



Lysosomal Enzyme Diagnostics

Disease		Enzyme defect		Sample Material		
				Filter Card	EDTA	
				Enzyme assay		
Pompe			α -glucosidase	X	X	
Gaucher			β -glucosidase	X	X	
Niemann-Pick A/B			Acid Sphingomyelinase	X		
Fabry			α -Galactosidase	X	X	
Mucopolysaccharidosis	MPS I		α -iduronidase	X	X	
	MPS II		Iduronate-2-sulfatase	X	X	
	Sanfilippo A-D	MPS IIIA		Heparan-N-sulfatase		X
		MPS IIIB		N-Acetylglucosaminidase		X
		MPS IIIC		Acetyl-CoA: α -glucosamine N-acetyltransferase		X
		MPS IIID		N-Acetylglucosamine-6-sulfatase		X
	MPS IV A		N-Acetylglucosamine-6-sulfatase		X	
	MPS VI		Arylsulfatase B	X		
	MPS VII		β -Glucuronidase	X	X	
Mucolipidoses II/III			X	Plasma		
Neuronal Ceroid Lipofuscinosis		CLN 1	PPT 1	X	X	
		CLN 2	TPP1	X	X	
Oligosaccharidoses			α -Mannosidase	X	X	
			β -Mannosidase	X		
		Asparetyl-glucosaminuria	Aspartyl-glucosidase	X	Plasma	
			α -Fucosidase	X	X	

In case of any questions please contact FREIBURG MEDICAL LABORATORY www.fml-dubai.com

Tel: +971-4-396 2227, Fax: +971-4-396 2228, E-mail: info@fml-dubai.com



Disease		Enzyme defect	Sample Material	
			Filter Card	EDTA
			Enzyme assay	
Gangliosidosis	GM1-gangliosidosis	β -Galactosidase	X	X
	GM2-gangliosidosis	Total hexosaminidase	X	X
		Hexosaminidase	X	X
Leukodystrophy	Metachromatic Leukodystrophy	Arylsulfatase A	serum & urine	X
	M. Krabbe	β -galactocerebrosidase	X	
Others	M. Wolman (CESD)	Acid Lipase	X	

Sample Material:

Filter Card

Transfer native blood onto filter paper card and let dry for at least 2 hours at room temperature (RT) and send at RT

EDTA = EDTA-blood

Store and ship the EDTA-blood at RT.

Note: Contact FML for more details

In case of any questions please contact FREIBURG MEDICAL LABORATORY www.fml-dubai.com
 Tel: +971-4-396 2227, Fax: +971-4-396 2228, E-mail: info@fml-dubai.com