qSeq Easy® · Neonatal

Disorder group	Disease	OMIM	Gene	Band	Spanish Neonatal Screening
Amino Acid Metabolism	Phenylketonuria	261600	PAH	12q23.2	✓
Amino Acid Metabolism	Alkaptonuria	203500	HGD	3q13.33	
Amino Acid Metabolism	Arginase deficiency	207800	ARG1	6q23.2	
Amino Acid Metabolism	Argininosuccinate Lyase Deficiency	207900	ASL	7q11.21	
Amino Acid Metabolism	Citrullinemia	215700	ASS1	9q34.11	
Amino Acid Metabolism	Cystathioninuria	219500	CTH	1p31.1	
Amino Acid Metabolism	Cystinuria	220100	SLC3A1	2p21	
Amino Acid Metabolism	Cystinuria	220100	SLC7A9	19q13.11	
Amino Acid Metabolism	Histidinemia	235800	HAL	12q23.1	
Amino Acid Metabolism	Homocystinuria B6-responsive and nonresponsive types	236200	CBS	21q22.3	
Amino Acid Metabolism	Hypermethioninemia	613752	AHCY	20q11.22	
Amino Acid Metabolism	Hypermethioninemia	606664	GNMT	6p21.1	
Amino Acid Metabolism	Hypermethioninemia	250850	MAT1A	10q23.1	
Amino Acid Metabolism	Hyperprolinemia	239510	ALDH4A1	1p36.13	
Amino Acid Metabolism	Hyperprolinemia	239500	PRODH	22q11.21	
Amino Acid Metabolism	Maple syrup urine disease type Ia	248600	BCKDHA	19q13.2	
Amino Acid Metabolism	Maple syrup urine disease type Ib	248600	BCKDHB	6q14.1	
Amino Acid Metabolism	Maple syrup urine disease type II	248600	DBT	1p21.1	
Amino Acid Metabolism	Maple syrup urine disease type III	248600	DLD	7q31.1	
Amino Acid Metabolism	Tyrosinemia type I	276700	FAH	15q25.1	
Biotinidase Deficiency	Biotinidase deficiency	253260	BTD	3p25.1	
CFTR-related disorders	Cystic Fibrosis	219700	CFTR	7q31.2	✓
Deafness	Deafness Autosomal Dominant 3A	601544	GJB2	13q11	
Deafness	Pendred syndrome	274600	SLC26A4	7q22.3	
Fatty Acid Metabolism	Acyl-CoA dehydrogenase, medium chain deficiency of	201450	ACADM	1p31.1	✓
Fatty Acid Metabolism	Glutaric acidemia IIA	231680	ETFA	15q24.2-3	✓
Fatty Acid Metabolism	Glutaric acidemia IIB	231680	ETFB	19q13.41	✓
Fatty Acid Metabolism	Glutaric acidemia IIC	231680	ETFDH	4q32.1	✓
Fatty Acid Metabolism	LCHAD deficiency	609016	HADHA	2p23.3	✓
Fatty Acid Metabolism	Adrenal hyperplasia due to 21-hydroxylase deficiency	201910	CYP21A2	6p21.3	
Fatty Acid Metabolism	Carnitine deficiency, systemic primary	212140	SLC22A5	5q31.1	
Fatty Acid Metabolism	CPT deficiency, hepatic	255120	CPT1A	11q13.3	
Fatty Acid Metabolism	CPT II deficiency, lethal neonatal	608836	CPT2	1p32.3	
Fatty Acid Metabolism	Galactokinase deficiency with cataracts	230200	GALK1	17q25.1	
Fatty Acid Metabolism	Galactose epimerase deficiency	230350	GALE	1p36.11	
Fatty Acid Metabolism	Glucose-6-phosphate dehydrogenase deficiency	305900	G6PD	Xq28	
Fatty Acid Metabolism	Short-chain acyl-CoA dehydrogenase deficiency	201470	ACADS	12q24.31	
Fatty Acid Metabolism	VLCAD deficiency	201475	ACADVL	17p13.1	
Galactosemia	Galactosemia	230400	GALT	9p13.3	
Hypothyroidism	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	610978	NKX2-1	14q13.3	✓
Hypothyroidism	Diabetes mellitus, neonatal, with congenital hypothyroidism	610199	GLIS3	9p24.2	✓
Hypothyroidism	Hypothyroidism, congenital nongoitrous 5	225250	NKX2-5	5q35.1	✓
Hypothyroidism	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	PAX8	2q13	✓
Hypothyroidism	Hypothyroidism, congenital, nongoitrous 1	275200	TSHR	14q31.1	✓
Hypothyroidism	Hypothyroidism, congenital, nongoitrous 4	275100	TSHB	1p13.2	✓
Hypothyroidism	Pseudopseudohypoparathyroidism	612463	GNAS	20q13.32	✓
Hypothyroidism	Thryoid dyshormonogenesis 6	607200	DUOX2	15q21.1	✓
Hypothyroidism	Thyroid dyshormonogenesis 1	274400	SLC5A5	19p13.11	✓
Hypothyroidism	Thyroid dyshormonogenesis 2A	274500	TPO	2p25.3	✓
Hypothyroidism	Thyroid dyshormonogenesis 3	274700	TG	8q24.22	✓
Hypothyroidism	Thyroid dyshormonogenesis 4	274800	IYD	6q25.1	✓
Hypothyroidism	Thyroid dyshormonogenesis 5	274900	DUOXA2	15q21.1	✓
Hypothyroidism	Bamforth-Lazarus syndrome	241850	TTF2	9q22.33	
Organic Acid Metabolism	3-methylcrotonyl-CoA carboxylase deficiency	210200	MCCC1	3q27.1	
Organic Acid Metabolism	3-methylcrotonyl-CoA carboxylase deficiency	210210	MCCC2	5q13.2	
Organic Acid Metabolism	Alpha-methylacetoacetic aciduria	203750	ACAT1	11q22.3	
Organic Acid Metabolism	Glutaric aciduria, type I	231670	GCDH	19p13.2	
Organic Acid Metabolism	HMG-CoA lyase deficiency	246450	HMGCL	1p36.11	
Organic Acid Metabolism	Isovaleric acidemia	243500	IVD	15q15.1	
Organic Acid Metabolism	Methylmalonic aciduria and homocystinuria, cblC type	277400	MMACHC	1p34.1	
Organic Acid Metabolism	Methylmalonic aciduria and homocystinuria, cblC type	277410	MMADHC	2q32.2	
Organic Acid Metabolism	Methylmalonic aciduria, mut(0) type	251000	MUT	6p12.3	
Organic Acid Metabolism	Methylmalonic aciduria, vitamin B12-responsive	251100	MMAA	4q31.21	
Organic Acid Metabolism	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	MMAB	12q24.11	
Organic Acid Metabolism	Mevalonate kinase deficiency	260920	MVK	12q24.11	
Organic Acid Metabolism	Propionic acidemia	606054	PCCA	13q32.3	
Organic Acid Metabolism	Propionic acidemia	606054	PCCB	3q22.3	
Organic Acid Metabolism	Vitamin B12-unresponsive methylmalonic acidemia	251120	MCEE	2p13.3	
Thalassemias	Alpha Thalassemias	604131	HBA1,HBA2	16p13.3	✓
Thalassemias	Beta Thalassemias	613985	HBB	11p15.4	✓