Counsyl

CPT code reference guide

Current procedural terminology codes for the Foresight™ Carrier Screen and Prelude™ Prenatal Screen

Foresight™ Carrier Screen (Universal Panel)

Gene	Associated syndromes	CPT code
ABCC8	ABCC8-Related Hyperinsulinism	81401, 81407
ABCD1	Adrenoleukodystrophy: X-Linked	81405
ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency	81400, 81401
ACADS	Short Chain Acyl-CoA Dehydrogenase Deficiency	81404, 81405
ACADVL	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	81406
ADA	Adenosine Deaminase Deficiency	81479
AGA	Aspartylglycosaminuria	81479
AGL	Glycogen Storage Disease, Type III	81407
AGXT	Primary Hyperoxaluria, Type 1	81479
AIRE	Polyglandular Autoimmune Syndrome, Type 1	81406
ALDH3A2	Sjogren-Larsson Syndrome	81479
ALDOB	Hereditary Fructose Intolerance	81479
ALG6	Congenital Disorder of Glycosylation, Type Ic	81479
ALMS1	Alstrom Syndrome	81479
ALPL	Hypophosphatasia, Autosomal Recessive	81479
AMT	AMT-Related Glycine Encephalopathy	81479
ARG1	Argininemia	81479
ARSA	Metachromatic Leukodystrophy	81405
ASL	Argininosuccinic Aciduria	81479

Gene	Associated syndromes	CPT code
ASPA	Canavan Disease	81200
ASS1	Citrullinemia, Type 1	81406
ATM	Ataxia-Telangiectasia	81408
ATP7A	ATP7A-Related Disorders	81479
ATP7B	Wilson Disease	81406
BBS1	Bardet-Biedl Syndrome, BBS1-Related	81406
BBS10	Bardet-Biedl Syndrome, BBS10-Related	81404
BBS12	Bardet-Biedl Syndrome, BBS12-Related	81479
BBS2	Bardet-Biedl Syndrome, BBS2-Related	81406
BCKDHA	Maple Syrup Urine Disease, Type Ia	81400, 81405
BCKDHB	Maple Syrup Urine Disease, Type IB	81205, 81406
BCS1L	GRACILE Syndrome	81405
BLM	Bloom Syndrome	81209
BTD	Biotinidase Deficiency	81404
CAPN3	Calpainopathy	81406
CBS	Homocystinuria caused by Cystathionine Beta-Synthase Deficiency	81401, 81406
CFTR	Cystic Fibrosis	81220, 81221, 81222, 81223, 81224
CLN3	CLN3-Related Neuronal Ceroid Lipofuscinosis	81479
CLN5	CLN5-Related Neuronal Ceroid Lipofuscinosis	81479

Gene	Associated syndromes	CPT code
CLN5	CLN5-Related Neuronal Ceroid Lipofuscinosis	81479
CLN6	CLN6-Neuronal Ceroid Lipofuscinosis, Type 6	81479
CLN8	Northern Epilepsy	81479
CLRN1	Usher Syndrome, Type 3	81400, 81404
COL4A3	COL4A3-Related Alport Syndrome	81408
COL4A4	COL4A4-Related Alport Syndrome	81407
COL4A5	Alport Syndrome, X-Linked	81407, 81408
CPS1	Carbamoylphosphate Synthetase I Deficiency	81479
CPT1A	Carnitine Palmitoyltransferase IA Deficiency	81406
CPT2	Carnitine Palmitoyltransferase II Deficiency	81404
CTNS	Cystinosis	81479
CTSK	Pycnodysostosis	81479
CYP11B1	11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	81405
CYP21A2	21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	81402, 81405
CYP27A1	Cerebrotendinous Xanthomatosis	81479
DBT	Maple Syrup Urine Disease, Type II	81405, 81406
DHCR7	Smith-Lemli-Opitz Syndrome	81405
DLD	Lipoamide Dehydrogenase Deficiency	81406
DMD	Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)	81161, 81408
DYSF	Dysferlinopathy	81408
ERCC6	ERCC6-Related Disorders	81479
ERCC8	ERCC8-Related Disorders	81479
EVC	EVC-Related Ellis-Van Creveld Syndrome	81479
EVC2	EVC2-Related Ellis-Van Creveld Syndrome	81479
FAH	Tyrosinemia, Type I	81406
FANCA	Fanconi Anemia Complementation, Group A	81479
FANCC	Fanconi Anemia, Type C	81242

Gene	Associated syndromes	CPT code
FKRP	FKRP-Related Disorders	81404
FKTN	FKTN-Related Disorders	81400, 81405
FMR1	Fragile X Syndrome	81243, 81244
G6PC	Glycogen Storage Disease, Type Ia	81250
GAA	Pompe Disease	81406
GALC	Krabbe Disease	81401, 81406
GALK1	Galactokinase Deficiency	81479
GALT	Galactosemia	81401, 81406
GBA	Gaucher Disease	81251
GCDH	Glutaric Acidemia, Type 1	81406
GJB2	GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness	81252, 81253
GJB6	DFNB1B (aka GJB6-Related Non-syndromic Hearing Loss and Deafness)	81254
GLA	Fabry Disease	81405
GLB1	GLB1-Related Disorders	81479
GLDC	GLDC-Related Glycine Encephalopathy	81479
GNE	Inclusion Body Myopathy 2	81400, 81046
GNPTAB	GNPTAB-Related Disorders	81479
GNPTG	Mucolipidosis III Gamma	81479
GRHPR	Primary Hyperoxaluria, Type 2	81479
HADHA	HADHA-Related Disorders	81406
HBA1/ HBA2	Alpha Thalassemia	81257, 81404, 81405
HBB	Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	81401, 81403, 81404
HEXA	Hexosaminidase A Deficiency (including Tay-Sachs Disease)	81255, 81406
HEXB	Sandhoff Disease	81479
HGSNAT	Mucopolysaccharidosis, Type IIIC	81479
HLCS	Holocarboxylase Synthetase Deficiency	81406

Gene	Associated syndromes	CPT code
HMGCL	HMG-CoA Lyase Deficiency	81479
HOGA1	Primary Hyperoxaluria, Type 3	81479
HSD17B4	D-Bifunctional Protein Deficiency	81479
HYLS1	Hydrolethalus Syndrome	81479
IDS	Mucopolysaccharidosis, Type II	81405
IDUA	Mucopolysaccharidosis, Type I	81406
IKBKAP	Familial Dysautonomia	81260
IL2RG	X-linked Severe Combined Immunodeficiency	81405
IVD	Isovaleric Acidemia	81400, 81406
KCNJ11	KCNJ11-Related Familial Hyperinsulinism	81403
LAMA2	LAMA2-Related Muscular Dystrophy	81408
LAMA3	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	81479
LAMB3	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	81479
LAMC2	Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	81479
LIPA	Lysosomal Acid Lipase Deficiency	81479
LRPPRC	Leigh Syndrome, French-Canadian Type	81479
MAN2B1	Alpha-Mannosidosis	81479
MCOLN1	Mucolipidosis IV	81290
MEFV	Familial Mediterranean Fever	81402, 81404
MESP2	Spondylothoracic Dysostosis	81479
MKS1	MKS1-Related Disorders	81479
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts	81479
MMAA	Methylmalonic Acidemia, cblA Type	81405
MMAB	Methylmalonic Acidemia, cblB Type	81405
MMACHC	Methylmalonic Aciduria and Homocystinuria, cblC Type	81404
MPI	Congenital Disorder of Glycosylation, Type Ib	81405
MTM1	X-Linked Myotubular Myopathy	81405, 81406

Gene	Associated syndromes	CPT code
MUT	MUT-Related Methylmalonic Acidemia	81406
MYO7A	MYO7A-Related Disorders	81407
NAGLU	Mucopolysaccharidosis, Type IIIB	81479
NBN	Nijmegen Breakage Syndrome	81479
NEB	NEB-Related Nemaline Myopathy	81400, 81408
NPC1	Niemann-Pick Disease, Type C	81406
NPC2	Niemann-Pick Disease, Type C2	81404
NPHS1	Congenital Finnish Nephrosis	81407
NPHS2	Steroid-Resistant Nephrotic Syndrome	81405
NR0B1	X-Linked Congenital Adrenal Hypoplasia	81404
OPA3	Costeff Optic Atrophy Syndrome	81479
OTC	Ornithine Transcarbamylase Deficiency	81405
PAH	Phenylalanine Hydroxylase Deficiency	81406
PC	Pyruvate Carboxylase Deficiency	81406
PCCA	PCCA-Related Propionic Acidemia	81405, 81406
PCCB	PCCB-Related Propionic Acidemia	81406
PCDH15	PCDH15-Related Disorders (including Usher Syndrome, Type IF)	81400, 81406, 81407
PEX1	PEX1-Related Zellweger Syndrome Spectrum	81479
PEX10	Peroxisome Biogenesis Disorder, Type 6	81479
PEX12	Peroxisome Biogenesis Disorder, Type 3	81479
PEX2	Peroxisome Biogenesis Disorder, Type 5	81479
PEX6	Peroxisome Biogenesis Disorder, Type 4	81479
PEX7	Rhizomelic Chondrodysplasia Punctata, Type 1	81479
PKHD1	PKHD1-Related Autosomal Recessive Polycystic Kidney Disease	81408
PMM2	Congenital Disorder of Glycosylation, Type Ia	81479
POMGNT1	Muscle-Eye-Brain Disease	81406
PPT1	PPT1-Related Neuronal Ceroid Lipofuscinosis	81479

Gene	Associated syndromes	CPT code
PROP1	PROP1-Related Combined Pituitary Hormone Deficiency	81404
PTS	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	81479
RMRP	Cartilage-Hair Hypoplasia	81479
RS1	X-Linked Juvenile Retinoschisis	81479
RTEL1	RTEL1-Related Disorders	81479
SACS	ARSACS	81479
SGCA	Alpha-Sarcoglycanopathy	81405
SGCB	Beta-Sarcoglycanopathy	81405
SGCD	Delta-Sarcoglycanopathy	81405
SGCG	Gamma-Sarcoglycanopathy	81404, 81405
SGSH	Mucopolysaccharidosis, Type IIIA	81479
SLC12A6	Andermann Syndrome	81479
SLC17A5	Salla Disease	81479
SLC22A5	Primary Carnitine Deficiency	81405
SLC26A2	Sulfate Transporter-Related Osteochondrodysplasia	81479
SLC26A4	Pendred Syndrome	81406

Gene	Associated syndromes	CPT code
SLC37A4	Glycogen Storage Disease, Type Ib	81406
SMN1	Spinal Muscular Atrophy	81400, 81401
SMPD1	Niemann-Pick Disease, SMPD1-Associated	81330
STAR	Lipoid Congenital Adrenal Hyperplasia	81479
TAT	Tyrosinemia, Type II	81479
TCIRG1	Autosomal Recessive Osteopetrosis, Type 1	81479
TGM1	TGM1-Related Autosomal Recessive Congenital Ichthyosis	81479
TH	Segawa Syndrome	81406
TMEM216	Joubert Syndrome 2	81479
TPP1	TPP1-Related Neuronal Ceroid Lipofuscinosis	81479
TTPA	Ataxia with Vitamin E Deficiency	81404
USH1C	USH1C-Related Disorders	81407
USH2A	USH2A-Related Disorders	81408
VPS13B	Cohen Syndrome	81407, 81408
XPA	Xeroderma Pigmentosum, Group A	81479
XPC	Xeroderma Pigmentosum, Group C	81479
ZFYVE26	Spastic Paraplegia, Type 15	81479

Prelude™ Prenatal Screen

Non-invasive prenatal screen (cell-free fetal DNA)

Conditions covered	CPT code
Common aneuploidies including Trisomy 21, Trisomy 18, and Trisomy 13	81420
Microdeletions including 22q11.2 deletion syndrome, 1p36 deletion syndrome, 15q11.2 deletion syndrome, 5p deletion (Cri du chat syndrome), 4p deletion (Wolf-Hirschhorn syndrome)	81422

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