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 also)		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
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
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 Panel**		6112	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
Custom Mutation Analysis (requires approval by UCGS Lab staff)				
Custom Mutation Analysis (targeted analysis for known sequence change for first family member)		7101	Gene specific	4 weeks
Custom targeted analysis (for known sequence change for additional family members)		7102	Gene specific	3 weeks
Custom deletion/duplication analysis for known familial deletion/duplication by real-time q-pcr		7103	Gene specific	6 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
Cytogenomic SNP array (postnatal)		8100	81229	4 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 Exome**		6102	81443	6 weeks
Cytogenomic SNP array (postnatal)		8100	81229	4 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 weeks
STAT Exome Sequencing (Trio)**		6108	81415, 81416x2, 99060	Prelim result in 2 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
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
Cytogenomic SNP array (postnatal)		8100	81229	4 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
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
Multiple Congenital Anomaly testing				
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks

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Movement Disorder testing				
Ataxia Exome**		6101	81443	6 weeks
Ataxia Repeat Expansion Panel		4101	81177, 81178, 81179, 81184, 81284, 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 - *RFC1*		4103	81479	2-4 weeks
Single Gene Repeat Expansion Analysis - *TBP*		4103	81344	2-4 weeks
Comprehensive Ataxia Testing (concurrent testing option)**		4102	81443	6 weeks
Comprehensive Ataxia Testing (reflex testing option)**		6111	81443	6-8 weeks
Hereditary Spastic Paraplegia Exome**		6110	81443	6 weeks
Dystonia Chorea Parkinson Exome**		6115	81443	6 weeks
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
Noonan syndrome testing				
Noonan Syndrome Panel**		1312	81442	6 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
Renal Disorders testing				
Hereditary Cystic and Glomerular Disorders Panel**		2176	81443	6 weeks
Hereditary Tubulopathy and Nephrolithiasis Panel**		2177	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
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

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***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 mutation analysis (known familial mutation)		7102	81403 (unless gene specific)	3 weeks
Testing for a known familial mutation by deletion/duplication analysis (MLPA)		gene specific	81402 (unless gene specific)	4 weeks
Single Gene Testing				
Single Gene Mutation 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
STAT fee for known mutations, MS-MLPA or UPD***		n/a	99060	MAY CUT TAT IN HALF
STAT fee for full gene mutation analysis (single gene, requires prior approval)***		n/a	99060	MAY CUT TAT IN HALF
STAT fee for Next-Generation Sequencing panel (requires prior approval)***		n/a	99060	MAY CUT TAT IN HALF
Technical lab charge for initial processing of failed sample		n/a	99199	n/a

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/purple to

***Prices listed are applicable for institutional/patient self pay billing options only. Please contact us for prices for direct insurance billing.**

****Please note that we do not bill insurance directly for these specific tests. We apologize for the inconvenience.**

*****Please note that STAT testing is not available for all tests. Please contact the laboratory for more information.**

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