CENTOGENE's Biomarker - A Growing Portfolio

	DISORDER COVERED BY CENTOGENE	ENZYMATIC TESTING (ENZYME PANEL)	BIOMARKER TESTING[9]	SINGLE GENE ANALYSIS
SPHINGOLIPIDOSES	Fabry disease	Alpha-galactosidase (CentoLSD, CentoSphingo®)	Globotriaosylsphingosine (Lyso-Gb3) + others in development[9]	GLA
	Farber disease	—	C26-Ceramide	ASAH1
	Gaucher disease type I, II, III	Beta-glucocerebrosidase (CentoLSD, CentoSphingo®)	Glucosylsphingosine (Lyso-Gb1)[10]	GBA
	Gaucher disease, atypical	—	Glucosylsphingosine (Lyso-Gb1)[10]	PSAP
	Krabbe disease	Galactocerebrosidase	In development[9]	GALC
	Metachromatic Leukodystrophy	Arylsulfatase A	In development[9]	ARSA
	Niemann-Pick disease A/B	Acidic sphingomyelinase (CentoLSD, CentoSphingo®)	Lyso-SM-509 & Lyso-SM-465	SMPD1
	Niemann-Pick disease C1/C2	—	Lyso-SM-509 & Lyso-SM-465	NPC1/NPC2
OTHERS (EXCERPT)	Aromatic L-amino acid decarboxylase (AADC) deficiency	—	3-O-Methyldopa (3-OMD)	DDC
	Hereditary angioedema (SERPING1 mutation)	—	C4 and C1-INH complement protein, cleaved high-molecular-weight kininogen (cHMWK) + others in development[9]	SERPING1
	Transthyretin Amyloidosis (TTR)	—	In development[9]	TTR
	Duchenne	—	In development[9]	DMD

(Status: March. 2020)

[9] Further biomarker developments upon request. Over 50 additional biomarkers in development.

[10] Lyso-Gb1 has been identified as the most effective biomarker for Gaucher disease (Elstein, Deborah, et al. "Reductions in glucosylsphingosine (lyso-Gb1) in treatment-naïve and previously treated patients receiving velaglucerase alfa for type 1 Gaucher disease: Data from phase 3 clinical trials." Molecular genetics and metabolism 122.1-2 (2017): 113-120.)