Approved by the Nemours IRB: Valid from: 02/16/2015 through 02/15/2016 IRBnet 83142 V 2-2015

Abbreviated Study Title: **Primordial Registry at A.I. duPont Hospital for Children**

Nemours
Informed Consent for Participation in an Observational / Non-Interventional Research Study

You have been asked to be in a research study. This form explains the research, your rights as a research participant, and any responsibilities that you may have as a result of your participation. You should understand the research study before you agree to be in it. ***You will receive a copy of this form. Read this permission form carefully. You may also talk with your family or friends about it. A research team member will answer any questions you have before you make a decision.***

1. WHAT IS THE TITLE OF THE STUDY?

Primordial Registry at A.I. duPont Hospital for Children

2. WHO IS IN CHARGE OF THE STUDY AT NEMOURS?

If you have a question, complaint, or problem related to the study, you can call the investigator anytime at the numbers listed below.

	Nemours - WIL
Principal Investigator	Michael B. Bober, MD, PhD, AIDHC-Medical Genetics, 302-651-5916
Co-Investigator(s)	William Mackenzie, Tim Niiler, Divya Moodalbail, Joshua Zaritsky, Frances Zappalla, Dinesh Choudhry, Divya Dixit, S. Charles Bean, Magee DeFelice, Vinay Kandula
Study Coordinator(s)	Angela L. Duker, MS, CGC AIDHC-Medical Genetics, 302-651-4181
Address	Nemours/Alfred I. duPont Hospital for Children 1600 Rockland Rd Wilmington, DE 19803

3. WHO SHOULD RESEARCH PARTICIPANTS CONTACT ABOUT THEIR RIGHTS?

If you have questions about your rights as a research participant, what to do if you are injured, if you would like to offer input or obtain information, or if you cannot reach the investigator or want to talk to someone else who is not involved with this research, you may contact the persons listed below.

Carlos Rosé, MD, CIP, Chairperson, Nemours IRB 1 at 302-651-5970
Paul Garfinkel, MSH, CIP, Director, Nemours Office of Human Subjects Protection at 904-697-4023
Email address: NOHSP@nemours.org

4. WHAT IS THE PURPOSE OF THE STUDY?

You are being invited to participate in a data collection effort called a registry. The goal of this registry is to collect information on individuals with Microcephalic Osteodysplastic Primordial Dwarfism Type II (also called MOPDII) and other forms of microcephalic primordial dwarfism. The study team hopes to learn more about these conditions and improve the care of people with them by establishing this registry.

5. WHO IS SPONSORING OR PAYING FOR THE STUDY?

The Potentials Foundation is a sponsor of this study, by ensuring that all participants will have no cost associated to mailing information to Nemours. If needed, a pre-paid mailing envelope can be forwarded to participants by a representative from the Potentials Foundation.

6. WHO CAN BE IN THE STUDY?

Individuals with MOPDII, MOPDI/III, Meier-Gorlin syndrome, and unclassified or closely related forms of microcephalic primordial dwarfism as diagnosed by a physician are eligible for this registry.

7. HOW MANY OTHER PEOPLE WILL BE IN THE STUDY?

Approximately 100 individuals with MOPDII (and/or other forms of primordial dwarfism) will be enrolled in the study.

8. HOW LONG WILL PARTICIPATION IN THE STUDY LAST?

This study is limited to chart review. There will be no additional visits or time in clinic because of your participation in this registry. The study team believes participation will last for at least 5 years.

9. WHAT ARE THE RESEARCH PROCEDURES?

This study involves only the collection and storage of data extracted from the medical record. There are no special procedures, visits, or expectations of you as a result of participation in this registry. You will not be asked to have any specific testing for the sole purposes of research.

Patient at AIDHC

If you have had lab work or imaging studies performed at AIDHC these records may be reviewed to gain additional information about this disease. Records that may be reviewed as a part of this study include but may not be limited to x-rays of teeth and other bones, results of routine blood and urine tests, results of genetic testing and neurovascular imaging (images of blood vessels in the brain).

Patient outside of AIDHC

You may have heard about this study by viewing information at the Potentials Foundation website (www.PotentialsFoundation.org), Walking with Giants Foundation (www.walkingwithgiants.org), Geneclinics.org, or the Skeletal Dysplasia program site within the Nemours organization website (http://www.nemours.org/hospital/de/aidhc/service/skeletal.html). You downloaded the study information, medical records release, and informed consent forms from one of these sites. You reviewed the information and contacted the study team to have any questions or concerns answered to your satisfaction. Dr. Michael Bober was contacted at (302) 651-5916 or mbober@nemours.org to enroll in the study. You may make records and/or x-rays from other hospitals and doctor's offices available to the study team if you wish. Records that may be reviewed as a part of this study include but may not be limited to x-rays of teeth and other bones, results of routine blood and urine tests, results of genetic testing and neurovascular imaging (images of blood vessels in the brain).

The following will be mailed to the following person and address:

Michael B. Bober, MD, PhD
Alfred I duPont Hospital for Children
Medical Genetics
1600 Rockland Road
Wilmington, DE 19803
302-651-5916

Items mailed:

- ❑ 1 copy of informed consent with original signature
- ❑ 1 copy of medical release form with original signature
- ❑ Medical records
- ❑ Medical images (x-ray and/or MRI)

By agreeing to be in the registry, you allow study team members to review your medical records and collect information about your condition.

10. WHAT ARE POSSIBLE RISKS OF BEING IN THIS STUDY?

The risks involved in this study are the same as the risks you would ordinarily encounter in daily life or during a routine physical examination. This research is observational which means that there is no change to any treatment that you may be receiving. The most common risk of participation in a registry is the chance that your private information (ex: insurance coverage, status of health, treatments prescribed) may be used for purposes other than those described in this permission form. Loss of privacy may affect your insurability, employability, or may result in labeling of a person with a chronic illness. The protection of your confidential information is described in Section 16 of this form.

Genetic Research

A Federal law, called the Genetic Information Nondiscrimination Act (GINA), reduces the risk of discrimination by health insurance companies, group health plans, and most employers based on your child's genetic information.

11. WHAT ARE POSSIBLE BENEFITS OF BEING IN THIS STUDY?

There is no direct benefit to you from participating in this registry. Participation in this registry may help doctors to better understand the disease process and identify a standard of care for individuals with MOPDII and related forms of microcephalic primordial dwarfism.

12. IS BEING IN THE STUDY VOLUNTARY?

Being in this study is totally voluntary. Anyone who takes part in the study can stop being in it at any time. There will be no change to your usual medical care if you decide not to be in the study or decide to stop being in the study. No one will be angry with you, or treat you any differently than before you were asked to be in the study.

In the event that you withdraw from the study, the study doctor may ask your permission to continue study follow-up, and all clinical data related to the study may continue to be collected from your medical records. You may ask the researcher to destroy your information. Your request must be in writing. The researcher will tell you if this is possible. There may be legal reasons for keeping your information or samples.

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13. WHAT ARE THE COSTS OF BEING IN THIS STUDY?

There are no direct costs to families for participating in this registry if you are a patient at Alfred I duPont Hospital for Children. If you are not a patient at Alfred I duPont Hospital for Children, you may incur costs related to obtaining and mailing medical records if you choose this option. Costs related to your regular care will still be your or your insurance company's responsibility.

14. WILL I BE PAID FOR BEING IN THIS STUDY?

Participants will not be paid for participating in this study. No arrangement exists that would allow participants to share in any profit generated from this study or future research.

15. WILL I BE TOLD OF ANY NEW INFORMATION THAT MIGHT AFFECT MY WILLINGNESS TO STAY IN THE STUDY?

Any new information that may change your mind about being in this study will be given to you. A committee called the Institutional Review Board (IRB) will review this study at least once per year. If the IRB finds that there is new information that you should know about while you are taking part in this study, the IRB will ask the study doctor to tell you about it. You may be asked to sign a new version of this form after discussing the new information with a member of the research team.

16. WHAT INFORMATION ABOUT ME WILL BE USED OR DISCLOSED? (AUTHORIZATION TO USE AND / OR DISCLOSE PROTECTED HEALTH INFORMATION)

Identifiable health information about you will be used by Nemours researchers and may be given to people outside of Nemours for this research. This is done to conduct the research study, to monitor the safety of research participants and for auditing. Federal law requires us to tell you about, and get your approval for research use and disclosure of health information that includes "identifiers" that can connect the health information to you. (Names, initials, date of birth, addresses, phone numbers, and social security numbers are examples of identifiers.) This Identifiable health information is called Protected Health Information (PHI).

Use of Health Information by Nemours Staff

The health information that will be used within Nemours includes all data collected for this study, as described in the section called "What Are the Research Procedures?"

Your identity will be protected as much as possible. Nemours protects your health information by storing records in files or computers that can only be used by authorized Nemours staff.

The people within Nemours that may use this health information include:

- The investigators listed on the first page of this permission form and their staff,
- The Nemours Institutional Review Board (IRB). (The IRB is a group of people that reviews research activities. The IRB is responsible for the safety and rights of research participants), and;
- Nemours internal audit staff.

Disclosure of Health Information to Others

Identifiable health information will be disclosed (given) to the following individuals or groups:

- Carol Wise, PhD, Co-Investigator (Genetics, Texas Scottish Rite Hospital for Children)
- Andrew Jackson, PhD, MRCP, Team Member (Consultant Clinical Geneticist, Western General Hospital, Edinburgh, Scotland)

- Jennie Murray, MBBS, Team Member (Clinical Genetics Registrar, Western General Hospital, Edinburgh, Scotland)
- Kenneth Rogers, PhD, ATC,Team Member (AIDHC-Orthopedics)
- Grant Eldridge, Team Member (AIDHC-Research)

The PHI that will be disclosed (given) to people or groups outside of Nemours for research purposes are listed in the table below*: (Use the Table tools to add rows as needed. Delete the rows that do not apply.).*

Type of Identifiable Health Information:	***Disclosed:***
History and Physical	☒
Results of Procedures	☒
X-Ray Reports	☒
Surgery Reports	☒
Genetics Studies	☒
Demographics (information about race, ethnicity, gender, age)	☒
Questionnaires	☒
Other: Medical Images (x-ray or MRI or ultrasound or CT)	☒

Limits on Protection of Privacy and Confidentiality

Only health care organizations have to follow laws and rules about protecting the privacy of health information. If health information containing peoples' identities is given to other kinds of companies or organizations, they are not required by law to safeguard the privacy and confidentiality of that information. Nemours expects these companies and organization to protect the privacy and confidentiality of research participants, but it is not possible for Nemours researchers to assure that this happens.

Government agencies that may look at records for this research study, including the above health information, include:

- The U.S. Food and Drug Administration
- The U.S. Department of Health and Human Services
- Other agencies of State and local government as required by law
- Governmental agencies in other countries

The research results may be presented at scientific meetings or in print. Participants' identities will not be disclosed in those presentations.

17. SIGNATURES:

I am making a decision whether or not to participate in this study. I have read this form, or had it read to me in a language that I understand. I have been given enough time to make this decision. I have asked questions and received answers about things I did not understand. I willingly consent to participate in this study. By signing this form, I am not giving up any rights to which I am entitled under law.

I understand that:

- I can withdraw consent for participation in this study and for the use and / or disclosure of my PHI by contacting the person in charge of the study listed on the first page of this form.

- The use and / or disclosure of my PHI will stop after Nemours receives the withdrawal notice. Information that is used or disclosed before the withdrawal may still be used.
- Unless I withdraw consent, the use and / or disclosure of my PHI described in this form will not have an expiration date.
- My PHI may be disclosed again by the person or organization (other than Nemours) that receives it. If this happens, Federal or state law may not protect the information.
- I have the right to refuse to sign this permission form.
- If I refuse to sign this consent form, I will not be allowed to be in this research study.
- I have the right to ask Nemours to tell me who has received my protected health information.
- I have the right to revoke my permission for the use and disclosure of my health information at any time, which would end my participation in this study.
- I will receive a signed and dated copy of this form.

My signature indicates that:

- I give my consent to participate in the research study described in this form.
- I give the researchers and Nemours permission to use and /or disclose my individually identifiable health information for this research study as described in the section on use and disclosure of PHI.

Name of Participant (**Print)** Date

Signature of Participant Date

I the undersigned, certify that to the best of my knowledge the participant signing this informed consent form had the study fully and carefully explained and that he / she understands the nature, risks and benefits of participation in this research study.

Name of Person Obtaining Permission (**Print**)
(Investigator or Designee)

Signature of Person Obtaining Permission
(Investigator or Designee) Date

A copy of the signed form was provided to Participant ☐ Date