



**Most children with Holoprosencephaly have many other challenges, some related to the HPE and some related to the underlying cause of their HPE.**

## **Cleft Lip or Palate**

Because HPE is a midline defect, some infants are born with a cleft lip and/or cleft palate. Surgical repair of the lip and/or palate may be considered, particularly to make feeding easier. It should be noted, however, that feeding problems might not be eliminated by the repair of the clefting, and the repair surgery can often affect the child's breathing. Repair of a cleft lip &/or palate is suggested between 6-12 months of age, to allow the tissue to grow enough to complete the repair. Consideration should also be given to stability of airway, risk of infection, etc.

## **Feeding**

Feeding can be a major challenge for many infants and children with HPE. Many show slowness in eating, frequent pauses while eating, and rapid loss of interest. Spitting up or vomiting may occur after feedings, and there is a risk of aspiration into the lungs. Alternate feeding methods may be very helpful such as tube feedings. For some children, swallowing and chewing can be improved through an intensive course of VitalStim therapy performed by a qualified speech or feeding therapist.

## **Gastrostomy Tube (G-Tube)**

When oral feeding becomes too difficult, a g-tube (gastrostomy tube) may be considered. This is a tube placed through the abdominal wall into the stomach so that liquid feedings and medications can be put directly into the stomach. If gastroesophageal reflux or vomiting is present, another procedure called a nissen fundoplication may be performed to tighten the inlet to the stomach. These two surgeries are typically performed at the same time.

## **Intestinal Gas**

Infants and children with HPE commonly have problems handling intestinal gas due to excessive air swallowing. Frequent burping during and following feedings can often relieve this discomfort. A variety of medications can help to decrease the gas and/or promote stomach emptying.

## **Elimination**

Constipation can be common, especially for those children with high muscle tone, spasticity, and for those who do not move as much as a typical child. This problem can often be improved by medication, altering the diet or by using suppositories.

## **Fluid Balance – Diabetes Insipidus**

Many children with HPE have a condition known as Diabetes Insipidus (DI) which is when the kidneys are unable to conserve water. Symptoms to watch for are: very heavy diapers (high urine output) and excessive thirst. Once diagnosed, the signs of DI are irritability, water retention and dehydration. In some children with HPE, DI is able to be managed by strict control of fluid intake. In other children, hormone replacement therapy is effective.

## **Temperature Control**

Temperature control can be quite erratic in infants and children with HPE. Elevated temperatures may occur in the absence of infection or other definable cause. At other times, the body temperature may be sub-normal for no apparent reason. Adjustments in the child's environment may be helpful in keeping the child's temperature within a normal range.

## **Motor Skills**

Virtually all infants and children with HPE have deficiencies in motor skills. There are some children with HPE who are successful in walking or crawling. Others are able to use assistive devices (walkers, gait trainers, etc.) to achieve independent mobility while others are wheelchair dependent. Head control can be difficult for infants and children. Reaching and grabbing are seen in some infants and children. Most children with HPE who do improve their motor skills do so at a slow and quite delayed pace compared to typical children. Physical and occupational therapies are commonly recommended and can bring improvement.

## **Hypertonicity**

Often, children with HPE have increased muscle tone to the point of spasticity. Spasticity is often evident when the child is stimulated, excited, or in distress. Arching of the back, thrusting of the legs and flexing of the arms are common in these circumstances. When relaxed, they may appear "floppy" due to poorly developed control of their muscles.

## **Microcephaly/Hydrocephaly**

The brain of an infant or child with HPE is often quite small. If there is no excess of cerebrospinal fluid around the brain, the head is also small (microcephaly). When there is excess cerebrospinal fluid, the head size may appear to be normal or enlarged. Head enlargement can make caring for the child quite difficult and excess fluid can result in pain and discomfort to the child. An operation may be recommended to shunt the fluid by way of fine tubing from the head to another part of the body, such as the abdomen, where the fluid can be absorbed back into the blood stream.

## **Seizures**

Many children with HPE are susceptible to seizures. There are various types of seizures, and frequency of seizures may change considerably over time. Many children with HPE who are affected by seizures respond well to anticonvulsant medications. There are many such medications on the market, and the child's neurologist will determine which medication or combination of medications will be best for treating the child's seizures.

## **Sleeping Problems**

Children with HPE frequently have difficulties sleeping, falling asleep or staying asleep. The hypothalamus helps to set the awake/sleep cycles. Bedtime sedatives have been beneficial for some children, but many families have an ongoing struggle with this problem. Many children with HPE have some degree of sleep apnea as well, either central (caused by the brain) or obstructive apnea that may or may not be able to be controlled.

## **Breathing Issues**

Many children with HPE have issues with breathing of some variety. This can range from a mild case of tracheomalasia (floppy windpipe) that the child might outgrow as he/she ages, to a more advanced case of stridor (noisy breathing), to a severe case of breathing obstruction in the nose or throat requiring a tracheostomy (breathing tube).

## **Smell**

It is often said that children with HPE do not have a sense of smell. This is perhaps due to the improper formation of the olfactory nerves and nasal cavities. This is not always the case, however, and many children with HPE are able to enjoy "smelling the roses."

**A child with Holoprosencephaly might have none, some or many of these additional challenges, or they might have other issues not listed here**

**If you think your child has any of these additional conditions, please see the appropriate specialist to help with diagnosis and treatment**

## **Language**

Full development of language is not usually seen in children with HPE; however, there are children who can communicate with a limited vocabulary of words and phrases and/or body language. Some children with HPE can understand sign language and use simple signs to express themselves. A variety of high and low tech augmentative communication devices can also be effective for a child with HPE to help communicate with others.

## **Hearing**

Virtually all children with HPE demonstrate that they can hear. They react to noises, learn to turn their heads to sound, and in time develop the ability to recognize certain voices and noises and react to those sounds in predictable ways. The children with HPE who are deaf or hard of hearing usually have additional conditions that might have caused the hearing loss.

## **Vision**

Unless the eyes are involved in the malformation, children with HPE usually demonstrate the ability to see. They can focus on faces and objects, develop the ability to track moving objects, and respond to facial expressions. The time at which these abilities appear is often delayed from the normal schedule of development. Some children with HPE have a condition called Cortical Vision Impairment (CVI). This is where the optic nerves are small and underdeveloped, causing less information to be sent to the brain, thus causing more of a challenge for the brain to interpret what is being seen. Many children with CVI and HPE make visual progress with the help of vision therapy.

## **Startle Reflex**

Many children with HPE show signs of an overdeveloped startle reflex. They are startled easily and jump what might be considered a small scare. This is often confused with infantile spasms and children can have a combination of both.

## **Cognition**

Many children with HPE have shown evidence that they can learn and remember. They can anticipate games that involve touching or tickling and can recognize familiar voices and sounds. Some children have proven the ability to learn colors and shapes, and other basic information. This capability is somewhat determined by the severity of the HPE and related formation of the brain, but can also be affected by the amount and intensity of early intervention. Most children with HPE have shown that they comprehend speech and can communicate this understanding thru simple facial expressions, vocalizations and some speech or movement. Also, many children are able to use simple switch devices to help communicate, facilitate play and other interactions.

## **Disclaimer**

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