

NewbornGeneID Carrier Test Genes and Conditions Tested for:

DISEASE	GENE
Alpha Thalassemia	HBA1
	HBA2
Arterial Tortuosity Syndrome	SLC2A10
Beta Thalassemia	HBB
Bloom Syndrome	BLM
Canavan Disease	ASPA
Classical Galactosemia	GALT
Congenital aneurysms	COL4A1
Cystic Fibrosis	CFTR
Cystic Fibrosis Related - CA12	CA12
Cystic Fibrosis Related - SCNN1A	SCNN1A
Cystic Fibrosis Related - SCNN1B	SCNN1B
Cystic Fibrosis Related - SCNN1G	SCNN1G
Dihydrolipoamide Dehydrogenase Deficiency	DLD
Ehlers Danlos Syndrome Type 4	COL3A1
Familial Dysautonomia	IKBKAP
Familial TAA - ACTA2-related	ACTA2
Familial TAA - MYH11-related	MYH11
Familial TAA - MYLK-related	MYLK
Fanconi Anemia: Type A	FANCA
Fanconi Anemia: Type C	FANCC
Fanconi Anemia: Type F	FANCF
Fanconi Anemia: Type G	FANCG
Fragile X Syndrome (available upon request)	FMR1
Gaucher disease	GBA
Glycogen Storage Disease II - Pompe Disease	GAA
Glycogen Storage Disease IV	GBE1
Jervell and Lange-Nielsen - LQT5	KCNE1
Jervell and Lange-Nielsen - LQT11	KCNQ1
Loeys-Dietz syndrome type I	TGFBR1
Loeys-Dietz syndrome type II	TGFBR2

DISEASE	GENE
Loeys-Dietz syndrome type III	SMAD3
Long QT Syndrome 3	SCN5A
Long QT Syndrome 6	KCNE2
Long QT Syndrome 11	AKAP9
Maple Syrup Urine Disease Type 1A	BCKDHA
Maple Syrup Urine Disease Type 1B	BCKDHB
Maple Syrup Urine Disease Type II	DBT
Marfan syndrome	FBN1
Mucopolidosis IV	MCOLN1
Niemann-pick Disease Type C1	NPC1
Niemann-pick Disease Type C2	NPC2
Nonsyndromic Hearing Loss (connexin 26)	GJB2
Nonsyndromic Hearing Loss (connexin 30)	GJB6
Nonsyndromic Hearing Loss (connexin 31)	GJB3
Nonsyndromic Hearing Loss (DFNA2)	KCNQ4
Nonsyndromic Hearing Loss (DFNA13)	COL11A2
Ornithine Transcarbamylase Deficiency	OTC
Pendred Syndrome	SLC26A4
Phenylketonuria	PAH
Sickle Cell Disease	HBB
Spinal Muscular Atrophy (Werdnig-Hoffman)	SMN1
Spinal Muscular Atrophy - Modifier	SMN2
Spinal Muscular Atrophy - DYNC1H1 related	DYNC1H1
Spinal Muscular Atrophy - UBA1-related	UBA1
Spinal Muscular Atrophy - VAPB-related	VAPB
Tay-Sachs	HEXA
Usher Syndrome Type 1B	MYO7A
Usher Syndrome Type 1C	USH1C
Usher Syndrome Type 1D	CDH23
Usher Syndrome Type 1F	PCDH15
Usher Syndrome Type 2A	USH2A

