Metabolic Disorders & Newborn Screening

NEW GENES AND PANELS, APRIL 2017

INVITAE

Invitae has added 54 new panels and expanded 21 panels for metabolic disorders and newborn screening. The panels are based on the latest research—one more way in which we are working to provide you and your patients the answers that you need.

LYSOSOMAL STORAGE DISORDERS PANELS - **NEW AND UPDATED**

The new Invitae Comprehensive Lysosomal Storage Disorders Panel includes 52 genes covering both common and rare lysosomal storage disorders (LSDs), which can be helpful for diagnosing patients with suspected LSDs in whom previous workup has been negative. Both pediatric and later onset disorders are included in this panel.

For patients with positive biochemical findings, we offer more focused LSD panels for molecular diagnosis. Overall, our menu includes testing for Fabry, Pompe, Krabbe, Niemann-Pick types A, B, C, mucopolysaccharidoses (MPSs), mucolipidoses, oligosaccharidoses, neuronal ceroid lipofuscinoses, and more. While Gaucher's disease is not currently included on our menu, we are continuing our work to include testing for this condition.

INVITAE METABOLIC DISORDERS NEWBORN SCREENING CONFIRMATION PANEL - **UPDATED**

This 90-gene panel has been updated to meet the growing needs of expanded newborn screening. Key updates include testing for X-linked adrenoleukodystrophy, lysosomal storage disorders, and organic acidemias. Furthermore, we have expanded testing for the combined methylmalonic acidemia and homocystinuria disorders to be comprehensive for diseases of cobalamin absorption, transport, and intracellular metabolism.

DID YOU KNOW?

Pathogenic copy number and large indel variants make up 10% of all pathogenic variants, yet are traditionally considered difficult to identify using next-generation sequencing. Invitae's methods have been shown to routinely identify these variants, in large part because we include deletion/duplication analysis with all testing at no additional charge.

Learn more in Rebecca Truty's ACMG presentation, Tracing the dark matter: Prevalence of copy number and structural variants across Mendelian disorders, available at: www.invitae.com/presentations

INVITAE TREATABLE NEUROMETABOLIC DISORDERS PANEL - **NEW**

Our new 92-gene Invitae Treatable Neurometabolic Disorders Panel covers a wide range of genes enabling an early diagnosis critical to clinical intervention and includes the option to add 41 additional genes if desired.

INVITAE NEUROTRANSMITTER DISORDERS PANEL - **NEW**

The 27-gene Invitae Neurotransmitter Disorders Panel can be used as an alternative to an invasive lumbar puncture when patients present with symptoms suggestive of a neurotransmitter disorder, and includes the option to add 10 additional genes if desired.

PEROXISOMAL DISORDERS PANEL - **NEW**

Four new tests have been designed to cover peroxisomal disorders including the X-linked Adrenoleukodystrophy Test, the Invitae Zellweger Spectrum Disorder Panel, the Invitae Adult Refsum Disease Panel, and the Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel.

PANELS BY ANALYTE - **NEW AND UPDATED**

Along with updates to several existing analyte panels, we have added several new panels including the Invitae Low Citrulline Panel, Invitae Elevated Glycine Panel, Invitae Elevated Methionine Panel, and Invitae Elevated Tyrosine Panel. All of these panels cover a range of analytes and can be used to ensure full differential testing for a given abnormal newborn screening result or abnormal biochemical result.

UREA CYCLE DISORDERS PANELS - **NEW AND UPDATED**

We offer comprehensive testing for urea cycle disorders, which now includes testing for N-acetylglutamate synthase (NAGS) deficiency with the choice to add hereditary orotic aciduria (UMPS deficiency), carbonic anhydrase deficiency, lysinuric protein intolerance, and other rare causes of hyperammonemia at no additional charge.

PANELS FOCUSED ON DEFECTS OF CARBOHYDRATE METABOLISM INCLUDING GLYCOGEN STORAGE DISORDERS - **NEW**

Our new offerings include the Invitae Comprehensive Glycogen Storage Disease Panel, plus separate glycogen storage disease panels for liver and muscle isoforms, testing for hereditary fructose intolerance, glucose-galactose malabsorption, and fructose-1, 6-bisphosphatase deficiency associated with hypoglycemia and metabolic acidosis.

PURINE METABOLISM DISORDERS PANELS - **NEW**

The Invitae Purine Metabolism Disorders Panel covers 11 genes including HPRT1 testing for Lesch-Nyhan Syndrome. HPRT1 can be ordered as part of the comprehensive panel, or as a single-gene test.

PYRUVATE METABOLISM AND TRICARBOXYLIC ACID CYCLE DEFECTS PANELS - **NEW**

We have added a new test for the pyruvate carboxylase (PC) gene associated with pyruvate carboxylase deficiency as well as the new eight-gene Invitae Pyruvate Dehydrogenase Deficiency Panel that includes sequencing as well as deletion and duplication testing for DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, and PDP1

KETOGENESIS AND KETOLYSIS DISORDERS PANELS - **NEW AND UPDATED**

Both the new Invitae Ketogenesis Disorders Panel and Invitae Ketolysis Disorders Panel provide comprehensive tests for disorders of ketone metabolism.

All new and updated panels are now available. Visit www.invitae.com to order online or download a paper order form.

If you have any questions, please do not hesitate to contact Client Services at 800-436-3037 or clientservices@invitae.com, or reach out to your regional manager.

.

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact |

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: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

METABOLIC DISORDERS NEWBORN SCREENING CONFIRMATION

Invitae Metabolic Disorders Newborn Screening Confirmation Panel (up to 229 genes)

Primary panel (90 genes)

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	SMPD1
SPR	SUCLA2	SUCLG1	TAT	TAZ	TMEM70						

Add-on 2,4-dienoyl-CoA reductase deficiency genes (2 genes)

DECR1 NADK2

Add-on cerebral creatine deficiency genes (3 genes)

GAMT GATM SLC6A8

Add-on congenital disorders of glycosylation genes (102 genes)

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	SEC23A
SEC23B	SLC26A2	SLC35A1	SLC35A2	SLC35A3	SLC35C1	SLC35D1	SRD5A3	SSR4	ST3GAL3	ST3GAL5	STT3A
STT3B	TMEM165	TMEM5	TRIP11	TUSC3	XYLT1						

Add-on generalized leukodystrophies genes (6 genes)

ARSA ASPA GALC GM2A HEXA HEXB

Add-on glucose transporter type 1 deficiency gene (1 gene)

SLC2A1

Add-on glycine encephalopathy genes (6 genes)

AMT GCSH GLDC LIAS NFU1 SLC6A9

Add-on mucopolysaccharidosis type II gene (1 gene)

IDS

Add-on Niemann-Pick type C genes (2 gene)

NPC1 NPC2

Add-on pyridoxal 5'-phosphate-dependent epilepsy gene (1 gene)

PNPO

Add-on pyridoxine-responsive epilepsy gene (1 gene)

ALDH7A1

Add-on Smith-Lemli-Opitz syndrome gene (1 gene)

DHCR7

Add-on cerebrotendinous xanthomatosis gene (1 gene)

CYP27A1

Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene (1 gene)

HMGCS2

Add-on neuronal ceroid lipofuscinosis genes (10 genes)

ATP13A2 CLN2 (TPP1) CLN3 CLN5 CLN6 CLN8 CTSD KCTD7 MFSD8 PPT1

Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene (1 gene)

OXCT1

Invitae Lysosomal Storage Disorders Newborn Screening Panel (6 genes)

Primary panel (6 genes)

GAA GALC GLA IDS IDUA SMPD1

Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test (1 gene)

Primary panel (1 gene)

ABCD1

PANELS BY ANALYTE

Invitae Low C0 Test (1 gene)

Primary panel (1 gene)

SLC22A5

Invitae Elevated C0/(C16+C18) Test (1 gene)

Primary panel (1 gene)

CPT1A

Invitae Elevated C3 Panel (9 genes)

Primary panel (9 genes)

BTD HLCS MMAA MMAB MMACHC MMADHC MUT PCCA PCCB

Invitae Elevated C3-DC Test (1 gene)

Primary panel (1 gene)

MLYCD

INVITAE

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

PANELS BY ANALYTE (continued)

Invitae Elevated C4 Panel (3 genes)	**Primary panel (3 genes)** ACAD8 ACADS ETHE1
Invitae Elevated C4-OH Panel (2 genes)	**Primary panel (2 genes)** HADH HIBCH
Invitae Elevated C4 & C5 Panel (7 genes)	**Primary panel (7 genes)** ETFA ETFB ETFDH ETHE1 SLC52A1 SLC52A2 SLC52A3
Invitae Elevated C5 Panel (2 genes)	**Primary panel (2 genes)** ACADSB IVD
Invitae Elevated C5-DC Test (1 gene)	**Primary panel (1 gene)** GCDH
Invitae Elevated C5-OH Panel (12 genes)	**Primary panel (12 genes)** ACAT1 AUH BTD DNAJC19 HLCS HMGCL HSD17B10 MCCC1 MCCC2 OPA3 SERAC1 TAZ
Invitae Elevated C6, C8 & C10 Test (1 gene)	**Primary panel (1 gene)** ACADM
Invitae Elevated C14 & C14:1 Test (1 gene)	**Primary panel (1 gene)** ACADVL
Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel (2 genes)	**Primary panel (2 genes)** HADHA HADHB
Invitae Elevated C16, C16:1, C18, & C18:1 Panel (2 genes)	**Primary panel (2 genes)** CPT2 SLC25A20
Invitae Elevated Arginine Test (1 gene)	**Primary panel (1 gene)** ARG1
Invitae Elevated Citrulline Panel (up to 5 genes)	**Primary panel (4 genes)** ASL ASS1 PC SLC25A13 **Add-on dihydrolipoamide dehydrogenase deficiency gene (1 gene)** DLD
Invitae Low Citrulline Panel (3 genes)	**Primary panel (3 genes)** CPS1 NAGS OTC
Invitae Elevated Glycine Panel (up to 62 genes)	**Primary panel (6 genes)** AMT GLDC GCSH LIAS NFU1 SLC6A9 **Add-on organic acidemia genes (56 genes)** 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 genes)	**Primary panel (5 genes)** BCKDHA BCKDHB DBT DLD PPM1K
Invitae Elevated Methionine Panel (up to 6 genes)	**Primary panel (4 genes)** AHCY CBS GNMT MAT1A **Add-on additional causes of elevated methionine genes (2 genes)** FAH SLC25A13
Invitae Elevated Phenylalanine Panel (6 genes)	**Primary panel (6 genes)** GCH1 PAH PCBD1 PTS QDPR SPR
Invitae Elevated Proline Panel (2 genes)	**Primary panel (2 genes)** ALDH4A1 PRODH
Invitae Elevated Succinylacetone Test (1 gene)	**Primary panel (1 gene)** FAH
Invitae Elevated Tyrosine Panel (3 genes)	**Primary panel (3 genes)** FAH HPD TAT

AMINOACIDOPATHIES

Invitae Alkaptonuria Test (1 gene)	**Primary panel (1 gene)** HGD
Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel (11 genes)	**Primary panel (11 genes)** ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMACHC MMADHC TCN1 TCN2
Invitae Cystinuria Panel (3 genes)	**Primary panel (3 genes)** PREPL SLC3A1 SLC7A9
Invitae Disorders of Serine Biosynthesis Panel (3 genes)	**Primary panel (3 genes)** PHGDH PSAT1 PSPH
Invitae Glycine Encephalopathy Panel (6 genes)	**Primary panel (6 genes)** AMT GCSH GLDC LIAS NFU1 SLC6A9

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

AMINOACIDOPATHIES (continued)

Invitae Homocystinuria Panel (up to 19 genes)

Primary panel (4 genes)
CBS MTHFR MTR MTRR

Add-on combined methylmalonic acidemia and homocystinuria genes (11 genes)
ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMACHC MMADHC TCN1 TCN2

Add-on elevated methionine genes (4 genes)
AHCY CBS GNMT MAT1A

Invitae Hyperphenylalaninemia Panel (6 genes)

Primary panel (6 genes)
GCH1 PAH PCBD1 PTS QDPR SPR

Invitae Hyperprolinemia Panel (2 genes)

Primary panel (2 genes)
ALDH4A1 PRODH

Invitae Maple Syrup Urine Disease Panel (up to 5 genes)

Primary panel (4 genes)
BCKDHA BCKDHB DBT PPM1K

Add-on DLD gene (1 gene)
DLD

Invitae Tyrosinemia Panel (3 genes)

Primary panel (3 genes)
FAH HPD TAT

CARBOHYDRATE DISORDERS

Invitae Galactosemia Panel (3 genes)

Primary panel (3 genes)
GALE GALK1 GALT

Invitae Glucose-6-Phosphate Dehydrogenase Deficiency Test (1 gene)

Primary panel (1 gene)
G6PD

Invitae Glucose Transporter Type 1 Deficiency Syndrome Test (1 gene)

Primary panel (1 gene)
SLC2A1

Invitae Comprehensive Glycogen Storage Disease Panel (23 genes)

Primary panel (23 genes)
AGL ALDOA ENO3 FBP1 G6PC GAA GBE1 GYG1 GYS1 GYS2 LAMP2 LDHA
PFKM PGAM2 PHKA1 PHKA2 PHKB PHKG2 PYGL PYGM RBCK1 SLC2A2 SLC37A4

Invitae Liver Glycogen Storage Disease Panel (11 genes)

Primary panel (11 genes)
AGL FBP1 G6PC GBE1 GYS2 PHKA2 PHKB PHKG2 PYGL SLC2A2 SLC37A4

Invitae Muscle Glycogen Storage Disease Panel (up to 15 genes)

Primary panel (14 genes)
ALDOA ENO3 GAA GBE1 GYG1 GYS1 LAMP2 LDHA PFKM PGAM2 PHKA1 PHKB
PYGM RBCK1

Add-on PGM1 gene (1 gene)
PGM1

Invitae Hereditary Fructose Intolerance Test (1 gene)

Primary panel (1 gene)
ALDOB

Invitae Rare Carbohydrate Disorders Panel (2 genes)

Primary panel (2 genes)
FBP1 SLC5A1

CEREBROTENDINOUS XANTHOMATOSIS

Invitae Cerebrotendinous Xanthomatosis Test (up to 3 genes)

Primary panel (1 gene)
CYP27A1

Add-on sitosterolemia genes (2 genes)
ABCG5 ABCG8

CONGENITAL DISORDERS OF GLYCOSYLATION

Invitae Congenital Disorders of Glycosylation Panel (up to 103 genes)

Primary panel (49 genes)
ALG1 ALG11 ALG12 ALG13 ALG2 ALG3 ALG6 ALG8 ALG9 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 TMEM165 TRIP11
TUSC3

Add-on preliminary-evidence genes (11 genes)
ALG14 B4GALT1 DDOST NUS1 PIGM RPN2 SEC23A SLC35A3 ST3GAL3 STT3A STT3B

Add-on disorders of O-mannosylation genes (13 genes)
B3GALNT2 B4GAT1 FKRP FKTN GNE ISPD LARGE1 POMGNT1 POMGNT2 POMK POMT1 POMT2
TMEM5

Add-on glycosylation genes not involved in N-glycosylation genes (30 genes)
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 ST3GAL5 XYLT1

CREATINE BIOSYNTHESIS AND TRANSPORT DISORDERS

Invitae Cerebral Creatine Deficiency Panel (3 genes)

Primary panel (3 genes)
GAMT GATM SLC6A8

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

CYSTIC FIBROSIS

Invitae Cystic Fibrosis Newborn Screening Confirmation Test (1 gene)	**Primary panel (1 gene)** CFTR

FATTY ACID OXIDATION DEFECTS

Invitae Fatty Acid Oxidation Defects Panel (up to 22 genes)	**Primary panel (18 genes)** ACADM ACADS ACADSB ACADVL CPT1A CPT2 ETFA ETFB ETFDH HADH HADHA HADHB HMGCL HMGCS2 MLYCD NADK2 SLC22A5 SLC25A20 **Add-on preliminary-evidence gene (1 gene)** DECR1 **Add-on riboflavin transporter deficiency genes (3 genes)** SLC52A1 SLC52A2 SLC52A3
Invitae Ketogenesis Disorders Panel (2 genes)	**Primary panel (2 genes)** HMGCL HMGCS2
Invitae Ketolysis Disorders Panel (2 genes)	**Primary panel (2 genes)** ACAT1 OXCT1
Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test (1 gene)	**Primary panel (1 gene)** ACADM
Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel (up to 6 genes)	**Primary panel (3 genes)** ETFA ETFB ETFDH **Add-on riboflavin transporter deficiency genes (3 genes)** SLC52A1 SLC52A2 SLC52A3
Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test (1 gene)	**Primary panel (1 gene)** ACADVL

LYSOSOMAL STORAGE DISORDERS

Invitae Comprehensive Lysosomal Storage Disorders Panel (up to 53 genes)	**Primary panel (52 genes)** AGA ARSA ARSB ASAH1 ATP13A2 CLN2 (TPP1) CLN3 CLN5 CLN6 CLN8 CTNS CTSA CTSD CTSF CTSK DNAJC5 FUCA1 GAA GALC GALNS GLA GLB1 GM2A GNPTAB GNPTG GNS GRN 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 gene)** CHIT1
Invitae Cystinosis Test (1 gene)	**Primary panel (1 gene)** CTNS
Invitae Farber Lipogranulomatosis Test (1 gene)	**Primary panel (1 gene)** ASAH1
Invitae Fabry Disease Test (1 gene)	**Primary panel (1 gene)** GLA
Invitae GM2 Gangliosidosis Panel (3 genes)	**Primary panel (3 genes)** GM2A HEXA HEXB
Invitae Krabbe Disease Panel (up to 2 genes)	**Primary panel (1 gene)** GALC **Add-on prosaposin deficiency gene (1 gene)** PSAP
Invitae Lysosomal Acid Lipase Deficiency Test (1 gene)	**Primary panel (1 gene)** LIPA
Invitae Metachromatic Leukodystrophy Panel (up to 7 genes)	**Primary panel (3 genes)** ARSA PSAP SUMF1 **Add-on generalized leukodystrophies genes (4 genes)** ASPA GALC HEXA HEXB
Invitae Mucolipidosis Panel (4 genes)	**Primary panel (4 genes)** GNPTAB GNPTG MCOLN1 NEU1
Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel (up to 23 genes)	**Primary panel (11 genes)** ARSB GALNS GLB1 GNS GUSB HGSNAT HYAL1 IDS IDUA NAGLU SGSH **Add-on mucolipidosis and oligosaccharidoses genes (12 genes)** AGA CTSA CTSK FUCA1 GNPTAB GNPTG MAN2B1 MANBA MCOLN1 NAGA NEU1 SLC17A5
Invitae Mucopolysaccharidosis Type I (MPS I) Test (up to 6 genes)	**Primary panel (1 gene)** IDUA **Add-on clinically overlapping lysosomal storage disorder genes (5 genes)** ARSB GNPTAB GUSB IDS SUMF1
Invitae Mucopolysaccharidosis Type II Test (up to 5 genes)	**Primary panel (1 gene)** IDS **Add-on clinically overlapping genes (4 gene)** GNPTAB GUSB IDUA SUMF1

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

LYSOSOMAL STORAGE DISORDERS (continued)

Invitae Mucopolysaccharidosis Type III (MPS III) Panel (4 genes)	**Primary panel (4 genes)**										
	GNS	HGSNAT	NAGLU	SGSH							
Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel (up to 3 genes)	**Primary panel (2 genes)**										
	GALNS	GLB1									
	Add-on multiple sulfatase deficiency gene (1 gene)										
	SUMF1										
Invitae Multiple Sulfatase Deficiency Test (up to 16 genes)	**Primary panel (1 gene)**										
	SUMF1										
	Add-on mucolipidosis and mucopolysaccharidosis genes (15 genes)										
	ARSB	GALNS	GLB1	GNPTAB	GNPTG	GNS	GUSB	HGSNAT	HYAL1	IDS	IDUA	MCOLN1
	NAGLU	NEU1	SGSH									
Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel (up to 13 genes)	**Primary panel (9 genes)**										
	CLN2 (TPP1)	CLN3	CLN5	CLN6	CLN8	CTSD	KCTD7	MFSD8	PPT1		
	Add-on preliminary-evidence gene (1 gene)										
	ATP13A2										
	Add-on adult-onset neuronal ceroid lipofuscinoses genes (3 gene)										
	CTSF	DNAJC5	GRN								
Invitae Niemann-Pick Disease Types A and B Test (up to 2 genes)	**Primary panel (1 gene)**										
	SMPD1										
	Add-on chitotriosidase deficiency gene (1 gene)										
	CHIT1										
Invitae Niemann-Pick Type C Panel (up to 3 genes)	**Primary panel (2 genes)**										
	NPC1	NPC2									
	Add-on lysosomal acid lipase deficiency gene (1 gene)										
	LIPA										
Invitae Oligosaccharidoses Panel (up to 23 genes)	**Primary panel (8 genes)**										
	AGA	CTSA	CTSK	FUCA1	MAN2B1	MANBA	NAGA	SLC17A5			
	Add-on mucolipidosis and mucopolysaccharidosis genes (15 genes)										
	ARSB	GALNS	GLB1	GNPTAB	GNPTG	GNS	GUSB	HGSNAT	HYAL1	IDS	IDUA	MCOLN1
	NAGLU	NEU1	SGSH									
Invitae Pompe Disease Test (up to 3 genes)	**Primary panel (1 gene)**										
	GAA										
	Add-on Danon disease gene (1 gene)										
	LAMP2										
	Add-on primary carnitine deficiency gene (1 gene)										
	SLC22A5										
Invitae Prosaposin Deficiency Test (1 gene)	**Primary panel (1 gene)**										
	PSAP										
Invitae Sandhoff Disease Test (up to 2 genes)	**Primary panel (1 gene)**										
	HEXB										
	Add-on Tay-Sachs disease gene (1 gene)										
	HEXA										
Invitae Tay-Sachs Disease Test (up to 2 genes)	**Primary panel (1 gene)**										
	HEXA										
	Add-on Sandhoff disease gene (1 gene)										
	HEXB										

METAL TRANSPORT DISORDERS

Invitae ATP7A-Related Disorders (1 gene)	**Primary panel (1 gene)**										
	ATP7A										
Invitae Copper Metabolism Disorders Panel (5 genes)	**Primary panel (5 genes)**										
	AP1S1	ATP7A	ATP7B	CP	SLC33A1						
Invitae Wilson Disease Test (1 gene)	**Primary panel (1 gene)**										
	ATP7B										

NEUROTRANSMITTER DISORDERS

Invitae Neurotransmitter Disorders Panel (up to 37 genes)	**Primary panel (27 genes)**										
	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 genes)										
	ATP13A2	C19orf12	COASY	CP	DCAF17	FA2H	FTL	PANK2	PLA2G6	WDR45		
Invitae Hereditary Hyperekplexia Panel (6 genes)	**Primary panel (6 genes)**										
	ARHGEF9	CLPB	GLRA1	CLRB	GPHN	SLC6A5						

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

ORGANIC ACIDEMIAS

Invitae Organic Acidemias Panel (up to 56 genes)	**Primary panel (49 genes)**											
	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 genes)											
	DHTKD1	DLD	FH	NFU1	OGDH	SLC13A5	SLC25A19					
Invitae 2-Hydroxyglutaric Aciduria Panel (4 genes)	**Primary panel (4 genes)**											
	D2HGDH	IDH2	L2HGDH	SLC25A1								
Invitae 3-Methylcrotonyl CoA Carboxylase Panel (2 genes)	**Primary panel (2 genes)**											
	MCCC1	MCCC2										
Invitae 3-Methylglutaconic Aciduria Panel (7 genes)	**Primary panel (7genes)**											
	AUH	CLPB	DNAJC19	OPA3	SERAC1	TAZ	TMEM70					
Invitae Barth Syndrome Test (1 gene)	**Primary panel (1 gene)**											
	TAZ											
Invitae Biotinidase Deficiency Test (1 gene)	**Primary panel (1 gene)**											
	BTD											
Invitae Canavan Disease Test (1 gene)	**Primary panel (1 gene)**											
	ASPA											
Invitae Glutaric Acidemia Type I Test (1 gene)	**Primary panel (1 gene)**											
	GCDH											
Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel (11 genes)	**Primary panel (11 genes)**											
	ABCD4	AMN	CD320	CUBN	GIF	HCFC1	LMBRD1	MMADHC	MMADHC	TCN1	TCN2	
Invitae Methylmalonic Acidemia Panel (up to 18 genes)	**Primary panel (7 genes)**											
	MMAA	MMAB	MMADHC	MCEE	MUT	SUCLA2	SUCLG1					
	Add-on combined malonic and methylmalonic acidemia (1 gene)											
	ACSF3											
	Add-on combined methylmalonic acidemia and homocystinuria genes (10 genes)											
	ABCD4	AMN	CD320	CUBN	GIF	HCFC1	LMBRD1	MMACHC	TCN1	TCN2		
Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel (up to 6 genes)	**Primary panel (3 genes)**											
	ETFA	ETFB	ETFDH									
	Add-on riboflavin transporter deficiency genes (3 genes)											
	SLC52A1	SLC52A2	SLC52A3									
Invitae Multiple Carboxylase Deficiency Panel (2 genes)	**Primary panel (2 genes)**											
	BTD	HLCS										
Invitae Propionic Acidemia Panel (up to 9 genes)	**Primary panel (2 genes)**											
	PCCA	PCCB										
	Add-on methylmalonic acidemia genes (5 genes)											
	MMAA	MMAB	MMADHC	MMACHC	MUT							
	Add-on multiple carboxylase deficiency genes (2 genes)											
	BTD	HLCS										

PEROXISOMAL DISORDERS

Invitae Adult Refsum Disease Panel (2 genes)	**Primary panel (2 genes)**											
	PEX7	PHYH										
Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel (3 genes)	**Primary panel (3 genes)**											
	AGPS	GNPAT	PEX7									
Invitae X-linked Adrenoleukodystrophy Test (up to 15 genes)	**Primary panel (1 gene)**											
	ABCD1											
	Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene (1 gene)											
	ACOX1											
	Add-on elevated very long chain fatty acids genes (13 genes)											
	HSD17B4	PEX1	PEX2	PEX3	PEX5	PEX6	PEX10	PEX12	PEX13	PEX14	PEX16	PEX19
	PEX26											
Invitae Zellweger Spectrum Disorder Panel (15 genes)	**Primary panel (15 genes)**											
	ACOX1	AMACR	HSD17B4	PEX1	PEX10	PEX12	PEX13	PEX14	PEX16	PEX19	PEX2	PEX26
	PEX3	PEX5	PEX6									

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

PURINE METABOLISM DISORDERS

Invitae Purine Metabolism Disorders Panel (up to 10 genes)

Primary panel (9 genes)

ADA	ADSL	AMPD1	HPRT1	GPHN	MOCOS	MOCS1	PNP	XDH

Add-on sulfite oxidase deficiency gene (1 gene)

SUOX

Invitae Lesch-Nyhan Syndrome Test (1 gene)

Primary panel (1 gene)

HPRT1

PYRUVATE METABOLISM AND TRICARBOXYLIC ACID CYCLE DEFECTS

Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel (up to 4 genes)

Primary panel (3 genes)

DLD	OGDH	SLC25A19

Add-on alpha-ketoadipic acid dehydrogenase deficiency gene (1 gene)

DHTKD1

Invitae Citrate Transporter Deficiency Test (1 gene)

Primary panel (1 gene)

SLC13A5

Invitae Dihydrolipoamide Dehydrogenase Deficiency Test (1 gene)

Primary panel (1 gene)

DLD

Invitae Fumarase Deficiency Test (1 gene)

Primary panel (1 gene)

FH

Invitae Pyruvate Carboxylase Deficiency Test (1 gene)

Primary panel (1 gene)

PC

Invitae Pyruvate Dehydrogenase Deficiency Panel (8 genes)

Primary panel (8 genes)

DLAT	DLD	LIAS	MPC1	PDHA1	PDHB	PDHX	PDP1

TREATABLE DISORDERS

Invitae Treatable Neurometabolic Disorders Panel (up to 133 genes)

Primary panel (92 genes)

ABCD1	ACAT1	AGA	ALDH5A1	ALDH7A1	AMN	AMT	ARG1	ARSA	ASL	ASS1	ATP7A
ATP7B	AUH	BCKDHA	BCKDHB	BTD	CBS	CLN2 (TPP1)	CP	CPS1	CUBN	CYP27A1	DBT
DHCR7	DLAT	DLD	ETFA	ETFB	ETFDH	ETHE1	GAMT	GATM	GCDH	GCH1	GCSH
GIF	GLA	GLDC	GLUD1	GUSB	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	SLC6A8	SPR	TAT	TH				

Add-on neurometabolic conditions genes (41 genes)

ABAT	ADSL	AP1S1	ATP13A2	BCKDK	C19orf12	CLN3	CLN5	CLN6	CLN8	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 gene)

Primary panel (1 gene)

SLC19A3

UREA CYCLE DISORDERS

Invitae Urea Cycle Disorders Panel (up to 15 genes)

Primary panel (10 genes)

ALDH18A1	ARG1	ASL	ASS1	CPS1	NAGS	OAT	OTC	SLC25A13	SLC25A15

Add-on hyperammonemia genes (4 genes)

CA5A	GLUD1	GLUL	SLC7A7

Add-on hereditary orotic aciduria gene (1 gene)

UMPS

Invitae Arginase Deficiency Test (1 gene)

Primary panel (1 gene)

ARG1

Invitae Ornithine Transcarbamylase (OTC) Deficiency Test (up to 4 genes)

Primary panel (1 gene)

OTC

Add-on hereditary orotic aciduria gene (1 gene)

UMPS

Add-on low citrulline genes (2 genes)

CPS1	NAGS

INVITAE

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact |