THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Quick Guide To Genetic Testing

TEST	DISORDER	TEST CODE	CPT	TAT
Albinism testing				
Albinism Panel		1101	81443	6 weeks
Angelman syndrome				
MS-MLPA (detects methylation and deletions in 15q11-13)	Angelman syndrome	1104	81331	4 weeks
UPD 15 testing (requires samples from both parents)		1105	81402	4 weeks
Imprinting center deletion analysis		1106	81331	4 weeks
Rett/Angelman Syndrome Panel		1112	81443	6 weeks
Aniridia testing				
PAX6 Mutation Analysis		2105	81479	4 weeks
IMAGe syndrome/Beckwith-Wiedemann syndrome testing				
CDKN1C Mutation Analysis	Beckwith-Wiedemann syndrome	1116	81479	4 weeks
Brain Malformation testing				
Comprehensive Brain Malformation Panel	Cerebral Cortical Malformations, Polymicrogyria, Cerebellar Hypoplasia, Lissencephaly	1130	81443	6 weeks
Holoprosencephaly Panel	Holoprosencephaly	1129	81405, 81479	6 weeks
Hydrocephalus Panel	Hydrocephalus	1321	81407, 81479	6 weeks
Cardiac disease testing				
Arrhythmia Comprehensive Panel		2181	81443	6 weeks
Aortopathy Comprehensive Panel		2182	81443	6 weeks
Arrhythmia and Cardiomyopathy Comprehensive Panel		2183	81443	6 weeks
Arrhythmogenic Cardiomyopathy Panel		2184	81443	6 weeks
Brugada Syndrome Panel		2185	81407	6 weeks
Cardiomyopathy Comprehensive Panel		2187	81439	6 weeks
Congenital Heart Disease Panel		2189	81443	6 weeks
Catecholaminergic Polymorphic Ventricular Tachycardia Panel		2190	81403, 81405, 81408, 81479	6 weeks
Dilated Cardiomyopathy and Left Ventricular Noncompaction Panel		2191	81439	6 weeks
Hereditary Transthyretin-mediated amyloidosis (hATTR amyloidosis) Mutation Analysis		2192	81404	4 weeks
Hypertrophic Cardiomyopathy Panel		2193	81443	6 weeks
Long QT Syndrome Panel		2194	81403, 81406, 81407, 81479	6 weeks
Pulmonary Arterial Hypertension Panel		2195	81405, 81406, 81479	6 weeks
Short QT Syndrome Panel		2196	81403, 81406	6 weeks
CHARGE syndrome testing				
CHD7 Mutation Analysis	CHARGE syndrome	1158	81407	4 weeks
CHILD syndrome testing				
NSDHL Mutation Analysis	CHILD syndrome	1160	81479	4 weeks
CHIME syndrome testing				
PIGL Mutation Analysis	CHIME syndrome	1162	81479	4 weeks
Chondrodysplasia punctata testing				
Rhizomelic Chondrodysplasia Punctata Panel		1170	81479	4 weeks
Ciliopathy testing				
Bardet-Biedl Syndrome Panel	Bardet-Biedl syndrome	2107	81443	6 weeks
Joubert/Meckel Gruber Syndrome Panel	Joubert syndrome and Meckel Gruber syndrome	1171	81443	6 weeks
Meckel Gruber Syndrome Panel	Meckel Gruber syndrome	1173	81443	6 weeks
Nephronophthisis Panel	Nephronophthisis	1175	81443	6 weeks
Coffin-Siris syndrome testing				
Coffin-Siris Syndrome Panel		1178	81479	6 weeks
Connective Tissue Disorder testing				
Ehlers-Danlos Syndrome Panel		2197	81443	6 weeks
Loeys-Dietz Syndrome Panel		2198	81405, 81408, 81479	6 weeks
Marfan Syndrome Mutation Analysis		2199	81408	4 weeks

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Congenital Muscle Disease testing				
Congenital Myopathy Panel		3100	81443	6 weeks
Congenital Muscular Dystrophy Panel		3102	81443	6 weeks
Congenital Myasthenic Syndrome Panel		3108	81443	6 weeks
Neuromuscular Disorders Exome		3118	81443	6 weeks
Cornelia de Lange syndrome testing				
Cornelia de Lange and Related Disorders Panel		1192	81443	6 weeks
Craniofacial testing				
Craniofacial Panel		1193	81443	6 weeks
Facial Dysostosis Panel		1195	81443	6 weeks
Currarino syndrome				
MNX1 Mutation Analysis		2175	81479	4 weeks
Disorders of Sex Development testing				
Abnormal/Ambiguous Genitalia Panel		2109	81443	6 weeks
Hypogonadotropic Hypogonadism/Kallmann Syndrome Panel		2110	81443	6 weeks
46,XX Disorders of Sex Development/Complete Gonadal Dysgenesis Panel		2112	81405x2, 81479	6 weeks
46,XY Disorders of Sex Development/Complete Gonadal Dysgenesis Panel		2113	81443	6 weeks
Distal arthrogryposes testing				
Distal Arthrogryposes Panel		1198	81479	6 weeks
Donnai-Barrow syndrome testing				
LRP2 Mutation Analysis	Donnai-Barrow syndrome	1200	81479	4 weeks
Dyslipidemia testing				
Dyslipidemia Panel		2186	81443	6 weeks
Hypercholesterolemia Panel		2188	81401, 81405, 81406x2, 81479	6 weeks
Epilepsy testing				
Early Infantile Epileptic Encephalopathy Panel		1326	81443	6 weeks
Epilepsy Panel		6102	81419	6 weeks
Exome Sequencing				
Exome Sequencing (Proband only)		6113	81415	6 weeks
Exome Sequencing (Trio)		6104	81415, 81416x2	6 weeks
STAT Exome Sequencing (Proband only)		6107	81415, 99060	Prelim result in 2-4 weeks
STAT Exome Sequencing (Trio)		6108	81415, 81416x2, 99060	Prelim result in 2-4 weeks
Exome Select		6105	81415	6 weeks
Floating Harbor syndrome testing				
SRCAP Mutation Analysis	Floating Harbor syndrome	1202	81479	4 weeks
Gastrointestinal/Liver disease testing				
Liver Disease Panel		1134	81443	6 weeks
Digestive Disease Panel		1135	81443	6 weeks
Glucose transporter type 1 deficiency testing				
SLC2A1 Mutation Analysis	Glucose transporter type 1 deficiency	4111	81405	4 weeks
Goldberg-Shprintzen megacolon syndrome testing				
KIAA1279 Mutation Analysis	Goldberg-Shprintzen megacolon	1204	81479	4 weeks

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Hereditary Cancer testing				
Comprehensive Hereditary Cancer Panel		5144	81443	6 weeks
Hereditary Breast and Ovarian Cancer Panel		5150	81432, 81433	6 weeks
Lynch Syndrome Panel		5151	81292, 81295, 81298, 81317, 81479	6 weeks
Hereditary Colorectal Cancer Panel		5154	81435, 81436	6 weeks
Hereditary Prostate Cancer Panel		5155	81162, 81292, 81295, 81298, 81317, 81479	6 weeks
Hereditary Myeloid Malignancy Panel		5165	81443	6 weeks
Hereditary Leukemia and Breast Cancer Panel		5142	81443	6 weeks
Hereditary Hematopoietic Malignancy/Immunodeficiency Predisposition Panel		5166	81443	6 weeks
Hereditary Lymphoid Malignancy/Immunodeficiency Predisposition Panel		5158	81443	6 weeks
Diamond-Blackfan Anemia Panel		5159	81405, 81479	6 weeks
Telomere Biology Disorder/Dyskeratosis Congenita Panel		5114	81345, 81479	6 weeks
Fanconi Anemia Panel		5160	81443	6 weeks
Mesothelioma Panel		5164	81443	6 weeks
Hereditary Melanoma Panel		5161	81162, 81404, 81405, 81479	6 weeks
Hereditary Gastric Cancer Panel		5121	81443	6 weeks
Hereditary Pheochromocytoma and Paraganglioma Panel		5122	81437, 81404, 81406, 81408, 81479	6 weeks
Hereditary Thyroid Cancer Panel		5145	81201, 81203, 81321, 81323, 81479	4 weeks
Inherited Bone Marrow Failure Panel		5133	81443	6 weeks
Severe Congenital Neutropenia Panel		5162	81406, 81479	6 weeks
SMARCA4 Mutation Analysis	Rhabdoid tumor predisposition syndrome testing	5163	81479	4 weeks
Hereditary Hemorrhagic Telangictasia (HHT)				
Hereditary Hemorrhagic Telangectasia (HHT) Panel		1314	81405x2, 81406x2, 81479	6 weeks
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum				
SLC12A6 Mutation Analysis	Hereditary Motor and Sensory Neuropathy with ACC	1208	81479	4 weeks
Hydroxyglutaric Aciduria				
2-Hydroxyglutaric Aciduria Panel		1325	81121, 81479	6 weeks
Hyperinsulinism, Congenital testing				
Tier 1 Panel: Diazoxide Unresponsive Hyperinsulinism	Familial hyperinsulinism	2116	81403, 81406, 81407, 81479	1 week
Hyperinsulinism Panel		2118	81443	4 weeks
Immunodeficiency				
Primary Immunodeficiency Panel		5169	81443	6 weeks
Infertility				
Male Infertility Panel		1132	81443	6 weeks
Female Infertility Panel		1133	81443	6 weeks
Intellectual Disability (ID) testing				
Autosomal Recessive Non-Specific ID Panel		1210	81443	6 weeks
X-Linked Non-Specific ID Panel		1211	81470	6 weeks
Non-Specific ID Panel		1212	81443	6 weeks
Intellectual Disability Exome (Trio)		6114	81415, 81416x2	6 weeks
Kabuki syndrome testing				
Kabuki Syndrome Panel		1327	81479	6 weeks
Laminopathy testing				
LMNA Mutation Analysis (Lamin A/C)	Laminopathies	1218	81406	4 weeks
Lipodystrophy testing				
Lipodystrophy Panel		2123	81443	6 weeks

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Macrocephaly testing				
Macrocephaly Panel		1220	81443	6 weeks
***MCT8* testing: (NOTE: 3-10cc of blood in a red top tube AND 3-10cc of blood in a purple top EDTA tube)**				
Tier 1: Thyroid testing only	Allan-Herndon-Dudley syndrome/*MCT8*-related thyroid hormone cell transporter deficiency	2129	84436, 84480, 84481, 84443, 84482	2 weeks
Tier 2: *SLC16A2* (MCT8) Mutation Analysis (performed after abnormal thyroid testing)		2130	81404, 81405	4 weeks
Menkes disease testing				
ATP7A Mutation Analysis	Menkes disease	1227	81479	4 weeks
Microcephalic Osteodysplastic Primordial Dwarfism				
Primordial Dwarfism Panel		1233	81479	6 weeks
Microcephaly testing				
Microcephaly Panel		1237	81443	6 weeks
Movement Disorder testing				
Ataxia Exome		6101	81443	6 weeks
Ataxia Repeat Expansion Panel		4101	0378U, 81177, 81178, 81179, 81180, 81181, 81182, 81183, 81184, 81243, 81284, 81343, 81344, 81479	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATN*		4103	81177	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN1*		4103	81178	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN2*		4103	81179	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN3*		4103	81180	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN7*		4103	81181	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN8OS*		4103	81182	2-4 weeks
Single Gene Repeat Expansion Analysis - *ATXN10*		4103	81183	2-4 weeks
Single Gene Repeat Expansion Analysis - *CACNA1A*		4103	81184	2-4 weeks
Single Gene Repeat Expansion Analysis - *FMR1*		4103	81243	2-4 weeks
Single Gene Repeat Expansion Analysis - *FXN*		4103	81284	2-4 weeks
Single Gene Repeat Expansion Analysis - *PPP2R2B*		4103	81343	2-4 weeks
Single Gene Repeat Expansion Analysis - *FGF14*		4103	81479	2-4 weeks
Single Gene Repeat Expansion Analysis - *RFC1*		4103	0378U	2-4 weeks
Single Gene Repeat Expansion Analysis - *TBP*		4103	81344	2-4 weeks
Single Gene Repeat Expansion Analysis - *NOP56*		4103	81479	2-4 weeks
Comprehensive Ataxia Testing (concurrent testing option)		4102	81443, 0378U, 81177, 81178, 81179, 81180, 81181, 81182, 81183, 81184, 81243, 81284, 81343, 81344, 81479	6 weeks
Comprehensive Ataxia Testing (reflex testing option)		6111	81443, 0378U, 81177, 81178, 81179, 81180, 81181, 81182, 81183, 81184, 81243, 81284, 81343, 81344, 81479	6 weeks
Hereditary Spastic Paraplegia Exome		6110	81443	6 weeks
Dystonia Chorea Parkinson Exome		6115	81443	6 weeks

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Neonatal Diabetes Mellitus, Maturity-Onset Diabetes of the Young and Monogenic Obesity testing				
Comprehensive Neonatal Diabetes Mutation Analysis		2136	81443	4 weeks
6q24 Methylation-Specific MLPA	Transient neonatal diabetes	2137	81402	4 weeks
Neonatal Diabetes Mellitus (NDM) Panel		2138	81443	4 weeks
Monogenic Diabetes Panel		2140	81443	6 weeks
MODY Panel		2141	81443	6 weeks
GCK Mutation Analysis	MODY type 2	2142	81406	4 weeks
Bardet-Biedl Syndrome Panel		2107	81443	6 weeks
Non-Syndromic Monogenic Obesity Panel	Monogenic Obesity	2169	81403, 81406, 81479	6 weeks
Monogenic Obesity Panel		2151	81443	6 weeks
Neurodegeneration with brain iron accumulation (NBIA) testing				
Neurodegeneration with Brain Iron Accumulation Panel		1254	81479	6 weeks
Neuronal Ceroid- Lipofuscinoses testing				
Neuronal Ceroid-Lipofuscinoses Panel		4113	81406, 81479	6 weeks
Nicolaides-Baraitser syndrome testing				
SMARCA2 Mutation Analysis	Nicolaides-Baraitser syndrome	1262	81479	4 weeks
Oculodentodigital dysplasia testing				
GJA1 Mutation Analysis		1264	81479	4 weeks
Oral-facial-digital syndrome testing				
OFD1 Mutation Analysis	Oral-facial-digital syndrome	1265	81479	4 weeks
Pancreatic Agenesis				
Pancreatic Agenesis Panel	Pancreatic Agenesis	2179	81404, 81479	6 weeks
Pitt-Hopkins syndrome testing				
TCF4 Mutation Analysis	Pitt-Hopkins syndrome	1267	81405, 81406	4 weeks
Prader-Willi syndrome testing				
Methylation Specific MLPA		1104	81331	4 weeks
UPD15 (requires samples from both parents also)		1105	81402	4 weeks
Imprinting center deletion analysis		1106	81331	4 weeks
MAGEL2 Mutation Analysis	Shaaf-Yang syndrome	1272	81479	4 weeks
Premature Ovarian Failure testing				
Premature Ovarian Failure Panel		2160	81406, 81479	6 weeks
RASopathies				
Noonan Syndrome Panel		1312	81442	6 weeks
RASopathies and Noonan Spectrum Disorders Panel		1313	81442	6 weeks
Renal Disorders testing				
Hereditary Cystic and Glomerular Disorders Panel		2176	81443	6 weeks
Hereditary Tubulopathy and Nephrolithiasis Panel		2177	81443	6 weeks
Congenital Anomalies of Kidney and Urinary Tract (CAKUT)		2180	81443	6 weeks
Atypical Hemolytic Uremic Syndromes Panel		2178	81443	6 weeks
Rett syndrome testing				
Rett/Angelman Syndrome Panel		1112	81443	6 weeks
Rickets testing				
Hypophosphatemic Rickets Panel		2173	81404, 81406, 81479	4 weeks
Roberts syndrome testing				
ESCO2 Mutation Analysis		1282	81479	4 weeks
Robinow syndrome testing				
Robinow Syndrome Panel		1319	81479	6 weeks
Rubinstein-Taybi syndrome testing				
Rubenstein-Taybi Syndrome Panel		1320	81406, 81407, 81479	6 weeks
Schinzel-Giedion syndrome testing				
SETBP1 Mutation Analysis		1292	81479	4 weeks
SHORT syndrome testing				
PIK3R1 Mutation Analysis		1294	81479	4 weeks
Sotos syndrome testing				
NSD1 Mutation Analysis		1317	81405, 81406	4 weeks
Thrombocytopenia testing				
Thrombocytopenia Panel		5106	81443	6 weeks
Temple-Baraitser syndrome testing				
KCNH1 Mutation Analysis		1146	81479	4 weeks

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Thyroid Disorders testing				
Hypothyroidism Panel		2170	81443	4 weeks
Hyperparathyroidism Panel		2171	81404; 81405x2; 81406, 81479	4 weeks
Hypoparathyroidism Panel		2172	81443	4 weeks
Type A Insulin Resistant Diabetes with Acanthosis Nigricans testing				
INSR Mutation Analysis	Type A Insulin Resistant Diabetes with Acanthosis Nigricans	2163	81479	4 weeks
***UGT1A1* testing**				
UGT1A1 genotyping for Gilbert syndrome		1295	81350	2 weeks
UGT1A1 genotyping for irinotecan dosing		1296	81350	1 week
UGT1A1 Mutation Analysis	Crigler-Najjar syndrome	1297	81404	4 weeks
UPD testing: (NOTE: 3-10cc of blood in an EDTA/purple top tube from patient AND BOTH parents)				
UPD6 testing	Transient neonatal diabetes	1299	81402	4 weeks
UPD7 testing	Russell-Silver syndrome	1300	81402	4 weeks
UPD14 testing	UPD14	1301	81402	4 weeks
UPD15 testing	Angelman and Prader-Willi syndromes	1105	81402	4 weeks
Warburg Micro syndrome testing				
Warburg Micro Syndrome Panel		1316	81479	6 weeks
Weaver syndrome testing				
EZH2 Mutation Analysis		1318	81236	4 weeks
Wiedemann-Steiner syndrome testing				
KMT2A (*MLL*) Mutation Analysis	Wiedemann-Steiner syndrome	1305	81479	4 weeks
Wilson Disease testing				
ATP7B Mutation Analysis	Wilson disease	1307	81406	4 weeks
Wolfram syndrome testing				
Wolfram Syndrome Panel		2167	81479	4 weeks
Targeted Testing				
Targeted analysis (sequencing)		7102	81403 (unless gene specific)	3 weeks
Targeted analysis (deletion/duplication)		7103	81402 (unless gene specific)	4 weeks
Single Gene Testing				
Single Gene Analysis		gene specific	gene specific	4 weeks
Prenatal Testing				
Prenatal testing for known mutation by sequence analysis		7104	81403 (unless gene specific)	1 week
MISCELLANEOUS FEES				
Cell culture service (skin fibroblasts)		1147	88233	2-3 weeks
ROUTINE TESTS: 3-10cc of blood in a purple top (EDTA) tube				
PRENATAL: 20cc amniotic fluid, 25mgs chorionic villi, or 2 T25 flasks cultured cells, along with 3-10cc of mother's blood in an EDTA tube				
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