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

**Estimated incidence rates based only on U.S .population*

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