

Fact Sheet: Optic Nerve Hypoplasia

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Definition

Optic Nerve Hypoplasia (ONH) is a congenital eye condition in which the optic nerve did not develop completely. An underdeveloped optic nerve cannot take visual messages to the brain adequately. The result may be poor visual acuity, fluctuating vision and the loss of visual fields or spots of no vision within the fields. Poor acuity may be corrected to some degree with prescription glasses (Liefert, 2008).

ONH commonly occurs in both eyes, often with one eye worse than the other, but it can also occur in only one eye. Instead of having over one million connections from each eye to the brain, people with ONH have far fewer connections. The more connections there are between the eye and the brain, the better the vision. Many children with ONH experience some improvement in their vision over the first few years of life (MAGIC Foundation, n.d.).

Most cases of ONH have no clearly identifiable cause. There are no known factors in the development of ONH, nor is there a known association with exposure to pesticides (AAPOS, 2016). Optic Nerve Hypoplasia is a leading cause of childhood blindness and is diagnosed with direct ophthalmoscopy, finding a small optic disc (Borchert, 2012).

History

The first description of ONH was generally ascribed to Magnus in 1884, but the first artistic rendering of the optic disc appearance was by Schwarz in 1915. The first recognition of an association of ONH with agenesis of the septum pellucidum was by Dr. David Reeves at Children's Hospital Los Angeles in 1941 (Borchert, 2012).

Optic Nerve Hypoplasia was an interest to a neurologist named Georges de Morsier in the early twentieth century. De Morsier discovered that a few of the brains he examined from people who had been diagnosed with ONH also had compromised development of the septum pellucidum. The septum pellucidum is the part of the brain which divides the two halves of the brain. He named a syndrome Septo-Optic Dysplasia (SOD) which includes both ONH and the absence of the septum pellucidum (Liefert, 2008).

The association of ONH with absence of the septum pellucidum was later erroneously attributed to Georges de Morsier, who coined the term "la dysplasia septo-optique (septo-optic dysplasia). The "optic dysplasia" recognized by de Morsier was not ONH (Borchert, 2012).

Absence of the septum pellucidum is not associated with the major neurologic or systemic consequences (Borchert, 2012). When referring to ONH, the terms septo-optic dysplasia and de Morsier's syndrome are historically inaccurate and diagnostically misleading (Borchert, 2012). Apparently, it makes little difference if the septum pellucidum is not developed fully. Children who are diagnosed with both Optic Nerve Hypoplasia and an underdeveloped septum pellucidum are likely to have other anomalies as well (Liefert, 2008). Borchert suggests that since absence of the septum pellucidum is not a problem itself, we would do well to drop the diagnosis of septo-optic dysplasia and concentrate on determining what other anomalies a person who is diagnosed with ONH has.

Visual Functioning

Most children with ONH initially present with vision problems. Nystagmus usually develops at 1 to 3 months of age, followed by strabismus, typically esotropia, in the first year of life. Approximately 80% of children with ONH are bilaterally affected, and two-thirds of those are asymmetrically affected (Borchert, 2012).

Visual acuity ranges from no light perception to near normal acuity. More than 80% of bilateral cases are legally blind. Most affected children enjoy some improvement in their vision during the first few years of life. It is difficult to quantify improvement in this age group; however, improvement from only light perception behavior to quantifiably functional vision is not rare (Borchert, 2012).

Additional Challenges

Several problems may accompany ONH whether or not the septum-pellucidum is absent:

- Hypothalamic Dysfunction: loss of regulation of homeostatic mechanisms controlling behavior and pituitary gland function (Borchert, 2012).
 - Hypopituitarism: dysfunction of the pituitary gland.
 - Thirst/Hunger: frequently exhibit abnormally increased appetite with obesity, or reduction in food intake and eating without wasting. Some children also have an aversion to certain textures of food. Water-seeking behavior is also common and may be mistakenly attributed to diabetes insipidus.
 - Sleep: many children with ONH have primary clock lesions with loss of rhythmicity and sleep or wakefulness distributed over the 24-hour day. These sleep irregularities commonly result in behavioral difficulties and disruption to family life.
 - Temperature Regulation: since the hypothalamus is involved in body temperature regulation, it is not surprising that many infants and children with ONH have problems with body temperature regulation and may be frequently hospitalized to rule out sepsis.

- Poor development of the corpus callosum: without a properly developed corpus callosum, it is difficult to do tasks that require both hands. Children without a properly developed corpus callosum do not cross midline of their bodies without a lot of effort. Emotions and thoughts are not communicated between the sides of the brain and so many not influence each other. Mistaking letters as their mirror images in print or braille is an example of a “reversal” mistake related to poor development of the corpus callosum (Liefert, 2008).

- Developmental delay: range from isolated focal defects to global delay (Borchert, 2012).
 - Motor delays
 - Communication delays
 - Social Skill delays
 - Autism Spectrum Disorders
 - Rigid thought processes: Some children have difficulty doing the same activity in more than one way or following different rules in different situations. They are called “first time learners”. They may learn something one way and not be able to alter their thinking about it when more information is available. For example, if a “first time learner” learns to call using the bathroom “going potty” at three years old, he may be talking about “going potty” in into his teens. Using language that

will be appropriate for life is important from the beginning when working with “first time learners” (Liefert, 2008).

People who have ONH have a wide range of abilities. Each person is unique!

Prenatal Risk Factors

Lack of definitive genetic associations has led to a search for prenatal environmental or biological risk factors for the development of ONH. According to Borchert (2012), the most commonly reported associations include:

- Young maternal age and or primiparity.
- Maternal use of recreational drugs
- Anticonvulsants, antidepressants, viral infections during pregnancy
- Children with fetal alcohol syndrome

Accommodations/Strategies

- Refractive errors should be treated when the visual acuity reaches a functional level
- Patching of the better eye can result in improvement of vision in the worse eye
- Braille or other tactile systems in place of print as learning medium (Liefert, 2008).
- Avoid glare
- High contrast
- Avoid visual clutter
- Enlarged materials as appropriate
- Preferential seating
- Allow breaks due to visual fatigue as needed
- Model language that will be appropriate for life
- Re-teach skills in new environments

References

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