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Center for Metabolic Diseases Heidelberg
Metabolic Laboratory

Department of Pediatrics I
(General Pediatrics, Metabolism, Gastroenterology, Nephrology, Neurology)

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Lysosomal diagnostics

Legend

SA	**40 ml** 24-hrs. collected urine, please cool during collection period and mix well before taking an aliquot
RT	room temperature
Urine / SA	**10-20 ml** send frozen on dry ice or add 2-3 droplets of dichlor methane and send at RT within 3-4 days
Serum / plasma (S /P)	**1 ml** send frozen on dry ice
Leukocytes (Leu)	**5-10 ml EDTA-Vollblut** send at RT per Express (within 24 hrs); please indicate sampling time and date or isolate leukocytes in your own laboratory and send isolated leukocytes frozen on dry ice
Dried blood spot (DBS)	transfer blood onto filter paper card and let dry for at least 2 hrs at RT before sending at RT
Skin biopsy	transfer skin biopsy into sterile 0.9% NaCl solution; send immediately at RT within 24 hrs.
Fibroblasts (Fib)	please fill up the flask with media + 10% FBS (fetal bovine / calf serum); send immediately at RT within 24 hrs.
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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	new x	x	x	x
	IIIA	Sanfilippo A	sulfamate-sulfatase	1		x		x
	IIIB	Sanfilippo B	α-N-Acetyl-glucosaminidase	1	new x	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	x
		Multiple sulfatase deficiency (MSD)	several sulfatases	1 / 4		x	x	x
Sphingolipidoses		Metachromatic Leukodystrophy (MLD)	arylsulfatase A	4		x		x
		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	GM2-gangliosidosis Type Sandhoff	β-hexosaminidase (N-Acetyl-β-D-glukosaminidase)	2	x	x	x	x
		GM2-gangliosidosis Type Tay-Sachs	β-hexosaminidase A (N-Acetyl-β-D-glukosaminidase A)		x	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	x
		Krabbe disease	galactocerebrosidase			x		x
Muco-lipidoses	I	Sialidosis	sialidase; neuraminidase	2 / 3				x
	II	Mukolipidose II (I-Cell-disease)	transport defect	(1) / (2) / (3)	x		x	x
	III	Mukolipidose III						
Neuronal ceroid lipofus-cinosis (NCL)		infantile (INCL; NCL 1)	palmitoyl protein thioesterase 1 (PPT1)		x	x		x
		late infantile (LINCL; NCL 2)	tripeptidyl peptidase 1 (TPP1)		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
cholesterlester storage disease		Wolman disease (CESD)	acid lipase		new x	x		x
Screening		e.g. Gaucher and NP A/B/C	chitotriosidase		x		x	