Fact Sheet
Norrie’s Disease

Information retrieved from:
www.ssc.education.ed.ac.uk/resources/vi&multi/eyeconds/Norr.html

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What is Norrie's Disease? Very rarely when a baby is growing in the womb not all the parts of the eye grow in the right way. In Norries' disease the back part of the eye stops growing while the unborn child is still in the womb. This happens about half way through pregnancy. As a result when the child is born the light sensitive film at the back of the eye (retina) is not ready to work and the vision is poor.

What is the Cause of Norrie's Disease? Growth factors help the unborn child to grow in the womb: When an unborn child is growing in the womb special chemicals called growth factors are needed. Certain parts of the body need certain growth factors to grow. In Norrie's Disease the growth factor that helps the back part of the eye to grow does not work.

Genes hold the plan for growth factors: Genes are a chemical alphabet held in all the parts of the body. The body has a "built-in" plan to make sure all the growth factors work. This plan is written in our genes. Genes are stored as long threads folded up into thick ropes called chromosomes. Every cell of the body has exactly the same number and kind of chromosomes. There are 23 different chromosomes but each chromosome has a copy of itself so that there are 46 in total (23 pairs). One pair of chromosomes is called the sex chromosomes and holds the plan for making a person grow into a boy or a girl. There are two types of sex chromosomes called X or Y. The letters just describe the shape of the chromosome when seen under a
microscope. If a child has an X and a Y sex chromosome then the child will develop into a boy (XY). If the sex chromosomes are both X then the child will develop in the womb into a girl (XX). Children inherit a sex chromosome from each parent. Mothers (XX) always pass on X chromosomes.

**Only Boys Develop Norrie's Disease:** The gene that has the wrong plan for the important growth factor in Norrie's Disease is on the X chromosome. If a girl (XX) has one X chromosome that does not work she will be protected from developing Norrie's Disease because she has a protective spare. This is because her 'spare' working X chromosome makes enough growth factor to make the back part of the eye grow in the womb. Boys (XY) however have only one X chromosome, with no protective spare. If boys have an X chromosome that does not work then they do not make any growth factor to help the back part of the eye to grow. They develop Norrie's disease. Most children with Norrie's disease have inherited the affected X chromosome from their mother.

**Might Another Child from the Same Mother Develop Norrie's Disease?** There is a 1 in 2 chance that another son would develop Norrie's Disease and a 1 in 2 chance that he would not be affected. All daughters will not be affected by Norrie's Disease but there is a 1 in 2 chance that a daughter would carry an affected X chromosome like her mother. This daughter would have a 1 in 2 chance of having a son with Norrie's Disease.

**Screening for Norrie's Disease Can be Performed on the Unborn Child if the Mother is a Known Carrier of an Affected X Chromosome:** Norrie's disease is very rare and is not routinely screened for in the unborn child. If however a family already has a son with the condition then a special test can be performed on the Mother and an unborn son to see if the child will also develop the condition.

**How does Norrie's Disease Affect a Child's Vision?** Norrie's Disease can affect vision in different ways depending on the degree to which the eyes are affected. If only mildly affected then the overall vision may be good enough for the child to see large print and recognize faces. However in Norrie's Disease the back part of the eye or the retina is often affected to a severe level. The vision can be very poor. The child may have reduced sharpness of vision and may only see bright lights and large shapes.

After a few years the lens in the eye can become hazy (cataract). This can reduce the vision further. Sometimes the pressure in the eye can also go up (glaucoma). If the pressure cannot be reduced with drops then an operation may need to be performed.

**How does Norrie's Disease Affect a Child?** About half of children with Norrie's disease have learning difficulties. After a few years about one third will also develop poor hearing. Many children also develop epilepsy (seizure disorder).

**How is Norrie's Disease Diagnosed?** Most children can be diagnosed by an examination from an ophthalmologist. A special test of the genes can be performed which can help confirm the diagnosis.

**How can Doctors and Other Specialists Help?** Special low visual aids may improve vision. Eye care specialists can treat any conditions of the eyes which sometimes develop after a few years.
The epilepsy can be treated with medication. If the child has a hearing loss, the child should be evaluated by an audiologist to determine if hearing aids will be of benefit. If the child has a hearing loss, a teacher certified in deafness will be an important team member of the educational team. Teachers certified in the area of visual impairment and certified Orientation and Mobility Specialists can address both educational, movement, and travel needs. When a learner has both a visual and a hearing loss, the family and service providers will also benefit from the services of their state project serving children with deafblindness.

**Resources:**

**Norrie Disease Association (NDA)**

Neurogenetics Simches Research Building, 5-238185 Cambridge St.

Boston, MA 02114

http://www.norries.org/

The Norrie Disease Association is a voluntary not-for-profit service organization dedicated to providing support and information to people with Norrie Disease and their families. Norrie Disease is a rare inherited disorder characterized by blindness in both eyes at birth; some affected children may later develop mild to profound hearing loss and varying degrees of mental retardation.

Established in 1994, the Norrie Disease Association compiles and disseminates educational and research information on the disorder; maintains a patient registry; and provides referrals to medical and/or genetic services. The Association also provides educational and supportive information through its database, directory, and flyers.

For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project and the project’s services specific to Colorado children who are deafblind, please contact:

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**Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 10/10**