

Cell3™ Target: Carrier Screening Panel

Detects 448 childhood recessive disorders

DISORDER	GENE	DISEASE TYPE
2-Methylbutryl-CoA dehydrogenase deficiency	ACADSB	Metabolic
3-Hydroxy-3-methylglutaryl- CoA lyase deficiency	HMGCL	Metabolic
3-Hydroxyacyl- CoA dehydrogenase deficiency	HADH	Metabolic
3-Methylcrotonyl- CoA carboxylase 2 deficiency	MCCC2	Metabolic
3-Methylglutaconic aciduria, i	AUH	Metabolic
3-Methylglutaconic aciduria, iii	OPA3	Metabolic
3-Methylglutaconic aciduria, v	DNAJC19	Metabolic
α -Thalassemia/mental retardation syndrome, nondeletion, XLR ATRX	ATRX	Hematologic
ABCD syndrome	EDNRB	Cutaneous
Abetalipoproteinemia; ABL	MTTP	Metabolic
Achalasia-Addisonianism-Alacrima syndrome; AAA	AAAS	Endocrine
Achondrogenesis, Ib; ACG1b	SLC26A2	Skeletal
Achromatopsia 3; ACHM3	CNGB3	Ocular
Acrocallosal syndrome; ACLS	GLI3	Developmental
Acyl-CoA dehydrogenase family, member 9, deficiency of	ACAD9	Metabolic
Acyl-CoA dehydrogenase, long-chain, deficiency of	ACADL	Metabolic
Acyl-CoA dehydrogenase, medium-chain, deficiency of	ACADM	Metabolic
Acyl-CoA dehydrogenase, short-chain, deficiency of	ACADS	Metabolic
Acyl-CoA dehydrogenase, very long-chain, deficiency of	ACADVL	Metabolic
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	CYP21A2	Endocrine
Adrenoleukodystrophy; ALD	ABCD1	Neurological
Afibrinogenemia, congenital	FGA	Hematologic
Afibrinogenemia, congenital	FGB	Hematologic

Afibrinogenemia, congenital	FGG	Hematologic
Agammaglobulinemia, XLR, XLA	BTK	Immunodeficiency
Agenesis of the corpus callosum with peripheral neuropathy; ACCPN	SLC12A6	Neurological
Aicar transylase/imp cyclohydrolase, deficiency of	ATIC	Metabolic
Aicardi-Goutieres syndrome 1; AGS1	TREX1	Neurological
Alkaptonuria	HGD	Metabolic
Allan-Herndon-Dudley syndrome AHDS	SLC16A2	Neurological
Alpers diffuse degeneration of cerebral gray matter with hepatic cirrhosis	POLG	Neurological
Alport syndrome, AR	COL4A3	Renal
Alport syndrome, AR	COL4A4	Renal
Alstrom syndrome; ALMS	ALMS1	Neurodegenerative
Amegakaryocytic thrombocytopenia, congenital; CAMT	MPL	Hematologic
Amish infantile epilepsy syndrome	ST3GAL5	Neurological
Amyotrophic lateral sclerosis 2, juvenile; ALS2	ALS2	Neurological
Anauxetic dysplasia	RMRP	Skeletal
Angelman syndrome AS	MECP2	Neurological

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Aniridia, cerebellar ataxia, and mental deficiency	PAX6	Neurological
Antley-Bixler syndrome; ABS	FGFR2	Skeletal
Argininosuccinic aciduria	ASL	Metabolic
Aromatic L-amino acid decarboxylase deficiency	DDC	Metabolic
Arterial calcification, generalized, of infancy; GACL	ENPP1	Cardiac
Arthrogyrosis, renal dysfunction, and cholestasis	VPS33B	Developmental
Arts syndrome	PRPS1	Neurological
Aspartylglucosaminuria	AGA	Metabolic
Ataxia-telangiectasia; AT	ATM	Immunodeficiency
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia;	APTX	Neurological
Atelosteogenesis, ii; AOII	SLC26A2	Developmental
Autoimmune polyendocrine syndrome, i; APS1	AIRE	Immunodeficiency
Barth syndrome; BTHS	TAZ	Cardiac
Bartter syndrome, antenatal, 1	SLC12A1	Renal
Bartter syndrome, antenatal, 2	KCNJ1	Renal
Beta-hydroxyisobutyryl-CoA deacylase, deficiency of	HIBCH	Metabolic
Bile acid synthesis defect, congenital, 4	AMACR	Metabolic
Biotinidase deficiency	BTD	Metabolic
Bloom syndrome; BLM	BLM	Immunodeficiency
Brittle cornea syndrome; BCS	ZNF469	Cutaneous
C syndrome	CD96	Developmental
C-like syndrome	CD96	Developmental
Canavan disease	ASPA	Metabolic
Carbamoyl phosphate synthetase I deficiency, hyperammonemia	CPS1	Metabolic
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase	SCO2	Metabolic
Cardiomyopathy, dilated, 3a; CMD3a	TAZ	Cardiac
Cardiomyopathy, dilated, 3b; CMD3b	DMD	Cardiac

Carnitine palmitoyltransferase i deficiency	CPT1A	Metabolic
Carnitine palmitoyltransferase ii deficiency, infantile	CPT2	Metabolic
Carnitine palmitoyltransferase ii deficiency, late-onset	CPT2	Metabolic
Carnitine palmitoyltransferase ii deficiency, lethal neonatal	CPT2	Metabolic
Carpenter syndrome	RAB23	Developmental
Cartilage-hair hypoplasia; CHH	RMRP	Immunodeficiency
Cerebellar hypoplasia and mental retardation with/without quadrupedal	VLDLR	Neurological
Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma	SNAP29	Neurological
Cerebrooculofacioskeletal syndrome 1; COFS1	ERCC6	Developmental
Cerebrotendinous xanthomatosis	CYP27A1	Metabolic
Ceroid lipofuscinosis, neuronal, 1; CLN1	PPT1	Neurological
Ceroid lipofuscinosis, neuronal, 10; CLN10	CTSD	Neurological
Ceroid lipofuscinosis, neuronal, 2; CLN2	TPP1	Neurological
Ceroid lipofuscinosis, neuronal, 3; CLN3	CLN3	Neurological
Ceroid lipofuscinosis, neuronal, 5; CLN5	CLN5	Neurological
Ceroid lipofuscinosis, neuronal, 6; CLN6	CLN6	Neurological

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Ceroid lipofuscinosis, neuronal, 7; CLN7	MFSD8	Neurological
Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy	CLN8	Neurological
Ceroid lipofuscinosis, neuronal, 8; CLN8	CLN8	Neurological
Charcot-Marie-tooth disease, 4h; CMT4h	FGD4	Neurological
Chediak-Higashi syndrome; CHS	LYST	Immunodeficiency
Cholestasis, progressive familial intrahepatic 1; PFIC1	ATP8B1	Metabolic
Cholestasis, progressive familial intrahepatic 2; PFIC2	ABCB11	Metabolic
Chondrodysplasia punctata 1, xlr recessive; CDPX1	ARSE	Developmental
Chondrodysplasia, blomstrand; BOCD	PTHIR	Skeletal
Choroideremia; CHM	CHM	Ocular
Cirrhosis, familial	KRT18	Hepatic
Cirrhosis, familial	KRT8	Hepatic
Citrullinemia, classic	ASS1	Metabolic
Cockayne syndrome, a; CSA	ERCC8	Neurological
Cockayne syndrome, b; CSB	ERCC6	Neurological
Coenzyme Q10 deficiency	APTX	Neurological
Coenzyme Q10 deficiency	CABC1	Neurological
Coenzyme Q10 deficiency	COQ2	Neurological
Coenzyme Q10 deficiency	PDSS1	Neurological
Coenzyme Q10 deficiency	PDSS2	Neurological
Cohen syndrome; COH1	VPS13B	Neurological
Colorectal adenomatous polyposis, AR	MUTYH	Gastroenterologic
Combined immunodeficiency, XLR; CIDX	IL2RG	Immunodeficiency
Combined oxidative phosphorylation deficiency 1; COXPD1	GFM1	Metabolic
Combined oxidative phosphorylation deficiency 2; COXPD2	MRPS16	Metabolic
Combined oxidative phosphorylation deficiency 3; COXPD3	TSFM	Metabolic
Combined oxidative phosphorylation deficiency 5; COXPD5	MRPS22	Metabolic
Combined saposin deficiency	PSAP	Metabolic
Congenital disorder of glycosylation, 1a; CDG1a	PMM2	Metabolic

Congenital disorder of glycosylation, Ib; CDG1b	MPI	Metabolic
Congenital disorder of glycosylation, Ic; CDG1c	ALG6	Metabolic
Congenital disorder of glycosylation, Ie; CDG1e	DPM1	Metabolic
Congenital disorder of glycosylation, IIa; CDG2A	MGAT2	Metabolic
Congenital disorder of glycosylation, IIb; CDG2B	MOGS	Metabolic
Congenital disorder of glycosylation, IIc; CDG2C	SLC35C1	Metabolic
Congenital disorder of glycosylation, IId; CDG2D	B4GALT1	Metabolic
Congenital disorder of glycosylation, IIf; CDG2F	SLC35A1	Metabolic
Congenital disorder of glycosylation, II; CDG1j	DPAGT1	Metabolic
Congenital disorder of glycosylation, IIk; CDG1k	ALG1	Metabolic
Congenital disorder of glycosylation, Im; CDG1m	DOLK	Metabolic
Corneal dystrophy and perceptive deafness	SLC4A11	Ocular
Corpus callosum, agenesis of, with abnormal genitalia	ARX	Neurological
Corpus callosum, agenesis of, with mental retardation, ocular coloboma,	IGBP1	Developmental
Corpus callosum, partial agenesis of, XLR	LICAM	Neurological

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Creatine deficiency syndrome, XLR	SLC6A8	Neurological
Crisponi syndrome	CRLF1	Developmental
Cutis laxa, AR, i	EFEMP2	Cutaneous
Cutis laxa, AR, i	FBLN5	Cutaneous
Cutis laxa, AR, ii	ATP6V0A2	Cutaneous
Cystic fibrosis; CF	CFTR	Respiratory
Cystinosis, adult nonnephropathic	CTNS	Metabolic
Cystinosis, late-onset juvenile or adolescent nephropathic	CTNS	Metabolic
Cystinosis, nephropathic; CTNS	CTNS	Metabolic
D-2-hydroxyglutaric aciduria	D2HGDH	Metabolic
D-bifunctional protein deficiency	HSD17B4	Metabolic
De sanctis-cacchione syndrome	ERCC6	Cutaneous
De sanctis-cacchione syndrome	XPA	Cutaneous
Deafness, AR 1a	GJB2	Deafness
Deafness, neurosensory, AR 2; DFNB2	MYO7A	Deafness
Desmosterolosis	DHCR24	Developmental
Diarrhea 4, malabsorptive, congenital	NEUROG3	Gastroenterologic
Diastrophic dysplasia	SLC26A2	Developmental
Dihydropyrimidine dehydrogenase; DPYD	DPYD	Metabolic
Donnai-Barrow syndrome	LRP2	Developmental
Donohue syndrome	INSR	Endocrine
Dosage-sensitive sex reversal; DSS	NR0B1	Developmental
Down syndrome	GATA1	Developmental
Dyssegmental dysplasia, Silverman-Handmaker ; DDSH	HSPG2	Skeletal
Ectodermal dysplasia, hypohidrotic, with immune deficiency	IKBKG	Immunodeficiency
Ectodermal dysplasia, hypohidrotic, XLR; XHED	EDA	Cutaneous
Ehlers-Danlos syndrome, AR, cardiac valvular	COL1A2	Cutaneous
Ehlers-Danlos syndrome, vii, AR	ADAMTS2	Neurological
Elejalde disease	MYO5A	Neurological

Encephalopathy, ethylmalonic	ETHE1	Metabolic
Encephalopathy, neonatal severe, due to MECP2 mutations	MECP2	Neurological
Epidermolysis bullosa dystrophica, AR; RDEB	COL7A1	Cutaneous
Epidermolysis bullosa junctionalis with pyloric atresia	ITGA6	Cutaneous
Epidermolysis bullosa junctionalis with pyloric atresia	ITGB4	Cutaneous
Epidermolysis bullosa simplex with muscular dystrophy	PLEC1	Cutaneous
Epidermolysis bullosa, junctional, Herlitz	LAMA3	Cutaneous
Epidermolysis bullosa, junctional, Herlitz	LAMB3	Cutaneous
Epidermolysis bullosa, junctional, Herlitz	LAMC2	Cutaneous
Epidermolysis bullosa, junctional, non-Herlitz	COL17A1	Cutaneous
Epidermolysis bullosa, junctional, non-Herlitz	ITGB4	Cutaneous
Epidermolysis bullosa, junctional, non-Herlitz	LAMA3	Cutaneous
Epidermolysis bullosa, junctional, non-Herlitz	LAMB3	Cutaneous
Epidermolysis bullosa, junctional, non-Herlitz	LAMC2	Cutaneous

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Epidermolysis bullosa, lethal acantholytic	DSP	Cutaneous

Epilepsy, progressive myoclonic 3; EPM3	KCTD7	Neurological
Epileptic encephalopathy, early infantile, 1	ARX	Neurological
Epileptic encephalopathy, early infantile, 2	CDKL5	Neurological
Epileptic encephalopathy, early infantile, 3	SLC25A22	Neurological
Epileptic encephalopathy, early infantile, 4	STXBPI	Neurological
Epileptic encephalopathy, lennox-gastaut	MAPK10	Neurological
Epiphyseal dysplasia, multiple, with early-onset diabetes mellitus	EIF2AK3	Skeletal
Fabry disease	GLA	Metabolic
Factor xi deficiency	F11	Hematologic
Familial mediterranean fever; FMF	MEFV	Immunodeficiency
Fetal akinesia deation sequence; FADS	RAPSN	Developmental
Fibromatosis, juvenile hyaline	ANTXR2	Cutaneous
Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and	WNT7A	Skeletal
Fraser syndrome	FRAS1	Developmental
Fraser syndrome	FREM2	Developmental
Fructose intolerance, hereditary	ALDOB	Metabolic
Fucosidosis	FUCA1	Metabolic
Fukuyama congenital muscular dystrophy; FCMD	FKTN	Neurological
Fumarase deficiency	FH	Metabolic
Galactosemia	GALT	Metabolic
Gaucher disease, i	GBA	Metabolic
Gaucher disease, ii	GBA	Metabolic
Gaucher disease, iii	GBA	Metabolic
Gaucher disease, perinatal lethal	GBA	Metabolic
Geleophysic dysplasia	ADAMTSL2	Skeletal
Glucose-6-phosphate dehydrogenase; G6PD	G6PD	Metabolic
Glutaric acidemia i	GCDH	Metabolic
Glutathione synthetase deficiency	GSS	Hematologic
Glycine encephalopathy; GCE	AMT	Metabolic

Glycine encephalopathy; GCE	GCSH	Metabolic
Glycine encephalopathy; GCE	GLDC	Metabolic
Glycogen storage disease i	G6PC3	Metabolic
Glycogen storage disease ib	SLC37A4	Metabolic
Glycogen storage disease ic	SLC37A4	Metabolic
Glycogen storage disease ii	GAA	Metabolic
Glycogen storage disease iii	AGL	Metabolic
Glycogen storage disease iv	GBE1	Metabolic
Glycogen storage disease of heart, lethal congenital	PRKAG2	Metabolic
Gm1-gangliosidosis, i	GLB1	Neurological
Gm1-gangliosidosis, ii	GLB1	Metabolic
Gracile syndrome	BCSIL	Metabolic
Griscelli syndrome, 1; GS1	MYO5A	Neurological
Griscelli syndrome, 2; GS2	RAB27A	Immunodeficiency

DISORDER	GENE	DISEASE TYPE
Hemochromatosis, 3	TFR2	Hematologic

Hemochromatosis, juvenile; JH	HAMP	Hematologic
Hemochromatosis, juvenile; JH	HFE2	Hematologic
Hemochromatosis; HFE	HFE	Metabolic
Hemochromatosis; HFE	HFE2	Metabolic
Hemoglobin--alpha locus 1; HBA1	HBA1	Hematologic
Hemoglobin--beta locus; HBB	HBB	Hematologic
Hepatic venoocclusive disease with immunodeficiency; VODI	SPI10	Immunodeficiency
Heterotaxy, visceral, 1, XLR; HTX1	ZIC3	Developmental
Homocystinuria	CBS	Metabolic
Homocystinuria due to deficiency of n(5,10)-methylenetetrahydrofolate	MTHFR	Metabolic
Hoyeraal-Hreidarsson syndrome; HHS	DKC1	Immunodeficiency
Hurler syndrome	IDUA	Metabolic
Hyalinosis, infantile systemic	ANTXR2	Skeletal
Hydrocephalus due to congenital stenosis of aqueduct of sylvius; HSAS	LICAM	Neurological
Hydrolethalus syndrome 1	HYLS1	Developmental
Hydrops-ectopic calcification-moth-eaten skeletal dysplasia	LBR	Skeletal
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	SLC25A15	Metabolic
Hyperoxaluria, primary, i	AGXT	Metabolic
Hyperoxaluria, primary, ii	GRHPR	Metabolic
Hypertrophic neuropathy of Dejerine-Sottas. CMT3, CMT4F	EGR2	Neurological
Hypertrophic neuropathy of Dejerine-Sottas. CMT3, CMT4F	MPZ	Neurological
Hypertrophic neuropathy of Dejerine-Sottas. CMT3, CMT4F	PMP22	Neurological
Hypertrophic neuropathy of Dejerine-Sottas. CMT3, CMT4F	PRX	Neurological
Hypomagnesemia, renal, with ocular involvement	CLDN19	Renal
Hypoparathyroidism-retardation-dysmorphism syndrome; HRD	TBCE	Neurological
Hypophosphatasia, childhood	ALPL	Skeletal
Hypophosphatemic rickets, AR	DMP1	Skeletal
Hypoplastic left heart syndrome	GJA1	Cardiac
Hypothyroidism, congenital, nongoitrous, 4; CHNG4	TSHB	Endocrine

Hypotonia-cystinuria syndrome	PREPL	Renal
Hypotonia-cystinuria syndrome	SLC3A1	Renal
Ichthyosis congenita, harlequin fetus	ABCA12	Cutaneous
Ichthyosis, lamellar, 1; il1	TGM1	Cutaneous
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	CLDN1	Cutaneous
Icos deficiency; LCCS2	ERBB3	Developmental
Immunodeficiency with hyper-IGM, 1; HIGM1	CD40LG	Immunodeficiency
Immunodeficiency-centromeric instability-facial anomalies syndrome	DNMT3B	Immunodeficiency
Immunodysregulation, polyendocrinopathy, and enteropathy, XLR	FOXP3	Immunodeficiency
Inclusion body myopathy 2, AR; IBM2	GENE	Neurological
Infantile sialic acid storage disorder	SLC17A5	Metabolic
Infantile-onset spinocerebellar ataxia; IOSCA	C10ORF2	Neurological
Infertile male syndrome	AR	Endocrine

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Insensitivity to pain, congenital, with anhidrosis; CIPA	NTRK1	Neurological
Insulin-like growth factor i, resistance to	IGF1	Endocrine

Intestinal pseudoobstruction, neuronal, chronic idiopathic, XLR	FLNA	Gastroenterologic
Isovaleric acidemia; IVA	IVD	Metabolic
Jervell and Lange-Nielsen syndrome 1; JLNS1	KCNQ1	Cardiac
Johanson-Blizzard syndrome; JBS	UBR1	Developmental
Joubert syndrome 3; JBTS3	AHI1	Neurological
Joubert syndrome 4; JBTS4	NPHP1	Neurological
Joubert syndrome 5; JBTS5	CEP290	Neurological
Joubert syndrome 6; JBTS6	TMEM67	Neurological
Kenny-Caffey syndrome, 1; KCS	TBCE	Endocrine
Krabbe disease	GALC	Metabolic
Krabbe disease, atypical, due to saposin A deficiency	PSAP	Neurological
Lactic acidosis, fatal infantile	SUCLG1	Metabolic
Laryngoonychocutaneous syndrome; LOCS	LAMA3	Cutaneous
Lathosterolosis	SC5DL	Metabolic
Leigh syndrome, French-Canadian ; LSFC	LRPPRC	Metabolic
Leigh syndrome, XLR	PDHA1	Neurological
Lesch-nyhan syndrome; LNS	HPRT1	Neurological
Lethal congenital contracture syndrome 1; LCCS1	GLE1	Developmental
Leukodystrophy, hypomyelinating, 2	GJC2	Neurological
Leukodystrophy, hypomyelinating, 5	FAM126A	Neurological
Lipoid congenital adrenal hyperplasia	CYP11A1	Endocrine
Lipoid congenital adrenal hyperplasia	STAR	Endocrine
Lissencephaly 2; LIS2	RELN	Developmental
Lissencephaly, XLR, 1; LISX1	DCX	Neurological
Lissencephaly, XLR, 2 LISX2	ARX	Developmental
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	Metabolic
Lowe oculocerebrorenal syndrome; OCRL	OCRL	Developmental
Lujan-Fryns syndrome	MEDI2	Neurological
Lymphoproliferative syndrome, XLR, 1; XLPI	SH2D1A	Immunodeficiency

Mandibuloacral dysplasia with b lipodystrophy; MADB	ZMPSTE24	Developmental
Mannosidosis, alpha B, lysosomal	MAN2B1	Metabolic
Maple syrup urine disease ia	BCKDHA	Metabolic
Maple syrup urine disease iii	DLD	Metabolic
Maple syrup urine disease, classic, ib	BCKDHB	Metabolic
Marinesco-Sjogren syndrome	SIL1	Neurological
Martsof syndrome	RAB3GAP2	Neurological
Masa syndrome	LICAM	Neurological
Meckel syndrome, 1; MKS1	MKS1	Developmental
Meckel syndrome, 5; MKS5	RPGRIP1L	Developmental
Megalencephalic leukoencephalopathy with subcortical cysts; MLC	MLC1	Neurological
Menkes disease	ATP7A	Neurological

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Mental retardation, XLR, syndromic 10; Herlitz	HSD17B10	Neurological
Mental retardation, XLR, syndromic, Christianson	SLC9A6	Neurological

Metachromatic leukodystrophy	ARSA	Neurological
Metachromatic leukodystrophy due to saposin b deficiency	PSAP	Neurological
Methylmalonic aciduria and homocystinuria, CBLC	MMACHC	Metabolic
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	MUT	Metabolic
Methylmalonic aciduria, CBLB	MMAB	Metabolic
Mevalonic aciduria	MVK	Metabolic
Microphthalmia, syndromic 9; MCOPS9	STRA6	Developmental
Mitochondrial complex iii deficiency	BCSIL	Metabolic
Mitochondrial complex iii deficiency	UQCRB	Metabolic
Mitochondrial complex iii deficiency	UQCRQ	Metabolic
Mitochondrial DNA depletion syndrome, hepatocerebral	C10ORF2	Neurological
Mitochondrial DNA depletion syndrome, hepatocerebral	DGUOK	Neurological
Mitochondrial DNA depletion syndrome, hepatocerebral	MPV17	Neurological
Mitochondrial DNA depletion syndrome, myopathic	TK2	Neurological
Molybdenum cofactor deficiency	MOCS1	Neurological
Molybdenum cofactor deficiency	MOCS2	Neurological
Mucopolipidosis ii alpha/beta	GNPTAB	Metabolic
Mucopolipidosis iii alpha/beta	GNPTAB	Metabolic
Mucopolipidosis iv	MCOLN1	Metabolic
Mucopolysaccharidosis iiia	SGSH	Metabolic
Mucopolysaccharidosis iiic	HGSNAT	Metabolic
Mucopolysaccharidosis vi	ARSB	Metabolic
Mucopolysaccharidosis vii	GUSB	Metabolic
Mucopolysaccharidosis viii	GNS	Metabolic
Mulibrey nanism	TRIM37	Developmental
Multiple acyl-CoA dehydrogenase deficiency; MADD	ETFA	Metabolic
Multiple acyl-CoA dehydrogenase deficiency; MADD	ETFB	Metabolic
Multiple acyl-CoA dehydrogenase deficiency; MADD	ETFDH	Metabolic
Multiple pterygium syndrome, Escobar	CHRNA3	Developmental

Multiple pterygium syndrome, lethal	CHRNA1	Developmental
Multiple pterygium syndrome, lethal	CHRND	Developmental
Multiple pterygium syndrome, lethal	CHRNA3	Developmental
Muscle-eye-brain disease; MEB	FKRP	Neurological
Muscle-eye-brain disease; MEB	POMGNT1	Neurological
Muscular dystrophy, congenital merosin-deficient, 1a; MDC1a	LAMA2	Neurological
Muscular dystrophy, congenital, 1c; MDC1c	FKRP	Neurological
Muscular dystrophy, congenital, 1d	LARGE	Neuromuscular
Muscular dystrophy, duchenne ; DMD	DMD	Neurological
Muscular dystrophy, limb-girdle, 2d; LGMD2d	SGCA	Neurological
Myoadenylate deaminase deficiency, myopathy due to	AMPD1	Neuromuscular
Myoclonic epilepsy of Lafora	EPM2A	Neurological

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Myoclonic epilepsy of Lafora	NHLRC1	Neurological
Myoclonic epilepsy of Unverricht and Lundborg	CSTB	Neurological

Myopathy, early-onset, with fatal cardiomyopathy	TTN	Cardiac
Myotubular myopathy 1; MTM1	MTM1	Neurological
Myxoma, intracardiac	PRKARIA	Cardiac
N-acetylglutamate synthase deficiency	NAGS	Metabolic
Navajo neurohepatopathy; NN	MPV17	Neurological
Nemaline myopathy 2; NEM2	NEB	Neurological
Nemaline myopathy 5; NEM5	TNNT1	Neurological
Nephronophthisis 1; NPHP1	NPHP1	Renal
Nephronophthisis 2; NPHP2	INVS	Renal
Nephronophthisis 4; NPHP4	NPHP4	Renal
Nephrosis 1, congenital, Finnish; NPHS1	NPHS1	Renal
Nephrotic syndrome, 3; NPHS3	PLCE1	Renal
Nephrotic syndrome, early-onset, with diffuse mesangial sclerosis	WT1	Renal
Neuraminidase deficiency	NEU1	Metabolic
Neuroaxonal dystrophy, infantile; inad1	PLA2G6	Neurological
Neuropathy, hereditary sensory and autonomic, iii; HSAN3	IKBKAP	Neurological
Neuropathy, hypomyelinating/Charcot-Marie-tooth disease, 4e	EGR2	Neurological
Neuropathy, hypomyelinating/Charcot-Marie-tooth disease, 4e	MPZ	Neurological
Nevo syndrome	PLOD1	Developmental
Newborn pulmonary hypertension, familial persistent	CPS1	Respiratory
Niemann-pick disease, a	SMPD1	Metabolic
Niemann-pick disease, b	SMPD1	Metabolic
Niemann-pick disease, c1; NPC1	NPC1	Metabolic
Niemann-pick disease, c2	NPC2	Metabolic
Nijmegen breakage syndrome	NBN	Immunodeficiency
Norrie disease; ND	NDP	Developmental
Odontoonychodermal dysplasia; OODD	WNT10A	Cutaneous
Omenn syndrome	DCLRE1C	Immunodeficiency
Omenn syndrome	RAG1	Immunodeficiency

Omenn syndrome	RAG2	Immunodeficiency
Opticoacoustic nerve atrophy with dementia	TIMM8A	Developmental
Ornithine transcarbamylase deficiency, hyperammonemia due to	OTC	Metabolic
Osteogenesis imperfecta, IIb	CRTAP	Skeletal
Osteogenesis imperfecta, viii	LEPRE1	Skeletal
Osteopetrosis, AR 1; OPTB1	TCIRG1	Skeletal
Osteopetrosis, AR 3; OPTB3	CA2	Skeletal
Osteopetrosis, AR 5; OPTB5	OSTM1	Skeletal
Osteopetrosis, lymphedema, ectodermal dysplasia, anhidrosis, immunodeficiency, OLEDAID	IKBKG	Immunodeficiency
Osteoporosis-pseudoglioma syndrome; OPPG	LRP5	Skeletal
Otospondylomegaepiphyseal dysplasia; OSMED	COL11A2	Skeletal
Otospondylomegaepiphyseal dysplasia; OSMED	COL2A1	Skeletal

DISORDER	GENE	DISEASE TYPE
Paget disease, juvenile	TNFRSF11B	Skeletal
Pelizaeus-merzbacher disease; PMD	PLP1	Neurological
Pendred syndrome; PDS	SLC26A4	Endocrine
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	Metabolic

Phenylketonuria; PKU	PAH	Metabolic
Phosphoserine aminotransferase deficiency	PSAT1	Metabolic
Pierson syndrome	LAMB2	Renal
Pituitary dwarfism iii	HESX1	Endocrine
Pituitary dwarfism iii	LHX3	Endocrine
Pituitary dwarfism iii	POU1F1	Endocrine
Pituitary dwarfism iii	PROPI	Endocrine
Plasminogen deficiency, i	PLG	Hematologic
Polycystic kidney disease, AR; ARPKD	PKHD1	Renal
Pontocerebellar hypoplasia 2a; PCH2A	TSEN54	Neurological
Pontocerebellar hypoplasia 4; PCH4	TSEN54	Neurological
Porphyria, congenital erythropoietic	UROS	Cutaneous
Primary lateral sclerosis, juvenile; PLSJ	ALS2	Neurological
Properdin deficiency, XLR	CFP	Immunodeficiency
Protease inhibitor 1; PI	SERPINA1	Hepatic
Pseudohypaldosteronism, i, ar; PHA1	SCNNIA	Renal
Pseudohypaldosteronism, i, ar; PHA1	SCNNIB	Renal
Pseudohypaldosteronism, i, ar; PHA1	SCNNIG	Renal
Pulmonary alveolar microlithiasis	SLC34A2	Respiratory
Pulmonary venoocclusive disease; PVOD	BMPR2	Respiratory
Pycnodysostosis	CTSK	Metabolic
Pyridoxamine 5-prime-phosphate oxidase deficiency	PNPO	Metabolic
Pyruvate carboxylase deficiency	PC	Metabolic
Pyruvate dehydrogenase E3-binding protein deficiency	PDHX	Metabolic
Pyruvate dehydrogenase phosphatase deficiency	PDPI	Metabolic
Pyruvate kinase deficiency of red cells	PKLR	Hematologic
Raine syndrome; RNS	FAM20C	Skeletal
Renal tubular dysgenesis; RTD	ACE	Renal
Renal tubular dysgenesis; RTD	AGT	Renal

Renal tubular dysgenesis; RTD	AGTR1	Renal
Renal tubular dysgenesis; RTD	REN	Renal
Renal-hepatic-pancreatic dysplasia; RHPD	NPHP3	Renal
Renpenning syndrome 1; RENS1	PQBPI	Developmental
Respiratory distress syndrome in premature infants	SFTPA1	Respiratory
Respiratory distress syndrome in premature infants	SFTPB	Respiratory
Respiratory distress syndrome in premature infants	SFTPC	Respiratory
Retinoschisis 1, XLR, juvenile; RS1	RS1	Ocular
Rett syndrome; RTT	MECP2	Neurological
Rhizomelic chondrodysplasia punctata, 1; RCDP1	PEX7	Developmental
Rhizomelic chondrodysplasia punctata, 3; RCDP3	AGPS	Metabolic

DISORDER	GENE	DISEASE TYPE
Rigid spine muscular dystrophy 1; RSMD1	SEPN1	Neurological
Roberts syndrome; RBS	ESCO2	Developmental
Sandhoff disease	HEXB	Neurological
Schindler disease, i	NAGA	Metabolic

Schneckenbecken dysplasia	SLC35D1	Skeletal
Seckel syndrome 1	ATR	Skeletal
Segawa syndrome, AR	TH	Metabolic
Senior-Loken syndrome 1; SLSN1	NPHP1	Renal
Senior-Loken syndrome 5; SLSN5	IQCB1	Renal
Severe combined immunodeficiency, AR, T-cell negative,	ADA	Immunodeficiency
Severe combined immunodeficiency, AR, T-cell negative,	RAG1	Immunodeficiency
Severe combined immunodeficiency, AR, T-cell negative,	RAG2	Immunodeficiency
Severe combined immunodeficiency, AR, T-cell negative, B-cell positive, NK cell negative	JAK3	Immunodeficiency
Severe combined immunodeficiency, XLR; SCIDX1	IL2RG	Immunodeficiency
Shwachman-diamond syndrome; SDS	SBDS	Hematologic
Sialuria, Finnish	SLC17A5	Metabolic
Sickle cell anemia	HBB	Hematologic
Simpson-Golabi-Behmel syndrome, 2	OFD1	Developmental
Sjogren-Larsson syndrome; SLS	ALDH3A2	Neurological
Skin fragility-woolly hair syndrome	DSP	Cutaneous
Smith-lemli-opitz syndrome; SLOS	DHCR7	Developmental
Spastic ataxia, charlevoix-saguenay ; SACS	SACS	Neurological
Spastic paraplegia 2, XLR; SPG2	PLP1	Neurological
Spinal muscular atrophy, distal, AR, 1; DSMA1	IGHMBP2	Neurological
Spinal muscular atrophy, distal, AR, 4; DSMA4	PLEKHG5	Neurological
Spinal muscular atrophy, i; SMA1	SMN1	Neurological
Spinal muscular atrophy, ii; SMA2	SMN1	Neurological
Spinal muscular atrophy, iii; SMA3	SMN1	Neurological
Spinal muscular atrophy, XLR 2; SMAX2	UBA1	Neurological
Spondylocostal dysostosis, AR 1; SCDO1	DLL3	Skeletal
Striatonigral degeneration, infantile; SNDI	NUP62	Neurological
Stuve-Wiedemann syndrome	LIFR	Skeletal
Succinic semialdehyde dehydrogenase deficiency	ALDH5A1	Metabolic

Sudden infant death with dysgenesis of the testes syndrome; SIDDT	TSPYL1	Respiratory
Sulfocysteinuria	SUOX	Metabolic
Surfactant metabolism dysfunction, pulmonary, 1; SMDP1	SFTPB	Metabolic
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1	Immunodeficiency
Tay-Sachs disease; TSD	HEXA	Neurological
Tetra-Amelia, AR	WNT3	Developmental
Thrombophilia due to activated protein c resistance	F5	Hematologic
Thrombophilia, hereditary, due to protein C deficiency, autosomal	PROC	Hematologic
Thrombotic thrombocytopenic purpura, congenital; TTP	ADAMTS13	Hematologic
Tight skin contracture syndrome, lethal	LMNA	Cutaneous

DISORDER	GENE	DISEASE TYPE
Tight skin contracture syndrome, lethal	ZMPSTE24	Cutaneous
Trichothiodystrophy, photosensitive; TTDP	ERCC2	Cutaneous
Trichothiodystrophy, photosensitive; TTDP	ERCC3	Cutaneous
Trichothiodystrophy, photosensitive; TTDP	GTF2H5	Cutaneous
Trifunctional protein deficiency	HADHA	Metabolic

Trifunctional protein deficiency	HADHB	Metabolic
Tyrosinemia, i	FAH	Metabolic
Ulna and fibula, absence of, with severe limb deficiency	WNT7A	Skeletal
Usher syndrome, i	MYO7A	Ocular
Usher syndrome, ic; USH1c	USH1C	Ocular
Usher syndrome, id; USH1d	CDH23	Ocular
Usher syndrome, if; USH1f	PCDH15	Ocular
Usher syndrome, ig; USH1g	USH1G	Ocular
Usher syndrome, iia; USH2a	USH2A	Ocular
Usher syndrome, iic; USH2c	GPR98	Ocular
Usher syndrome, iii; USH3	CLRN1	Ocular
Vacterl association with hydrocephalus, XLR	FANCB	Developmental
Vitamin D-dependent rickets, i	CYP27B1	Endocrine
Vitamin D-dependent rickets, ii	VDR	Skeletal
Vitamin E, familial isolated deficiency of; VED	TTPA	Neurological
Waardenburg-Shah syndrome	EDN3	Cutaneous
Waardenburg-Shah syndrome	EDNRB	Cutaneous
Waardenburg-Shah syndrome	SOX10	Cutaneous
Walker-Warburg syndrome; WWS	POMT1	Neurological
Walker-Warburg syndrome; WWS	POMT2	Neurological
Warburg micro syndrome; WARBM	RAB3GAP1	Developmental
Wilson disease	ATP7B	Metabolic
Wiskott-Aldrich syndrome; WAS	WAS	Immunodeficiency
Xeroderma pigmentosum, complementation group a; XPA	XPA	Cutaneous
Xeroderma pigmentosum, complementation group b; XPB	ERCC3	Cutaneous
Xeroderma pigmentosum, complementation group d; XPD	ERCC2	Cutaneous
Xeroderma pigmentosum, complementation group e; XPE	DDB2	Cutaneous
Xeroderma pigmentosum, complementation group f; XPF	ERCC4	Cutaneous
Xeroderma pigmentosum, complementation group g; XPG	ERCC5	Cutaneous

Yemenite deaf-blind hypopigmentation syndrome	SOX10	Cutaneous
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