Protocol ALL-127: Combination Retrospective and Prospective Natural History Study of Patients with Sanfilippo Syndrome Type D (MPS IIID) Disease

What Is The Purpose of This Research Study?

This study is being conducted to learn more about Sanfilippo Syndrome Type D (also called Mucopolysaccharidosis type IIID or MPS IIID). This is a natural history and observational study, which means only medical information will be collected. No experimental drugs will be given and there will be no changes to participants' regular medical care. Information from this study will help doctors, patient caregivers, and scientists learn more about the health issues in patients with Sanfilippo Syndrome Type D, which may help pharmaceutical companies design future studies.

How Many People Will Participate?

About 10 people from different parts of the world will take part in on-site visits and video capture of daily living activities. Additionally, we will be remotely collecting medical record information on up to 15 people with MPS IIID (both living and deceased) some of who will not participate in the on-site clinical visits.

Where Does the Study Take Place?

On-site visits will take place at Columbia University, in the United States. More information about travel (including international travel) and reimbursement will be provided.

What is Involved in the Study?

This study will have 6 study visits in total over approximately 2 years. Four of the visits will take place on-site at Columbia University. The other 2 visits will be conducted over the phone. On-site visits may occur over 2 days, if needed.

In the study, we will collect information from patient medical records starting from birth through the end of the study. At study visits there will be tests and other tasks that participants will be asked to do.

	Screening Visit 1	Baseline Visit 1	Visit 3	Visit 4	Visit 5	Visit 6
Visit Timing	Up to 4 weeks before Baseline (Day 0)	Day 0	Month 6	Month 12	Month 18	Month 24
Location	On-site	On-site	Phone	On-site	Phone	On-site

What Are Some of the Study Tests and Procedures?

Some of the procedures that take place during the study are listed below. Many of these procedures are part of regular patient care for people with Sanfilippo Syndrome Type D.

- Blood samples
- Hip X-ray
- Urine samples
- Physical exam
- Vital signs
- Growth assessments (height, weight, and head circumference)
- Cerebrospinal fluid sample (also called a CSF sample or a spinal tap)
- Observational and language skills testing
- Mobility skills and intellectual skills testing
- Parent questionnaires about the participant's quality of life, mobility, language, and observational function
- Parent(s)/legal guardian(s) stress assessments
- Neurologic assessments
- 25-foot walk test
- Vision assessments
- Imaging of the brain and cervical spine (an MRI)

What is RARE-App?

RARE-app is a real time, real world, home video application that was specifically designed for this study. Participant caregivers will capture predetermined video segments of daily living activities on their own mobile device and upload them using RARE-app. Participants who participate in the observational site visits will also have the option to participate in this video collection portion of the study which will coincide with completion around the time of site visits.

What Will this Study Cost?

Study-related care, tests, and other study-related procedures are all provided at no cost.

Why Participate?

There are currently no disease-modifying therapies available for Sanfilippo Syndrome Type D, only therapies that help to manage certain symptoms. This study will help doctors and scientists learn more about the condition and how it affects people over time. This may help researchers design other studies in the future for potential treatments. Your family's participation could be an important part of that process.

Participation in the study is completely voluntary, and participants can leave the study at any time and for any reason.

Who is Sponsoring This Study?

The study is sponsored by Phoenix Nest, Inc., a biotechnology company focused on treating Sanfilippo Syndrome. Through funding from the United States National Institutes of Health (NIH), Phoenix Nest has been able to facilitate research and meet the challenges of pioneering treatments for ultra-rare and untreatable diseases.

How Can I Learn More About The Study?

To learn more about the study please contact the Columbia University Site Coordinator:
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https://clinicaltrials.gov/ct/show/NCT05648851 ■ NCT05648851