

Natural History Study of Participants With Sanfilippo Syndrome Type IIIC

NCT05825131

Status RECRUITING
Sponsor Phoenix Nest
Enrollment 30 participants

Key Eligibility Criteria

Inclusion (12)

- Confirmed diagnosis of Sanfilippo syndrome type C disease by all of the following:
- Deficiency in heparan-alpha-glucosaminide N-acetyltransferase enzyme activity
- Has presented with signs/symptoms consistent with Sanfilippo syndrome type C, or, for individuals who have not presented with signs/symptoms of disease (eg, siblings of known patients), the determination of eligibility will be at the discretion of the Sponsor in conjunction with the site Investigator
- Genomic DNA analysis demonstrating homozygous or compound heterozygous, pathogenic and/or potentially pathogenic variants in the HGSNAT gene
- Accumulated GAG HS in urine
- ... and 7 more (see full listing online)

Exclusion (4)

- Patients who meet any of the following criteria will not be eligible to participate in the study:
- Have received an investigational drug within 30 days prior to the Baseline Visit
- Concomitant illness or medical condition or extenuating circumstance that, in the opinion of the Investigator, might compromise the patient's ability to comply with protocol requirements, the patient's well-being or safety, or the interpretability of the patient's clinical data
- The presence of significant non-MPS IIIC-related CNS impairment or behavioral disturbances that would confound the scientific rigor or interpretation of results of the study

Locations (2 total)

The University of Texas Southwestern Medical Center, Dallas, Texas, United States
Hospices Civils De Lyon, Bron, France

<https://clinicaltrials.gov/study/NCT05825131>

DISCLAIMER: This document is for informational purposes only and does not constitute medical advice. Always consult your healthcare provider before enrolling in any clinical trial. Information may not be up to date — verify details at [ClinicalTrials.gov](https://clinicaltrials.gov). Generated by [ClinicalTrialsFinder.org](https://clinicaltrialsfinder.org).