INVITAE

ORDER ONLINE at www.invitae.com/signin.

ORDER ID
For Invitae internal use only

Requisition Form
METABOLIC TRF913-8

PATIENT INFORMATION

First name | MI | Last name

Date of birth (MM/DD/YYYY) | Sex ◯ M ◯ F | MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other:

Email address (for report access after release by medical professional)

Phone | Is this patient deceased? ◯ Yes ◯ No Deceased date:

Address | City

State | ZIP code | Country

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date.
A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva ◯ DNA - source:

DNA must be extracted in a CLIA or other suitably certified laboratory

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Collection date (MM/DD/YYYY) *If not provided, date will be 1 day prior to our receipt of specimen. For DNA, provide date retrieved from archive.*

Special cases : ◯ History of/current hematologic malignancy ◯ Resubmission

REASON FOR TESTING

Primary indication:

ICD-10 codes | **Previous results**

Testing for a personal history of disease? ◯ **Yes** ◯ **No** If yes, describe below.
Age at diagnosis: _____

ORGANIZATION INFORMATION

Organization name and address

Organization name

Phone | Fax

Address | City

State | ZIP code | Country

Primary clinical contact

Name | Role/title

Phone | NPI

Email address (for report access)

Ordering physician

◯ **Same as primary clinical contact**

Name | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

Letter of Medical Necessity (LMN)

◯ I have attached an LMN and/or other documentation for insurance billing purposes.

◯ I agree to allow Invitae to transfer the information from this requisition to an LMN and/or other documentation using the ordering physician's name as the signature for insurance billing.

Family history? ◯ **Yes** ◯ **No** If yes, describe in detail below or attach pedigree. If there is a known familial variant, indicate here.

◯ INSURANCE BILLING (U.S. ONLY)

◯ **I have attached a copy of the patient's card**

Insurance company name | Member ID#

Patient relation to policy holder: ◯ Self ◯ Child ◯ Spouse ◯ Other

Policy holder name | Prior-authorization #

◯ PATIENT PAY BILLING

Invitae will send an electronic invoice to the patient email listed above

◯ INSTITUTIONAL BILLING

◯ **Send invoice to organization address above**

Billing contact name | Phone | Fax

Billing email address

Billing address | City

State | ZIP code | Country

◯ OTHER BILLING

Invitae partner code:

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/patient-consent), and has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated). The Patient has further been informed and hereby authorizes Invitae Corporation ("Invitae") and its designees to release information concerning testing to their insurer in order to process and/or appeal claims on behalf of the Patient. For amounts received directly, the Patient agrees to remit payment to Invitae for testing services rendered. I acknowledge that I offered pre-test Genetic Counseling to the Patient, if required by their insurer. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test.

Medical professional signature | **Date**

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 |

ORDER INSTRUCTIONS

Select a pre-curated test, combine multiple tests, or customize your own test for each patient. Invitae's pricing is per clinical area for initial order and re-requisition. All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send in two sample tubes and your order will represent two billable events. Your test results will be delivered as two reports. Please contact Client Services with any questions. For Invitae's full test menu, please visit **www.invitae.com**.

RE-REQUISITION

Invitae offers one re-requisition at no additional charge within 90 days for genes within the original clinical area. For more information and to request online, please visit **www.invitae.com/re-requisition**.

FAMILY VARIANT TESTING

Invitae offers Family Variant Testing at no additional charge within 90 days for the genes in which the original family member's variant was identified. In such cases, please use the Family Variant Testing/VUS Resolution requisition form (TRF920), available at **www.invitae.com/forms**.

To request a complimentary specimen collection kit visit **www.invitae.com/request-a-kit**

SHIPPING INSTRUCTIONS

Please ship specimen overnight in insulated containers:

Attn: Invitae Client Services
1400 16th Street
San Francisco, CA 94103
USA

PRELIMINARY-EVIDENCE GENES

Invitae's primary panels contain genes for which there is definitive evidence that variants in these genes cause specific diseases. Preliminary-evidence genes are genes for which there is only early evidence of a relationship between variants in these genes and specific diseases. All preliminary-evidence genes are indicated as such on the requisition form below.

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at **www.invitae.com/physician/search**.

Test continued on next page

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s). To avoid confusion, please consider placing your order using our online test catalog.

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name		# gene(s)	Gene list
Metabolic Disorders Newborn Screening Confirmation				
○ 06102	Invitae Metabolic Disorders Newborn Screening Confirmation Panel		90	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
	○ 06102.01	Add-on 2,4-dienoyl-CoA reductase deficiency genes	2	DECR1, NADK2
	○ 06102.02	Add-on cerebral creatine deficiency genes	3	GAMT, GATM, SLC6A8
	○ 06102.03	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, SEC23A, SEC23B, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TMEM165, TMEM5, TRIP11, TUSC3, XYLT1
	○ 06102.04	Add-on generalized leukodystrophies genes	6	ARSA, ASPA, GALC, GM2A, HEXA, HEXB
	○ 06102.05	Add-on glucose transporter type 1 (GLUT1) deficiency gene	1	SLC2A1
	○ 06102.06	Add-on glycine encephalopathy genes	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
	○ 06102.07	Add-on mucopolysaccharidosis type II (MPSII) gene	1	IDS
	○ 06102.08	Add-on Niemann-Pick type C genes	2	NPC1, NPC2
	○ 06102.09	Add-on pyridoxal 5'-phosphate-dependent epilepsy gene	1	PNPO
	○ 06102.10	Add-on pyridoxine-responsive epilepsy gene	1	ALDH7A1
	○ 06102.11	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
	○ 06102.12	Add-on cerebrotendinous xanthomatosis gene	1	CYP27A1
	○ 06102.13	Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene	1	HMGCS2
	○ 06102.14	Add-on neuronal ceroid lipofuscinosis genes	10	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
	○ 06102.15	Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene	1	OXCT1
○ 06171	Invitae Lysosomal Storage Disorders Newborn Screening Panel		6	GAA, GALC, GLA, IDS, IDUA, SMPD1
○ 06210	Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test		1	ABCD1

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Panels by Analyte			
○ 06103	Invitae Low C0 Test	1	SLC22A5
○ 06104	Invitae Elevated C0/(C16+C18) Test	1	CPT1A
○ 06105	Invitae Elevated C3 Panel	15	ABCD4, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, TCN2
	○ 06105.1 Add-on ACSF3 gene	1	ACSF3
○ 06106	Invitae Elevated C3-DC Test	1	MLYCD
○ 06107	Invitae Elevated C4 Panel	3	ACAD8, ACADS, ETHE1
○ 06108	Invitae Elevated C4-OH Panel	2	HADH, HIBCH
○ 06109	Invitae Elevated C4 & C5 Panel	7	ETFA, ETFB, ETFDH, ETHE1, SLC52A1, SLC52A2, SLC52A3
○ 06110	Invitae Elevated C5 Panel	2	ACADSB, IVD
○ 06111	Invitae Elevated C5-DC Test	1	GCDH
○ 06112	Invitae Elevated C5-OH Panel	13	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, MCCC1, MCCC2, OPA3, SERAC1, TAZ
○ 06113	Invitae Elevated C6, C8 & C10 Test	1	ACADM
○ 06114	Invitae Elevated C14 & C14:1 Test	1	ACADVL
○ 06115	Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	2	HADHA, HADHB
○ 06116	Invitae Elevated C16, C16:1, C18, & C18:1 Panel	2	CPT2, SLC25A20
○ 06117	Invitae Elevated Arginine Test	1	ARG1
○ 06118	Invitae Elevated Citrulline Panel	4	ASL, ASS1, PC, SLC25A13
	○ 06118.1 Add-on dihydrolipoamide dehydrogenase deficiency gene	1	DLD
○ 06123	Invitae Low Citrulline Panel	3	CPS1, NAGS, OTC
○ 06124	Invitae Elevated Glycine Panel	6	AMT, GLDC, GCSH, LIAS, NFU1, SLC6A9
	○ 06124.1 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
○ 06119	Invitae Elevated Leucine Panel	5	BCKDHA, BCKDHB, DBT, DLD, PPM1K
○ 06125	Invitae Elevated Methionine Panel	4	AHCY, CBS, GNMT, MAT1A
	○ 06125.1 Add-on additional causes of elevated methionine genes	2	FAH, SLC25A13
○ 06120	Invitae Elevated Phenylalanine Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
○ 06121	Invitae Elevated Proline Panel	2	ALDH4A1, PRODH
○ 06122	Invitae Elevated Succinylacetone Test	1	FAH
○ 06126	Invitae Elevated Tyrosine Panel	3	FAH, HPD, TAT

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Aminoacidopathies (continued)			
○ 06140	Invitae Alkaptonuria Test	1	HGD
○ 06141	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
○ 06142	Invitae Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9
○ 06148	Invitae Disorders of Serine Biosynthesis Panel	3	PHGDH, PSAT1, PSPH
○ 06143	Invitae Glycine Encephalopathy Panel	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
○ 06144	Invitae Homocystinuria Panel	4	CBS, MTHFR, MTR, MTRR
	○ 06144.1 Add-on combined methylmalonic acidemia and homocystinuria genes	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
	○ 06144.2 Add-on elevated methionine genes	4	AHCY, CBS, GNMT, MAT1A
○ 06145	Invitae Hyperphenylalaninemia Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
○ 06146	Invitae Hyperprolinemia Panel	2	ALDH4A1, PRODH
○ 06147	Invitae Maple Syrup Urine Disease Panel	4	BCKDHA, BCKDHB, DBT, PPM1K
	○ 06147.1 Add-on DLD gene	1	DLD
○ 06149	Invitae Tyrosinemia Panel	3	FAH, HPD, TAT
Carbohydrate Disorders			
○ 06152	Invitae Galactosemia Panel	3	GALE, GALK1, GALT
○ 06153	Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	1	G6PD
○ 06154	Invitae Glucose Transporter Type 1 (GLUT1) Deficiency Syndrome Test	1	SLC2A1
○ 06156	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
○ 06157	Invitae Liver Glycogen Storage Disease Panel	11	AGL, FBP1, G6PC, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4
○ 06158	Invitae Muscle Glycogen Storage Disease Panel	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
	○ 06158.1 Add-on PGM1 gene	1	PGM1
○ 06159	Invitae Hereditary Fructose Intolerance Test	1	ALDOB
○ 06160	Invitae Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1
Cerebrotendinous Xanthomatosis			
○ 06161	Invitae Cerebrotendinous Xanthomatosis Test	1	CYP27A1
	○ 06161.1 Add-on sitosterolemia genes	2	ABCG5, ABCG8
Congenital Disorders of Glycosylation			
○ 06155	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
	○ 06155.1 Add-on preliminary-evidence genes	11	ALG14, B4GALT1, DDOST, NUS1, PIGM, RPN2, SEC23A, SLC35A3, ST3GAL3, STT3A, STT3B
	○ 06155.2 Add-on disorders of O-mannosylation genes	13	B3GALNT2, B4GAT1, FKRP, FKTN, GNE, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5
	○ 06155.3 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

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Creatine Biosynthesis Disorders			
○ 06162	Invitae Cerebral Creatine Deficiency Panel	3	GAMT, GATM, SLC6A8
Cystic Fibrosis			
○ 06220	Invitae Cystic Fibrosis Newborn Screening Confirmation Test	1	CFTR
Fatty Acid Oxidation Defects			
○ 06165	Invitae Fatty Acid Oxidation Defects Panel	18	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20
	○ 06165.1 Add-on preliminary-evidence gene	1	DECR1
	○ 06165.2 Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
○ 06168	Invitae Ketogenesis Disorders Panel	2	HMGCL, HMGCS2
○ 06169	Invitae Ketolysis Disorders Panel	2	ACAT1, OXCT1
○ 06166	Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADM
○ 06197	Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel	3	ETFA, ETFB, ETFDH
	○ 06197.1 Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
○ 06167	Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADVL
Lysosomal Storage Disorders			
○ 06170	Invitae Comprehensive Lysosomal Storage Disorders Panel	52	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
	○ 06170.1 Add-on chitotriosidase deficiency gene	1	CHIT1
○ 06172	Invitae Cystinosis Test	1	CTNS
○ 06179	Invitae Farber Lipogranulomatosis Test	1	ASAH1
○ 02266	Invitae Fabry Disease Test	1	GLA
○ 06180	Invitae GM2 Gangliosidosis Panel	3	GM2A, HEXA, HEXB
○ 06173	Invitae Krabbe Disease Test	1	GALC
	○ 06173.1 Add-on prosaposin deficiency gene	1	PSAP
○ 06181	Invitae Lysosomal Acid Lipase Deficiency Test	1	LIPA
○ 06174	Invitae Metachromatic Leukodystrophy Panel	3	ARSA, PSAP, SUMF1
	○ 06174.1 Add-on generalized leukodystrophies genes	4	ASPA, GALC, HEXA, HEXB
○ 06184	Invitae Mucolipidosis Panel	4	GNPTAB, GNPTG, MCOLN1, NEU1
○ 06185	Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel	11	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH
	○ 06185.1 Add-on mucolipidosis and oligosaccharidoses genes	12	AGA, CTSA, CTSK, FUCA1, GNPTAB, GNPTG, MAN2B1, MANBA, MCOLN1, NAGA, NEU1, SLC17A5

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Lysosomal Storage Disorders (continued)			
○ 06186	Invitae Mucopolysaccharidosis Type I (MPS I) Test	1	IDUA
○ 06186.1	Add-on clinically overlapping lysosomal storage disorder genes	5	ARSB, GNPTAB, GUSB, IDS, SUMF1
○ 06175	Invitae Mucopolysaccharidosis Type II Test	1	IDS
○ 06175.1	Add-on clinically overlapping genes	4	GNPTAB, GUSB, IDUA, SUMF1
○ 06187	Invitae Mucopolysaccharidosis Type III (MPS III) Panel	4	GNS, HGSNAT, NAGLU, SGSH
○ 06188	Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel	2	GALNS, GLB1
○ 06188.1	Add-on multiple sulfatase deficiency gene	1	SUMF1
○ 06189	Invitae Multiple Sulfatase Deficiency Test	1	SUMF1
○ 06189.1	Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
○ 03405	Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel	9	CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
○ 03405.1	Add-on preliminary-evidence gene	1	ATP13A2
○ 03405.2	Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
○ 06190	Invitae Niemann-Pick Disease Types A and B Test	1	SMPD1
○ 06190.1	Add-on chitotriosidase deficiency gene	1	CHIT1
○ 06176	Invitae Niemann-Pick Type C Panel	2	NPC1, NPC2
○ 06176.1	Add-on lysosomal acid lipase deficiency gene	1	LIPA
○ 06176.2	Add-on chitotriosidase deficiency gene	1	CHIT1
○ 06200	Invitae Oligosaccharidoses Panel	8	AGA, CTSA, CTSK, FUCA1, MAN2B1, MANBA, NAGA, SLC17A5
○ 06200.1	Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
○ 06177	Invitae Pompe Disease Test	1	GAA
○ 06177.1	Add-on Danon disease gene	1	LAMP2
○ 06177.2	Add-on primary carnitine deficiency gene	1	SLC22A5
○ 06201	Invitae Prosaposin Deficiency Test	1	PSAP
○ 06178	Invitae Sandhoff Disease Test	1	HEXB
○ 06178.1	Add-on Tay-Sachs disease gene	1	HEXA
○ 04719	Invitae Tay-Sachs Disease Test	1	HEXA
○ 04719.1	Add-on Sandhoff disease gene	1	HEXB
Metal Transport Disorders			
○ 06182	Invitae ATP7A-Related Disorders Test	1	ATP7A
○ 06202	Invitae Copper Metabolism Disorders Panel	5	AP1S1, ATP7A, ATP7B, CP, SLC33A1
○ 06183	Invitae Wilson Disease Test	1	ATP7B

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Neurotransmitter Disorders			
○ 06203	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
	○ 06203.1 Add-on neurodegeneration with brain iron accumulation genes	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
○ 06204	Invitae Hereditary Hyperekplexia Panel	6	ARHGEF9, CLPB, GLRA1, GLRB, GPHN, SLC6A5
Organic Acidemias			
○ 06191	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
	○ 06191.1 Add-on Kreb cycle defect genes	7	DHTKD1, DLD, FH, NFU1, OGDH, SLC13A5, SLC25A19
○ 06205	Invitae 2-Hydroxyglutaric Aciduria Panel	4	D2HGDH, IDH2, L2HGDH, SLC25A1
○ 06192	Invitae 3-Methylcrotonyl CoA Carboxylase Panel	2	MCCC1, MCCC2
○ 06206	Invitae 3-Methylglutaconic Aciduria Panel	7	AUH, CLPB, DNAJC19, OPA3, SERAC1, TAZ, TMEM70
○ 06193	Invitae Barth Syndrome Test	1	TAZ
○ 06194	Invitae Biotinidase Deficiency Test	1	BTD
○ 04713	Invitae Canavan Disease Test	1	ASPA
○ 06195	Invitae Glutaric Acidemia Type I Test	1	GCDH
○ 06142	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
○ 06196	Invitae Methylmalonic Acidemia Panel	7	MMAA, MMAB, MMADHC, MCEE, MUT, SUCLA2, SUCLG1
	○ 06196.1 Add-on combined malonic and methylmalonic acidemia gene	1	ACSF3
	○ 06196.2 Add-on combined methylmalonic acidemia and homocystinuria genes	10	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, TCN1, TCN2
○ 06197	Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel	3	ETFA, ETFB, ETFDH
	○ 06197.1 Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
○ 06198	Invitae Multiple Carboxylase Deficiency Panel	2	BTD, HLCS
○ 06199	Invitae Propionic Acidemia Panel	2	PCCA, PCCB
	○ 06199.1 Add-on methylmalonic acidemia genes	5	MMAA, MMAB, MMADHC, MMACHC, MUT
	○ 06199.2 Add-on multiple carboxylase deficiency genes	2	BTD, HLCS
Peroxisomal Disorders			
○ 06207	Invitae Adult Refsum Disease Panel	2	PEX7, PHYH
○ 06208	Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7
○ 06209	Invitae X-linked Adrenoleukodystrophy Test	1	ABCD1
	○ 06209.1 Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
	○ 06209.2 Add-on elevated very long chain fatty acids genes	13	HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
○ 06211	Invitae Zellweger Spectrum Disorder Panel	15	ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Purine Metabolism Disorders			
○ 06213	Invitae Purine Metabolism Disorders Panel	9	ADA, ADSL, AMPD1, HPRT1, GPHN, MOCOS, MOCS1, PNP, XDH
	○ 06213.1 Add-on sulfite oxidase deficiency gene	1	SUOX
○ 06214	Invitae Lesch-Nyhan Syndrome Test	1	HPRT1
Pyruvate Metabolism and Tricarboxylic Acid Cycle Defects			
○ 06215	Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel	3	DLD, OGDH, SLC25A19
	○ 06215.1 Add-on alpha-ketoadipic acid dehydrogenase deficiency gene	1	DHTKD1
○ 06216	Invitae Citrate Transporter Deficiency Test	1	SLC13A5
○ 06217	Invitae Dihydrolipoamide Dehydrogenase Deficiency Test	1	DLD
○ 06218	Invitae Fumarase Deficiency Test	1	FH
○ 06219	Invitae Pyruvate Carboxylase Deficiency Test	1	PC
○ 06221	Invitae Pyruvate Dehydrogenase Deficiency Panel	8	DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, PDP1
Treatable Disorders			
○ 06222	Invitae Treatable Neurometabolic Disorders Panel	92	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
	○ 06222.1 Add-on neurometabolic conditions genes	41	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
○ 06223	Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test	1	SLC19A3
Urea Cycle Disorders			
○ 06212	Invitae Urea Cycle Disorders Panel	10	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15
	○ 06212.1 Add-on hyperammonemia genes	4	CA5A, GLUD1, GLUL, SLC7A7
	○ 06212.2 Add-on hereditary orotic aciduria gene	1	UMPS
○ 06224	Invitae Arginase Deficiency Test	1	ARG1
○ 06225	Invitae Ornithine Transcarbamylase (OTC) Deficiency Test	1	OTC
	○ 06225.1 Add-on hereditary orotic aciduria gene	1	UMPS
	○ 06225.2 Add-on low citrulline genes	2	CPS1, NAGS

METABOLIC DISORDERS AND NEWBORN SCREENING INDIVIDUAL GENES

○ ABAT	○ AMPD1	○ CHST6	○ DLAT	○ GALT	○ HEXB	○ MGAT2	○ PCCA
○ ABCD1	○ AMT	○ CHSY1	○ DLD	○ GAMT	○ HGD	○ MLYCD	○ PCCB
○ ABCD4	○ AP1S1	○ CLN2 (TPP1)	○ DNAJC19	○ GATM	○ HGSNAT	○ MMAA	○ PDHA1
○ ABCG5	○ ARG1	○ CLN3	○ DNAJC5	○ GBE1	○ HIBCH	○ MMAB	○ PDHB
○ ABCG8	○ ARHGEF9	○ CLN5	○ DOLK	○ GCDH	○ HLCS	○ MMACHC	○ PDHX
○ ACAD8	○ ARSA	○ CLN6	○ DPAGT1	○ GCH1	○ HMGCL	○ MMADHC	○ PDP1
○ ACADM	○ ARSB	○ CLN8	○ DPM1	○ GCSH	○ HMGCS2	○ MOCOS	○ PEX1
○ ACADS	○ ASAH1	○ CLPB	○ DPM2	○ GFPT1	○ HPD	○ MOCS1	○ PEX10
○ ACADSB	○ ASL	○ COASY	○ DPM3	○ GIF	○ HPRT1	○ MOGS	○ PEX12
○ ACADVL	○ ASPA	○ COG1	○ DSE	○ GLA	○ HSD17B10	○ MPC1	○ PEX13
○ ACAT1	○ ASS1	○ COG2	○ ENO3	○ GLB1	○ HSD17B4	○ MPDU1	○ PEX14
○ ACOX1	○ ATP13A2	○ COG4	○ EOGT	○ GLDC	○ HYAL1	○ MPI	○ PEX16
○ ACSF3	○ ATP6V0A2	○ COG5	○ ETFA	○ GLRA1	○ IDH2	○ MTHFR	○ PEX19
○ ADA	○ ATP7A	○ COG6	○ ETFB	○ GLRB	○ IDS	○ MTR	○ PEX2
○ ADSL	○ ATP7B	○ COG7	○ ETFDH	○ GLUD1	○ IDUA	○ MTRR	○ PEX26
○ AGA	○ AUH	○ COG8	○ ETHE1	○ GLUL	○ ISPD	○ MUT	○ PEX3
○ AGL	○ B3GALNT2	○ CP	○ EXT1	○ GM2A	○ IVD	○ NADK2	○ PEX5
○ AGPS	○ B3GALT6	○ CPS1	○ EXT2	○ GMPPA	○ KCTD7	○ NAGA	○ PEX6
○ AHCY	○ B3GAT3	○ CPT1A	○ FA2H	○ GMPPB	○ L2HGDH	○ NAGLU	○ PEX7
○ ALDH18A1	○ B3GLCT	○ CPT2	○ FAH	○ GNE	○ LAMP2	○ NAGS	○ PFKM
○ ALDH4A1	○ B4GALNT1	○ CTNS	○ FBP1	○ GNMT	○ LARGE1	○ NEU1	○ PGAM2
○ ALDH5A1	○ B4GALT1	○ CTSA	○ FH	○ GNPAT	○ LDHA	○ NFU1	○ PGM1
○ ALDH7A1	○ B4GALT7	○ CTSD	○ FKRP	○ GNPTAB	○ LFNG	○ NGLY1	○ PGM3
○ ALDOA	○ B4GAT1	○ CTSF	○ FKTN	○ GNPTG	○ LIAS	○ NPC1	○ PHGDH
○ ALDOB	○ BCKDHA	○ CTSK	○ FTCD	○ GNS	○ LIPA	○ NPC2	○ PHKA1
○ ALG1	○ BCKDHB	○ CUBN	○ FTL	○ GPHN	○ LMBRD1	○ NUS1	○ PHKA2
○ ALG11	○ BCKDK	○ CYP27A1	○ FUCA1	○ GRN	○ MAGT1	○ OAT	○ PHKB
○ ALG12	○ BTD	○ D2HGDH	○ G6PC	○ GSS	○ MAN1B1	○ OGDH	○ PHKG2
○ ALG13	○ C19orf12	○ DBH	○ G6PC3	○ GUSB	○ MAN2B1	○ OPA3	○ PHYH
○ ALG14	○ C1GALT1C1	○ DBT	○ G6PD	○ GYG1	○ MANBA	○ OPLAH	○ PIGA
○ ALG2	○ CA5A	○ DCAF17	○ GAA	○ GYS1	○ MAOA	○ OTC	○ PIGL
○ ALG3	○ CBS	○ DDC	○ GAD1	○ GYS2	○ MAT1A	○ OXCT1	○ PIGM
○ ALG6	○ CD320	○ DDOST	○ GALC	○ HADH	○ MCCC1	○ PAH	○ PIGN
○ ALG8	○ CFTR	○ DECR1	○ GALE	○ HADHA	○ MCCC2	○ PANK2	○ PIGO
○ ALG9	○ CHIT1	○ DHCR7	○ GALK1	○ HADHB	○ MCEE	○ PAPSS2	○ PIGQ
○ AMACR	○ CHST14	○ DHDDS	○ GALNS	○ HCFC1	○ MCOLN1	○ PC	○ PIGT
○ AMN	○ CHST3	○ DHTKD1	○ GALNT3	○ HEXA	○ MFSD8	○ PCBD1	○ PIGV

METABOLIC DISORDERS AND NEWBORN SCREENING INDIVIDUAL GENES

○ PIGW	○ POMT2	○ RBCK1	○ SLC22A5	○ SLC35A1	○ SLC6A3	○ ST3GAL3	○ TCN2
○ PLA2G6	○ PPM1K	○ RFT1	○ SLC25A1	○ SLC35A2	○ SLC6A5	○ ST3GAL5	○ TH
○ PMM2	○ PPT1	○ RPN2	○ SLC25A13	○ SLC35A3	○ SLC6A8	○ STT3A	○ TMEM165
○ PNP	○ PREPL	○ SLC52A1	○ SLC25A15	○ SLC35C1	○ SLC6A9	○ STT3B	○ TMEM5
○ PNPO	○ PRODH	○ SEC23A	○ SLC25A19	○ SLC35D1	○ SLC7A7	○ SUCLA2	○ TMEM70
○ POFUT1	○ PSAP	○ SEC23B	○ SLC25A20	○ SLC37A4	○ SLC7A9	○ SUCLG1	○ TRIP11
○ POGLUT1	○ PSAT1	○ SERAC1	○ SLC25A22	○ SLC3A1	○ SMPD1	○ SUMF1	○ TUSC3
○ POLG	○ PSPH	○ SGSH	○ SLC26A2	○ SLC52A1	○ SPR	○ SUOX	○ UMPS
○ POMGNT1	○ PTS	○ SLC13A5	○ SLC2A1	○ SLC52A2	○ SRD5A3	○ TAT	○ WDR45
○ POMGNT2	○ PYGL	○ SLC17A5	○ SLC2A2	○ SLC52A3	○ SGSH	○ TAZ	○ XDH
○ POMK	○ PYGM	○ SLC19A3	○ SLC33A1	○ SLC5A1	○ SSR4	○ TCN1	○ XYLT1
○ POMT1	○ QDPR						

