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Fact Sheet

Anophthalmia / Microphthalmia

Information retrieved from: International Children's Anophthalmia Network - http://anophthalmia.org/ *and the National Eye Institute -* *http://www.nei.nih.gov/health/anoph/anophthalmia.asp*

What is Anophthalmia? Anophthalmia, also known as anophthalmos (Greek: ανόφθαλμος, "without eye"), is the congenital (at birth) absence of one or both eyes. It is a medical term that describes the lack of ocular tissue and globe from the eye(s). The absence of the eye(s) will cause a small bony orbit, a constricted mucosal socket, shortened eyelids, reduced palpebral fissure, and the underdevelopment of the high cheek area of the face. Anophthalmia can be unilateral (one eye) or bilateral (both eyes).

What is Microphthalmia? Microphthalmia (microphthalmos) is also a congenital condition in which the eye(s) started to form during pregnancy, but for various reasons, stopped developing. The infant is born with atypically small eye(s). The size of the eye(s) can vary from one child to the next. If very mild, microphthalmia may not be overly noticeable, but more often than not, this condition is obvious due to the small nature of the eye(s). In extreme situations, the microphthalmia may present as almost as anophthalmia where the eyes appear to be almost absent.

Complex microphthalmos describes a condition in addition to very small eyes, there are additional structural findings that may interfere with the child's vision. These findings may include cataract (clouding of the lens), coloboma (absence of a portion of a structure of the eye such as the iris, retina, and/or optic nerve), and/or orbital cyst. The degree of visual abilities is dependent on the extent and location of the other eye findings.

Microphthalmia, anophthalmia and coloboma are on a spectrum of eye abnormalities referred to as MAC.

What is the Prevalence of Anophthalmia / Microphthalmia? Both of these conditions are rare, though the exact incidence is unknown. One report from a prospective study of 50,000 newborns found an incidence of microphthalmia of 0.22 per 1,000 live births. Another study indicated that the prevalence of anophthalmia and microphthalmia was 1.0 per 10,000 births. The combined birth prevalence of these conditions is estimated to be about 30 per 100,000 births, with microphthalmia reported up to 11% of children who were blind in another study.

What are the Causes? Anophthalmia or microphthalmia are congenital (present at birth). Genetic mutations and abnormal chromosomes may be causes for either condition, which can occur alone or along with other health problems / disabilities. Anophthalmia or microphthalmia may result from

inherited genetic mutations, sporadic genetic mutations, chromosome abnormalities, prenatal environmental insult, or other unknown factors. Anophthalmia can also be associated with other syndromes.

What are the Functional Vision implications? Children with microphthalmia may have functional vision with varying ranges of visual abilities (light perception to low vision). A prosthesis can be made to cap the microphthalmic eye to help with cosmetic appearance, while preserving the remaining sight. A prosthesis is an artificial eye.

What Medical Treatments? An infant with anophthalmia or microphthalmia should be evaluated by an ophthalmologist (medical eye doctor), ocularist, and, as needed, an oculoplastic surgeon. An ocularist is a health professional who specializes in prosthetic devices for the eye.

The orbits (eye sockets) are very important for proper growth and development of the developing child's face. If an eye is missing or too small, the bones around the eye may not grow properly. A conformer is a plastic shell-like device, which is made by an ocularist that can be placed inside the orbit to help support the growth of the eye socket and the bones in the face. As the child grows and the orbits and facial bones continue to develop, the ocularist can make prosthetic (artificial) eyes. Since every child is an individual, the age when the child will be ready for a prosthetic eye will vary. The ocularist will work closely with the ophthalmologist and oculoplastic surgeon to make conformers and prosthetic eyes that are optimally customized for each child. It may be recommended to implant a prosthetic device deep into the orbit to make it easier to fit the prosthetic eye through a surgical procedure. There are other surgical options available when the conformers are alone are not enough.

There is no treatment that can reverse or improve visual impairment associated with microphthalmia. The condition should be monitored by an eye doctor. If a cataract(s) is present that causes further visual impairment, the child may be a candidate for surgery to remove the cataract. There is increased risk of detached retina in eyes that are microphthalmic. It is likely that the child may need to be seen by an ophthalmologist more frequently during the early years to monitor for treatable conditions that might develop, but will depend on the individual needs of the child. The child may need to be followed up periodically when the condition is seen to be stable and there are no other complications. If the eye is small or seriously malformed, then an artificial eye or lens (sometimes called a scleral shell or prosthesis) can be fitted for cosmetic reasons and/or for promoting socket growth.

A medical specialist will need to evaluate the child's needs with prosthesis or a conformer. A prosthesis or conformer must be regularly enlarged to expand the socket in a very young child who is still actively growing. This is necessary to stimulate the socket tissues and bony orbit to grow at a normal rate. In the case of a congenitally small orbit, the socket and lid opening is actually smaller than the companion eye. Due to the difference in size, a series of conformers may be necessary to stretch the surrounding tissues and form a socket into which prosthetic eye can later be fit. The conformer may need to be enlarged as often as once a month or as necessary to increase orbital volume as circumstances allow.

As the child grows the prosthesis will need to be checked regularly for size, comfort and fit (usually 2-3 times a year). The prosthesis will also need to be polished and checked for scratches or sharp edges.

When a child's eyes are fully developed, usually by the preteen years, the prosthesis will generally remain the same shape and size for a longer period of time. It may still be necessary, however, to enlarge the artificial eye periodically or fabricate a new prosthesis from time to time. This procedure may need to be done every three to five years depending on the child's age.

If the microphthalmia is unilateral (only one eye is involved) and causing reduced vision in that eye, then the eye care specialist may prescribe protective glasses with polycarbonate lenses to protect the better eye from injury. Such glasses will be especially important during sports and recreation activities.

What are Educational Implications? Children with anophthalmia or microphthalmia may be eligible for early childhood or school-age special education services, if the associated vision loss prevents the child from receiving reasonable educational benefit from general education. The child may benefit from specially designed instruction and equipment. A certified teacher of student with visual impairments and a certified orientation and mobility specialist will likely be members of an educational team, if the child is determined to be eligible for special education services as a child with visual impairment, including blindness. The child's hearing should be screened to ensure that there are no further sensory complications to learning.

Resources

ican (International Children's Anophthalmia/Microphthalmia Network) is a support group made up of families and professionals, for families dealing with these issues. The support group can put you in touch with other families or medical professionals who can help. Phone: 800-580-ican or http://anophthalmia.org/

For more information about the Colorado Services for Children and Youth with Combined Vision and Hearing Loss Project 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. The contents of this Fact Sheet were developed under a grant from the United States Department of Education (US DOE), #H326C080044. However, these contents do not necessarily represent the policy of the US DOE and you should not assume endorsement by the Federal Government. 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: 5/12