

Is optic nerve hypoplasia increasing in prevalence?

Devon Schuyler

SOME ophthalmologists say they're seeing more children than ever with optic nerve hypoplasia (ONH). But is the rise due to more cases of the disease, or better diagnosis?

"I'm absolutely seeing more cases," said Mark Borchert MD, a paediatric neuro-ophthalmologist at the University of Southern California in Los Angeles, in an interview with *EuroTimes*. He said he believes that the increase started in the 70s, and has been growing every year since.

"I think that everyone's experiencing an increase in cases," agreed Creig Hoyt MD, chair of the department of ophthalmology at the University of California, San Francisco.

As for whether this represents a true increase in prevalence, nobody knows for sure.

"The impression in the United States and Europe is that it definitely is more common than in the past," said Dr Hoyt, although he stressed that this impression remains unproven.

Robert Egan MD, a neuro-ophthalmologist from the Casey Eye Institute at Oregon Health & Science University, was even more sceptical. "I don't think that there is enough data out there to suggest that it's increasing in prevalence," he said.

He attributed the increase in cases to greater awareness of the disease over the past 20 years, and improved skill in diagnosing it.

Evidence of increased incidence

Dr Borchert agreed that data are lacking, but proposed several arguments to support the hypothesis that the disease is on the rise. First, he pointed out that ONH

was first diagnosed and described more than 120 years ago, and that the best instrument for diagnosis - the ophthalmoscope - has been in common use for 150 years. Second, he pointed to the experience of ophthalmologists who have been in practice since the 1970s and have noticed an increase in cases since then.

"They don't believe that they were missing it in the past and are more sensitive now," he said.

Finally, he pointed out that there are very few people over 30 who have the condition. By comparison, many children in schools for the visually impaired have been diagnosed with ONH.

Could Dr Borchert be diagnosing more cases than he used to because he's now recognised as an expert on ONH? That's unlikely, he said. Most of his patients first visit him because they have vision impairment, not because they have suspected ONH.

True prevalence unclear

Optic nerve hypoplasia is the most common ocular cause of vision impairment among children in Sweden and one of the top two causes (along with retinopathy of prematurity) in the United States, but its prevalence is unclear. Data from the Swedish Register of Visually Impaired Children have put the number at 6.3 cases per 100,000 people aged 19 and under in that country (*Acta Ophthalmologica Scandinavica*, December 1997 75: 681-687). Dr Hoyt estimated that the number of cases worldwide is in the tens or hundreds of thousands.

Ophthalmologists are working to understand what's causing the disease. They do have an idea what isn't causing it.

"It's not genetic, it doesn't

run in families, and it doesn't seem to be related to infection," said Dr Hoyt.

"The impression in the United States and Europe is that it definitely is more common than in the past."

Creig Hoyt MD

He said that although heavy alcohol use during pregnancy increases the risk slightly, there is no history of alcohol use in the vast majority of cases. Other factors linked to the condition include young maternal age, first parity, maternal smoking, preterm birth, and factors associated with preterm birth. Some people have proposed that contaminants in the water or air, food additives, or medications may be behind the increase, but the cause remains a mystery.

Optic nerve hypoplasia is a congenital abnormality marked by a reduced number of axons in one or both optic nerves. An ophthalmoscopic exam reveals a small, grey, or pale optic nerve head, often with a "double-ring sign" representing the canal through which a full-sized nerve would have passed. Vision can range from 20/20 to the inability to perceive light; legal blindness occurs in less than 10% of cases.

Association with other pathologies

Diagnosis is important because people with ONH often have coexisting health conditions. Many children with ONH experience neurologic problems, which may be caused by abnormalities in the corpus

callosum. An MRI or CT scan may reveal abnormalities in the septum pellucidum as well. These abnormalities are not believed to cause neurologic problems.

Dr Borchert said that in 50% to 70% of cases, children with ONH have endocrine dysfunction stemming from poor regulation of the pituitary gland. This can lead to deficiencies in growth hormone, thyroid-stimulating hormone, gonadotropin, corticotropin hormone, vasopressin, or prolactin.

Dr Borchert is currently carrying out three studies to

"It's not genetic, it doesn't run in families, and it doesn't seem to be related to infection."

Creig Hoyt MD

learn more about ONH. First is a cohort study of patients aged two and under designed to identify clinical and prenatal factors associated with the disease. Second is an epidemiologic study designed to identify seasonal or geographic variations associated with the condition. Third is a randomised trial designed to determine whether early treatment with growth hormones lead to improved outcomes in children with ONH and a deficiency of the hormone.

Although there is no way to restore the optic nerve, some children experience improvements in vision as they learn to adjust to the condition. However, the three experts interviewed by *EuroTimes* agreed that eye exercises are unproven and do not recommend them.

Creig Hoyt MD
choyt@itsa.ucsf.edu

Robert A. Egan MD
eganr@ohsu.edu

Mark Borchert MD
mborchert@chla.usc.edu