

Hereditary Neuropathy Foundation (HNF) Expands Charcot-Marie-Tooth (CMT) Biobank Inventory Now Available to Researchers

HNF expands CMT Biobank with immediate access to biospecimens to accelerate biomarker and translational research. Calling researchers, industry and patients.

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The Hereditary Neuropathy Foundation (HNF) is excited to announce the significant expansion of its [CMT Biobank](#), in partnership with COMBINEDBrain, now offering an extensive range of biospecimens for researchers. This biobank serves a critical role in advancing CMT research by providing samples like blood, plasma, and iPSCs, linked to GRIN's clinical and genomic data. These resources are essential for biomarker discovery, understanding disease progression, and accelerating therapeutic development. With no treatments currently available for CMT, this biobank fills a crucial gap by offering a much-needed resource to researchers worldwide.



[Upcoming roadshows](#) will take place in Nashville, TN, on November 11th, and Los Angeles, CA, on December 5th and 6th, where researchers can learn more about the CMT Biobank, explore collaborations, and discuss sample access.

Patients interested in donating specimens can sign up [here](#).

These samples are available immediately for research use, with all details and guidelines accessible through the HNF and COMBINEDBrain websites.

- CMT1A (PMP22 dup) - PBMC, P, WB, U, BS
- CMT2A (MFN2) - PBMC, P, WB, U, BS
- HNPP (PMP22 del) - PBMC, P, WB, U, BS
- CMT4A (GDAP1) - iPSC, PBMC, P, WB, U, BS
- CMT-SORD - PBMC, P, WB, U, BS
- CNTNAP1 - PBMC, P, WB, U, BS
- CMT2P (LRSAM1) - P, WB, U, BS
- CMT2O (DYNC1H1) - P, WB, U, BS
- CMT1X (GJB1) - P, WB, U, BS
- HMNR1 (IGHMBP2) - P, WB, U, BS
- CMT2T (MME) - P, WB, U, BS
- CMT1B (MPZ) - P, WB, U, BS
- SPG7 - P, WB, U, BS
- CMT2DD - P, WB, U, BS

CMT type	Sample type
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CMT1B (MPZ)	P, WB, U, BS
SPG7	P, WB, U, BS
CMT2DD	P, WB, U, BS
Pending genomic diagnosis	P, WB, U, BS
Control (unaffected family)	iPSC, PBMC, P, WB, U, BS

PBMC - peripheral blood mononuclear cells; iPSC – induced pluripotent stem cells; P – Plasma; WB – Whole blood; U – Urine; BS – Blood Spot

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There are over 130 subtypes of CMT. HNF aims to continue the expansion of the CMT Biobank to include as many mutations as possible.

Interested in partnership opportunities?

Contact Allison Moore or Matt Jarpe, PhD at biobank@hnf-cure.org

About the Hereditary Neuropathy Foundation (HNF):

HNF's mission is to increase awareness and accurate diagnosis of Charcot-Marie-Tooth (CMT) and related inherited neuropathies, support people living with CMT and their families with critical information to improve quality of life, and fund research that will lead to treatments and cures. HNF's Therapeutic Research in Accelerated Discovery (TRIAD) is a collaborative effort with academia, government, and industry to develop treatments for CMT. As part of TRIAD's research consortium, the Global Registry for Inherited Neuropathies (GRIN) was established as a patient registry to collect and analyze patient-reported data and clinical scales, including the ONLS, CMT-FOM, CMTpedS, CMTInfS, Sensor Wearables, and the collection and curation of genetic reports. The data has been instrumental in identifying the burden, and diagnostic journey of disease while providing vital resources for clinical researchers and the biopharmaceutical industry for clinical trials. For more information, visit www.hnf-cure.org.

What is Charcot-Marie-Tooth (CMT)?

CMT is one of the most common inherited nerve disorders and is also referred to as hereditary sensory and motor neuropathy (HSMN). CMT affects an estimated 1 in 2,500 people in the United States and although considered a rare disease (under 200,000 people), experts believe the number to be much higher. There are an estimated 2.6 million people affected worldwide. CMT is considered to be one of the most common of the 7000 rare diseases affecting children. Often, the muscle loss happens unevenly, which causes foot and hand deformities, muscle wasting, and atrophy leading to mobility issues. It can also have serious impacts on vision, hearing, breathing, speech, and swallowing. Some patients experience hip dysplasia, scoliosis, and blindness.

For those patients interested in learning what type of CMT they have or if you are one of the million patients not diagnosed yet, HNF's CMT Genie, a patient-initiated genetic testing program is available to you.

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