ADOLESCENT PARTICIPANT ASSENT FORM

(Age 14-18 years)

The International Pyridoxine-Dependent Epilepsy Registry

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Introduction

I have been invited to participate in this study because I have Pyridoxine dependent epilepsy.

Your participation is voluntary

My participation in this study is entirely voluntary and it is up to me to decide whether or not to participate in this study. Before I decide, it is important for me to understand what the research involves. This document will tell me about the study, why the research is being done, what will happen to me during the study and the possible benefits, risks and discomforts.

If I decide to participate, I will be asked to sign this form. If I do decide to take part in this study, I am still free to stop my participation at any time and without giving any reasons for my decision.

Please take time to read the following information carefully and to discuss it with your family, friends, and doctor before you decide.

For any questions related to the study, please contact your doctor.

What is the purpose of this study?

The doctors are collecting medical data from people who have Pyridoxine Dependent Epilepsy. This information will be put into a database that doctors can use to improve medical care for people with Pyridoxine Dependent Epilepsy. For example, using a large amount of data, they can find out which type of treatment works best for different people with the same disease and whether the same treatment could be used for different patients with the same disease.

Why are we asking you to participate?

I am being asked to participate in this research study because I have a confirmed diagnosis of Pyridoxine dependent epilepsy (PDE) due to antiquitin (*ATQ)* deficiency. PDE is a rare form of epilepsy characterized by seizures that begin in infancy or, in some cases, before birth. Researchers recently discovered that PDE is caused by a defect in a gene (*ATQ*) that affects the body's ability to break down a substance called lysine, an important protein building block. The chemicals that accumulate in the body because of this defect are thought to be toxic to brain cells and therefore responsible for the developmental delays experienced by 75-80% of PDE patients.

What kind of medical data will be collected?

The doctors want to collect information about my symptoms due to PDE related seizures, effects on development and which treatments were used to treat the condition and its symptoms. We also want to collect data about clinical and laboratory findings in blood, urine and spinal fluid which may be associated with these conditions.

The following information will be collected:

- Diagnosis
- Year of birth
- Age (month and year only)
- symptoms of my condition (seizures, learning problems that I may have)
- Clinical and laboratory test results such as EEG, MRI, behavior tests, blood/urine laboratory results)

Will also genetic data be collected?

The doctors also want to collect information about the gene test that was done to confirm the diagnosis of PDE due to ATQ deficiency.

Are there any risks or discomforts from being in the study?

There are no physical risks or discomforts because the study only involves collecting information. All information obtained for the registry will come from a review of my medical chart . This means that there is no testing needed.

Can I choose NOT to participate in this project? Yes. Participating in the research is completely up to me.

Will participating or NOT participating in this study negatively affect my clinical care? No.

Will I receive any direct benefits from participating?

There is no direct benefit from participation in this study. However the results of this study may help scientists to learn more about early recognition of damaged functions in people with PDE and the ways to prevent the problems.

How will my information be used?

The doctors involved with this study are asking for permission to include my information in the PDE database. The database includes information collected from clinical visits and patient charts. At each centre the patient and their designated parent are being asked to provide consent to participate in the study. Usually this consent is discussed after a clinic visit. If I decide to consent, my medical information will be entered into the PDE research registry (database). This will occur each time I visit my doctor's clinic until 2026. Information from previous clinic visits if required will be collected by reviewing my

medical chart. Information will be entered to a secure server that uses the same kind of secure internet connections that banks use. If other researchers wish to use the information from the registry, the information requested will be available to them only in a manner that will not identify a person: all information that may potentially identify me will be removed (such as name, address, birth date, and other personal information). This de-identified information will be released only if approved by the doctors working on this study.

Who can access the database?
Only the Principal Investigators or their appointed designees as well as the Institutional Review Board will be granted direct access to my original medical and research records. Investigators from other participating centers will not have access to my original records.

How will my information be protected?

Protecting the security and privacy of my information is very important to the doctors. My name and other personal information will not be used with the exception of my month and year of birth in order to calculate my age each time your physician enters new data.

Can I stop participating in the study at any time in the future? Yes, I can decide to stop participating in the study at any time by writing or calling my doctor. I will not lose any benefits which I am using. If I decide to stop participating, all information that was already included in the database will continue to be used, but no new information will be added to the database for research purposes.

Are there any costs to participate? No.

Will I be paid or given anything extra for participating? Iwill not be paid for participating in this study.

Are there risks in the study?

Appropriate care will be taken to make sure that information about our health is safely and securely stored. However there is a small risk that my personal information will be identifiable due to the rarity of the disease.

Who gives the consent?
If I was consented to be enrolled in this study while I was a child, the consent would have been given by my parents / legal custody. Should I turn 18 years of age during the study period I will be contacted by a study doctor and given the opportunity to provide my individual consent to continue my participation in this study.

Who can I contact for more information if I have questions during the study?

If Ihave any questions about this study, I can contact the study doctor or the project director **Dr. Clara van Karnebeek** at **604-875-2628** or email: cvankarnebeek@cw.bc.ca.

Who do I contact if I have questions about my rights as a subject during the study?

If I have any concerns or complaints about my rights as a research participant and/or my experiences while participating in this study, I can contact the Research Participant Complaint Line at the University of British Columbia Office of Research Ethics by e-mail at RSIL@ors.ubc.ca or by phone at 604-822-8598 (Toll Free: 1-877-822-8598).

Please take time to read the following information carefully and to discuss it with your family, friends, and doctor before you decide.

ASSENT TO PARTICIPATE

PARTICIPANTASSENT TO PARTICIPATE:

- I have read (or have had it read to me) and understand the subject information and assent form.
- I have had sufficient time to consider the information provided and to ask for advice if necessary.
- I have had the opportunity to ask questions and have had satisfactory responses to my questions.
- I understand that my data will be used to increase the knowledge of PDE.
- I understand that all the data collected will be kept confidential and that the results will only be used for scientific objectives.
- I understand that my participation in this study is voluntary and that I am completely free to refuse to participate or to withdraw from this study at any time without changing in any way the quality of care that I receive.
- I understand that there is no guarantee that this study will provide any benefits to me.
- I understand that my coded data will be shared among the investigators listed on this study.
- I understand that I am not waiving any of my rights as a result of signing this assent form.
- I have read this form and I freely assent to participate in this study.
- I have been told that I will receive a dated and signed copy of this form.

Additional options. With m y signature I assent to participate in the study as described above. Additionally I have the option to be contacted if I want to learn more about other research studies relevant to my medical condition and / or to be asked for feedback on my experiences as a participant in this study.

Option 1: Future Research Studies

☐ YES, I agree to be contacted in the future, by my own physician who will be informed by the research team, to learn more about any future research studies that may be relevant to my family.

☐ NO, I DO NOT wish to be contacted about any future research study that may be relevant to my family.

Option 2:Future Contact Feedback

☐ YES, I agree to be contacted in the future, by my own physician who will be informed by the research team, to provide feedback on my experiences as a research subject in this study.

☐ NO, I DO NOT agree to be contacted in the future to provide feedback on my experiences as a research subject in this study.

I ASSENT BY SIGNING BELOW TO PARTICIPATE IN THIS STUDY

By signing I hereby assent to participate in this study. _____ *Name of study participant*

_____
Signature of Participant Date: _____

_____
Name of Person who Obtained Assent

_____ Date: _____
Signature of Person who Obtained Assent

Note to Physician: After consent, can you please write the PDE Registry REDCap® web-page generated patient data code here: _____