What is Septo-Optic Dysplasia?

Septo-Optic Dysplasia (SOD) is a rare condition in which there is a malformation in the central part of the brain. This malformation can affect the structures of the eyes; parts of the brain which may include the hypothalamus and pituitary gland.

What causes septo-optic dysplasia?
The cause of SOD is not known and it is thought to occur randomly. It has been seen within families, however, it is not thought to be hereditary. SOD occurs during the first trimester of pregnancy when the brain of the fetus is developing.

What are the possible effects of septo-optic dysplasia?
Every child with SOD has special differences depending on which area of the brain has been affected. Most individuals have some disturbance to the optic nerve, which results in visual changes. These can range from blindness to slight visual problems in one eye. Other structures within the central portion (midline) of the brain may be underdeveloped. This can cause developmental, educational and muscular delays.

The hypothalamus and pituitary gland located within the brain are also usually affected. The hypothalamus helps the body regulate temperature and coordinates the function of the pituitary. The pituitary gland produces several hormones and releases them directly into the bloodstream. As a result, these individuals can have the inability to regulate body temperature, hypersensitivity to sound, and deficiencies of certain hormones. Children with hormone deficiencies may present with unquenchable thirst and heavy urination, low blood sugar, poor growth, difficulty sleeping, small genitalia, or abnormal puberty.

How is septo-optic dysplasia diagnosed?
In infants, the most common symptom first seen is involuntary rapid eye movements and partial blindness. Abnormally small or crossed eyes may be present. If there is pituitary deficiency the infant may present with low blood sugar. In childhood, the child may seem unusually small or frail and have difficulties in school. Several specialized tests may be used to confirm SOD.

- Opthalmoscopy - The doctor will look inside the eye to examine the retina and optic disc.
- Visual Field - This machine flashes a light into different areas of the eye to determine how the range of vision has been affected.
- Electroencephalograph (EEG) - Records the brains electrical impulses in response to light patterns and assesses the way the brain functions.
- Magnetic Resonance Imaging (MRI) - Magnetic fields and radio waves are used to create cross-section views of the brain to determine affected structure.

- Computerized Tomography (CT) – A computer and X-rays are used to create a film showing cross-sections of the brains tissue and function.
- Bone Age –An X-ray of the hand to determine the skeletal age of the child as compared to chronologic or "birthday" age.
- Blood tests– May be done to check for deficient levels of hormones in the blood. Listed are some of the levels that are checked to screen for hypopituitarism.
    - Growth hormone (GH)
    - Luteinizing hormone (LH)
    - Glucose levels
    - Antidiuretic hormone (ADH)
    - Cortisol
    - Follicle-stimulating hormone (FSH)
    - Thyroid-stimulating hormone (TSH)

What is the treatment of septo-optic dysplasia?

The treatment of SOD is directed towards the specific symptoms of each individual. Corrective lenses, surgery and other supportive measures are used to help correct vision. Hormones are replaced as needed to correct associated secondary conditions such as growth hormone deficiency, adrenal insufficiency, hypothyroidism, diabetes insipidus, hypoglycemia, and/or delayed puberty. If the child has seizures medication may be needed. Vision therapy, mobility training and physical therapy may be beneficial. Early intervention and child specific therapies are important to ensure that each child reaches his or her potential.

When should my child see the doctor?

Treatment will require care from a team of specialists. Most children with SOD will see a primary care physician, an ophthalmologist (eye doctor), a neurologist (examines the nerves and brain), an endocrinologist (examines the glands and hormones), and multiple other health professionals to coordinate therapies. If taking hormone replacement, doctor visits will be every three to four months. Remember, to follow up with your child's primary care doctor for routine health care. If you have additional questions, contact your health care professional.

Resources:

Resources are listed on the next page

RESOURCES FOR CHILDREN WITH SEPTO-OPTIC DYSPLASIA

National Organization
For Rare Disorders, Inc. (NORD)
P.O. Box 8923
New Fairfield, CT 06812-8923
Telephone: (203) 724-6518
Fax: (203) 746-6481
Toll free: (800) 999-6673
TDD: (203) 746-6927
E-mail: orphan@nord-rdb.com
http://www.nord-rdb.com/~orphan

The Arc
(A national organization on mental retardation)
500 East Border Street, Ste. 300
Arlington, TX 76010
Telephone: (817) 261-6003
E-mail: thearc@metronet.com
http://thearc.org/welcome.html

Brain and Pituitary Foundation of America
281 East Moody Avenue
Fresno, CA 93720-1524
Telephone: (209) 434-0610

National Association for Parents
of the Visually Impaired)
P.O. Box 317
Watertown, MA 02272-0317
Telephone: (617) 972-7441

American Foundation for the Blind
11 Penn Plaza, Ste. 300
New York, NY 10001
Telephone: (212) 502-7600
E-mail: afbinfo@afb.org
http://www.afb.org

Blind Children's Fund
4740 Okemos Road
Okemos, MI 48864-1637
Telephone: (517) 347-1357
E-mail: blindchfnd@aol.com
http://www.blindchildrensfund.org

Council of Families with Visual Impairment
6212 West Franklin Street
Richmond, VA 23226
Telephone: (804) 288-0395

Diabetes Insipidus Foundation, Inc.
4533 Ridge Drive
Baltimore, MD 21229
Telephone: (410) 247-3953
E-mail: diabetesinsipidus@amdyne.net
http://diabetesinsipidus.amdyne.net

Foundation Fighting Blindness
Executive Plaza 1
11350 McCormick Road, Ste. 800
Hunt Valley, MD 21031-1014
Telephone: (410) 785-1414
http://www.blindness.org

Human Growth Foundation
7777 Leesburg Pike, Ste. 202 South
Falls Church, VA 22043
Telephone: (703) 883-1773
E-mail: hgfound@erols.com
http://www.hgfound.org

MAGIC Foundation for Children's Growth
1327 North Harlem Avenue
Oak Park, IL 60302
Telephone (708) 383-0808
E-mail: mary@magicfoundation.org
http://www.magicfoundation.org

National Federation of the Blind
1800 Johnson Street
Baltimore, MD 21230
Telephone (410) 659-9314
E-mail: nfb@access.digex.net
http://www.nfb.org

NIH/National Digestive Diseases
Information Clearinghouse
2 Information Way
Bethesda, MD 20892-3570
Telephone (301) 654-3810

What is Hypopituitarism?

Hypopituitarism is the decrease or absence of one or more of the pituitary hormones. The lack of all anterior and posterior pituitary hormones is known as panhypopituitarism. The pituitary is a small gland located at the base of the brain and is controlled by the hypothalamus. It is referred to as the "master gland" because its chemical messengers or hormones signal other endocrine glands to produce their own hormones. The anterior or front, of the pituitary produces hormones that are needed for normal body functions. These hormones include:

- Growth Hormone (GH)
- Adrenocorticotropic Hormone (ACTH)
- Thyroid Stimulating Hormone (TSH)
- Gonadotropins or sex hormones(LH/FSH)

The posterior, or back, of the pituitary produces:

- Antidiuretic Hormone (ADH)
- Oxytocin

What causes hypopituitarism?

Congenital hypopituitarism refers to the condition being present at birth, even though it may present later in life. This means the pituitary or hypothalamus did not develop normally before birth. Acquired hypopituitarism refers to damage to the pituitary or hypothalamus during or after birth. Certain kinds of tumors may affect the pituitary. Other causes of pituitary problems include radiation treatment for cancer, severe head trauma, infections, or surgical removal of the pituitary gland.

What are the possible effects of hypopituitarism?

Children with congenital hypopituitarism may have hypoglycemia (low blood sugar) and prolonged jaundice at birth. Male infants may have a small penis. Children with growth hormone deficiency may grow normally until 6-18 months, and then growth may slow down. The loss of ACTH or TSH may cause adrenal insufficiency (not enough cortisol) or hypothyroidism (not enough thyroid hormone), respectively. A summary of the hormones and there function is described below.

Growth Hormone Deficiency (GH): Growth hormone affects the growth of bone and body tissues, as well as, muscle, fat, and sugar metabolism. Without growth hormone a child will be normally proportioned, but grow very slowly. The child will be smaller and younger-looking than their peers. Some infants or children with growth hormone deficiency may have low blood sugar.

Adrenocorticotropic Hormone (ACTH) Deficiency: Normally this hormone stimulates the adrenal gland (located on top of the kidneys) to produce cortisol. If the pituitary gland does not produce ACTH then cortisol will not be produced. Cortisol keeps

the body's blood sugar at a normal level and helps the body deal with physical stress, such as fever or injury. Children with too little cortisol may experience the following:

- tiredness, weakness
- diarrhea
- dizziness
- lower than normal body temperature
- frequent illnesses, such as ear infections
- nausea/ vomiting
- fast pulse, fast breathing
- confusion
- pain in abdomen, lower back and legs
- seizures from low blood sugar

Thyroid Stimulating Hormone (TSH) Deficiency:

TSH stimulates the thyroid gland to release thyroxin, which is important for brain development, growth, and metabolic balance. Children with too little thyroxin may experience the following:

- tiredness and increased sleep
- weight gain
- poor appetite
- constipation
- cold intolerance
- slow growth
- coarse, dry, thin hair
- dry skin

Gonadotropin Deficiency (FSH, LH)

LH and FSH stimulate the sex organs of the body to produce puberty hormones. In females, these hormones stimulate the ovaries to make estrogen and progesterone, causing development of sexual characteristics. In males, these hormones stimulate the testes to make testosterone, causing the development of sexual characteristics. Lack of gonadotropins will prevent the body from developing into physical maturity.

ADH Deficiency

ADH helps the body maintain the water and salt balance by acting on the kidneys to save more water when necessary. When there is not enough ADH the child drinks a lot but also urinates frequently, causing the body to dehydrate.

How is hypopituitarism diagnosed?

Magnetic resonance imagery (MRI) can detect tumors and structural problems in the area of the hypothalamus and pituitary. Bone age x-rays are used to estimate the maturity of the bones and growth potential. Blood tests measure pituitary function and target gland hormones (Cortisol, TSH, GH, LH, and FSH).

How is hypopituitarism treated?

Since your child is unable to make some or all of these hormones, replacement therapy will be life long. Treatment includes replacement of the hormones normally made by the target glands.

Growth Hormone is replaced with man-made growth hormone given by an injection.

Adrenocorticotropin Hormone replacement is with hydrocortisone a form of cortisol. This is available in pill or liquid form and is taken 2-3 times a day throughout life. The

dose is increased for illness, fever, and injury. If the medicine cannot be taken by mouth, it must be given by injection. Ask your doctor for instructions should this occur.

Thyroid replacement is with levothyroxin. It is available in pill form and is taken daily throughout life.

Gonadotropin deficiency requires the replacement of estrogen or testosterone at the appropriate time to begin puberty. Adolescent females are given estrogen replacement. Estrogen hormone is taken daily and progesterone is added after several months. These hormones mimic the way the ovaries would produce hormones. There will be a menstrual period at the end of each cycle. Males are given testosterone replacement by an injection every 2-4 weeks.

ADH is replaced with vasopressin (DDAVP), which can be given in intranasal form, by mouth, or by injection.

When will the doctor see my child?

The extent of the pituitary deficiency will determine how often your child needs to see the pediatric endocrinologist. Replacement therapy needs to be monitored and adjusted. Your child's doctor or nurse will give you guidelines for contacting them if your child becomes ill between visits. Remember that you will also need to follow up with your child's primary care doctor for routine health care. If you have additional questions, contact your health care professional.

Resources:

Pituitary Network Association
Website: www.pituitary.org
(805) 499-9973
(805) 480-0633

Magic Foundation
Website: www.MagicFoundation.org
(708) 383-0808