

# **Neurocognitive Assessment for Children with Holoprosencephaly & Severe Motor Impairments**

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# Introduction

## Abstract

**Children born with Holoprosencephaly (HPE), a midline developmental brain disorder, often have severe motor and language impairments, making traditional assessment tools impractical and uninformative. The Carter Neurocognitive Assessment (CNA, 2001) was developed to assess children with HPE and other related brain malformations whose cognitive and language levels fall into the 0-2 year range. Four cognitive domains were examined: Social Awareness, Auditory Attention and Comprehension, Visual Attention, and Vocal Communication. To date, 18 children with HPE have been assessed using this tool. As a group, performance was variable in each domain. However, the Vocal Communication domain was consistently lower than other areas for all children. The CNA seems to be a useful tool to establish baseline level of ability, to monitor changes in development, and to provide constructive feedback to parents and professionals in order to plan appropriate intervention.**

## What is holoprosencephaly?

**Holoprosencephaly (HPE) is a midline developmental brain disorder with mild to severe agenesis of the brain. Incomplete cleavage of the embryonic forebrain into cerebral hemispheres results in an under specified ventricular system, fused frontal lobes, and is often associated with microcephaly and hydrocephaly (Kinsman, et. al. 2000, for review).**

# Characteristics

- **HPE is associated with cranio-facial malformations including abnormalities such as: cebocephaly, hypotelorism, cleft lip or palate, and single maxillary incisor.**
- **Children with HPE have severe motor impairments with varying degrees of hypertonicity or hypotonicity.**
- **Few of these children develop functional expressive language.**
- **Incidence of HPE in the general population is reported to be 5-12/100,000 live births.**

# Etiology

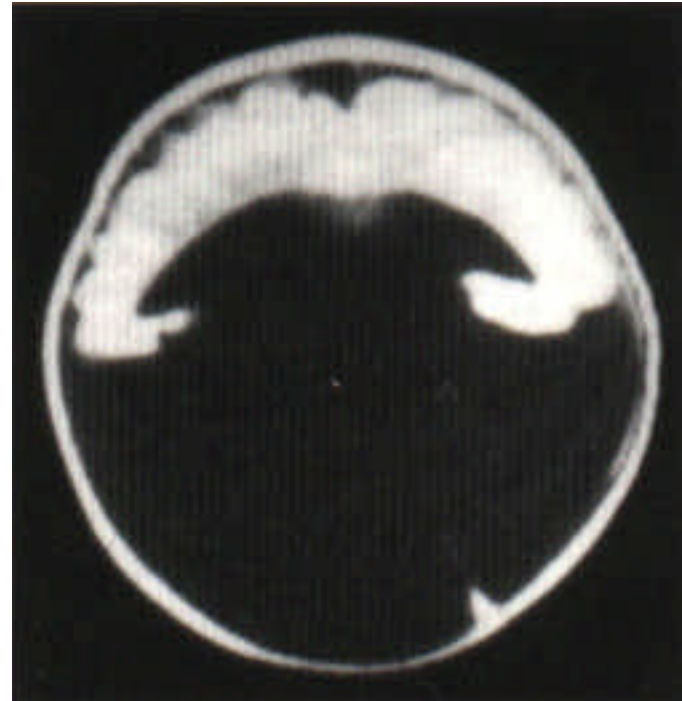
- **A complex signaling pathway involving many genes, including Sonic Hedgehog (SHH), ZIC2 and SIX3, is believed to be affected in HPE (Ming & Muenke, 1998; Brown et al, 1998).**
- **Teratogens and deficits in cholesterol biosynthesis can also impact this pathway (Wallis and Muenke, 1999).**
- **Possible environmental factors include maternal diabetes, infections during pregnancy or drug use during pregnancy.**
- **Chromosomal abnormalities have been identified in approximately 40% of live births of children with HPE.**

# Classification

Currently, classification of HPE is based on three levels of brain agenesis:

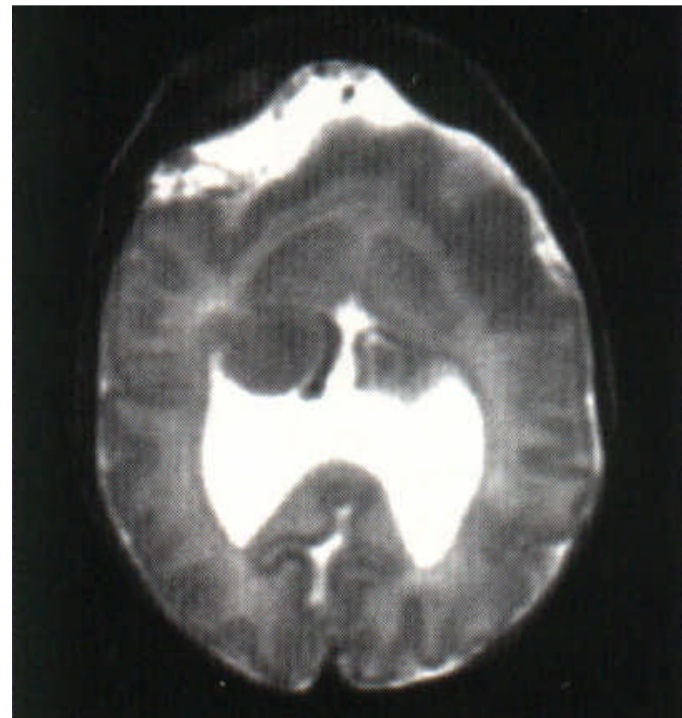
## Alobar

No significant interhemispheric fissure; single ventricle; fused thalamic nuclei and basal ganglia.



## Semi-Lobar

Hemispheres partially formed in posterior with single ventricle cavity; incomplete cleavage of anterior brain structures; small dorsal cyst possible.



## Lobar

well-developed interhemispheric fissure, but incomplete cleavage of structures; dysmorphic ventricles; posterior corpus callosum and third ventricle often formed.



# Goals

- To develop a tool that assesses the cognitive and early language abilities (0-24 months) of children with HPE and other severe motor and language impairments.
- To generate a meaningful description of skills attained in specific cognitive and language domains.
- To give caregivers and professionals feedback regarding the child's developmental range and abilities, and to assist them in determining appropriate therapeutic goals.
- To monitor gains (or losses) in each child's development over time.
- To determine relationship of cognitive performance with structural hierarchy.

# Participants

	Alobar	Semi-Lobar	Lobar	Total
Male	0	8	2	10
Female	1	6	1	8
Total	1	14	3	n=18
Mean Age	64 mos.	24 mos.	15 mos.	

# Methods

## [The Carter Neurocognitive Assessment \(CNA\)](#)

- **A 103 item scale requiring minimal motor and expressive language abilities.**
- **Tasks are presented in hierarchical order with age approximations based on commonly recognized developmental milestones.**
- **Areas assessed include:**
  - Social Awareness**
  - Auditory Attention/Comprehension**
  - Visual Attention**
  - Vocal Communication**
- **Skills are elicited, observed spontaneously, or reported by caregivers.**

**See following pages for excerpts from the CNA.**

### Excerpts from Social Awareness Scale (29 items)

Age in Months	Score +/-, PR	Behaviors to be elicited, observed, or reported.
1		<b>S4. Comforted by being held</b> (quiets or stills when upset)
2		<b>S8. Smiles or vocalizes in response to adult</b> attention, smile, or voice ( <i>Also credit S7</i> )
6		<b>S16. Anticipates</b> an event (e.g., approach and lightly tickle child with a stuffed toy or your fingers to elicit a pleasurable reaction. Repeat this sequence four times. Note the child's reaction, as he/she is approached but not yet in contact with you or the toy. There should be a response to the pause in the activity, not a general increase in excitability due to the sequence. Incidental observation is acceptable.)
8		<b>S18. Makes social reference</b> (child directs gaze from an object or environmental event, to an adult, and maybe, but not necessarily, back to object or event, as if to make a comment. <i>Also credit S2</i> )

### Excerpts from Visual Attention Scale (27 items)

Age in Months	Score +/-, PR	Behaviors to be elicited, observed, or reported.
1		<b>V5. Horizontal tracking</b> (suspend object, such as a tennis ball or small toy, in front of child while standing to the side, can jiggle it to get attention, slowly move to left then right, then back to mid-line. Credit if child's eyes follow object in both directions - can attract child's attention back to object. Can repeat twice if necessary. <i>Also credit V3, V4</i> )
3		<b>V10. Alternates glance between 2 objects</b> (present 2 objects in child's field of vision simultaneously, credit if child alternates glance between objects. Can score from picture presentations.)
9		<b>V19. Looks for object in 1 of 2 places</b> (hide object under 1 of 2 cups, call child so that he or she shifts gaze from cup, ask, "Where is it?" look in cup which child looks at, show correct place if necessary, repeat twice. Child must look

		correctly to left and right. Can use food like goldfish or cheerios)
<b>18</b>		<b>V25. Maintains attention</b> to a person or an object for <b>3 minutes</b> (using toy or book engage child in simple play activity, or child can play independently, not watching video. <i>Also credit V15, V23</i> )

**Excerpts from Auditory Attention/Comprehension Scale (14 items)**

<b>Age in Months</b>	<b>Score +/-, PR</b>	<b>Behaviors to be elicited, observed, or reported.</b>
<b>1</b>		<b>A1. Responds to sounds</b> (e.g., startles to loud noise, activity arrested when approached by sound, quiets to sound or voice, looks around)
<b>6</b>		<b>A5. Recognizes 2 words: emphasized</b> (e.g., “daddy,” “bye-bye,” “mama,” or child’s own name; child responds when own name is called or specific object or person is named, child may perk up and look for object or person when named, can be a similar reaction to similar sounding words)
<b>8</b>		<b>A7. Follows any direction</b> with no gestural cue (e.g., “Where’s mama/light/bottle?”, “Wave bye bye”, “Look up” etc., ask caregiver for appropriate request or routine, caregiver can elicit)

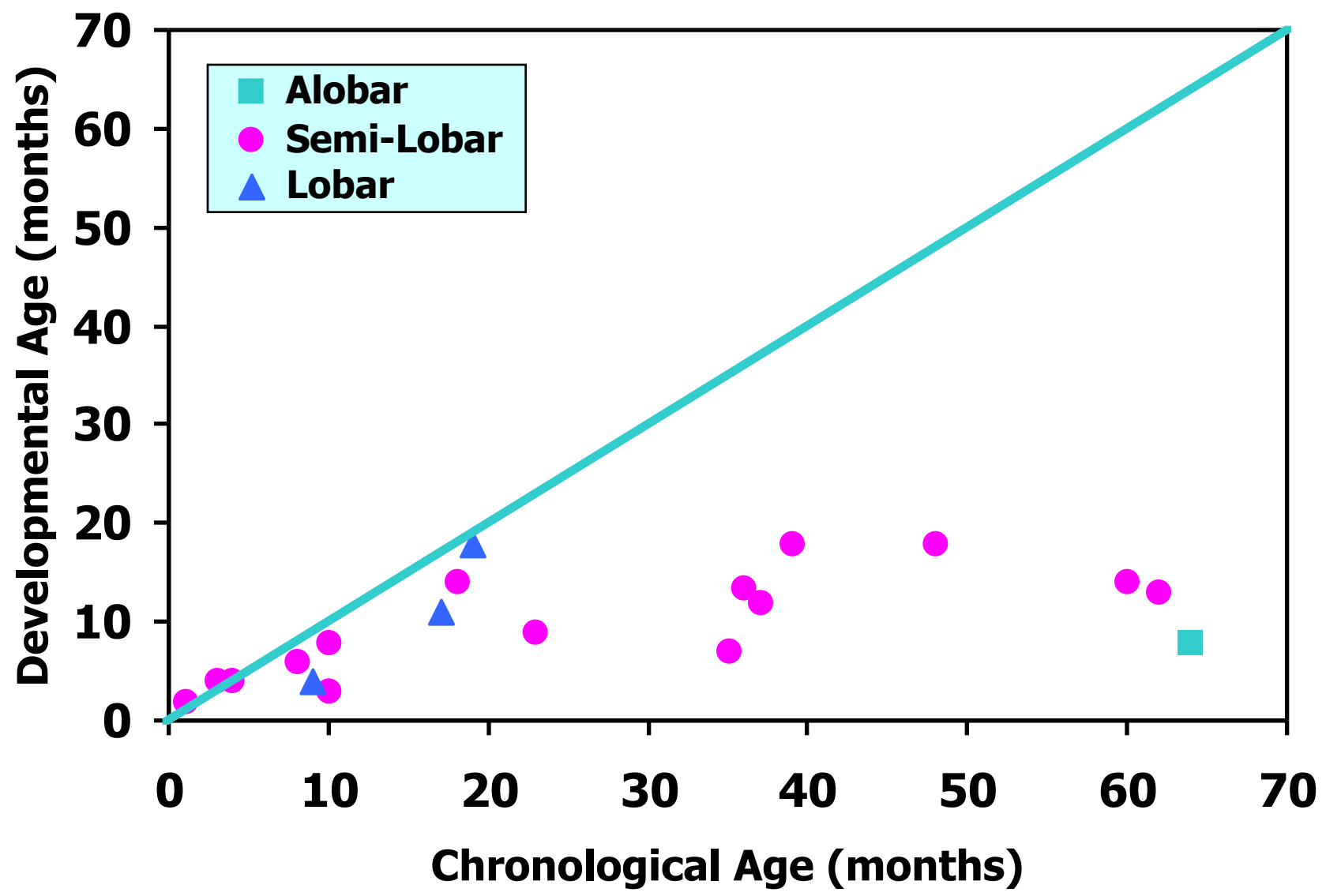
**Excerpts from Vocal Communication Scale (33 items)**

<b>Age in Months</b>	<b>Score +/-, PR</b>	<b>Behaviors to be elicited, observed, or reported.</b>
<b>2</b>		<b>C4. Vocalizes when adult smiles or speaks</b>
<b>2</b>		<b>C5. Uses intonation to express feelings</b> (e.g., hunger, pain, social attention)
<b>4</b>		<b>C11. Vocalizes 1 consonant</b> (can try to elicit through imitation - ask caregiver for suggestions)
<b>8</b>		<b>C15. Reduplicative babbling</b> (credit for any chain containing at least two consonants and two vowels, e.g., “baba”, “baga”. <i>Also credit C11</i> )

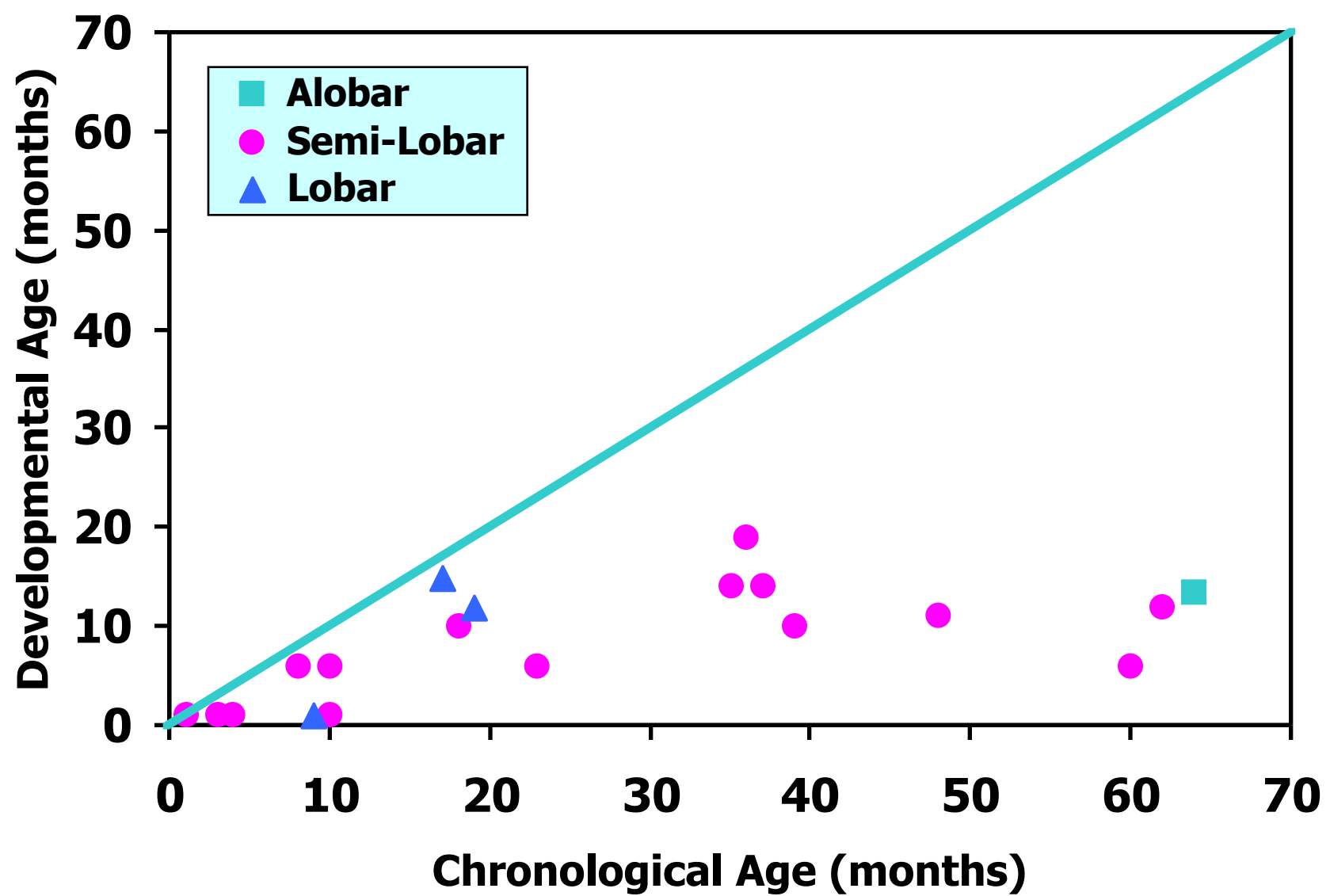
All CNA items 2001

# Analysis

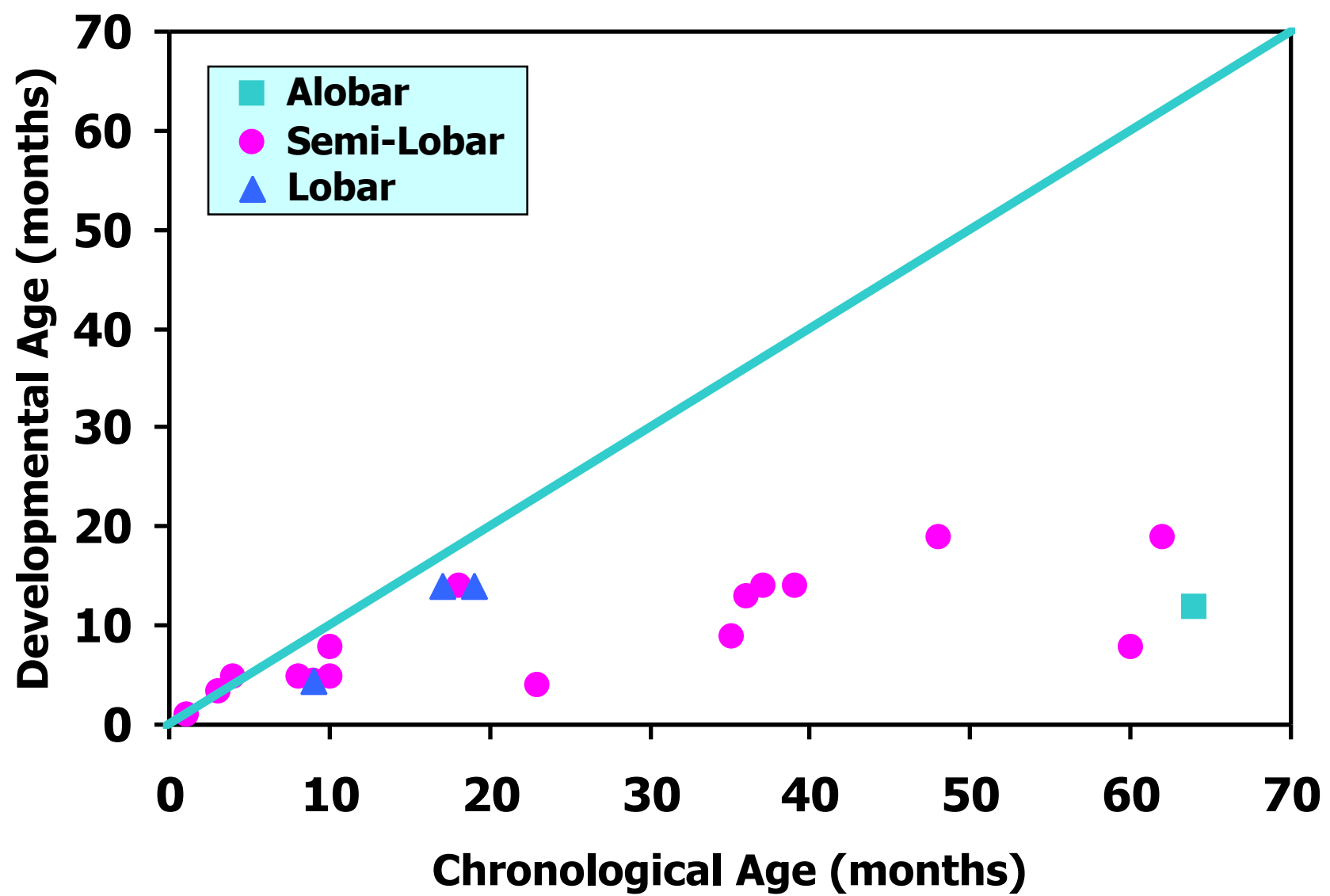
## Social Awareness Scale



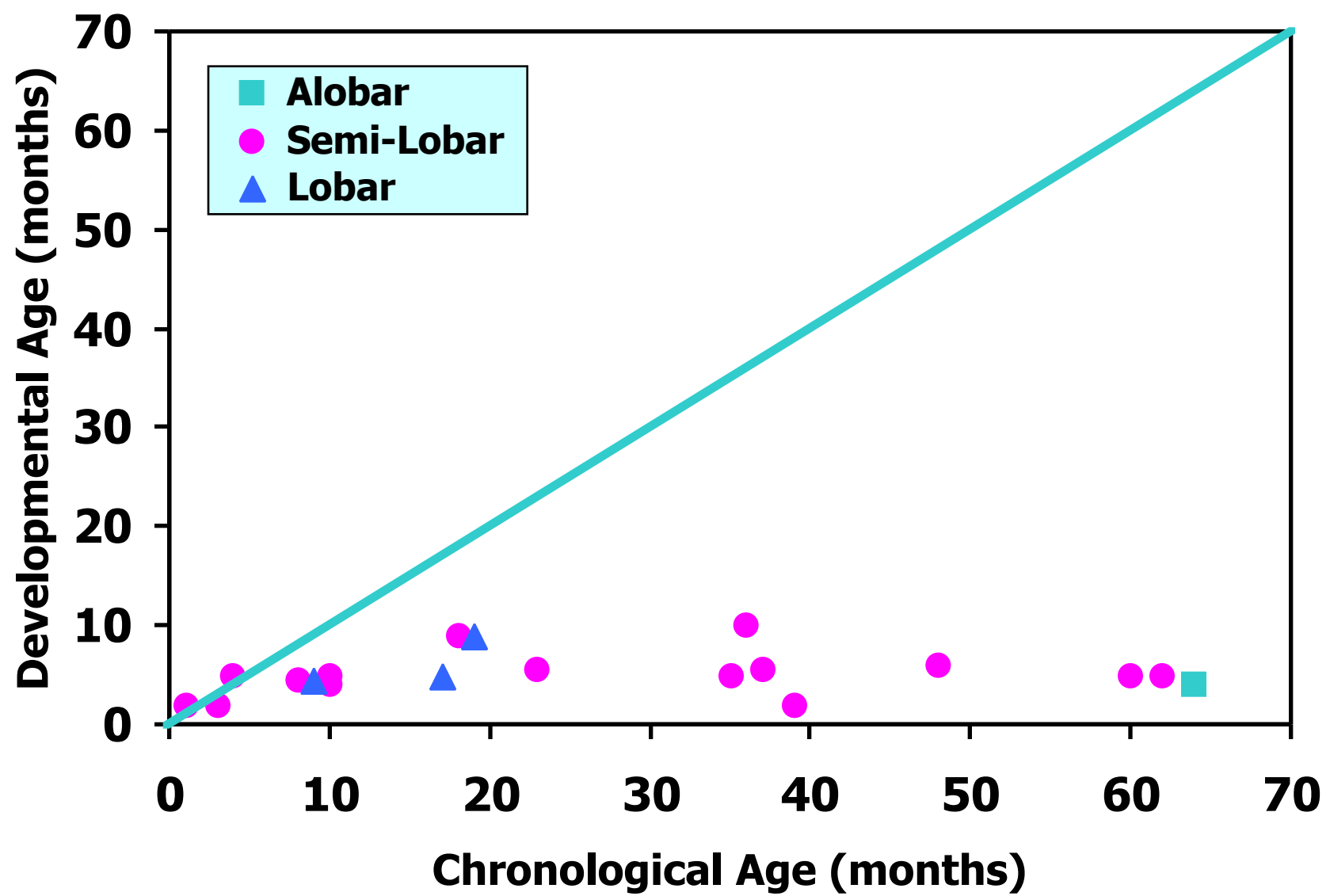
## Visual Attention Scale



### Auditory Attention/Comprehension Scale



### Vocal Communication Scale



# Results

- **Performance was variable in the areas of Social Awareness, Visual Attention, and Auditory Comprehension regardless of chronological age or HPE classification.**
- **Although not reflected in our results, the majority of children with HPE demonstrated strong social engagement skills with regard to social referencing and joint attention.**
- **For the children under one year of age, skills in these areas reflected a more age-appropriate trajectory. However, progress then appeared to slow in the chronologically older children and then plateau. Future longitudinal assessments and examination of individual trajectories will allow us to evaluate whether this is, indeed, the case.**
- **Vocal communication was the area of greatest weakness for all children. In general, scores for vocal communication plateau at the 4-6 month level of development. While all of the children produced vowel-like vocalizations, one child was observed to produce consonant-vowel combinations**
- **Inter-rater reliability ranged from 79-97% accuracy with a mean of 88% accuracy (N= 100 items co-scored).**

# Conclusions

- **The Carter Neurocognitive Assessment (CNA) provides information about specific skills that children with HPE have achieved at the 0-2 year level of development.**
- **The CNA allows us to provide parents and professionals with positive, constructive feedback regarding areas of strength and weakness in order to establish intervention goals.**
- **The CNA can be used to establish a baseline of behavioral function and to monitor change in development over time for individual children.**
- **At this time, structural classification of HPE does not appear to be related to specific patterns of cognitive performance. This is an unexpected result as it had been thought that the cognitive continuum varied according to the severity of brain agenesis.**

# Future Direction

**As we collect more CNA data on children with HPE and others with severe motor and expressive language impairments, we will:**

- **Modify the CNA to include only the items that provide the most useful information for monitoring progress**

- **Generate items that might provide information about the quality of response**
- **Re-evaluate children in the study to examine individual trajectories (as well as item-by-item analysis of progress)**
- **Add items to help identify children who have the necessary prerequisites to begin training for an augmentative or alternative system of communication**
- **Combine our data with that of the other Carter Center sites also piloting the CNA**
- **Begin to relate these CNA profiles to the structural and functional information we receive from imaging and electrophysiological measures currently being used at the Carter Centers**

## Acknowledgments

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