

Consensus document
Biological sample wish list

Version	1
Status	Approved
Date	6-3-2023
Purpose of this document	List the biological samples that are preferably and optionally collected in routine care in MLD patients.

Version control

Version number	Revision date	Summary of major changes

Background

- Currently, the MLD initiative (MLDi) does not comprise a central biobank for sample collection and storage.
- There is an urgent need for biomarker research in MLD.
- As a workaround the MLDi members decided to make a consensus-based wishlist of samples to be regularly or optionally collected in MLD patients.
- In the MLDi registry it is captured whether samples can be shared or used for research.
- In this way, steps towards a federated biobank model are made.

Preferably routinely* collected and stored in all MLD patients

* Yearly

Material	Specification	Pros	Cons	Proposed measurements/biomarkers
Blood - Plasma	EDTA	<ul style="list-style-type: none"> • Easy to collect • Often leftover material because enzyme activity is measured in EDTA plasma 		<ul style="list-style-type: none"> • Neurofilament light • Sulfatides • Metabolomics • Chitotriosidase • Inflammatory markers
Blood - Serum		<ul style="list-style-type: none"> • Easy to collect • Often leftover material when NFL is measured 		<ul style="list-style-type: none"> • Neurofilament light • Chitotriosidase
Blood - Dried blood spots		<ul style="list-style-type: none"> • Easy to collect • Easy to store 		<ul style="list-style-type: none"> • Neurofilament light • Sulfatides • Metabolomics • Inflammatory markers

Optional collection in all MLD patients

Material	Specification	Pros	Cons	Proposed measurements/biomarkers
Blood - Plasma	Heparine	<ul style="list-style-type: none"> • Easy to collect 	<ul style="list-style-type: none"> • Extra tube, so additional consent/IRB approval needed 	<ul style="list-style-type: none"> • Neurofilament light • Sulfatides • Metabolomics
Blood	Pax-gene tube	<ul style="list-style-type: none"> • Easy to collect • RNA analysis 	<ul style="list-style-type: none"> • Extra tube, so additional consent/IRB approval needed 	
Skin - Fibroblasts		<ul style="list-style-type: none"> • iPSC can be made • RNA analysis • Skin biopsy can be done simultaneously with gastrostomy or intrathecal baclofen pump placement 	<ul style="list-style-type: none"> • Somewhat burdensome collection • IRB approval (specific research question needed) 	
Urine		<ul style="list-style-type: none"> • Easy to collect 		<ul style="list-style-type: none"> • Sulfatides • Metabolomics
Cerebrospinal fluid			<ul style="list-style-type: none"> • Burdensome collection 	<ul style="list-style-type: none"> • Neurofilament light • (lyso)sulfatides • Metabolomics
Peripheral nerve tissue	Sural nerve		<ul style="list-style-type: none"> • Burdensome collection 	<ul style="list-style-type: none"> • (lyso)sulfatides • Metabolomics
Gall bladder				<ul style="list-style-type: none"> • Microscopic examination • Sulfatides in tissue and bile
Autopsy				
- Brain				
- Sural nerve tissue				
- Gall bladder				