INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS

A broad menu covering the vast majority of inherited metabolic disorders that are routinely tested as part of state newborn screening programs. Our curated panels are designed by medical genetics experts based on disorders and/or analyte results.

CLINICAL AREA: PORPHYRIAS

Test name	# gene(s)	Gene list
Porphyria		
Invitae Acute Hepatic Porphyrias Panel	4	ALAD, CPOX, HMBS, PPOX

CLINICAL AREA: PRIMARY HYPEROXALURIAS

Test name	# gene(s)	Gene list
Primary Hyperoxalurias		
Invitae Primary Hyperoxaluria Panel	3	AGXT, GRHPR, HOGA1

continued on page 2

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test name		# gene(s)	Gene list
Metabolic Disorders Newborn Screening Confirmation			
Invitae Metabolic Disorders Newborn Screening Confirmation Panel		93	ABCD1, ABCD4, ACAD8, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, AHCY, ALDH4A1, ARG1, ASL, ASS1, AUH, BCKDHA, BCKDHB, BTD, CBS, CD320, CFTR, CPS1, CPT1A, CPT2, DBT, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FAH, FTCD, G6PD, GAA, GALE, GALK1, GALT, GCDH, GCH1, GLA, GNMT, GSS, HADH, HADHA, HADHB, HCFC1, HLCS, HMGCL, HPD, HSD17B10, IDUA, IVD, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, OAT, OPA3, OTC, PAH, PCBD1, PC, PCCA, PCCB, PPM1K, PRODH, PTS, QDPR, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC52A1, SLC52A2, SLC52A3, SMPD1, SPR, SUCLA2, SUCLG1, TAT, TAZ, TMEM70
	Add-on 2,4-dienoyl-CoA reductase deficiency genes	1	NADK2
	Add-on cerebral creatine deficiency genes	3	GAMT, GATM, SLC6A8
	Add-on congenital disorders of glycosylation genes	102	ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, C1GALT1C1, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, FKRP, FKTN, G6PC3, GALNT3, GFPT1, GMPPA, GMPPB, GNE, ISPD, LARGE1, LFNG, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, NUS1, PAPSS2, PGM1, PGM3, PIGA, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RFT1, RPN2, RXYLT1, SEC23A, SEC23B, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TMEM165, TRIP11, TUSC3, XYLT1
	Add-on generalized leukodystrophies genes	6	ARSA, ASPA, GALC, GM2A, HEXA, HEXB
	Add-on glucose transporter type 1 (GLUT1) deficiency gene	1	SLC2A1
	Add-on glycine encephalopathy genes	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
	Add-on mucopolysaccharidosis type II (MPSII) gene	1	IDS
	Add-on Niemann-Pick type C genes	2	NPC1, NPC2
	Add-on pyridoxal 5'-phosphate-dependent epilepsy gene	1	PNPO
	Add-on pyridoxine-responsive epilepsy gene	1	ALDH7A1
	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
	Add-on cerebrotendinous xanthomatosis gene	1	CYP27A1
	Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene	1	HMGCS2
	Add-on neuronal ceroid lipofuscinosis genes	10	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
	Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene	1	OXCT1
	Add-on elevated very long chain fatty acids genes	14	ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
Invitae Lysosomal Storage Disorders Newborn Screening Panel		6	GAA, GALC, GLA, IDS, IDUA, SMPD1
Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test		1	ABCD1
	Add on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
	Add-on elevated very long chain fatty acids genes	13	HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test name	# gene(s)	Gene list
Panels by Analyte		
Invitae Low C0 Test	1	SLC22A5
Invitae Elevated C0/(C16+C18) Test	1	CPT1A
Invitae Elevated C3 Panel	15	ABCD4, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, TCN2
Add-on ACSF3 gene	1	ACSF3
Invitae Elevated C3-DC Test	1	MLYCD
Invitae Elevated C4 Panel	3	ACAD8, ACADS, ETHE1
Add-on limited evidence gene	1	FTCD
Invitae Elevated C4-DC Panel	2	SUCLA2, SUCLG1
Invitae Elevated C4-OH Panel	2	HADH, HIBCH
Invitae Elevated C4 & C5 Panel	7	ETFA, ETFB, ETFDH, ETHE1, SLC52A1, SLC52A2, SLC52A3
Invitae Elevated C5 Panel	2	ACADSB, IVD
Invitae Elevated C5-DC Test	1	GCDH
Invitae Elevated C5-OH Panel	13	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, MCCC1, MCCC2, OPA3, SERAC1, TAZ
Invitae Elevated C6, C8 & C10 Test	1	ACADM
Invitae Elevated C14 & C14:1 Test	1	ACADVL
Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	2	HADHA, HADHB
Invitae Elevated C16, C16:1, C18, & C18:1 Panel	2	CPT2, SLC25A20
Invitae Elevated Arginine Test	1	ARG1
Invitae Elevated Citrulline Panel	4	ASL, ASS1, PC, SLC25A13
Add-on dihydrolipoamide dehydrogenase deficiency gene	1	DLD
Invitae Low Citrulline Panel	3	CPS1, NAGS, OTC
Add-on limited evidence gene	1	ALDH18A1
Invitae Elevated Glycine Panel	6	AMT, GLDC, GCSH, LIAS, NFU1, SLC6A9
Add-on organic acidemia genes	56	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC13A5, SLC25A1, SLC25A19, SUCLA2, SUCLG1, TAZ, TMEM70
Invitae Elevated Leucine Panel	5	BCKDHA, BCKDHB, DBT, DLD, PPM1K
Invitae Elevated Methionine Panel	4	AHCY, CBS, GNMT, MAT1A
Add-on additional causes of elevated methionine genes	2	FAH, SLC25A13
Invitae Elevated Phenylalanine Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR

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Test name	# gene(s)	Gene list
Panels by Analyte (continued)		
Invitae Elevated Proline Panel	2	ALDH4A1, PRODH
Invitae Elevated Succinylacetone Test	1	FAH
Invitae Elevated Tyrosine Panel	3	FAH, HPD, TAT
Aminoacidopathies		
Invitae Alkaptonuria Test	1	HGD
Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
Invitae Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9
Invitae Disorders of Serine Biosynthesis Panel	3	PHGDH, PSAT1, PSPH
Invitae Glycine Encephalopathy Panel	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
Invitae Homocystinuria Panel	4	CBS, MTHFR, MTR, MTRR
Add-on combined methylmalonic acidemia and homocystinuria genes	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
Add-on elevated methionine genes	4	AHCY, CBS, GNMT, MAT1A
Invitae Hyperphenylalaninemia Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
Invitae Hyperprolinemia Panel	2	ALDH4A1, PRODH
Invitae Maple Syrup Urine Disease Panel	4	BCKDHA, BCKDHB, DBT, PPM1K
Add-on DLD gene	1	DLD
Invitae Tyrosinemia Panel	3	FAH, HPD, TAT
Carbohydrate Disorders		
Invitae Galactosemia Panel	3	GALE, GALK1, GALT
Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	1	G6PD
Invitae Glucose Transporter Type 1 (GLUT1) Deficiency Syndrome Test	1	SLC2A1
Invitae Comprehensive Glycogen Storage Disease Panel	23	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4
Add-on fatty acid oxidation genes	21	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3
Add-on limited evidence genes	2	PGM1, POLG
Invitae Liver Glycogen Storage Disease Panel	11	AGL, FBP1, G6PC, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4
Invitae Muscle Glycogen Storage Disease Panel	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
Add-on fatty acid oxidation genes	21	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3
Add-on limited evidence genes	2	PGM1, POLG
Invitae Hereditary Fructose Intolerance Test	1	ALDOB
Invitae Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1

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Test name	# gene(s)	Gene list
Cerebrotendinous Xanthomatosis		
Invitae Cerebrotendinous Xanthomatosis Test	1	CYP27A1
Add-on sitosterolemia genes	2	ABCG5, ABCG8
Congenital Disorders of Glycosylation		
Invitae Congenital Disorders of Glycosylation Panel	50	ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, B3GLCT, CHST14, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, G6PC3, GFPT1, GMPPA, GMPPB, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PGM3, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, ST3GAL5, TMEM165, TRIP11, TUSC3
Add-on preliminary-evidence genes	11	ALG14, B4GALT1, DDOST, NUS1, PIGM, RPN2, SEC23A, SLC35A3, ST3GAL3, STT3A, STT3B
Add-on disorders of O-mannosylation genes	13	B3GALNT2, B4GAT1, FKRP, FKTN, GNE, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1
Add-on glycosylation genes not involved in N-glycosylation	29	B3GALT6, B3GAT3, B4GALNT1, B4GALT7, C1GALT1C1, CHST3, CHST6, CHSY1, DSE, EOGT, EXT1, EXT2, GALNT3, LFNG, PAPSS2, PIGA, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, POFUT1, POGLUT1, SLC26A2, SLC35D1, XYLT1
Creatine Biosynthesis Disorders		
Invitae Cerebral Creatine Deficiency Panel	3	GAMT, GATM, SLC6A8
Cystic Fibrosis		
Invitae Cystic Fibrosis Newborn Screening Confirmation Test	1	CFTR
Fatty Acid Oxidation Defects		
Invitae Fatty Acid Oxidation Defects Panel	17	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, SLC22A5, SLC25A20
Add-on 2,4-dienoyl-CoA reductase deficiency gene	1	NADK2
Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
Add-on muscle glycogen storage disorders	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
Invitae Ketogenesis Disorders Panel	2	HMGCL, HMGCS2
Invitae Ketolysis Disorders Panel	2	ACAT1, OXCT1
Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADM
Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel	3	ETFA, ETFB, ETFDH
Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADVL
Lysosomal Storage Disorders		
Invitae Comprehensive Lysosomal Storage Disorders Panel	48	AGA, ARSA, ARSB, ASAH1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSK, FUCA1, GAA, GALC, GALNS, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1
Add-on chitotriosidase deficiency gene	1	CHIT1
Add-on preliminary-evidence gene	1	ATP13A2
Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN

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CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test name	# gene(s)	Gene list
Lysosomal Storage Disorders (continued)		
Invitae Cystinosis Test	1	CTNS
Invitae Farber Lipogranulomatosis Test	1	ASAH1
Invitae Fabry Disease Test	1	GLA
Invitae GM2 Gangliosidosis Panel	3	GM2A, HEXA, HEXB
Invitae Krabbe Disease Test	1	GALC
Add-on prosaposin deficiency gene	1	PSAP
Invitae Lysosomal Acid Lipase Deficiency Test	1	LIPA
Invitae Metachromatic Leukodystrophy Panel	3	ARSA, PSAP, SUMF1
Add-on generalized leukodystrophies genes	4	ASPA, GALC, HEXA, HEXB
Invitae Mucolipidosis Panel	4	GNPTAB, GNPTG, MCOLN1, NEU1
Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel	11	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH
Add-on mucolipidosis and oligosaccharidoses genes	12	AGA, CTSA, CTSK, FUCA1, GNPTAB, GNPTG, MAN2B1, MANBA, MCOLN1, NAGA, NEU1, SLC17A5
Invitae Mucopolysaccharidosis Type I (MPS I) Test	1	IDUA
Add-on clinically overlapping lysosomal storage disorder genes	5	ARSB, GNPTAB, GUSB, IDS, SUMF1
Invitae Mucopolysaccharidosis Type II Test	1	IDS
Add-on clinically overlapping genes	4	GNPTAB, GUSB, IDUA, SUMF1
Invitae Mucopolysaccharidosis Type III (MPS III) Panel	4	GNS, HGSNAT, NAGLU, SGSH
Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel	2	GALNS, GLB1
Add-on multiple sulfatase deficiency gene	1	SUMF1
Invitae Multiple Sulfatase Deficiency Test	1	SUMF1
Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel	9	CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
Add-on preliminary-evidence gene	1	ATP13A2
Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
Invitae Niemann-Pick Disease Types A and B Test	1	SMPD1
Add-on chitotriosidase deficiency gene	1	CHIT1
Invitae Niemann-Pick Type C Panel	2	NPC1, NPC2
Add-on lysosomal acid lipase deficiency gene	1	LIPA
Add-on chitotriosidase deficiency gene	1	CHIT1
Invitae Oligosaccharidoses Panel	8	AGA, CTSA, CTSK, FUCA1, MAN2B1, MANBA, NAGA, SLC17A5
Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH

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Test name	# gene(s)	Gene list
Lysosomal Storage Disorders (continued)		
Invitae Pompe Disease Test	1	GAA
Add-on Danon disease gene	1	LAMP2
Add-on primary carnitine deficiency gene	1	SLC22A5
Invitae Prosaposin Deficiency Test	1	PSAP
Invitae Sandhoff Disease Test	1	HEXB
Add-on Tay-Sachs disease gene	1	HEXA
Invitae Tay-Sachs Disease Test	1	HEXA
Add-on Sandhoff disease gene	1	HEXB
Metal Transport Disorders		
Invitae ATP7A-Related Disorders Test	1	ATP7A
Invitae Copper Metabolism Disorders Panel	5	AP1S1, ATP7A, ATP7B, CP, SLC33A1
Invitae Wilson Disease Test	1	ATP7B
Neurotransmitter Disorders		
Invitae Neurotransmitter Disorders Panel	27	ABAT, ALDH5A1, ALDH7A1, AMT, ARHGEF9, DBH, DDC, GAD1, GCH1, GCSH, GLDC, GLRA1, GLRB, GPHN, MAOA, PCBD1, PHGDH, PNPO, PSAT1, PSPH, PTS, QDPR, SLC25A22, SLC6A3, SLC6A5, SPR, TH
Add-on neurodegeneration with brain iron accumulation genes	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
Invitae Hereditary Hyperekplexia Panel	6	ARHGEF9, CLPB, GLRA1, GLRB, GPHN, SLC6A5
Organic Acidemias		
Invitae Organic Acidemias Panel	49	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC25A1, SUCLA2, SUCLG1, TAZ, TMEM70
Add-on Kreb cycle defect genes	7	DHTKD1, DLD, FH, NFU1, OGDH, SLC13A5, SLC25A19
Invitae 2-Hydroxyglutaric Aciduria Panel	4	D2HGDH, IDH2, L2HGDH, SLC25A1
Invitae 3-Methylcrotonyl CoA Carboxylase Panel	2	MCCC1, MCCC2
Invitae 3-Methylglutaconic Aciduria Panel	8	AUH, CLPB, CPS1, DNAJC19, OPA3, SERAC1, TAZ, TMEM70
Invitae Barth Syndrome Test	1	TAZ
Invitae Biotinidase Deficiency Test	1	BTD
Invitae Canavan Disease Test	1	ASPA
Invitae Glutaric Acidemia Type I Test	1	GCDH
Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
Invitae Methylmalonic Acidemia Panel	7	MMAA, MMAB, MMADHC, MCEE, MUT, SUCLA2, SUCLG1
Add-on combined malonic and methylmalonic acidemia gene	1	ACSF3
Add-on combined methylmalonic acidemia and homocystinuria genes	10	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, TCN1, TCN2

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Test name	# gene(s)	Gene list
Organic Acidemias (continued)		
Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel	3	ETFA, ETFB, ETFDH
Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
Invitae Multiple Carboxylase Deficiency Panel	2	BTD, HLCS
Invitae Propionic Acidemia Panel	2	PCCA, PCCB
Add-on methylmalonic acidemia genes	5	MMAA, MMAB, MMADHC, MMACHC, MUT
Add-on multiple carboxylase deficiency genes	2	BTD, HLCS
Peroxisomal Disorders		
Invitae Adult Refsum Disease Panel	2	PEX7, PHYH
Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7
Invitae X-linked Adrenoleukodystrophy (X-ALD) Test	1	ABCD1
Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
Add-on elevated very long chain fatty acids genes	13	HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
Invitae Zellweger Spectrum Disorder Panel	15	ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6
Purine Metabolism Disorders		
Invitae Purine Metabolism Disorders Panel	9	ADA, ADSL, AMPD1, HPRT1, GPHN, MOCOS, MOCS1, PNP, XDH
Add-on sulfite oxidase deficiency gene	1	SUOX
Invitae Lesch-Nyhan Syndrome Test	1	HPRT1
Pyruvate Metabolism and Tricarboxylic Acid Cycle Defects		
Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel	3	DLD, OGDH, SLC25A19
Add-on alpha-ketoadipic acid dehydrogenase deficiency gene	1	DHTKD1
Invitae Citrate Transporter Deficiency Test	1	SLC13A5
Invitae Dihydrolipoamide Dehydrogenase Deficiency Test	1	DLD
Invitae Fumarase Deficiency Test	1	FH
Invitae Pyruvate Carboxylase Deficiency Test	1	PC
Invitae Pyruvate Dehydrogenase Deficiency Panel	8	DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, PDP1

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels		
Invitae Hyperammonemia Panel	58	ABCD4, ACADM, ACADVL, ALDH18A1, ARG1, ASL, ASS1, BCKDHA, BCKDHB, BTD, CA5A, CPS1, CPT1A, CPT2, DBT, DLAT, DLD, ETFA, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, OAT, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, TAZ, TMEM70, UMPS
Invitae Mendelian Disorders with Psychiatric Symptoms Panel	88	ABCD1, ADSL, ALDH5A1, AMACR, AMT, AP1S1, ARG1, ARSA, ASL, ASS1, ATP7B, ATP13A2, BCKDHA, BCKDHB, BCKDK, C19orf12, CA5A, CBS, CLN3, CLN5, CLN6, CLN8, COASY, CP, CPS1, CTSD, CYP27A1, DBH, DBT, DCAF17, DDC, DLD, FAH, FTL, FUCA1, GALC, GAMT, GATM, GCH1, GCSH, GLA, GLB1, GLDC, GM2A, GNS, GSS, HEXA, HEXB, HGSNAT, HMGCL, HPRT1, MAN2B1, MANBA, MAOA, MFSD8, MMACHC, MTHFR, MTR, NAGLU, NAGS, NPC1, NPC2, OTC, PAH, PANK2, PCCA, PCCB, PLA2G6, POLG, PPT1, PRODH, PSAP, PTS, QDPR, SGSH, SLC6A8, SLC7A7, SLC25A13, SLC25A15, SLC52A1, SLC52A2, SLC52A3, SPR, SUMF1, TBX1, TH, TPP1, WDR45
Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
Invitae Metabolic Non-Immune Fetal Hydrops Panel	51	AHCY, ALG1, ALG12, ALG8, ALG9, ARSB, ASAH1, CTSA, DHCR7, G6PD, GAA, GALC, GALNS, GBE1, GLB1, GLUL, GNPTAB, GUSB, HADH, HADHA, HADHB, IDUA, LIPA, MVK, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIGA, PMM2, PSAT1, SLC17A5, SLC22A5, SLC26A2, SMPD1, SUMF1, TAZ, TRIP11
Add-on limited evidence gene	1	SEC23B
Treatable Disorders		
Invitae Treatable Neurometabolic Disorders Panel	102	ABCD1, ABCD4, ACAT1, AGA, ALDH5A1, ALDH7A1, AMN, AMT, ARG1, ARHGEF9, ARSA, ASL, ASS1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BTD, CBS, CD320, CLN2 (TPP1), CP, CPS1, CUBN, CYP27A1, DBT, DHCR7, DLAT, DLD, ETFA, ETFB, ETFDH, ETHE1, GALC, GAMT, GATM, GCDH, GCH1, GCSH, GIF, GLA, GLDC, GLRA1, GLRB, GLUD1, GUSB, HCFC1, HLCS, HMGCL, HMGCS2, HSD17B10, IDS, IDUA, IVD, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MTHFR, MTR, MTRR, MUT, NAGS, NPC1, NPC2, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PHGDH, PNPO, PPM1K, PSAT1, PSPH, PTS, QDPR, SGSH, SLC19A3, SLC25A13, SLC25A15, SLC2A1, SLC6A5, SLC6A8, SPR, TAT, TCN1, TCN2, TH
Add-on neurometabolic conditions genes	42	ABAT, ADSL, AP1S1, ATP13A2, BCKDK, C19orf12, CLN3, CLN5, CLN6, CLN8, CLPB, COASY, CTSD, D2HGDH, DBH, DCAF17, DDC, FA2H, FTL, GAD1, GNS, GPHN, HEXA, HEXB, HGSNAT, HPRT1, IDH2, KCTD7, L2HGDH, MAOA, MFSD8, MOCOS, NAGLU, PLA2G6, POLG, PPT1, SLC13A5, SLC33A1, SLC6A3, SUOX, WDR45, XDH
Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test	1	SLC19A3
Urea Cycle Disorders		
Invitae Urea Cycle Disorders Panel	10	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15
Add-on hyperammonemia genes	4	CA5A, GLUD1, GLUL, SLC7A7
Add-on hereditary orotic aciduria gene	1	UMPS
Invitae Arginase Deficiency Test	1	ARG1
Invitae Ornithine Transcarbamylase (OTC) Deficiency Test	1	OTC
Add-on hereditary orotic aciduria gene	1	UMPS
Add-on low citrulline genes	2	CPS1, NAGS

