# ONH 101: What Families Need to Know

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## Empowering Families

* A primary responsibility of the 0-3 Educator is to educate families
* Greater knowledge regarding a child’s eye conditions leads to increased confidence and ability to advocate
* As TVI/DTV, we must understand all aspects of ONH in order to address the development of the whole child, so we are in a position to share that knowledge with families

## Information Overload

* ONH involves ocular, neurological, and endocrine abnormalities
* Many doctor’s appointments, information coming in from several specialties
* Dealing with grief/adjusting to the diagnosis
* Young mothers/first-time mothers
* We must individualize our approach to sharing information

## Visual System

## Optic Nerve Hypoplasia (ONH)

* Congenital condition in which the optic nerve is underdeveloped
* Diagnosed by presence of small/pale optic nerve on ophthalmologist’s exam

## The “Syndrome of ONH”

* Wide variation in visual functioning
* Vision impairment ranges from mild to severe
* Stable/non-progressive condition
* May affect one or both eyes (and of varying degrees)
* Nystagmus
* May be noted when both eyes affected
* May be associated with significant bilateral reduced visual acuity
* Develops at 1-3 months of age

## The “Syndrome of ONH” (continued)

* Strabismus
* Noticed in first year of life
* Typically esotropia
* Those with unilateral ONH may present with strabismus rather than nystagmus
* Incidence of strabismus may increase with unilateral ONH

## Over first 5 years of life

* Vision may actually improve
* A mild improvement in visual function may occur as the result of maturation processes of the brain
* Nystagmus may decrease over time

## Functional Vision

* Vision can range from no light perception to good functional vision, or even full vision in one eye
* Vision is characterized by a lack of detail (depressed field), but this lack of detail is not comparable to the blurred reduction in vision when a person removes his/her glasses
* Poor visual acuity
* May be unable to focus
* May have difficulty recognizing faces and facial expressions
* May have difficulty accessing near and detailed information

## Functional Vision (continued)

* In certain cases of ONH a specific field defect occurs. Children may not be aware of people or objects in the periphery
* The effect on the visual field may range from generalized loss of detailed vision in both central and peripheral fields (depressed visual fields) to subtle peripheral field loss
* Loss of peripheral field(s)
* “Tunnel Vision”
* May be difficult to gather comprehensive visual information in environments

## Functional Vision (cont.)

* Variable visual field defects dependent on multiple factors:
* Extent of ONH
* Chiasmal involvement
* Optic tract involvement
* Other brain involvement such as schizencephaly or effects from hydrocephalus
* Depth perception may be more severe if vision loss is great and/or if unilateral
* Children with ONH may be unable to locate objects in space precisely due to a lack of depth perception.

## Functional Vision (cont’d)

* Mild light sensitivity (photophobia) may occur
* These children may squint, lower their head, avoid light by turning away, or resist participating in outdoor activities.
* Variable color vision deficiency
* When one eye is affected more than the other, an ophthalmologist may recommend a trial of patching the stronger eye, since the visual loss may be due to amblyopia

*It is very difficult to predict visual acuity potential!*

## Septo Optic Dysplasia (SOD)

* Considered to be a variation of ONH
* SOD has been generalized to mean any case of ONH plus hypopituitarism or midline brain differences
* De Morsier’s Syndrome
* SOD was a term created by DeMorsier (1956) to describe abnormalities of the optic tract accompanied by an absent septum pellucidum
* Small optic discs and double rings

## SOD/ONH

### Three main structures of the brain may be affected

* Corpus callosum
* Connects the right and left hemispheres of the brain, allowing communication
* Septum pellucidum
* Connected to corpus callosum, divides the lateral ventricles
* Hypothalamus
* Controls autonomic processes such as temperature regulation and sleep, as well as mood/behavior
* Controls pituitary gland, which directs release of many important hormones

### How Does This Manifest?

* Abnormal sleep pattern
* Difficulty with temperature regulation
* Severe mood changes
* Feeding difficulties
* Atypical sensory behaviors
* Low tone
* Poor motor planning

### Endocrine

* A number of hormone deficiencies are possible when a child has ONH
* These hormones affect growth, weight gain, hunger/thirst, and other important characteristics
* Hormone deficiencies have appeared for the first time **up to age 5**
* Hypothyroidism detected at mean age 15 months
* Without proper medication, these conditions can be severe

**Encourage parents to continue regular endocrine appointments and testing!**

### Endocrine (continued)

* Hypopituitarism (75-80%)
* Growth hormone insufficiency (70%)
* Hypothyroidism (43%)
* Adrenal insufficiency/low cortisol (27%)
* Diabetes insipidus (antidiuretic hormone insufficiency, 5%)

### How Does This Manifest?

* Developmental delay
* Small stature
* Unexpected weight gain
* Always hungry or thirsty
* Constipation
* Severe fatigue

### Behavior Issues

* Atypical social behaviors
* Language delays
* Sensory regulation
* Tactile selectivity/defensiveness
* Low frustration level
* Self stimulation
* Self injury
* Unable to self-soothe
* Sleep issues

### Behaviors (cont.)

* Social
* no joint attention,
* perseverative behaviors
* Self stim
* Twirling/spinning
* Flapping
* Self injury
* Head banging
* Biting
* Sensory regulation
* Sounds,
* Textures/feeding
* Temperature

### Sensory Dysregulation Implications

* Language differences
* May only be able to use one sense at a time
* Other senses are distracting
* Ex: sensitivity to the feel of clothes distracts from attention to language
* Auditory processing issues may make receptive language lower than expressive

### Social skills differences

* Visual cues
* Social language
* Shifting attention away from other stimuli

### Perseverative interests

* Routine is familiar (no new processing)
* Repetitive behavior is calming
* Repetitive events are easier to understand

### Autism

* Much controversy over diagnosis of children with visual impairments
* “blindisms” vs autistic behaviors
* Not enough expertise on ONH in the field of developmental pediatrics
* Borchert and colleagues added adaptations to the ADOS (Williams et al., 2013)
* More appropriate for children with VI, but still a screening
* Autistic-like behaviors may resolve after age 5 for child with ONH
* Social/communication challenges – there has to be a cluster of features from both social and language areas to be autism (not just hand flapping alone)
* Look at overall developmental level, then discern if social/language are different than other domains

### Autism (continued)

* Many children with ONH have delays in social and language skills
* Children with ONH are at higher risk for autism than the general population, but not every child with ONH will have autism
* Autism assessments must be adapted for VI
* Visually-based autism interventions need to be adapted for a child with ONH

## Conclusion

* ONH is a complex condition that causes visual impairment
* Non-visual associations include neurological, endocrine, and behavior differences
* These differences may manifest as developmental delays or sensory processing issues
* Sensory dysregulation may affect social and language development
* Careful assessment must be performed when autism is suspected

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