HOLOPROSENCEPHALY

THE CARTER CENTERS FOR BRAIN RESEARCH IN HOLOPROSENCEPHALY AND RELATED MALFORMATIONS

THE INTERNATIONAL HPE REGISTRY

The Carter Centers have established the International HPE Registry as a way to create a comprehensive list of patients diagnosed with holoprosencephaly not only in the United States but also worldwide. Holoprosencephaly (HPE) is a medical condition that is not well understood. The International HPE Registry provides research scientists with a more accurate understanding of the number of HPE cases diagnosed and serves as a vehicle to collect clinical information to share with other researchers. It also provides a means for families to contact other families and allows families to receive information regarding potential research initiatives in which they may want to participate.

Even if you have lost a child diagnosed with HPE, you may still register your child. Please complete the form according to what your child was able to accomplish and his/her limitations. We appreciate information on every child with HPE.

CONSENTS:

By registering my child in The Carter Centers' International HPE Registry, I give my permission to have my family's name and address and my child's clinical information included in the registry. Your permission to add your child's health information to the registry is greatly appreciated, but it is completely voluntary. If you choose not to allow us to add your child's health information to the registry, this will not affect your care at any of The Carter Centers.

I further give my permission to: (Check all that apply)

- ☐ **Mailing List Consent:** be included on the mailing list for items containing information pertaining to HPE.
- ☐ **Family Network Consent:** have my family's name and address shared with other families and caregivers affected by HPE.
- ☐ **Research Opportunities Consent:** receive information from the registry regarding potential research initiatives regarding HPE in which I may or may not wish to participate.
- ☐ **Researcher Contact Consent:** have my family's name and address provided to researchers requesting such information from the registry. I understand this may bring direct requests to me from researchers for participation of my family in other research projects.
- ☐ **Radiology Consent:** have information and images from my child's ultrasound, CAT scan or MRI entered into the database and shared with other researchers or used in educational presentations. I understand my child's name and personal information will be removed from anything that is shared with others.

I give my permission for the following information to be shared with other researchers:
(Please choose only one)

- ☐ **Clinical information only (NOT name and address)**
- ☐ **Name and address only (NOT clinical information)**
- ☐ **Clinical information AND name and address**

Signature: _____

Date: _____ Relationship to patient: _____

No matter what options you choose, you reserve the right to contact researchers independently and directly. You may also cancel or revise this release at any time by submitting a written notice to:

THE CARTER CENTERS FOR BRAIN RESEARCH IN
HOLOPROSENCEPHALY AND RELATED MALFORMATIONS

Texas Scottish Rite Hospital for Children • 2222 Welborn Street, Dallas, TX 75219

Phone: (214) 559-8411 Fax: (214) 559-8383 E-mail: hpe@tsrh.org

THE CARTER CENTERS INTERNATIONAL HPE REGISTRY

Today's date: __ __/__ __/__ __ __ __
Mo. Day Year

Registry No. _____

(For office use only)

Patient's full name (first/middle/last): _____

Date of birth: __ __/__ __/__ __ __ __
Mo. Day Year

Date of death (if applicable): __ __/__ __/__ __ __ __
Mo. Day Year

Birthplace (City/State/Country): _____

Gender: ☐ Male ☐ Female

Ethnicity: ☐ Black/African-American ☐ Hispanic/Latino ☐ Other: _____
☐ Asian ☐ Native American
☐ White/Non-Hispanic ☐ Pacific Islander

Address: _____ Apt. #: _____

City: _____ State: _____ Postal Code: _____ Country: _____

Home phone: ( ) _____ Fax: ( ) _____

Mom's work phone: ( ) _____ Dad's work phone: ( ) _____

E-mail: _____ Alternate phone: ( ) _____

Biological mother's name (first/middle/last): _____

Date of birth: __ __/__ __/__ __ __ __
Mo. Day Year

Date of death (if applicable): __ __/__ __/__ __ __ __
Mo. Day Year

Birthplace (City/State/Country): _____

Biological father's name (first/middle/last): _____

Date of birth: __ __/__ __/__ __ __ __
Mo. Day Year

Date of death (if applicable): __ __/__ __/__ __ __ __
Mo. Day Year

Birthplace (City/State/Country): _____

Name of legal guardian (first/middle/last): _____

Relationship to patient: ☐ Biological Parent ☐ Adoptive Parent ☐ Foster Parent ☐ Grandparent
☐ Family Member ☐ Other _____

Alternate contact name: _____ Phone: ( ) _____

Relationship to patient: _____

Primary language spoken in the household: _____

THE CARTER CENTERS INTERNATIONAL HPE REGISTRY

DIAGNOSIS AND TESTS

1. Has your child's HPE diagnosis been confirmed by a brain scan?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, check all types of scans your child has had:

- ☐ CT
- ☐ MRI
- ☐ Cranial ultrasound
- ☐ Prenatal MRI
- ☐ Prenatal ultrasound
- ☐ Autopsy

2. What type of HPE does your child have?

- ☐ Alobar
- ☐ Semilobar
- ☐ Lobar
- ☐ Middle Interhemispheric Variant (MIH)
- ☐ Unsure

3. When was your child diagnosed with HPE?

- ☐ During pregnancy
- ☐ Within the first month after birth
- ☐ Before the age of 1 year
- ☐ After the age of 1 year
- ☐ Unsure

4. Has your child had chromosomal testing?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, what were the results?

- ☐ Normal results
- ☐ Waiting for results
- ☐ Abnormal results (please describe): _____

5. Has your child had a gene test for HPE?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, what were the results?

- ☐ Normal results
- ☐ Waiting for results
- ☐ Abnormal results

If abnormal, what were the results?

- ☐ SHH
- ☐ ZIC2
- ☐ SIX3
- ☐ TGIF
- ☐ Other: _____

MEDICAL PROBLEMS

6. Does your child have any deformities of the face or head?

- ☐ No deformities
- ☐ Small head (microcephaly)
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Nose (single nostril, flat nose, etc.): _____
- ☐ Teeth (single incisor, missing teeth, etc.): _____
- ☐ Eyes (close together, wide apart, missing or central eye, etc.): _____
- ☐ Ears (low-set, absent, etc.): _____
- ☐ Other facial deformities (please describe): _____

7. Does your child have hydrocephalus (extra water in the brain)?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, does your child have a Shunt? ☐ No ☐ Yes

8. Has your child ever had convulsions/seizures/epilepsy?

- ☐ Unsure
- ☐ No
- ☐ Occasionally (fewer than 3 seizures in his/her lifetime)
- ☐ Some (more than 3 seizures in his/her lifetime)
- ☐ Often (difficult to control)

About how many seizures does your child have per ____ day ____ wk. ____ mo.

What was the approximate date of your child's last seizure? _____

Has your child taken or is your child currently taking medication to control seizures?

- ☐ No, never has taken medicine for seizures
- ☐ Yes, took medicine in the past, but not now.

Name of medicine(s): _____

- ☐ Yes, is currently taking medicine for seizures.

Name of medicine(s): _____

Please describe any other treatment for seizures:

- ☐ Vagal Nerve Stimulator (VNS)
- ☐ Ketogenic Diet
- ☐ Other: _____

9. Has your child been diagnosed with Diabetes Insipidus?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, how are you managing it?

- ☐ Managed with diet and fluids
- ☐ Managed with medication and sodium levels are well controlled
- ☐ Managed with medication, but sodium levels are NOT well controlled

10. Has your child been diagnosed with Growth Hormone Deficiency?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, how are you managing it?

- ☐ No medication needed
- ☐ Receives hormone replacement medication

11. Has your child been diagnosed with Cortisol Hormone Deficiency?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, how are you managing it?

- ☐ No medication needed
- ☐ Receives hormone replacement medication

12. Has your child been diagnosed with thyroid problems?

- ☐ Unsure
- ☐ No
- ☐ Yes

If yes, how are you managing it?

- ☐ No medication needed
- ☐ Receives hormone replacement medication

13. Does your child have feeding problems?

- ☐ No
- ☐ Some difficulty swallowing or chewing
- ☐ Has feeding tube

14. Does your child have sleeping problems?

- ☐ No
- ☐ Yes

If yes, how are you managing it?

- ☐ Occasional problems, but no medication needed
- ☐ Medication required – responds well
- ☐ Severe sleep problems, but medicine doesn't help

DEVELOPMENT

15. When was your child born?

- ☐ Full-term (at least 37 weeks gestation)
- ☐ Premature (less than 37 weeks gestation) About how many weeks early? _____

16. What is your child's current age? ☐ wks _____ ☐ months _____ ☐ yrs _____

17. Does your child have developmental delays?

- ☐ No
- ☐ Yes

18. What can your child do? (Check all that apply)

- ☐ Walks independently
- ☐ Walks with assistive devices (crutches, walker)
- ☐ Walks with parental assistance
- ☐ Stands independently
- ☐ Stands with support, cruises along furniture
- ☐ Crawls
- ☐ Sits independently
- ☐ Rolls over
- ☐ None of the above

19. How does your child reach for or handle objects? (Check all that apply)

- ☐ Normal for age
- ☐ Able to transfer objects from one hand/place to the other
- ☐ Able to reach for and attain objects
- ☐ Able to "bat" at objects
- ☐ Able to hold objects when placed in hands
- ☐ None of the above

20. Does your child speak? (Check all that apply)

- ☐ Normal for age
- ☐ Full sentences
- ☐ Single words
- ☐ Consonant sounds ("ma-ma," "da-da," "ba-ba," etc.)
- ☐ Vowel sounds ("baby talk" or babbling/gurgling)
- ☐ None of the above

21. Does your child have motor problems? (Check all that apply)

- ☐ No motor problems
- ☐ Coordination or balance problems
- ☐ Increased muscle tone (hypertonia), spasticity, dystonia
- ☐ Decreased muscle tone (hypotonia), floppy or weak muscles

22. Does your child receive treatment for motor problems?

- ☐ No
- ☐ Yes

If yes, check all types of treatment your child has received:

- ☐ Physical/Occupational Therapy
- ☐ Braces or splints
- ☐ Botox injections
- ☐ Surgery
- ☐ Baclofen pump
- ☐ Medicines (please list): _____
- ☐ Other: _____

23. Has your child been diagnosed with any additional syndromes/medical conditions/malformations?

- ☐ No
- ☐ Yes (please list): _____

FAMILY HISTORY

24. Does anyone else in your family have developmental delays, facial abnormalities (cleft lip/palate, close-set eyes, single front tooth), endocrine (hormone) problems, chromosomal abnormalities, gene mutations, or frequent miscarriages, etc.?

- ☐ No
- ☐ Yes

Relationship to child: _____ Medical condition: _____
(use as much space as necessary to list family members and conditions)

OTHER

25. Is there any other information or concerns you would like to share?

Thank you very much for sharing information about your child with The Carter Centers for Brain Research in Holoprosencephaly and Related Malformations.