

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Sex <input type="radio"/> M <input type="radio"/> F	MRN (medical record number)
Ancestry <input type="radio"/> Asian <input type="radio"/> Black/African American <input type="radio"/> White/Caucasian <input type="radio"/> Ashkenazi Jewish <input type="radio"/> Hispanic <input type="radio"/> Native American <input type="radio"/> Pacific Islander <input type="radio"/> Other:		
▶ Email address (for report access after release by medical professional)		
Phone	Is this patient deceased? <input type="radio"/> Yes <input type="radio"/> 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)	Special cases <input type="radio"/> History of/current hematologic malignancy <input type="radio"/> Resubmission
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REASON FOR TESTING

Primary indication:

ICD-10 codes	Previous results
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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)
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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: <input type="radio"/> Self <input type="radio"/> Child <input type="radio"/> Spouse <input type="radio"/> Other	
Policy holder name	Prior-authorization #

INSTITUTIONAL BILLING

Send invoice to organization address above

Billing contact name	Phone	Fax
Billing email address		
Billing address		City
State	ZIP code	Country

PATIENT PAY BILLING

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

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
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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 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.

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.

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

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			
<input type="radio"/> 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, BTBD, 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
<input type="radio"/> 06102.01	Add-on 2,4-dienoyl-CoA reductase deficiency genes	2	DECR1, NADK2
<input type="radio"/> 06102.02	Add-on cerebral creatine deficiency genes	3	GAMT, GATM, SLC6A8
<input type="radio"/> 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, FKR, 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, PIQ, 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
<input type="radio"/> 06102.04	Add-on generalized leukodystrophies genes	6	ARSA, ASPA, GALC, GM2A, HEXA, HEXB
<input type="radio"/> 06102.05	Add-on glucose transporter type 1 (GLUT1) deficiency gene	1	SLC2A1
<input type="radio"/> 06102.06	Add-on glycine encephalopathy genes	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
<input type="radio"/> 06102.07	Add-on mucopolysaccharidosis type II (MPSII) gene	1	IDS
<input type="radio"/> 06102.08	Add-on Niemann-Pick type C genes	2	NPC1, NPC2
<input type="radio"/> 06102.09	Add-on pyridoxal 5'-phosphate-dependent epilepsy gene	1	PNPO
<input type="radio"/> 06102.10	Add-on pyridoxine-responsive epilepsy gene	1	ALDH7A1
<input type="radio"/> 06102.11	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
<input type="radio"/> 06102.12	Add-on cerebrotendinous xanthomatosis gene	1	CYP27A1
<input type="radio"/> 06102.13	Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene	1	HMGCS2
<input type="radio"/> 06102.14	Add-on neuronal ceroid lipofuscinosis genes	10	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
<input type="radio"/> 06102.15	Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene	1	OXCT1
<input type="radio"/> 06171	Invitae Lysosomal Storage Disorders Newborn Screening Panel	6	GAA, GALC, GLA, IDS, IDUA, SMPD1
<input type="radio"/> 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			
<input type="radio"/> 06103	Invitae Low C0 Test	1	SLC22A5
<input type="radio"/> 06104	Invitae Elevated C0/(C16+C18) Test	1	CPT1A
<input type="radio"/> 06105	Invitae Elevated C3 Panel	15	ABCD4, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, TCN2
	<input type="radio"/> 06105.1 Add-on ACSF3 gene	1	ACSF3
<input type="radio"/> 06106	Invitae Elevated C3-DC Test	1	MLYCD
<input type="radio"/> 06107	Invitae Elevated C4 Panel	3	ACAD8, ACADS, ETHE1
<input type="radio"/> 06108	Invitae Elevated C4-OH Panel	2	HADH, HIBCH
<input type="radio"/> 06109	Invitae Elevated C4 & C5 Panel	7	ETF A, ETFB, ETFDH, ETHE1, SLC52A1, SLC52A2, SLC52A3
<input type="radio"/> 06110	Invitae Elevated C5 Panel	2	ACADSB, IVD
<input type="radio"/> 06111	Invitae Elevated C5-DC Test	1	GCDH
<input type="radio"/> 06112	Invitae Elevated C5-OH Panel	13	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, MCCC1, MCCC2, OPA3, SERAC1, TAZ
<input type="radio"/> 06113	Invitae Elevated C6, C8 & C10 Test	1	ACADM
<input type="radio"/> 06114	Invitae Elevated C14 & C14:1 Test	1	ACADVL
<input type="radio"/> 06115	Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	2	HADHA, HADHB
<input type="radio"/> 06116	Invitae Elevated C16, C16:1, C18, & C18:1 Panel	2	CPT2, SLC25A20
<input type="radio"/> 06117	Invitae Elevated Arginine Test	1	ARG1
<input type="radio"/> 06118	Invitae Elevated Citrulline Panel	4	ASL, ASS1, PC, SLC25A13
	<input type="radio"/> 06118.1 Add-on dihydroliipoamide dehydrogenase deficiency gene	1	DLD
<input type="radio"/> 06123	Invitae Low Citrulline Panel	3	CPS1, NAGS, OTC
<input type="radio"/> 06124	Invitae Elevated Glycine Panel	6	AMT, GLDC, GCSH, LIAS, NFU1, SLC6A9
	<input type="radio"/> 06124.1 Add-on organic acidemia genes	56	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETF A, 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
<input type="radio"/> 06119	Invitae Elevated Leucine Panel	5	BCKDHA, BCKDHB, DBT, DLD, PPM1K
<input type="radio"/> 06125	Invitae Elevated Methionine Panel	4	AHCY, CBS, GNMT, MAT1A
	<input type="radio"/> 06125.1 Add-on additional causes of elevated methionine genes	2	FAH, SLC25A13
<input type="radio"/> 06120	Invitae Elevated Phenylalanine Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
<input type="radio"/> 06121	Invitae Elevated Proline Panel	2	ALDH4A1, PRODH
<input type="radio"/> 06122	Invitae Elevated Succinylacetone Test	1	FAH
<input type="radio"/> 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)			
<input type="radio"/> 06140	Invitae Alkaptonuria Test	1	HGD
<input type="radio"/> 06141	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
<input type="radio"/> 06142	Invitae Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9
<input type="radio"/> 06148	Invitae Disorders of Serine Biosynthesis Panel	3	PHGDH, PSAT1, PSPH
<input type="radio"/> 06143	Invitae Glycine Encephalopathy Panel	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
<input type="radio"/> 06144	Invitae Homocystinuria Panel	4	CBS, MTHFR, MTR, MTRR
<input type="radio"/> 06144.1	Add-on combined methylmalonic acidemia and homocystinuria genes	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
<input type="radio"/> 06144.2	Add-on elevated methionine genes	4	AHCY, CBS, GNMT, MAT1A
<input type="radio"/> 06145	Invitae Hyperphenylalaninemia Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
<input type="radio"/> 06146	Invitae Hyperprolinemia Panel	2	ALDH4A1, PRODH
<input type="radio"/> 06147	Invitae Maple Syrup Urine Disease Panel	4	BCKDHA, BCKDHB, DBT, PPM1K
<input type="radio"/> 06147.1	Add-on DLD gene	1	DLD
<input type="radio"/> 06149	Invitae Tyrosinemia Panel	3	FAH, HPD, TAT
Carbohydrate Disorders			
<input type="radio"/> 06152	Invitae Galactosemia Panel	3	GALE, GALK1, GALT
<input type="radio"/> 06153	Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	1	G6PD
<input type="radio"/> 06154	Invitae Glucose Transporter Type 1 (GLUT1) Deficiency Syndrome Test	1	SLC2A1
<input type="radio"/> 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
<input type="radio"/> 06157	Invitae Liver Glycogen Storage Disease Panel	11	AGL, FBP1, G6PC, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4
<input type="radio"/> 06158	Invitae Muscle Glycogen Storage Disease Panel	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
<input type="radio"/> 06158.1	Add-on PGM1 gene	1	PGM1
<input type="radio"/> 06159	Invitae Hereditary Fructose Intolerance Test	1	ALDOB
<input type="radio"/> 06160	Invitae Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1
Cerebrotendinous Xanthomatosis			
<input type="radio"/> 06161	Invitae Cerebrotendinous Xanthomatosis Test	1	CYP27A1
<input type="radio"/> 06161.1	Add-on sitosterolemia genes	2	ABCG5, ABCG8
Congenital Disorders of Glycosylation			
<input type="radio"/> 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
<input type="radio"/> 06155.1	Add-on preliminary-evidence genes	11	ALG14, B4GALT1, DDOST, NUS1, PIGM, RPN2, SEC23A, SLC35A3, ST3GAL3, STT3A, STT3B
<input type="radio"/> 06155.2	Add-on disorders of O-mannosylation genes	13	B3GALNT2, B4GAT1, FKRP, FKTN, GNE, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5
<input type="radio"/> 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, PIQQ, PIQT, 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			
<input type="radio"/> 06162	Invitae Cerebral Creatine Deficiency Panel	3	GAMT, GATM, SLC6A8
Cystic Fibrosis			
<input type="radio"/> 06220	Invitae Cystic Fibrosis Newborn Screening Confirmation Test	1	CFTR
Fatty Acid Oxidation Defects			
<input type="radio"/> 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
<input type="radio"/> 06165.1	Add-on preliminary-evidence gene	1	DECRI
<input type="radio"/> 06165.2	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
<input type="radio"/> 06168	Invitae Ketogenesis Disorders Panel	2	HMGCL, HMGCS2
<input type="radio"/> 06169	Invitae Ketolysis Disorders Panel	2	ACAT1, OXCT1
<input type="radio"/> 06166	Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADM
<input type="radio"/> 06197	Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel	3	ETF A, ETFB, ETFDH
<input type="radio"/> 06197.1	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
<input type="radio"/> 06167	Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test	1	ACADVL
Lysosomal Storage Disorders			
<input type="radio"/> 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
<input type="radio"/> 06170.1	Add-on chitotriosidase deficiency gene	1	CHIT1
<input type="radio"/> 06172	Invitae Cystinosis Test	1	CTNS
<input type="radio"/> 06179	Invitae Farber Lipogranulomatosis Test	1	ASAH1
<input type="radio"/> 02266	Invitae Fabry Disease Test	1	GLA
<input type="radio"/> 06180	Invitae GM2 Gangliosidosis Panel	3	GM2A, HEXA, HEXB
<input type="radio"/> 06173	Invitae Krabbe Disease Test	1	GALC
<input type="radio"/> 06173.1	Add-on prosaposin deficiency gene	1	PSAP
<input type="radio"/> 06181	Invitae Lysosomal Acid Lipase Deficiency Test	1	LIPA
<input type="radio"/> 06174	Invitae Metachromatic Leukodystrophy Panel	3	ARSA, PSAP, SUMF1
<input type="radio"/> 06174.1	Add-on generalized leukodystrophies genes	4	ASPA, GALC, HEXA, HEXB
<input type="radio"/> 06184	Invitae Mucopolipidosis Panel	4	GNPTAB, GNPTG, MCOLN1, NEU1
<input type="radio"/> 06185	Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel	11	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH
<input type="radio"/> 06185.1	Add-on mucopolipidosis 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)			
<input type="radio"/> 06186	Invitae Mucopolysaccharidosis Type I (MPS I) Test	1	IDUA
<input type="radio"/> 06186.1	Add-on clinically overlapping lysosomal storage disorder genes	5	ARSB, GNPTAB, GUSB, IDS, SUMF1
<input type="radio"/> 06175	Invitae Mucopolysaccharidosis Type II Test	1	IDS
<input type="radio"/> 06175.1	Add-on clinically overlapping genes	4	GNPTAB, GUSB, IDUA, SUMF1
<input type="radio"/> 06187	Invitae Mucopolysaccharidosis Type III (MPS III) Panel	4	GNS, HGSNAT, NAGLU, SGSH
<input type="radio"/> 06188	Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel	2	GALNS, GLB1
<input type="radio"/> 06188.1	Add-on multiple sulfatase deficiency gene	1	SUMF1
<input type="radio"/> 06189	Invitae Multiple Sulfatase Deficiency Test	1	SUMF1
<input type="radio"/> 06189.1	Add-on mucopolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
<input type="radio"/> 03405	Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel	9	CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
<input type="radio"/> 03405.1	Add-on preliminary-evidence gene	1	ATP13A2
<input type="radio"/> 03405.2	Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
<input type="radio"/> 06190	Invitae Niemann-Pick Disease Types A and B Test	1	SMPD1
<input type="radio"/> 06190.1	Add-on chitotriosidase deficiency gene	1	CHIT1
<input type="radio"/> 06176	Invitae Niemann-Pick Type C Panel	2	NPC1, NPC2
<input type="radio"/> 06176.1	Add-on lysosomal acid lipase deficiency gene	1	LIPA
<input type="radio"/> 06176.2	Add-on chitotriosidase deficiency gene	1	CHIT1
<input type="radio"/> 06200	Invitae Oligosaccharidoses Panel	8	AGA, CTSA, CTSK, FUCA1, MAN2B1, MANBA, NAGA, SLC17A5
<input type="radio"/> 06200.1	Add-on mucopolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
<input type="radio"/> 06177	Invitae Pompe Disease Test	1	GAA
<input type="radio"/> 06177.1	Add-on Danon disease gene	1	LAMP2
<input type="radio"/> 06177.2	Add-on primary carnitine deficiency gene	1	SLC22A5
<input type="radio"/> 06201	Invitae Prosaposin Deficiency Test	1	PSAP
<input type="radio"/> 06178	Invitae Sandhoff Disease Test	1	HEXB
<input type="radio"/> 06178.1	Add-on Tay-Sachs disease gene	1	HEXA
<input type="radio"/> 04719	Invitae Tay-Sachs Disease Test	1	HEXA
<input type="radio"/> 04719.1	Add-on Sandhoff disease gene	1	HEXB
Metal Transport Disorders			
<input type="radio"/> 06182	Invitae ATP7A-Related Disorders Test	1	ATP7A
<input type="radio"/> 06202	Invitae Copper Metabolism Disorders Panel	5	AP1S1, ATP7A, ATP7B, CP, SLC33A1
<input type="radio"/> 06183	Invitae Wilson Disease Test	1	ATP7B

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Neurotransmitter Disorders			
<input type="radio"/> 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
<input type="radio"/> 06203.1	Add-on neurodegeneration with brain iron accumulation genes	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
<input type="radio"/> 06204	Invitae Hereditary Hyperekplexia Panel	6	ARHGEF9, CLPB, GLRA1, GLRB, GPHN, SLC6A5
Organic Acidemias			
<input type="radio"/> 06191	Invitae Organic Acidemias Panel	49	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTM, 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
<input type="radio"/> 06191.1	Add-on Krebs cycle defect genes	7	DHTKD1, DLD, FH, NFU1, OGDH, SLC13A5, SLC25A19
<input type="radio"/> 06205	Invitae 2-Hydroxyglutaric Aciduria Panel	4	D2HGDH, IDH2, L2HGDH, SLC25A1
<input type="radio"/> 06192	Invitae 3-Methylcrotonyl CoA Carboxylase Panel	2	MCCC1, MCCC2
<input type="radio"/> 06206	Invitae 3-Methylglutaconic Aciduria Panel	7	AUH, CLPB, DNAJC19, OPA3, SERAC1, TAZ, TMEM70
<input type="radio"/> 06193	Invitae Barth Syndrome Test	1	TAZ
<input type="radio"/> 06194	Invitae Biotinidase Deficiency Test	1	BTD
<input type="radio"/> 04713	Invitae Canavan Disease Test	1	ASPA
<input type="radio"/> 06195	Invitae Glutaric Acidemia Type I Test	1	GCDH
<input type="radio"/> 06142	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
<input type="radio"/> 06196	Invitae Methylmalonic Acidemia Panel	7	MMAA, MMAB, MMADHC, MCEE, MUT, SUCLA2, SUCLG1
<input type="radio"/> 06196.1	Add-on combined malonic and methylmalonic acidemia gene	1	ACSF3
<input type="radio"/> 06196.2	Add-on combined methylmalonic acidemia and homocystinuria genes	10	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, TCN1, TCN2
<input type="radio"/> 06197	Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel	3	ETF A, ETFB, ETFDH
<input type="radio"/> 06197.1	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
<input type="radio"/> 06198	Invitae Multiple Carboxylase Deficiency Panel	2	BTD, HLCS
<input type="radio"/> 06199	Invitae Propionic Acidemia Panel	2	PCCA, PCCB
<input type="radio"/> 06199.1	Add-on methylmalonic acidemia genes	5	MMAA, MMAB, MMADHC, MMACHC, MUT
<input type="radio"/> 06199.2	Add-on multiple carboxylase deficiency genes	2	BTD, HLCS
Peroxisomal Disorders			
<input type="radio"/> 06207	Invitae Adult Refsum Disease Panel	2	PEX7, PHYH
<input type="radio"/> 06208	Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7
<input type="radio"/> 06209	Invitae X-linked Adrenoleukodystrophy Test	1	ABCD1
<input type="radio"/> 06209.1	Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
<input type="radio"/> 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
<input type="radio"/> 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			
<input type="radio"/> 06213	Invitae Purine Metabolism Disorders Panel	9	ADA, ADSL, AMPD1, HPRT1, GPHN, MOCOS, MOCS1, PNP, XDH
<input type="radio"/> 06213.1	Add-on sulfite oxidase deficiency gene	1	SUOX
<input type="radio"/> 06214	Invitae Lesch-Nyhan Syndrome Test	1	HPRT1
Pyruvate Metabolism and Tricarboxylic Acid Cycle Defects			
<input type="radio"/> 06215	Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel	3	DLD, OGDH, SLC25A19
<input type="radio"/> 06215.1	Add-on alpha-ketoadipic acid dehydrogenase deficiency gene	1	DHTKD1
<input type="radio"/> 06216	Invitae Citrate Transporter Deficiency Test	1	SLC13A5
<input type="radio"/> 06217	Invitae Dihydroliipoamide Dehydrogenase Deficiency Test	1	DLD
<input type="radio"/> 06218	Invitae Fumarase Deficiency Test	1	FH
<input type="radio"/> 06219	Invitae Pyruvate Carboxylase Deficiency Test	1	PC
<input type="radio"/> 06221	Invitae Pyruvate Dehydrogenase Deficiency Panel	8	DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, PDP1
Treatable Disorders			
<input type="radio"/> 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
<input type="radio"/> 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
<input type="radio"/> 06223	Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test	1	SLC19A3
Urea Cycle Disorders			
<input type="radio"/> 06212	Invitae Urea Cycle Disorders Panel	10	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15
<input type="radio"/> 06212.1	Add-on hyperammonemia genes	4	CA5A, GLUD1, GLUL, SLC7A7
<input type="radio"/> 06212.2	Add-on hereditary orotic aciduria gene	1	UMPS
<input type="radio"/> 06224	Invitae Arginase Deficiency Test	1	ARG1
<input type="radio"/> 06225	Invitae Ornithine Transcarbamylase (OTC) Deficiency Test	1	OTC
<input type="radio"/> 06225.1	Add-on hereditary orotic aciduria gene	1	UMPS
<input type="radio"/> 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	○ CASA	○ 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	○ MFSB8	○ PCBD1	○ PIGV

METABOLIC DISORDERS AND NEWBORN SCREENING INDIVIDUAL GENES

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