



Heidelberg University Hospital

University Children's Hospital
Department I

(General Pediatrics, Metabolism, Gastroenterology and Nephrology)

Chairman of Pediatrics

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Lysosomal Diagnostics

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Disease	Enzyme defect	Sample				
		Urine/SA	DBS	Leu	S / P	Fib
		MPS-EL (1) Oligo-TLC (2) Neuraminic acid (3) Sulfatides (SA) (4)	Enzyme assay			
Mucopolysaccharidoses	I Hurler/Scheie	α -iduronidase	1	x	x	x
	II Hunter	iduronate-sulfatase	1	x	x	x
	IIIA Sanfilippo A	sulfamate-sulfatase	1		x	x
	IIIB Sanfilippo B	α -N-Acetyl-glucosaminidase	1	x	x	x
	IIIC Sanfilippo C	Acetyl-CoA: α -glucosamine-acetyl-transferase	1		x	x
	IIID Sanfilippo D	α -N-Acetyl-glucosamine-6-sulfatase	1		x	x
	IVA Morquio A	galactose-6-sulfatase	1		x	x
	IVB Morquio B	β -galactosidase	1	x	x	x
	VI Maroteaux-Lamy	arylsulfatase B	1	x	x	x
	VII Sly	β -glucuronidase		x	x	x
	Multiple sulfatase deficiency (MSD)	several sulfatases	1 / 4		x	x
Sphingolipidoses	Metachromatic Leukodystrophy (MLD)	arylsulfatase A	4		x	x
	MLD, Saposin B defect, MSD	Sulfatides	4 (SA)			
	GM1 GM1-gangliosidosis	β -galactosidase	1 / 2	x	x	x
	Galactosialidosis	β -galactosidase + sialidase	2 / 3			x
	Fabry disease	α -D-galactosidase A		x	x	x
	GM2-gangliosidosis Type Sandhoff	β -hexosaminidase (N-Acetyl- β -D-glucosaminidase)	2	x	x	x
	GM2-gangliosidosis Type Tay-Sachs	β -hexosaminidase A (N-Acetyl- β -D-glucosaminidase A)		x	x	x
	Gaucher disease	β -glucosidase (glucocerebrosidase)			x	x
	Niemann-Pick (NP) type A/B disease	acid sphingomyelinase			x	x
	Schindler disease	N-acetyl- α -D-galactosaminidase	2		x	x
	Krabbe disease	galactocerebrosidase			x	x
	Fabry disease	lysoGL-3				x
	Gaucher disease	hexSph (lysoGL-1 =GlcSph)				x
	Krabbe disease	hexSph (galSph)				x
	Niemann-Pick (NP) type A/B disease	lyso-SM + lyso-SM509				x
	GM1-gangliosidosis	lysoGM1				x
	GM2-gangliosidosis Type Sandhoff	lysoGM2				x
	GM2-gangliosidosis Type Tay-Sachs	lysoGM2				x
Muco-lipidoses	I Sialidosis	sialidase; neuraminidase	2 / 3			x
	II Mukopolipidose II (I-Cell-disease)	transport defect	(1) / (2) / (3)	x		x
	III Mukopolipidose III					

Neuronal ceroid lipofuscinosis (NCL)	infantile (INCL)	palmitoyl protein thioesterase 1 (PPT1)		x	x		x
	late infantile (LINCL)	tripeptidyl peptidase 1		x	x		x
Oligo-saccharidoses	Fucosidosis	α -L-fucosidase	2	x	x	x	x
	α -Mannosidosis	α -mannosidase	2	x	x	x	x
	β -Mannosidosis	β -mannosidase	2	x	x	x	x
	Aspartyl-glucosaminuria	aspartyl-glucosaminidase	2		x		x
Glykogenosis type II (GSD II)	Pompe disease	α -glucosidase (acid maltase)	2	x	(x)		x
Sialic acid storage disease	ISSD, salla disease		3				x
Cholesteryl ester storage disease	Wolman disease	acid lipase		x	x		x
Screening	e.g. Gaucher and NP A/B/C	chitotriosidase		x		x	

Legend:

- ¹ **SA:** 24-hrs. collected urine please cool during collection period
- Urine/SA:** 5-40 ml
add 4-6 droplets of dichloro methane/10 ml urine/SA and send immediately at ambient temperature or store at -20°C and send frozen
- Serum/Plasma:** 1.0 ml (if plasma then EDTA plasma preferred)
(S/P) send immediately at ambient temperature or store at -20°C and send frozen
- Leukocytes:** 5-10 ml **EDTA blood**
(Leu) **or isolated leukocytes** (suspended in 1.0 ml RPMI media)
send at ambient temperature per Express (within 24 hrs; max. 48 hrs);
please indicate sampling time at the tube
or isolated leukocytes (pellet only) frozen on dry ice
- DBS** 5 circles filled with blood, let dry for at least 3 hours protected from light at ambient temperature. Send at ambient temperature by regular mail.
- Fibroblasts:** send immediately at ambient temperature within 24 hrs (max. 48 hrs).
(Fib) please fill up the flask with media + 10% FBS (fetal bovine/calf serum).
- skin biopsy:** transfer skin biopsy in sterile 0.9% NaCl solution; tube must be filled completely
(for fibroblasts) with 0.9 % NaCl solution!
send immediately at ambient temperature within 24 hrs (max. 48 hrs).
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