CENTOGENE
THE RARE DISEASE COMPANY

Findings in some specific metabolic disorders

METABOLIC DISORDERS COVERED BY CENTOGENE	COARSE FACIAL FEATURES	DYSOSTOSIS MULTIPLEX	ORGANO-MEGALY	INTELLECTUAL DISABILITY	SPASTICITY	PERIPHERAL NEUROPATHY	MYOCLONIC SEIZURES	HYDROPS FETALIS	ANGIO-KERATOMA	CORNEAL CLOUDING	CHERRY-RED SPOT	VISUAL LOSS	CARDIAC INVOLVEMENT	MACRO-GLOSSIA	VACUOLATED LYMPHOCYTES	GAGs ELEVATED	PATHOLOGICAL OLIGOSACCHARIDES	ENZYMATIC TESTING	BIOMARKER TESTING	SINGLE GENE ANALYSIS	DELETION/DUPLICATION ANALYSIS
SPHINGOLIPIDOSES																		TESTING AT CENTOGENE			
Fabry disease									●			○	○					○	○	○	○
Farber disease			●	●		●				○	○								○	○	○
Galactosialidosis	●	●	●	●	●		○	●	○	●	●	●	●		●					○	○
Gaucher type I			●														○	○	○	○	○
Gaucher type II			●	●	●		●	●									○	○	○	○	○
Gaucher type III			●	●	○		○	○				●	○				○	○	○	○	○
GM1-Gangliosidosis	●	●	●	●	○			●	●	●	●	●	○	●			●	○		○	○
Krabbe disease				●	●	●					○	●						○		○	○
Metachromatic Leukodystrophy				●	○	●					○	●						○ [1]		○	○
Multiple sulfatase deficiency	●	●	●	●	●		●			●	○		●	●		●		○		○	○
Niemann Pick A, B			●	○			○	●		○	○	●						○	○	○	○
Niemann Pick C1/C2			○	●	●	●					○				●				○	○	○
Wolman disease			●					●			○		○		●			○		○	○
MUCOPOLYSACCHARIDOSES																					
MPS I type IH	●	●	●	●						●		●	●	●		●		○		○	○
MPS I type IS	●	●	●							●		●	●	○		●		○		○	○
MPS type II	●	●	●	●								●	●	●		●		○		○	○
MPS type III a,b,c,d	○	○	○	●	●					●		○				○		○ [2]		○	○
MPS type IV a,b	●	●	●	○				●		○		●				○		○		○	○
MPS type VI	○	●	●							●		●	●	●		●		○		○	○
MPS type VII	●	●	●	●				●		●		●	○	●		●		○		○	○
NEURONAL CEROID LIPOFUSCINOSES																					
NCL Type I				●	●		●					●					●	○		○	○
NCL Type II				●	●		●					●					●	○		○	○
NCL Type III				●	●		●					●	●							○	○
NCL Type V				●	●		●					●								○	○
NCL Type VI				●			●					●								○	○
NCL Type VII, VIII, X				●	●		●					●								○	○
OLIGOSACCHARIDOSES																					
Fucosidosis	●	○	○	●	●		●		○				●		●		●	○		○	○
Mannosidosis α	●	○	○	●	○				○	●					●		●	○		○	○
Mannosidosis β	●			●	●	●			○								●	○		○	○
Sandhoff disease			●		●		●				●	●						○		○	○
Schindler disease				●	●		●					●					●	○		○	○
Sialidosis type I					●	●	●	●			●	●			●		●			○	○
Sialidosis type II	●	○	●	●			○	○	●		●	●	●		●		●			○	○
Tay Sachs disease			●		●		●				●	●						○		○	○

● = prominent feature ● = often present ○ = sometimes present ◯ = available for testing

1 = only from leukocytes, at least 5 mL freshly collected ETDA blood needed **2** = available for MPS III b

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