Eli screens for these conditions:

Condition	Gene(s)	Test
Congenital Cytomegalovirus (cCMV)		PCR
Duchenne Muscular Dystrophy (DMD)	*DMD*	Biochemical
Fabry Disease	*GLA*	Biochemical
Gaucher Disease	*GBA*	Biochemical
MPS II (Hunter Syndrome)	*IDS*	Biochemical
MPS I (Hurler-Scheie Syndrome)	*IDUA*	Biochemical
Pompe Disease	*GAA*	Biochemical
alpha-Mannosidosis	*MAN2B1*	Targeted Next-Generation Sequencing
ASAH1-related disorders (Farber Disease, SMA-PME)	*ASAH1*	Targeted Next-Generation Sequencing
Cerebral Creatine Deficiency Syndrome 2	*GAMT*	Targeted Next-Generation Sequencing
Cerebrotendinous Xanthomatosis (CTX)	*CYP27A1*	Targeted Next-Generation Sequencing
Congenital Hearing Loss (Cx26)	*GJB2*	Targeted Next-Generation Sequencing
Copper Transport Disorders:		Targeted Next-Generation Sequencing
Menkes, Occipital Horn Syndrome, etc.	*ATP7A*	Targeted Next-Generation Sequencing
Wilson Disease	*ATP7B*	Targeted Next-Generation Sequencing
Cystinosis	*CTNS*	Targeted Next-Generation Sequencing
Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)	*G6PD*	Targeted Next-Generation Sequencing
Hereditary Fructose Intolerance	*ALDOB*	Targeted Next-Generation Sequencing
Hypophosphatasia (HPP)	*ALPL*	Targeted Next-Generation Sequencing
Krabbe Disease	*GALC*	Targeted Next-Generation Sequencing
Lysinuric Protein Intolerance	*SLC7A7*	Targeted Next-Generation Sequencing
Lysosomal Acid Lipase Deficiency (LAL-D)	*LIPA*	Targeted Next-Generation Sequencing
Metachromatic Leukodystrophy	*ARSA*	Targeted Next-Generation Sequencing
MPS IIIA (Sanfilippo A Syndrome)	*SGSH*	Targeted Next-Generation Sequencing
MPS IIIB (Sanfilippo B Syndrome)	*NAGLU*	Targeted Next-Generation Sequencing
MPS VI (Maroteaux-Lamy Syndrome)	*ARSB*	Targeted Next-Generation Sequencing
MPS VII (Sly Syndrome)	*GUSB*	Targeted Next-Generation Sequencing
Neuronal Ceroid-Lipofuscinoses (CLN2, Batten disease)	*TPP1*	Targeted Next-Generation Sequencing
Niemann-Pick Disease Type A and Type B	*SMPD1*	Targeted Next-Generation Sequencing
Retinoblastoma	*RB1*	Targeted Next-Generation Sequencing
Smith-Lemli-Opitz Syndrome	*DHCR7*	Targeted Next-Generation Sequencing
Spinal Muscular Atrophy (SMA)	*SMN1/2*	Targeted Next-Generation Sequencing
Vascular Ehlers-Danlos Syndrome	*COL3A1*	Targeted Next-Generation Sequencing

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1-833-ELI-1234 (1-833-354-1234)

eliCares@eliScreen.com

eliScreen.com