Optic Nerve Hypoplasia / Hypopituitarism

(previously known as Septo-optic Dysplasia)

What is Optic Nerve Hypoplasia / Hypopituitarism?

Optic nerve hypoplasia (ONH) is a congenital condition characterized by underdevelopment or incomplete development of the optic nerve, which is the nerve that transmits visual information from the eyes to the brain. This underdevelopment can result in various degrees of visual impairment, ranging from mild to severe, and may affect one or both eyes.

Hypopituitarism, on the other hand, is a condition characterized by underactivity of the pituitary gland, which is a small gland located at the base of the brain. The pituitary gland produces and regulates several hormones that are essential for various bodily functions, including growth, metabolism, reproduction, and stress response.

In some cases, optic nerve hypoplasia and hypopituitarism can occur together as part of a broader condition known as septo-optic dysplasia (SOD) or De Morsier syndrome. Septo-optic dysplasia is characterized by the triad of optic nerve hypoplasia, midline brain abnormalities (such as absence or underdevelopment of the septum pellucidum), and hypopituitarism.

The pituitary gland secretes a variety of important hormones. A child with ONH/Hypopituitarism can be missing one or multiple pituitary hormones. Some children are not missing any hormones. Each potential hormone is listed below along with a description of symptoms associated with a deficiency of that hormone.

- Growth hormone: Symptoms of low growth hormone include slow growth and short stature. A baby can also have low blood sugar if this hormone is missing. This is the most commonly missing pituitary hormone.

- Thyroid-stimulating hormone or TSH: This hormone controls production of thyroid hormone which, if missing, can cause poor growth, low energy, dry skin, constipation, feeling cold frequently, more weight gain than height gain, and, in some cases, delayed intellectual and physical development.
- Gonadotropins (luteinizing hormone or LH and follicle-stimulating hormone or FSH): These hormones are important for the initiation of puberty. A young boy with a deficiency of these hormones can have a small penis size or testicles that haven't descended into the scrotum.
- Adrenocorticotropic hormone (ACTH): This hormone stimulates the release of cortisol from the adrenal glands. Cortisol is an important hormone when our bodies are in a state of stress. Cortisol helps maintain our blood pressure and blood sugar amongst other things. A child who is missing cortisol can be very weak, tired, dizzy, or have a poor appetite.
- Anti-diuretic hormone (ADH): This hormone helps the kidneys concentrate urine when a child is dehydrated. If this hormone is missing, a person will urinate frequently and be very thirsty. A child who is unable to communicate well (like an infant or a child with an intellectual disability) can become very dehydrated due to an inability to verbalize a sense of thirst.
- Prolactin: This hormone is ordinarily responsible for milk formation in mothers who breast-feed their infants. Lower levels are normally present in both girls and boys, with slight *elevations* found frequently in infants and children with ONH/Hypopituitarism.

What causes ONH/Hypopituitarism?

Optic nerve hypoplasia (ONH) and hypopituitarism are both complex conditions with multifactorial causes. Here's a brief overview:

1. **Optic Nerve Hypoplasia (ONH)**:

- ONH is a congenital condition where the optic nerve is underdeveloped. The exact cause is not always clear, but several factors may contribute:

Genetic factors

Maternal factors

Prenatal factors

Environmental factors

In both cases, the exact cause can vary from person to person, and in many cases, the cause may not be definitively identified. Treatment and management typically focus on addressing symptoms and complications associated with these conditions.

How is ONH/Hypopituitarism diagnosed?

Most commonly, the abnormal eye movement is the first clue for ONH/Hypopituitarism. Sometimes one of the pituitary hormones is missing and may provide an early clue that the child has ONH/Hypopituitarism. An MRI of the brain may be ordered to get an inside look into how the brain has developed. Finally, genetic testing can be used to diagnose ONH/Hypopituitarism.

A child does not have to have all 3 features (meaning optic nerve hypoplasia, abnormal midline structures of the brain, and pituitary gland hypoplasia) to be diagnosed with ONH/Hypopituitarism. A child can have 2 of the 3 and still be diagnosed with ONH/Hypopituitarism.

How is ONH/Hypopituitarism treated?

There is no treatment to cure ONH/Hypopituitarism.

Typically, each symptom that develops is treated by an appropriate specialist. For example, an ophthalmologist can prescribe glasses or recommend certain therapies to improve vision. If your child has seizures, a neurologist can provide anti-seizure medication. If your child is missing a pituitary hormone, an endocrinologist can prescribe treatment to replace the missing hormone. An affected child may not have to see each of these doctors for life; it depends on what problems the child has. It is important

to note that, even though a child may not have a pituitary hormone deficiency initially, one can still develop later on in life.