



Dear Members of the ULF Community Who are Undiagnosed,

At ULF's 2026 Family Conference in June, we plan to offer testing with long-read Whole Genome Sequencing (LR-WGS) for attendees who have been diagnosed with leukodystrophy but not diagnosed with a specific gene variant.

Dr. Josh Bonkowsky (University of Utah/Primary Children's Hospital) is offering enrollment into a research study using long-read WGS as a service for our undiagnosed community to participate in a study that he is leading together with Dr. Genevieve Bernard (McGill University) and Dr. Tomi Pastinen (Kansas Mercy Research Institute). The testing is research and would need clinical validation if a gene variant (mutation) is found.

Long-read Whole Genome Sequencing is a more effective method of diagnosing gene variants that are missed by standard tests and has a higher diagnostic efficacy, faster turnaround times, and provides comprehensive genetic information in a single test. It is not perfect, but there is a greater chance of diagnosis than most current testing options.

The risks of testing, which will be fully discussed in a formal consent process, include the blood draw; there is a chance that the testing will not find the diagnosis; a risk for loss of confidentiality; and risks associated with genetic testing.

The criteria for eligibility to participate is as follows:

- The patient and family **must** be registered to attend the ULF Family Conference <https://ulf.org/scientific-symposium-family-conference/>
- For most accurate results, blood should be drawn from the patient as well as both biological parents.
- The patient must have a diagnosis of leukodystrophy based on an MRI.
- Patients who are residents of the US and who reside outside the US who have a diagnosis of leukodystrophy may participate.

Clinical information that must be provided by June 15, 2026:

- Plan to provide electronic versions of clinical notes, an electronic copy of the patient's MRI result, and actual copy of the images of their MRI.
 - MRI Images and clinical notes must be provided ahead of the conference (No later than June 15).
 - If the information is not provided, you will not be able to participate.
- To send your information and to schedule a LR-WGS meeting during the conference, use the following email joshua.bonkowsky@hsc.utah.edu.
- **This email MUST have in the subject line "Long read WGS PHI"**
- Do not include any other wording in the subject line
- Send the email and required information to Dr. Bonkowsky **no later than June 15, 2026**

Additional information for your attention:

There is no cost to the undiagnosed patient who is eligible to participate in the study.



The undiagnosed patient and family must be **registered for the ULF Family Conference**. The ULF Family Conference will be held on June 26 and 27, 2026, at the Hilton Chicago/Oak Brook Hills Resort and Conference Center, 3500 Midwest Road, Oakbrook, IL 60526. Please plan to attend the entire Family Conference.

A private room will be set up for the testing at the conference. There will be signage directing you to this room.

For questions prior to the conference, please contact Dr. Josh Bonkowsky
joshua.bonkowsky@hsc.utah.edu