# Cognitive and Social Development of Children with Holoprosencephaly

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When parents are first told that their child has holoprosencephaly (HPE) they are often given low expectations for their child's future achievements. However, specialists working with children with HPE find that many children develop a range of skills and are able to communicate and interact with the world, although not always in conventional ways. In fact, some of the problems attributed to children with HPE are actually due to *our* difficulty in understanding them. As we learn to adapt our interactional style and become better and more flexible observers, we come to understand the children better. Sometimes incidental observations can reveal a lot. For instance, a child might look at his mother upon seeing an unfamiliar and unusual toy. This suggests to us that the child notices the new toy and must therefore be able to remember more familiar toys and to compare the different toys. Furthermore, the child is developing a social awareness of others as he looks to the mother to perhaps to reassure himself of her presence and to see if she looks happy about the new toy indicating that there is nothing to worry about. Here we see early communication between the mother and child. By asking parents and caregivers about their children's behaviors and by using new assessment techniques a more positive picture of the potential of children with HPE emerges.

In this chapter, we start by outlining what we mean by cognitive and social development and we look at the sorts of skills that are typically acquired in these areas. We then consider how these skills are traditionally assessed and how they can be examined using alternative techniques in children who are limited in their ability to physically interact with the world and to communicate using spoken language, but who may have relatively strong skills in other areas. We then outline what sort of cognitive and social skills may be evident in children with HPE.

**What is cognitive development?**

Cognitive development is the growth of skills used by infants and children to understand and interact with the world around them. For example, from birth, children learn how to organize the images they see in the visual world. Early skills include focusing on objects, being able to remember what they look like, and tracking objects as they move across the visual field. At later ages, children are able to understand basic physical rules that objects follow, for instance, that solid objects cannot pass through each other. One way to find out what children understand about objects is by watching them play. For instance, if a child searches for a toy that has been hidden in a cup then he or she knows that the toy still exists and remembers where it is even though it is out of sight. Children also learn a lot about people in their first year, continually developing their social awareness. From birth, babies enjoy interacting with others and are particularly sensitive to the smells and sounds of their mother. They rapidly increase their social awareness skills across the first years, making eye-contact with others, enjoying play, and starting to anticipate and understand the behaviors of others.

Another important area of early growth, closely linked to social awareness and cognition, is language. Children start learning about sounds when they are still in the womb. Over the first year, this attention towards sounds and particularly voices develops into an understanding of specific words and phrases. We can assess children's understanding of language by looking at

their reactions to familiar words and their ability to follow instructions. The ability to understand language begins well before children begin to speak their first words. We can also listen to what children are saying. Most children start to produce language-like babbling towards the end of the first year before they actually start speaking words.

There is a large range in the rate of acquisition of these skills among normally developing children, although the pattern of development is fairly consistent with skills emerging in the approximately the same order.

**Assessing cognitive development**

When working with children, psychologists frequently assess cognitive and social skills by using standardized tests that require children to understand spoken instructions and respond by speaking, writing, drawing, pointing, or manipulating objects. Even tests given to children who are not yet speaking often require children to understand language and nearly always require children to move or manipulate objects. For instance, a young child might be asked to find a toy that has been hidden under a cup: that child must be able to understand the request and either point to the cup or lift it up to reveal the toy. It is quite difficult to assess children who have little spoken language and whose motor abilities may limit their use of gesture (including pointing) and their manipulation of objects. This is a common profile of children with HPE. Previous research on HPE has rarely examined their cognitive development, possibly because of the difficulty of assessing cognition due to the limitations in the testing methods.

Parents of children with HPE often believe that their child understand a lot more than is obviously apparent. However, psychological assessment of their children frequently results in a report that seems to document disappointingly low levels of achievement. This could be because unsuitable tests have been given. First of all, a child may have been penalized for having relatively unsophisticated motor skills. For instance, if a child did not search for a hidden object we might think that it was because he or she did not know where to find the object, rather than because the child had difficulty performing the search action. Similarly, a child's difficulty in pointing to a picture may be interpreted as a lack of understanding the instructions, rather than a difficulty in coordinating a complex action. Furthermore, the results are often conveyed as a single number or as a single age-level of performance; this is inappropriate for children who show a large range in their development, with skills at a much higher level in some areas than others. Finally, there is a great deal of variability in what children can do on any given day, and this variability may be greater in children with HPE (possibly because of medical complications or medication fluctuations).

Sometimes a tester knows that a task is inappropriate for a child, but has to present it to the child anyway to get a valid score on a "standardized test" (standardized tests have strict administration guidelines which must be followed in order to generate a score or age-level). Although it can be very frustrating to watch children with HPE being tested in a way that seems unfair and inappropriate, it is often necessary to give children these tests in order to qualify for services.

Although there is sometimes a need to test children in a standardized manner, even if the results do not accurately represent children's abilities, it is also extremely important for us to be able to accurately assess children's skills and strengths. As we understand more about children with HPE we can provide parents with appropriate expectations of their children's development level and also discuss children's performance properly among the scientific and clinical communities. In addition, it is important to be able to monitor children's development over time, particularly in response to training, intervention, or medical treatments. Finally, it may be

important to accurately assess children's skills to know when a child is ready to try certain activities.

Thus it is important to be able to assess children's skills accurately and this may mean that we have to use "non-standardized" assessments. One way in which we can improve our understanding of children with HPE is to ask parents and caregivers what their children can do. People caring for children with HPE, on a day-to-day basis, are the best source of information about their performance. Parents and caregivers can be asked about their children's understanding and production of language. Although it is easier to assess the sounds or words that children make, much research suggests that parents are also good at assessing what words and phrases their children understand. We can also learn a lot about children from incidental observations. For instance, a mother might comment that her daughter becomes excited when she hears her father opening the front door. This tells us that the daughter has good hearing, can remember the order of a chain of events (door opening then father returning), anticipates an outcome before it occurs, and has a social and happy memory of her father. Another early and important skill is understanding cause and effect relations – we can often see evidence of this as children start trying to control situations. For instance, a little girl might scream if a video that she is watching is turned off, but stop screaming as soon as the video is turned back on. She didn't really miss the video that much, but she knew what to do to get it turned back on! Alternatively, a little boy might yell or scream when he was put to bed, but stop as soon as someone had gone in to check on him. Nothing was really the matter, he just wanted some attention. These children are starting to understand how two quite different events can be related and, importantly, they are realizing that they can cause things to happen.

Another way in which we can improve the assessment of children with HPE is to use assessments that are not dependent on sophisticated motor skills or language (unless language is actually what a particular test is meant to assess). Techniques have been developed which allow children to use simple responses, such as a shift in eye-gaze. These techniques have been used to look at developing skills in infant perception from birth and can be adapted to assess more sophisticated skills in older children.

**Performance of children with HPE**

In this section we will summarize the findings of contemporary work on children with HPE. This includes results from a questionnaire given to parents of 35 children with alobar HPE, published by Mason Barr and Michael Cohen (1999) as well as observations shared among clinicians specializing in children with HPE. We also draw on our experiences testing children with HPE at Rutgers University on a unique battery of tests designed specifically for them.

In general, children with HPE show stronger social skills and understanding of language and the physical world, as compared with their spoken language and ability to physically interact with objects. Children are good at maintaining active interest in people and what they see or hear around them. Children can usually see, focus on, and track faces and objects (i.e., move their eyes to keep things in their view). Children can also remember things. Some children were tested with pictures and showed that they could remember and tell the difference between different pictures (although they may not necessarily know what the pictures were of, they could remember and tell the difference between the shapes). Children also demonstrated memory in their anticipation and recognition of people, objects, or events. Some children were also given a test requiring "associative memory". All of the children tested learned to turn their heads or shift their eye-gaze when they heard a certain sound to see some toys light up and move. Furthermore, many children

understand some of the basic rules that objects follow (for instance, that they continue to exist when they are out of sight).

Nearly all children with HPE react to sound, including turning to localize sound and recognizing certain sounds or voices. Many children respond especially to voices, understand the emotion conveyed by the tone of voice, and, perhaps, some words and phrases. Most children with HPE produce vocalizations. These vocalizations include noises that children use to express themselves (happy, sad, hungry etc.), noises they use as words (e.g., a certain sound used only to indicate "mama"), and early words and phrases. However, progress in this area is often very slow. Children also use gestures in play (e.g., pretending to talk on the telephone or pushing a toy car) or in a communicative manner (e.g., waving good-bye or indicating yes or no). Children's use of vocalizations, words and gestures is often inconsistent, in that they may use them relatively infrequently. Many children also use eye-gaze to successfully communicate, for example, looking at their father and then a toy, as a request to be given the toy.

Some children with HPE use alternative communication systems, such as pressing a button to indicate "yes" or "no". Before children develop a communication system they need to have developed some fundamental skills or concepts (not all children who communicate have fully developed these skills, although most have). One of these skills is the ability to share attention with others, that is, to look where others are looking or to direct others to the child's point of interest (by eye-gaze or pointing). Similarly, children must want to communicate with others. Another critical skill is the ability to learn associations between things. In spoken language these associations are between spoken words (e.g., "dog") and what they represent (the animal). In other, non-spoken communicative systems the associations might be between buttons, symbols, pictures, or gestures and what we want them to represent. Children also need to have some concept of cause and effect. That is, they must be able to realize that an action or word can "make" another thing occur. Children ready to learn an alternative communication system to spoken language are often communicating their basic needs in other ways (such as, through eye-gaze, gesture, or vocalizations) and understand some words and phrases. Systems often start with very simple associations, such as "yes" and "no", "more" and "enough", or "food and "drink" and a choice of buttons.

In general, children with alobar HPE often have the most severe difficulties, and children with semi-lobar HPE have the more sophisticated skills. However, it is important to remember that there are many exceptions to this mapping of diagnosis to level of achievement and it cannot always be a good predictor of individual achievement.

**Conclusions**

In sum, we believe that techniques are available that will allow children with HPE to be assessed much more accurately than in the past. Many clinical and medical personnel first working with children with HPE have low expectations for their performance. These low expectations will be confirmed if the children are tested by conventional means. However, recent work suggests that children with HPE are capable of achieving much more than had been thought possible in the past. Many children develop skills in a range of areas, including understanding of people and objects. Furthermore, parents and caregivers of children with HPE are often able to understand their children's needs and communicate with them. As this information reaches the medical, clinical, and research communities, it should encourage a more accurate perception of the potential of children with HPE. Although we believe there is much cause for optimism, it should be noted that even the least affected children still show significant delays, although their performance can be much higher than previously thought.

It is important for parents and caregivers of children with HPE to have a realistic idea of their children's capabilities and of the skills they have the potential to acquire. This knowledge allows the planning of intervention goals and provides guidelines as to what sorts of activities to encourage. The goals that are set for children with HPE may involve smaller steps than those for normally developing children, but the achievement of these goals is just as important and perhaps even more exciting. While children can learn much through individual play, they are often helped when their play is "guided" by an adult with a goal in sight. For example, if your child is almost, but not quite, able to perform a task on his or her own, providing "just enough" help, brings your child a little closer to performing it independently. The rich, multi-sensory environment that you provide for your child, especially in your interactions together, also has a strong impact on how he or she develops and grows. Responding to your child in the loving ways that feel natural provides a type of therapy no less important than that provided by clinicians and specialists. These warm interactions can powerfully affect patterns of brain connectivity and may fine-tune complex neural circuits. The role of the Carter Centers as a group is to provide the information and support that you need, as parents and caregivers, to ensure that each of these special children reaches his or her own full potential.