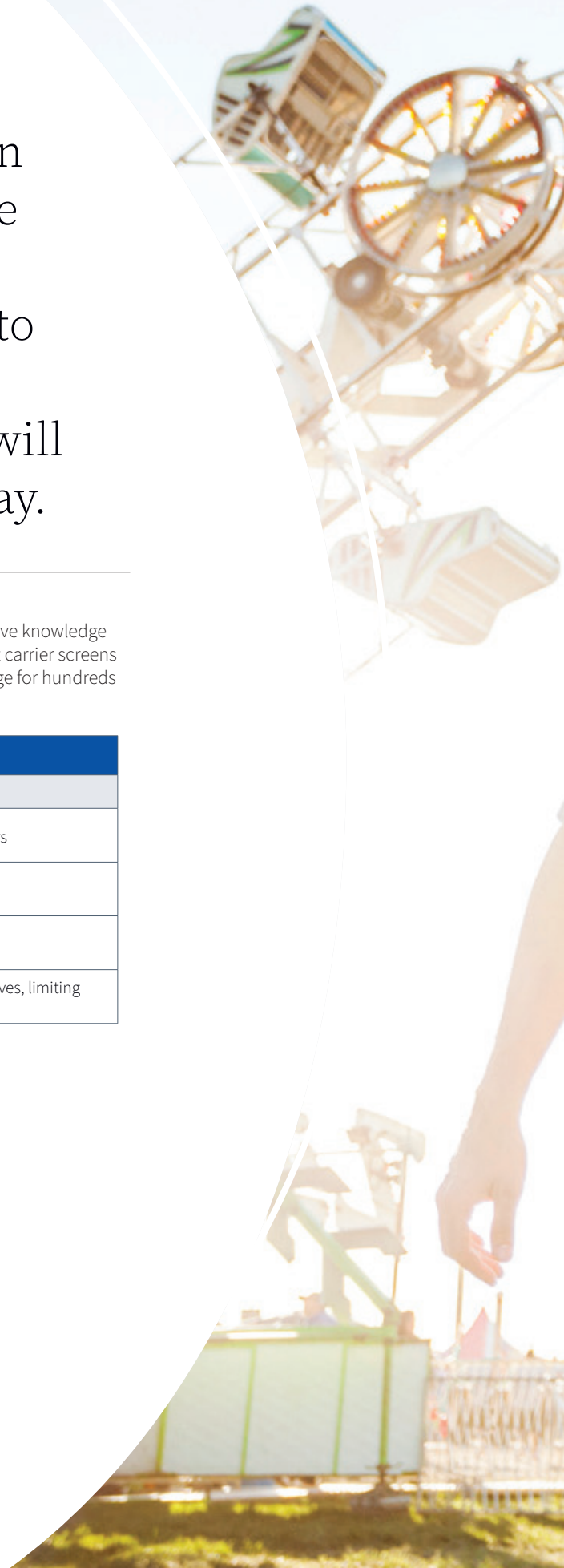
GENETICS & WOMEN'S HEALTH

Inheritest® Carrier Screen 500 PLUS Panel

Understanding more means empowering more

labcorp

Your patients are asking for even more insight as they plan for the future. Empower your patients with more genetic information to help guide family planning and treatment decisions. Our team will support you every step of the way.



In many cases, based on current and advanced treatment options, early, comprehensive knowledge can significantly enhance prenatal decision-making and neonatal care. One of the first carrier screens of this size and scope, the Inheritest® 500 PLUS Panel provides expanded gene coverage for hundreds of clinically-relevant genetic disorders.

Cutting-edge science and methodology	
Methodology/capability	**How you and your patients benefit**
Comprehensive full-exon sequencing with detection of deletions and duplications	99% detection rates for most disorders
Identification of patients at risk of being SMA silent carriers	Potentially more carriers identified
Integrated CGG and AGG analysis for fragile X syndrome	More precise risk information
Positive result confirmation using alternative technology per ACMG guidelines[1]	Helps to reduce the risk of false positives, limiting unnecessary diagnostic procedures

Extensive gene coverage

- Panel comprises more than 500 genes, each associated with a clinically-relevant genetic disorder.
- Thoughtful disorder selection based on ACOG and ACMG criteria and backed by our expert scientific and clinical team. These disorders are focused on severity, early childhood onset, identification of early intervention opportunities and increased childhood mortality.
- Many disorders may have treatment benefit with early medical intervention.

Excellent support for your practice and patients

- Pre- and post-test genetic counseling. National network of board-certified and state-licensed genetic counselors dedicated to patient care.
- Access to experts. In-house lab genetic counselors, medical geneticists and lab directors available to support results interpretation.
- Broad in-network coverage and access to multiple pricing options as well as our Patient Engagement Program. Send your patients to **www.integratedgenetics.com/transparency** or call **844.799.3243**.
- Simple, clear, and concise lab reports based on extensive customer insights. Combined reports for reproductive partners when tested simultaneously.

Types of disorders identified by the Inheritest® 500 PLUS Panel*†	
522	Associated with severe, early onset; increased child mortality; decreased life expectancy; degenerative and progressive disorders; affecting quality of life; and/or requiring medical management.
291	May cause intellectual disability in affected individuals.
284	May cause loss of vision/ eye problems in affected individuals – early identification may be beneficial.
180	Metabolic disorders; may have treatment benefit with early medical intervention.
152	May cause deafness/hearing loss in affected individuals – early identification could be beneficial.
36	X-linked genes, meaning only the mother has to be a carrier for the child to be at risk.

*Based on information on the relevant disorders compiled from Genetics Home Reference and GARD.2-3
†Due to category overlap, the total number of genes is greater than 522

One fast result for fragile X risk assessment

AGG analysis in women who have a premutation with 55-90 CGG repeats provides a more accurate risk assessment compared to CGG testing alone.[4-6]

Risk of expansion to a full mutation based on CGG repeat size and AGG data[7]

Maternal CGG repeat size range*	0 AGGs	1 AGG	2 or more AGGs
55-59	1.9%	<1%	<1%
60-64	5.4%	<1%	<1%
65-69	10%	<1%	<1%
70-74	51.9%	7.6%	<1%
75-79	71.7%	40%	10.7%
80-84	88.2%	65.2%	20.7%
85-90	86.1%	84.6%	29.4%

*AGG analysis is not performed for CGG repeats >90 because once the repeat length exceeds this number, there is no apparent effect of AGG interruptions.[6]

Example: In a patient with 75-79 CGG repeats, the risk of expansion to a full mutation is 10.7% for 2 AGG interruptions compared to 71.7% for no AGG interruptions.

Inheritest 500 PLUS Panel offers a turnaround time of ~21 to 24 days for a complete fragile X result, with both CGG and AGG repeats reported simultaneously.

National network of approximately 100 genetic counselors to deliver genetics expertise to you and your patients.

- Genetic results counseling and comprehensive counseling customized to meet your practice needs.
- Telegenetic counseling through an audio and video connection so patients can receive counseling in the comfort and privacy of their own home.
- Quick and convenient online scheduling and patient management platform via **integratedgenetics.com/genetic-counseling.**
- Genetic Education Video Series to help educate and inform patients about their testing options available on **integratedgenetics.com/videos**. Pediatric-specific options with a focus on minimum samples, alternative samples, and age-specific reference ranges.

GeneSeq® PLUS

Focused comprehensive single gene analysis

- Provides an option for partner testing when an analysis of a particular gene is desired.
- Valuable when a patient has a family history of a specific disorder or when prenatal diagnosis is requested.
- Available with or without VUS (variants of unknown significance), based on provider or patient preference.
- Detection of deletions and duplications contributes to high detection rates.

Disorders covered by Inheritest 500 PLUS Panel, with their related genes:

- 3M syndrome *(CCDC8)*
- 3M syndrome *(CUL7)*
- 3M syndrome *(OBSL1)*
- 3-Methylcrotonyl-CoA carboxylase deficiency *(MCCC1)*
- 3-Methylcrotonyl-CoA carboxylase deficiency *(MCCC2)*
- Abetalipoproteinemia *(MTTP)*
- Acute infantile liver failure *(LARS)*
- Acute infantile liver failure *(NBAS)*
- Acute infantile liver failure *(TRMU)*
- Adenosine deaminase deficiency *(ADA)*
- Adrenoleukodystrophy, X-linked *(ABCD1)*
- Agammaglobulinemia, X-linked *(BTK)*
- Aicardi-Goutières syndrome *(RNASEH2A)*
- Aicardi-Goutières syndrome *(RNASEH2B)*
- Aicardi-Goutières syndrome *(RNASEH2C)*
- Aicardi-Goutières syndrome *(SAMHD1)*
- Allan-Herndon-Dudley syndrome *(SLC16A2)*
- Alpha-mannosidosis *(MAN2B1)*
- Alpha-thalassemia *(HBA1)*
- Alpha-thalassemia *(HBA2)*
- Alpha-thalassemia, X-linked intellectual disability syndrome *(ATRX)*
- Alport syndrome *(COL4A3)*
- Alport syndrome, X-linked *(COL4A5)*
- Alström syndrome *(ALMS1)*
- Andermann syndrome *(SLC12A6)*
- Arginase deficiency *(ARG1)*
- Argininosuccinic aciduria *(ASL)*
- Aromatic l-amino acid decarboxylase deficiency *(DDC)*
- Arterial tortuosity syndrome *(SLC2A10)*
- Arthrogryposis, mental retardation, and seizures (AMRS) *(SLC35A3)*
- Asparagine synthetase deficiency *(ASNS)*
- Aspartylglucosaminuria *(AGA)*
- Ataxia with vitamin E deficiency *(TTPA)*
- Ataxia-telangiectasia *(ATM)*
- *(ATP7A)* copper transport disorders, includes Menkes syndrome
- Autoimmune polyglandular syndrome type 1 *(AIRE)*
- Autosomal recessive congenital ichthyosis (ARCI) *(ABCA12)*
- Autosomal recessive congenital ichthyosis (ARCI) *(ALOX12B)*
- Autosomal recessive congenital ichthyosis (ARCI) *(ALOXE3)*
- Autosomal recessive congenital ichthyosis (ARCI) *(CASP14)*
- Autosomal recessive congenital ichthyosis (ARCI) *(CERS3)*
- Autosomal recessive congenital ichthyosis (ARCI) *(CYP4F22)*
- Autosomal recessive congenital ichthyosis (ARCI) *(LIPN)*
- Autosomal recessive congenital ichthyosis (ARCI) *(NIPAL4)*
- Autosomal recessive congenital ichthyosis (ARCI) *(PNPLA1)*
- Autosomal recessive congenital ichthyosis (ARCI) *(SDR9C7)*
- Autosomal recessive congenital ichthyosis (ARCI) *(SLC27A4)*
- Autosomal recessive congenital ichthyosis (ARCI) *(TGM1)*
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) *(SACS)*
- Axonal neuropathy with neuromyotonia, autosomal recessive *(HINT1)*
- Bardet-Biedl syndrome *(ARL6)*
- Bardet-Biedl syndrome *(BBS1)*
- Bardet-Biedl syndrome *(BBS10)*
- Bardet-Biedl syndrome *(BBS12)*
- Bardet-Biedl syndrome *(BBS2)*
- Bardet-Biedl syndrome *(BBS4)*
- Bardet-Biedl syndrome *(BBS5)*
- Bardet-Biedl syndrome *(BBS7)*
- Bardet-Biedl syndrome *(BBS9)*
- Bardet-Biedl syndrome *(MKKS)*
- Bardet-Biedl syndrome *(SDCCAG8)*
- Bardet-Biedl syndrome *(TTC8)*
- Bare lymphocyte syndrome type II *(CIITA)*
- Bare lymphocyte syndrome type II *(RFX5)*
- Bare lymphocyte syndrome type II *(RFXANK)*
- Bare lymphocyte syndrome type II *(RFXAP)*
- Barth syndrome *(TAZ)*
- Bartter syndrome *(BSND)*
- Bartter syndrome *(KCNJ1)*
- Bartter syndrome *(SLC12A1)*
- Beta-hemoglobinopathies, includes sickle cell disease and beta-thalassemias *(HBB)*
- Beta-ketothiolase deficiency *(ACAT1)*
- Beta-mannosidosis *(MANBA)*
- Biotinidase deficiency *(BTD)*
- Bloom syndrome *(BLM)*
- Brittle cornea syndrome *(PRDM5)*
- Brittle cornea syndrome *(ZNF469)*
- Canavan disease *(ASPA)*
- Carbamoyl phosphate synthetase I deficiency *(CPS1)*
- Carnitine palmitoyltransferase I deficiency *(CPT1A)*
- Carnitine palmitoyltransferase II deficiency *(CPT2)*
- Carnitine-acylcarnitine translocase deficiency *(SLC25A20)*
- Carpenter syndrome *(MEGF8)*
- Carpenter syndrome *(RAB23)*
- Cartilage-hair hypoplasia *(RMRP)*
- Cerebellar hypoplasia, VLDLR-associated *(VLDLR)*
- Cerebral creatine deficiency syndromes *(GAMT)*
- Cerebral creatine deficiency syndromes *(GATM)*
- Cerebral creatine deficiency syndromes *(SLC6A8)*
- Cerebrotendinous xanthomatosis *(CYP27A1)*
- Chronic granulomatous disease *(CYBA)*
- Chronic granulomatous disease *(CYBB)*
- Chronic granulomatous disease *(NCF2)*
- Chronic granulomatous disease *(NCF4)*
- Ciliopathies *(CEP290)*
- Ciliopathies *(MKS1)*
- Citrullinemia *(ASS1)*
- Citrullinemia *(SLC25A13)*
- Coats plus syndrome and dyskeratosis congenita *(CTC1)*
- Cockayne syndrome *(ERCC6)*
- Cockayne syndrome *(ERCC8)*
- Coffin-Lowry syndrome *(RPS6KA3)*
- Cohen syndrome *(VPS13B)*
- Cold-induced sweating syndrome *(includes Crisponi syndrome (CLCF1)*
- Cold-induced sweating syndrome *(includes Crisponi syndrome (CRLF1)*
- Combined malonic and methylmalonic aciduria *(ACSF3)*
- Congenital adrenal hyperplasia *(CYP11B1)*
- Congenital adrenal hyperplasia *(CYP17A1)*
- Congenital adrenal hyperplasia *(CYP21A2)*
- Congenital adrenal hyperplasia *(HSD3B2)*
- Congenital adrenal hyperplasia *(POR)*
- Congenital adrenal hyperplasia *(STAR)*
- Congenital adrenal hypoplasia, X-linked *(NR0B1)*
- Congenital amegakaryocytic thrombocytopenia *(MPL)*
- Congenital disorder of deglycosylation *(NGLY1)*
- Congenital disorders of glycosylation type 1 *(ALG1)*
- Congenital disorders of glycosylation type 1 *(ALG6)*
- Congenital disorders of glycosylation type 1 *(MPI)*
- Congenital disorders of glycosylation type 1 *(PMM2)*
- Congenital generalized lipodystrophy *(AGPAT2)*
- Congenital generalized lipodystrophy *(CAVIN1)*
- Congenital insensitivity to pain with anhidrosis *(NTRK1)*
- Congenital myasthenic syndrome *(CHAT)*
- Congenital myasthenic syndrome *(COLQ)*
- Congenital myasthenic syndrome *(DOK7)*
- Congenital myasthenic syndrome *(GFPT1)*
- Congenital myasthenic syndrome *(RAPSN)*
- Corneal dystrophy and perceptive deafness *(SLC4A11)*
- Costeff optic atrophy syndrome, autosomal recessive *(OPA3)*
- Cutis laxa *(ATP6V0A2)*
- Cutis laxa *(ATP6V1E1)*
- Cutis laxa *(EFEMP2)*
- Cutis laxa *(LTBP4)*
- Cutis laxa *(PYCR1)*
- Cystic fibrosis *(CFTR)*
- Cystinosis *(CTNS)*
- Danon disease *(LAMP2)*
- D-bifunctional protein deficiency *(HSD17B4)*
- Deafness and hearing loss, nonsyndromic *(GJB2)*
- Deafness and hearing loss, nonsyndromic *(GJB6)*
- Deafness and hearing loss, nonsyndromic *(LOXHD1)*
- Deafness and hearing loss, nonsyndromic *(OTOF)*
- Deafness and hearing loss, nonsyndromic *(POU3F4)*
- Deafness and hearing loss, nonsyndromic *(SYNE4)*
- Dent disease *(CLCN5)*
- Dent disease *(OCRL)*
- Dihydrolipoamide dehydrogenase deficiency *(DLD)*
- Dihydropyrimidine dehydrogenase deficiency *(DPYD)*
- Distal spinal muscular atrophy, autosomal recessive *(PLEKHG5)*
- Donnai-Barrow syndrome *(LRP2)*
- Dystrophinopathies, including Duchenne and Becker muscular dystrophy and X-linked cardiomyopathy *(DMD)*
- Early infantile epileptic encephalopathy *(CAD)*
- Early infantile epileptic encephalopathy *(ITPA)*
- Ehlers-Danlos syndrome type VIIC *(ADAMTS2)*
- Emery-Dreifuss muscular dystrophy *(EMD)*
- Emery-Dreifuss muscular dystrophy *(FHL1)*
- Ethylmalonic encephalopathy *(ETHE1)*
- Fabry disease *(GLA)*
- Familial dysautonomia *(ELP1)*
- Familial hemophagocytic lymphohistiocytosis *(PRF1)*
- Familial hemophagocytic lymphohistiocytosis *(STX11)*
- Familial hemophagocytic lymphohistiocytosis *(STXBP2)*
- Familial hemophagocytic lymphohistiocytosis *(UNC13D)*
- Familial hyperinsulinism *(ABCC8)*
- Familial Mediterranean fever *(MEFV)*
- Fanconi anemia *(BRIP1)*
- Fanconi anemia *(FANCA)*
- Fanconi anemia *(FANCB)*
- Fanconi anemia *(FANCC)*
- Fanconi anemia *(FANCD2)*
- Fanconi anemia *(FANCE)*
- Fanconi anemia *(FANCF)*
- Fanconi anemia *(FANCG)*
- Fanconi anemia *(FANCI)*
- Fanconi anemia *(FANCL)*
- Fragile X syndrome *(FMR1)*
- Fraser syndrome *(FRAS1)*
- Fraser syndrome *(FREM2)*
- Fraser syndrome *(GRIP1)*
- Fucosidosis *(FUCA1)*
- Galactosemia *(GALE)*
- Galactosemia *(GALK1)*
- Galactosemia *(GALT)*
- Galactosialidosis *(CTSA)*
- Gaucher disease *(GBA)*
- Glutaric acidemia type I *(GCDH)*
- Glutaric acidemia type II *(ETFA)*
- Glutaric acidemia type II *(ETFB)*
- Glutaric acidemia type II *(ETFDH)*
- Glutathione synthetase deficiency *(GSS)*
- Glycine encephalopathy *(AMT)*
- Glycine encephalopathy *(GLDC)*
- Glycogen storage disease type I *(G6PC)*
- Glycogen storage disease type I *(SLC37A4)*
- Glycogen storage disease type III *(AGL)*
- Glycogen storage disease type IV *(GBE1)*
- Glycogen storage disease type IX *(PHKA1)*
- Glycogen storage disease type IX *(PHKA2)*
- Glycogen storage disease type IX *(PHKB)*
- Glycogen storage disease type IX *(PHKG2)*
- Glycogen storage disease type V *(PYGM)*
- Glycogen storage disease type VII *(PFKM)*
- GM1 gangliosidosis and mucopolysaccharidosis type IVB *(GLB1)*
- GRACILE syndrome *(BCS1L)*
- Gyrate atrophy of choroid and retina *(OAT)*
- Hepatic venoocclusive disease with immunodeficiency *(SP110)*
- Hereditary folate malabsorption *(SLC46A1)*
- Hereditary fructose Intolerance *(ALDOB)*
- Hereditary spastic paraplegia *(CYP7B1)*
- Hereditary spastic paraplegia *(SPG11)*
- Hereditary spastic paraplegia *(SPG21)*
- Hereditary spastic paraplegia *(TECPR2)*
- Hermansky-Pudlak syndrome *(AP3B1)*
- Hermansky-Pudlak syndrome *(AP3D1)*
- Hermansky-Pudlak syndrome *(BLOC1S3)*
- Hermansky-Pudlak syndrome *(BLOC1S6)*
- Hermansky-Pudlak syndrome *(DTNBP1)*
- Hermansky-Pudlak syndrome *(HPS1)*
- Hermansky-Pudlak syndrome *(HPS3)*
- Hermansky-Pudlak syndrome *(HPS4)*
- Hermansky-Pudlak syndrome *(HPS5)*
- Hermansky-Pudlak syndrome *(HPS6)*
- HMG-CoA lyase deficiency *(HMGCL)*
- Holocarboxylase synthetase deficiency *(HLCS)*
- Homocystinuria *(CBS)*
- HSD10 disease *(HSD17B10)*
- Hyaline fibromatosis syndrome *(ANTXR2)*
- Hydrolethalus syndrome *(HYLS1)*
- Hypomyelination and congenital cataract *(FAM126A)*
- Hypophosphatasia *(ALPL)*
- Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome *(CDCA7)*
- Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome *(DNMT3B)*
- Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome *(HELLS)*
- Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome *(ZBTB24)*
- Immunodysregulation, polyendocrinopathy, and enteropathy *(FOXP3)*
- Inclusion body myopathy 2 *(GNE)*
- Isovaleric acidemia *(IVD)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(AHI1)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(ARL13B)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(B9D1)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(B9D2)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(CEP104)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(CPLANE1)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(INPP5E)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(KIF14)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(NPHP1)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(NPHP3)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(RPGRIP1L)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TCTN1)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TCTN2)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TCTN3)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TMEM138)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TMEM216)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TMEM231)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TMEM237)*
- Joubert syndrome and related disorders, including Meckel-Gruber syndrome *(TMEM67)*
- Junctional epidermolysis bullosa *(LAMA3)*
- Junctional epidermolysis bullosa *(LAMB3)*
- Junctional epidermolysis bullosa *(LAMC2)*
- Juvenile hereditary hemochromatosis *(HAMP)*
- Juvenile hereditary hemochromatosis *(HJV)*

Krabbe disease *(GALC)*
L1 syndrome *(L1CAM)*
Leber congenital amaurosis *(AIPL1)*
Leber congenital amaurosis *(LCA5)*
Leber congenital amaurosis *(LRAT)*
Leber congenital amaurosis *(RD3)*
Leber congenital amaurosis *(RDH12)*
Leber congenital amaurosis *(RPE65)*
Leber congenital amaurosis *(RPGRIP1)*
Leber congenital amaurosis *(SPATA7)*
Leber congenital amaurosis *(TULP1)*
Leigh syndrome, autosomal recessive *(COX15)*
Leigh syndrome, autosomal recessive *(FBXL4)*
Leigh syndrome, autosomal recessive *(FOXRED1)*
Leigh syndrome, autosomal recessive *(LRPPRC)*
Leigh syndrome, autosomal recessive *(NDUFAF2)*
Leigh syndrome, autosomal recessive *(NDUFAF5)*
Leigh syndrome, autosomal recessive *(NDUFS4)*
Leigh syndrome, autosomal recessive *(NDUFS6)*
Leigh syndrome, autosomal recessive *(NDUFS7)*
Leigh syndrome, autosomal recessive *(NDUFV1)*
Leigh syndrome, autosomal recessive *(SURF1)*
Leukoencephalopathy with vanishing white matter *(EIF2B1)*
Leukoencephalopathy with vanishing white matter *(EIF2B2)*
Leukoencephalopathy with vanishing white matter *(EIF2B3)*
Leukoencephalopathy with vanishing white matter *(EIF2B4)*
Leukoencephalopathy with vanishing white matter *(EIF2B5)*
Limb-girdle muscular dystrophy, autosomal recessive *(CAPN3)*
Limb-girdle muscular dystrophy, autosomal recessive *(DYSF)*
Limb-girdle muscular dystrophy, autosomal recessive *(FKRP)*
Limb-girdle muscular dystrophy, autosomal recessive *(POMGNT1)*
Limb-girdle muscular dystrophy, autosomal recessive *(POMT1)*
Limb-girdle muscular dystrophy, autosomal recessive *(POMT2)*
Limb-girdle muscular dystrophy, autosomal recessive *(SGCA)*
Limb-girdle muscular dystrophy, autosomal recessive *(SGCB)*
Limb-girdle muscular dystrophy, autosomal recessive *(SGCD)*
Limb-girdle muscular dystrophy, autosomal recessive *(SGCG)*
Limb-girdle muscular dystrophy, autosomal recessive *(TRAPPC11)*
Limb-girdle muscular dystrophy, autosomal recessive *(TRIM32)*
Lipoprotein lipase deficiency, familial *(LPL)*
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency *(HADHA)*
Lysinuric protein intolerance *(SLC7A7)*
Lysosomal acid lipase deficiency *(LIPA)*
Maple syrup urine disease *(BCKDHA)*
Maple syrup urine disease *(BCKDHB)*
Maple syrup urine disease *(DBT)*
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency *(ACADM)*
Megalencephalic leukoencephalopathy with subcortical cysts type 1 *(MLC1)*
Metachromatic leukodystrophy *(ARSA)*
Metachromatic leukodystrophy *(PSAP)*
Methylmalonic acidemia *(MCEE)*
Methylmalonic acidemia *(MMAA)*
Methylmalonic acidemia *(MMAB)*
Methylmalonic acidemia *(MMUT)*
Methylmalonic acidemia with homocystinuria *(ABCD4)*
Methylmalonic acidemia with homocystinuria *(HCFC1)*
Methylmalonic acidemia with homocystinuria *(LMBRD1)*
Methylmalonic acidemia with homocystinuria *(MMACHC)*
Methylmalonic acidemia with homocystinuria *(MMADHC)*
Mitochondrial complex I deficiency *(ACAD9)*
Mitochondrial complex V deficiency *(TMEM70)*
Mitochondrial DNA depletion syndrome *(MPV17)*
Mitochondrial DNA depletion syndrome *(TK2)*
Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia *(PUS1)*
Mucolipidosis type II and III *(GNPTAB)*
Mucolipidosis type IV *(MCOLN1)*
Mucopolysaccharidosis type I *(IDUA)*
Mucopolysaccharidosis type II *(IDS)*
Mucopolysaccharidosis type III *(GNS)*
Mucopolysaccharidosis type III *(HGSNAT)*
Mucopolysaccharidosis type III *(NAGLU)*
Mucopolysaccharidosis type III *(SGSH)*
Mucopolysaccharidosis type IVA *(GALNS)*
Mucopolysaccharidosis type IX *(HYAL1)*
Mucopolysaccharidosis type VI *(ARSB)*
Mucopolysaccharidosis type VII *(GUSB)*
Multiple pterygium syndrome *(CHRNG)*
Multiple sulphatase deficiency *(SUMF1)*
Muscular dystrophy *(LAMA2)*
Myotubular myopathy *(MTM1)*
Nemaline myopathy *(NEB)*
Nephrogenic diabetes insipidus *(AVPR2)*
Nephrotic syndrome *(NPHS1)*
Nephrotic syndrome *(NPHS2)*
Neurodegeneration with brain iron accumulation disorder *(ATP13A2)*
Neurodegeneration with brain iron accumulation disorder *(C19orf12)*
Neurodegeneration with brain iron accumulation disorder *(COASY)*
Neurodegeneration with brain iron accumulation disorder *(CP)*
Neurodegeneration with brain iron accumulation disorder *(DCAF17)*
Neurodegeneration with brain iron accumulation disorder *(FA2H)*
Neurodegeneration with brain iron accumulation disorder *(PLA2G6)*
Neuronal ceroid-lipofuscinosis *(CLN3)*
Neuronal ceroid-lipofuscinosis *(CLN5)*
Neuronal ceroid-lipofuscinosis *(CLN6)*
Neuronal ceroid-lipofuscinosis *(CLN8)*
Neuronal ceroid-lipofuscinosis *(CTSD)*
Neuronal ceroid-lipofuscinosis *(CTSF)*
Neuronal ceroid-lipofuscinosis *(KCTD7)*
Neuronal ceroid-lipofuscinosis *(MFSD8)*
Neuronal ceroid-lipofuscinosis *(PPT1)*
Neuronal ceroid-lipofuscinosis *(TPP1)*
Niemann-Pick disease type C *(NPC1)*
Niemann-Pick disease type C *(NPC2)*
Niemann-Pick disease types A and B *(SMPD1)*
Nijmegen breakage syndrome *(NBN)*
Omenn syndrome *(DCLRE1C)*
Omenn syndrome *(RAG1)*
Omenn syndrome *(RAG2)*
Ornithine transcarbamylase deficiency *(OTC)*
Ornithine translocase deficiency *(SLC25A15)*
Osteogenesis imperfecta, autosomal recessive *(BMP1)*
Osteogenesis imperfecta, autosomal recessive *(CRTAP)*
Osteogenesis imperfecta, autosomal recessive *(FKBP10)*
Osteogenesis imperfecta, autosomal recessive *(P3H1)*
Osteogenesis imperfecta, autosomal recessive *(PLOD2)*
Osteogenesis imperfecta, autosomal recessive *(PPIB)*
Osteogenesis imperfecta, autosomal recessive *(SERPINF1)*
Osteogenesis imperfecta, autosomal recessive *(TMEM38B)*
Osteogenesis imperfecta, autosomal recessive *(WNT1)*
Osteopetrosis, autosomal recessive *(OSTM1)*
Osteopetrosis, autosomal recessive *(TCIRG1)*
Osteopetrosis, autosomal recessive *(TNFSF11)*
Pantothenate kinase-associated neurodegeneration *(PANK2)*
Pendred syndrome *(SLC26A4)*
Peroxisomal acyl-CoA oxidase deficiency *(ACOX1)*
Phenylalanine hydroxylase deficiency, includes phenylketonuria (PKU) *(PAH)*
Phosphoglycerate dehydrogenase deficiency *(PHGDH)*
Pitt-Hopkins-like syndrome 1 *(CNTNAP2)*
Polycystic kidney disease, autosomal recessive *(PKHD1)*
Pompe disease *(GAA)*
Pontocerebellar hypoplasia *(AMPD2)*
Pontocerebellar hypoplasia *(CHMP1A)*
Pontocerebellar hypoplasia *(CLP1)*
Pontocerebellar hypoplasia *(EXOSC3)*
Pontocerebellar hypoplasia *(RARS2)*
Pontocerebellar hypoplasia *(SEPSECS)*
Pontocerebellar hypoplasia *(TSEN2)*
Pontocerebellar hypoplasia *(TSEN34)*
Pontocerebellar hypoplasia *(TSEN54)*
Pontocerebellar hypoplasia *(VPS53)*
Pontocerebellar hypoplasia *(VRK1)*
Primary carnitine deficiency *(SLC22A5)*
Primary congenital glaucoma *(CYP1B1)*
Primary hyperoxaluria *(AGXT)*
Primary hyperoxaluria *(GRHPR)*
Primary hyperoxaluria *(HOGA1)*
Progressive familial intrahepatic cholestasis *(ABCB11)*
Progressive familial intrahepatic cholestasis *(ABCB4)*
Progressive familial intrahepatic cholestasis *(ATP8B1)*
Progressive pseudorheumatoid dysplasia *(CCN6)*
Propionic acidemia *(PCCA)*
Propionic acidemia *(PCCB)*
Pseudocholinesterase deficiency *(BCHE)*
Pycnodysostosis *(CTSK)*
Pyridoxal 5'-phosphate-dependent epilepsy *(PNPO)*
Pyridoxine-dependent epilepsy *(ALDH7A1)*
Pyruvate dehydrogenase deficiency *(DLAT)*
Pyruvate dehydrogenase deficiency *(PDHA1)*
Pyruvate dehydrogenase deficiency *(PDHB)*
Pyruvate dehydrogenase deficiency *(PDHX)*
Pyruvate dehydrogenase deficiency *(PDP1)*
Renal tubular acidosis and deafness *(ATP6V0A4)*
Renal tubular acidosis and deafness *(ATP6V1B1)*
Retinitis pigmentosa *(CERKL)*
Retinitis pigmentosa *(CWC27)*
Retinitis pigmentosa *(DHDDS)*
Retinitis pigmentosa *(EYS)*
Retinitis pigmentosa *(FAM161A)*
Retinitis pigmentosa *(IFT140)*
Retinitis pigmentosa *(MAK)*
Retinitis pigmentosa *(PRCD)*
Retinitis pigmentosa *(RLBP1)*
Retinitis pigmentosa *(RP2)*
Retinitis pigmentosa *(RPGR)*
Rhizomelic chondrodysplasia punctata *(AGPS)*
Rhizomelic chondrodysplasia punctata *(GNPAT)*
Rhizomelic chondrodysplasia punctata *(PEX7)*
Sandhoff disease *(HEXB)*
SELENON-related disorders
Severe combined immunodeficiency (SCID) *(AK2)*
Severe combined immunodeficiency (SCID) *(CD247)*
Severe combined immunodeficiency (SCID) *(CD3D)*
Severe combined immunodeficiency (SCID) *(CD3E)*
Severe combined immunodeficiency (SCID) *(CD3G)*
Severe combined immunodeficiency (SCID) *(CD8A)*
Severe combined immunodeficiency (SCID) *(CORO1A)*
Severe combined immunodeficiency (SCID) *(DOCK8)*
Severe combined immunodeficiency (SCID) *(FOXN1)*
Severe combined immunodeficiency (SCID) *(IKBKB)*
Severe combined immunodeficiency (SCID) *(IL2RA)*
Severe combined immunodeficiency (SCID) *(IL7R)*
Severe combined immunodeficiency (SCID) *(JAK3)*
Severe combined immunodeficiency (SCID) *(LCK)*
Severe combined immunodeficiency (SCID) *(LIG4)*
Severe combined immunodeficiency (SCID) *(MALT1)*
Severe combined immunodeficiency (SCID) *(MTHFD1)*
Severe combined immunodeficiency (SCID) *(NHEJ1)*
Severe combined immunodeficiency (SCID) *(PGM3)*
Severe combined immunodeficiency (SCID) *(PNP)*
Severe combined immunodeficiency (SCID) *(PRKDC)*
Severe combined immunodeficiency (SCID) *(PTPRC)*
Severe combined immunodeficiency (SCID) *(STK4)*
Severe combined immunodeficiency (SCID) *(TTC7A)*
Severe combined immunodeficiency (SCID) *(ZAP70)*
Severe combined Immunodeficiency *(SCID)*, X-linked *(IL2RG)*
Severe congenital neutropenia, autosomal recessive *(HAX1)*
Sialic acid storage disorders *(SLC17A5)*
Sialidosis *(NEU1)*
Sjogren-Larsson syndrome *(ALDH3A2)*
Smith-Lemli-Opitz syndrome *(DHCR7)*
Spinal muscular atrophy *(SMN1)*
Spondylothoracic dysostosis *(MESP2)*
Sulfate transporter) osteochondrodysplasias, includes achondrogenesis type 1B, atelosteogenesis type 2, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia *(SLC26A2)*
Sulfite oxidase deficiency *(SUOX)*
Tay-Sachs disease *(HEXA)*
Tetrahydrobiopterin deficiency *(PCBD1)*
Tetrahydrobiopterin deficiency *(PTS)*
Tetrahydrobiopterin deficiency *(QDPR)*
Trichohepatoenteric syndrome *(SKIV2L)*
Trichohepatoenteric syndrome *(TTC37)*
Trifunctional protein deficiency *(HADHB)*
Triple A syndrome *(AAAS)*
Tyrosine hydroxylase deficiency *(TH)*
Tyrosinemia type I *(FAH)*
Tyrosinemia type II *(TAT)*
Tyrosinemia type III *(HPD)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(ADGRV1)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(CDH23)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(CIB2)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(CLRN1)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(PCDH15)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(USH1C)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(USH1G)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(USH2A)*
Usher syndrome (hearing loss and retinitis pigmentosa) *(WHRN)*
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency *(ACADVL)*
Walker-Warburg syndrome and other FKTN related dystrophies
Werner syndrome *(WRN)*
Wilson disease *(ATP7B)*
Xeroderma pigmentosum *(DDB2)*
Xeroderma pigmentosum *(ERCC2)*
Xeroderma pigmentosum *(ERCC3)*
Xeroderma pigmentosum *(ERCC4)*
Xeroderma pigmentosum *(ERCC5)*
Xeroderma pigmentosum *(POLH)*
Xeroderma pigmentosum *(XPA)*
Xeroderma pigmentosum *(XPC)*
X-linked syndromic mental retardation *(NONO)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX1)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX10)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX11B)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX12)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX13)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX14)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX16)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX19)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX2)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX26)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX3)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX5)*
Zellweger spectrum disorder/ peroxisome biogenesis disorder *(PEX6)*

Test/Panel Name	Test No.	Turnaround Time*
Inheritest® 500 PLUS Panel	**630049**	21-24 days
Inheritest® 500 PLUS Panel with Repro Partners Report	**630217**	21-24 days
GeneSeq® PLUS	**630068**	14-21 days
GeneSeq® PLUS without VUS	**630085**	14-21 days
GeneSeq® PLUS, Prenatal	**630119**	14-21 days
GeneSeq® PLUS without VUS, Prenatal	**630102**	14-21 days

*From the date of pickup of a specimen for testing to when the result is released.

8.5 mL whole blood in a yellow-top (ACD-A) tube or lavender-top (EDTA) tube Applies to tests noted above except prenatal options

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