produces a hormone called **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 a 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.



Prolactin

Prolactin is another hormone made in the pituitary gland. Children with ONH often have abnormal levels of prolactin in their blood. This leads to no known health problems, but is often measured by the endocrinologist to test pituitary function.

Services

Children with ONH may need many special evaluations, tests, and services. As the parent of child with ONH, it is important to have a health care team that is knowledgeable about your child's condition. The medical team should include a primary care provider, an [ophthamologist](#), an [endocrinologist](#), a [psychologist](#), a [neurologist](#), and perhaps a [social worker](#).

During preschool years, your child will need a [developmental assessment](#). This must be done by someone who is skilled in working with children with poor vision. When your child is about to start school, testing for the most appropriate school placement should be performed. All children are entitled to receive education which meets their needs. To start the process for school placement, contact your local elementary school or local center for children with developmental problems (for example, regional center) both in person and in writing. You will need to request an [Individualized Education Plan \(IEP\)](#) for your child. This process includes an assessment of your child by either the school district or a center for children with developmental problems a specific educational program for your child. You will need to let the professionals know that your child has ONH and any other problems. Keep track of all letters and phone calls made to the district or developmental center. If your child is too young to start an in-school program (for example, if he or she is an infant), services may be provided on a weekly or monthly basis in your home via the local developmental disabilities center or visually impaired program. To make your child's educational experience the best it can be, repeat assessments must be done to insure that your child is learning and developing as expected.



MATTHEW, AGE 5 YEARS

Many children require placement in programs for children with poor sight ([visually handicapped](#)). As your child gets older, he or she may benefit from visual aids such as an enlarger (to increase the size of print) and/or a special computer for the visually impaired (e.g., a scanner which would take written words and turn them into spoken words).

If your child is not progressing well, additional services might be helpful. Other services may include language therapy (provided by a speech/ language pathologist), and vision therapy. If you have concerns about your child's development in any of these areas, contact the school or developmental disabilities center and request an assessment in these specific areas.

You should become aware of state and local programs that can help your child, such as [early intervention programs](#) (funded by state and federal funds), [supplemental insurance programs](#), and special resources. In addition, find out about the national agencies that have interest in ONH; these include **Focus Families** (For Our Children's Unique Sight), (866)-FOCUS-50; **The Magic Foundation** (Major Aspects of Growth In Children), (800)-3-MAGIC-3; **the Human Growth Foundation**, (800)-451-6434; **the Foundation for the Junior Blind**, (213)295-4555; **the Braille Institute**, (800) BRAILLE (272-4553); and **the Lions Clubs**.

Glossary

[ACTH](#)-A pituitary hormone that tells the adrenal gland to make cortisol.

[ADH \(Anti-Diuretic Hormone\)](#)- A pituitary hormone which keeps water in the body by controlling the amount of urine (pee) that is made.

[Adolescence](#)- The teenage years, when the child is becoming an adult physically and mentally.

[Adrenal Glands](#)- Two glands that sit on top of the kidneys and make several hormones, including cortisol.

[Axon](#)- Another name for nerve fibers.

[Blood Sugar Meter](#)- A machine that is used to test blood sugar levels.

[Bone Age X-Ray](#)- Bone age refers to the stage of development or maturity of the bones. In most children, bone age will be about the same as actual (chronological) age, but in some children it may be advanced (ahead) or delayed (behind). Bone age is measured by taking an X-ray, usually of the hand and wrist (or half of the body if a child is less than 2 years old) and then the bones are compared to standards for boys and girls of different ages.

[Computerized Tomography \(CT Scan\)](#)- A type of X-ray picture. The patient lies on a table that moves. There are clicking sounds while the pictures are being taken.

[Convulsion](#)- A strong spasm or series of twitches of the face, body, arms or legs.

[Corpus Callosum](#)- The connection between the two halves of the brain.

[Cortisol](#)- A hormone made by the adrenal glands after activation by the pituitary hormone, ACTH; Cortisol is needed to survive physical stress; maintain normal fluid, electrolyte, and blood sugar levels; and to maintain an energy supply.

[Cortisol Stimulating Test](#)- A test that measures how much cortisol is made by the adrenal glands in response to ACTH.

[Cyst](#)- An abnormal sac containing liquid.

[DDAVP](#)- A medicine that is used when a child does not make enough Anti-Diuretic Hormone; it is given in the nose, by mouth, and by injection.

[Dehydration](#)- Not enough water in the body.

[De Morsier's Syndrome](#)- A syndrome in children with Optic Nerve Hypoplasia who have problems with the formation of the septum pellucidum.

[Developmental Assessment](#)- A test of how a child is developing in the areas of thinking speaking , deciding things, using big and small muscles, learning, and performing in school.

[Early Intervention Program](#)- A program geared for children under the age of 5 to provide help in the development of language, motor skills and socialization.

[Endocrinologist \(Pediatric\)](#)- A doctor who specializes in treating children's hormones and growth problems.

[Estrogen](#)- Hormones that are made by ovaries in women and cause female sexual characteristics and control the menstrual (period) cycle and the ability to have a baby.

[Fine Motor](#)- Hand movements, used in writing and drawing.

[FSH \(Follicle Stimulating Hormone\)](#)- A hormone made by the pituitary gland. In women, it causes the development of eggs and the release of estrogen. In men, it helps produce sperm.

[Gonadotropins](#)- A hormone which causes gonadal growth (testes in males and ovaries in females).

[Gross Motor](#)- Movement of arms and legs (e.g., crawling, walking, and running).

[Growth Hormone](#)- A pituitary hormone which causes physical growth.

[Growth Hormone Stimulation Test](#)- This test measures the ability of the pituitary gland to make growth hormone.

[Hemispheres](#)- The two halves of the brain.

[Hormones](#)- A chemical substance made in a gland and carried in the blood to cause an organ to work; hormones act as "messengers" to control growth, reproduction, and body metabolism.

[Hypoglycemia](#)-An abnormally small amount of glucose (sugar) in the blood.

[Hypopituitarism](#)- A condition in which the pituitary gland does not make needed hormones.

[Hypothyroid](#)- Not enough thyroid hormone.

[IGF-1 \(Somatomedin C\) and IGBPs](#)- Growth factors that can measure in the blood to screen for growth hormone deficiency.

[LH \(Luteinizing Hormone\)](#)- A hormone that causes the release of sex hormones.

[Magnetic Resonance Imaging \(MRI\)](#)- Special pictures of the inside of the body. Like the CT Scan, the patient needs to lie still on a table. The table moves so that the patient's entire body is in the tunnel of the magnet. The machine makes grinding noises while the pictures are taken.

[Menstrual Cycle](#)- Female reproductive cycle starting at puberty; involve the build-up of the lining of the uterus (womb) for conception, followed by shedding of this lining (menstrual period) if conception does not occur.

[Metabolism](#)- The process by which substances needed for life are made and are broken down.

[Nerve Fibers](#)- Pathways for impulses between nerve centers and body parts.

[Neurologic Tests](#)- Tests used to reveal problems in the workings of the nervous system.

[Neurologist](#)- A specialist in the diagnosis and treatment of the disorders of the brain and spinal cord.

[Nystagmus](#)- To-and-fro shaking movements of the eyes.

[Ophthalmologist](#)- A medical doctor specializing in the eye and it's diseases.

[Optic Disc](#)- The front surface of the optic nerve that can be seen inside the eye with special instruments.

[Optic Nerve Hypoplasia](#)- A disease with poor formation of the optic nerves prior to birth. It can cause poor vision.

[Ovaries](#)- Females reproductive organs located in the lower abdomen on either side of the uterus (womb); they have eggs and make hormones that control sexual development and reproduction.

[Pituitary Gland](#)- The "master" gland that sits under the brain and makes hormones, most of which activate other glands to make hormones.

[Progesterone](#)- A female hormone that makes the uterus (womb) ready to accept a fertilized egg (developing baby).

[Prolactin](#)- A hormone made by the pituitary gland; children with Optic Nerve Hypoplasia often have abnormal levels of this hormone in their blood.

[Psychologist](#)- An expert in assessing overall development (including thinking, speaking, memory, reasoning skills, learning, and school achievement). A psychologist can also help to understand and cope with different feelings related to illnesses, hospitalization, and treatment.

[Puberty](#)- The stage of growth when the reproductive organs start to work, the person matures and develops adult sexual characteristics.

[Radiologist](#)- A physician trained in the use of x-rays and other ways to view into the body (imaging).

[Reproduce](#)- To make children.

[Septo-Optic Dysplasia](#)- A syndrome in children with Optic Nerve Hypoplasia who have a problem with the formation of the septum pellucidum.

[Septum Pellucidum](#)- A thin wall of brain tissue which divides the ventricles.

[Sexual Development](#)- When a child begins to form the characteristics of an adult male or female.

[Social Worker](#)- A professional who works with children and their families to help them understand and adjust to hospitalizations and long-term illness. The social worker provides counseling, help in getting financial assistance when needed, and provides information about community resources.

[Supplemental/ Social Security Insurance Program\(SSSI\)](#)- Federal program which gives money to some families to help care for children with handicaps.

ONH Families



"I found out I had Optic Nerve Hypoplasia after a visit to the ophthalmologist at age about six years old. When I first started getting shots I was very scared, but my mom helped by giving me my shots at night."
Brandon, age 13



"...having a child with Optic Nerve Hypoplasia can be very isolation. Trying to explain the condition to the many doctors and psychologists you see, trying to explain it to teachers, and at IEP meetings. It can be a lonely process... With all of that, my husband and I consider ourselves very lucky to have Zachary in our lives. He is such a happy child. He has a unique sensitivity for everything and everyone around him. He has changed our lives in ways I never thought possible. I can honestly say I would not change anything about my son. He truly is a joy and a gift."
Kimberly and Adam,
parents of Zachary, age 6



"When I first learned about my son's diagnosis I felt very overwhelmed. After my initial reaction, I quickly realized the best way for me to help him was to learn as much as I could about his medical condition. I had to adopt a new mind-set to **take one day at a time** and at the same time try to project what his future needs would entail. When my son brought home a letter home from school informing me that he was selected as Student of the Month and being recognized for his outstanding character traits of integrity and perseverance, it was then I knew it was going to be OK.

"Through every challenging moment I remind myself that beneath the diagnosis is a child full of hope and promise."
Alison, mother of Blake, age 9

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THANK YOU

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WILL GAIN KNOWLEDGE AND FIND SUPPORT.