THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Quick Guide To Genetic Testing

TEST	DISORDER	TEST CODE	CPT	TAT	INSTITUTIONAL COST*
Albinism testing					
Albinism Panel**		1101	81443	6 weeks	$2,500
Angelman syndrome					
MS-MLPA (detects methylation and deletions in 15q11-13)	Angelman syndrome	1104	81331	4 weeks	$525
UPD 15 testing (requires samples from both parents also)		1105	81402	4 weeks	$540
Imprinting center deletion analysis		1106	81331	4 weeks	$450
Rett/Angelman Syndrome Panel**		1112	81443	6 weeks	$2,500
Aniridia testing					
PAX6 Mutation Analysis		2105	81479	4 weeks	$1,000
IMAGe syndrome/Beckwith-Wiedemann syndrome testing					
CDKN1C Mutation Analysis	Beckwith-Wiedemann syndrome	1116	81479	4 weeks	$840
Brain Malformation testing					
Comprehensive Brain Malformation Panel**	Cerebral Cortical Malformations, Polymicrogyria, Cerebellar Hypoplasia, Lissencephaly	1130	81443	6 weeks	$3,000
Holoprosencephaly Panel**	Holoprosencephaly	1129	81405, 81479	6 weeks	$2,500
Hydrocephalus Panel**	Hydrocephalus	1321	81407, 81479	6 weeks	$2,500
CHARGE syndrome testing					
CHD7 Mutation Analysis	CHARGE syndrome	1158	81407	4 weeks	$1,000
CHILD syndrome testing					
NSDHL Mutation Analysis	CHILD syndrome	1160	81479	4 weeks	$1,000
CHIME syndrome testing					
PIGL Mutation Analysis	CHIME syndrome	1162	81479	4 weeks	$1,000
Chondrodysplasia punctata testing					
Rhizomelic Chondrodysplasia Punctata Panel**		1170	81479	4 weeks	$2,000
Ciliopathy testing					
Bardet-Biedl Syndrome Panel**	Bardet-Biedl syndrome	2107	81443	6 weeks	$2,500
Joubert/Meckel Gruber Syndrome Panel**	Joubert syndrome and Meckel Gruber syndrome	1171	81443	6 weeks	$3,000
Meckel Gruber Syndrome Panel**	Meckel Gruber syndrome	1173	81443	6 weeks	$2,500
Nephronophthisis Panel**	Nephronophthisis	1175	81443	6 weeks	$3,000
Coffin-Siris syndrome testing					
Coffin-Siris Syndrome Panel**		1178	81479	6 weeks	$2,000
Congenital Muscle Disease testing					
Congenital Myopathy Panel**		3100	81443	6 weeks	$2,000
Congenital Muscular Dystrophy Panel**		3102	81443	6 weeks	$2,000
Congenital Myasthenic Syndrome Panel**		3108	81443	6 weeks	$2,000
Neuromuscular Disorders Panel**		6112	81443	6 weeks	$2,000
Neuromuscular Disorders Exome**		3118	81443	6 weeks	$4,000
Cornelia de Lange syndrome testing					
Cornelia de Lange and Related Disorders Panel**		1192	81443	6 weeks	$3,000
Craniofacial testing					
Craniofacial Panel**		1193	81443	6 weeks	$3,000
Facial Dysostosis Panel**		1195	81443	6 weeks	$2,500
Currarino syndrome					
MNX1 Mutation Analysis		2175	81479	4 weeks	$1,000
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	$540
Custom targeted analysis (for known sequence change for additional family members)		7102	Gene specific	3 weeks	$500
Custom deletion/duplication analysis for known familial deletion/duplication by real-time q-pcr		7103	Gene specific	6 weeks	$1,000
Disorders of Sex Development testing					
Abnormal/Ambiguous Genitalia Panel**		2109	81443	6 weeks	$3,000
Hypogonadotropic Hypogonadism/Kallmann Syndrome Panel**		2110	81443	6 weeks	$3,000
46,XX Disorders of Sex Development/Complete Gonadal Dysgenesis Panel**		2112	81405x2, 81479	6 weeks	$2,000
46,XY Disorders of Sex Development/Complete Gonadal Dysgenesis Panel**		2113	81443	6 weeks	$3,000
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Distal arthrogryposes testing					
Distal Arthrogryposes Panel**		1198	81479	6 weeks	$2,000
Donnai-Barrow syndrome testing					
LRP2 Mutation Analysis	Donnai-Barrow syndrome	1200	81479	4 weeks	$1,000
Dyslipidemia testing					
Dyslipidemia Panel		2186	81443	6 weeks	$2,500
Hypercholesterolemia Panel		2188	81401, 81405, 81406x2, 81479	6 weeks	$2,000
Epilepsy testing					
Early Infantile Epileptic Encephalopathy Panel**		1326	81443	6 weeks	$3,000
Epilepsy Exome**		6102	81443	6 weeks	$4,000
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Exome Sequencing					
Exome Sequencing (Proband only)**		6113	81415	6 weeks	$4,000
Exome Sequencing (Trio)**		6104	81415, 81416x2	6 weeks	$5,800
STAT Exome Sequencing (Proband only)**		6107	81415, 99060	Prelim result in 2 weeks	$8,000
STAT Exome Sequencing (Trio)**		6108	81415, 81416x2, 99060	Prelim result in 2 weeks	$11,600
Exome Select**		6105	81415	6 weeks	$3,000
Floating Harbor syndrome testing					
SRCAP Mutation Analysis	Floating Harbor syndrome	1202	81479	4 weeks	$1,000
Gastrointestinal/Liver disease testing					
Liver Disease Panel**		1134	81443	6 weeks	$3,000
Digestive Disease Panel**		1135	81443	6 weeks	$3,000
Glucose transporter type 1 deficiency testing					
SLC2A1 Mutation Analysis	Glucose transporter type 1 deficiency	4111	81405	4 weeks	$1,000
Goldberg-Shprintzen megacolon syndrome testing					
KIAA1279 Mutation Analysis	Goldberg-Shprintzen megacolon	1204	81479	4 weeks	$1,000

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Hereditary Cancer testing					
Comprehensive Hereditary Cancer Panel**		5144	81443	6 weeks	$3,000
Hereditary Breast and Ovarian Cancer Panel**		5150	81432, 81433	6 weeks	$2,500
Lynch Syndrome Panel**		5151	81292, 81295, 81298, 81317, 81479	6 weeks	$2,000
Hereditary Colorectal Cancer Panel**		5154	81435, 81436	6 weeks	$2,500
Hereditary Prostate Cancer Panel**		5155	81162, 81292, 81295, 81298, 81317, 81479	6 weeks	$2,000
Hereditary Myeloid Malignancy Panel**		5165	81443	6 weeks	$3,000
Hereditary Leukemia and Breast Cancer Panel**		5142	81443	6 weeks	$3,000
Hereditary Hematopoietic Malignancy/Immunodeficiency Predisposition Panel**		5166	81443	6 weeks	$3,000
Hereditary Lymphoid Malignancy/Immunodeficiency Predisposition Panel**		5158	81443	6 weeks	$3,000
Diamond-Blackfan Anemia Panel**		5159	81405, 81479	6 weeks	$2,500
Telomere Biology Disorder/Dyskeratosis Congenita Panel**		5114	81345, 81479	6 weeks	$2,500
Fanconi Anemia Panel**		5160	81443	6 weeks	$2,500
Mesothelioma Panel**		5164	81443	6 weeks	$2,500
Hereditary Melanoma Panel**		5161	81162, 81404, 81405, 81479	6 weeks	$2,500
Hereditary Gastric Cancer Panel**		5121	81443	6 weeks	$2,500
Hereditary Pheochromocytoma and Paraganglioma Panel**		5122	81437, 81404, 81406, 81408, 81479	6 weeks	$2,500
Hereditary Thyroid Cancer Panel**		5145	81201, 81203, 81321, 81323, 81479	4 weeks	$2,000
Inherited Bone Marrow Failure Panel**		5133	81443	6 weeks	$3,000
Severe Congenital Neutropenia Panel**		5162	81406, 81479	6 weeks	$2,000
SMARCA4 Mutation Analysis	Rhabdoid tumor predisposition syndrome testing	5163	81479	4 weeks	$1,000
Hereditary Hemorrhagic Telangictasia (HHT)					
Hereditary Hemorrhagic Telangectasia (HHT) Panel**		1314	81405x2, 81406x2, 81479	6 weeks	$2,000
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	$1,000
Hydroxyglutaric Aciduria					
2-Hydroxyglutaric Aciduria Panel**		1325	81121, 81479	6 weeks	$2,000
Hyperinsulinism, Congenital testing					
Tier 1 Panel: Diazoxide Unresponsive Hyperinsulinism	Familial hyperinsulinism	2116	81403, 81406, 81407, 81479	1 week	$3,000
Hyperinsulinism Panel		2118	81443	4 weeks	$2,500
Infertility					
Male Infertility Panel**		1132	81443	6 weeks	$3,000
Female Infertility Panel**		1133	81443	6 weeks	$3,000
Premature Ovarian Failure Panel**		2160	81406, 81479	6 weeks	$3,000
Intellectual Disability (ID) testing					
Autosomal Recessive Non-Specific ID Panel**		1210	81443	6 weeks	$3,000
X-Linked Non-Specific ID Panel**		1211	81470	6 weeks	$3,000
Non-Specific ID Panel**		1212	81443	6 weeks	$3,000
Intellectual Disability Exome (Trio)**		6114	81415, 81416x2	6 weeks	$4,000
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Kabuki syndrome testing					
Kabuki Syndrome Panel**		1327	81479	6 weeks	$2,000
Laminopathy testing					
LMNA Mutation Analysis (Lamin A/C)	Laminopathies	1218	81406	4 weeks	$1,000
Lipodystrophy testing					
Lipodystrophy Panel**		2123	81443	6 weeks	$2,500
Macrocephaly testing					
Macrocephaly Panel**		1220	81443	6 weeks	$3,000
***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	$350
Tier 2: *SLC16A2* (MCT8) Mutation Analysis (performed after abnormal thyroid testing)		2130	81404, 81405	4 weeks	$1,000
Menkes disease testing					
ATP7A Mutation Analysis	Menkes disease	1227	81479	4 weeks	$1,000
Microcephalic Osteodysplastic Primordial Dwarfism					
Primordial Dwarfism Panel**		1233	81479	6 weeks	$3,000
Microcephaly testing					
Microcephaly Panel**		1237	81443	6 weeks	$3,000
Multiple Congenital Anomaly testing					
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500

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Movement Disorder testing					
Ataxia Exome**		6101	81443	6 weeks	$4,000
Ataxia Repeat Expansion Panel		4101	81177, 81178, 81179, 81184, 81284, 81479	2-4 weeks	$1,250
Single Gene Repeat Expansion Analysis - *ATN*		4103	81177	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN1*		4103	81178	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN2*		4103	81179	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN3*		4103	81180	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN7*		4103	81181	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN8OS*		4103	81182	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *ATXN10*		4103	81183	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *CACNA1A*		4103	81184	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *FMR1*		4103	81243	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *FXN*		4103	81284	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *PPP2R2B*		4103	81343	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *RFC1***		4103	81479	2-4 weeks	$750
Single Gene Repeat Expansion Analysis - *TBP*		4103	81344	2-4 weeks	$750
Comprehensive Ataxia Testing (concurrent testing option)**		4102	81443	6 weeks	$5,000
Comprehensive Ataxia Testing (reflex testing option)**		6111	81443	6-8 weeks	$1,250 (repeat exapansion) $3,750 (exome)
Hereditary Spastic Paraplegia Exome**		6110	81443	6 weeks	$4,000
Dystonia Chorea Parkinson Exome**		6115	81443	6 weeks	$4,000
Neonatal Diabetes Mellitus, Maturity-Onset Diabetes of the Young and Monogenic Obesity testing					
Comprehensive Neonatal Diabetes Mutation Analysis		2136	81443	4 weeks	$3,500
6q24 Methylation-Specific MLPA	Transient neonatal diabetes	2137	81402	4 weeks	$600
Neonatal Diabetes Mellitus (NDM) Panel		2138	81443	4 weeks	$3,000
Monogenic Diabetes Panel		2140	81443	6 weeks	$3,000
MODY Panel		2141	81443	6 weeks	$2,500
GCK Mutation Analysis	MODY type 2	2142	81406	4 weeks	$850
Bardet-Biedl Syndrome Panel**		2107	81443	6 weeks	$2,000
Non-Syndromic Monogenic Obesity Panel**	Monogenic Obesity	2169	81403, 81406, 81479	6 weeks	$2,000
Monogenic Obesity Panel**		2151	81443	6 weeks	$3,000
Neurodegeneration with brain iron accumulation (NBIA) testing					
Neurodegeneration with Brain Iron Accumulation Panel**		1254	81479	6 weeks	$2,000
Neuronal Ceroid- Lipofuscinoses testing					
Neuronal Ceroid-Lipofuscinoses Panel**		4113	81406, 81479	6 weeks	$2,000
Nicolaides-Baraitser syndrome testing					
SMARCA2 Mutation Analysis	Nicolaides-Baraitser syndrome	1262	81479	4 weeks	$1,000
Noonan syndrome testing					
Noonan Syndrome Panel**		1312	81442	6 weeks	$2,000
Oculodentodigital dysplasia testing					
GJA1 Mutation Analysis		1264	81479	4 weeks	$540
Oral-facial-digital syndrome testing					
OFD1 Mutation Analysis	Oral-facial-digital syndrome	1265	81479	4 weeks	$1,000
Pancreatic Agenesis					
Pancreatic Agenesis Panel**	Pancreatic Agenesis	2179	81404, 81479	6 weeks	$2,000
Pitt-Hopkins syndrome testing					
TCF4 Mutation Analysis	Pitt-Hopkins syndrome	1267	81405, 81406	4 weeks	$1,000
Prader-Willi syndrome testing					
Methylation Specific MLPA		1104	81331	4 weeks	$525
UPD15 (requires samples from both parents also)		1105	81402	4 weeks	$540
Imprinting center deletion analysis		1106	81331	4 weeks	$450
MAGEL2 Mutation Analysis	Shaaf-Yang syndrome	1272	81479	4 weeks	$1,000
Renal Disorders testing					
Hereditary Cystic and Glomerular Disorders Panel**		2176	81443	6 weeks	$3,000
Hereditary Tubulopathy and Nephrolithiasis Panel**		2177	81443	6 weeks	$3,000
Atypical Hemolytic Uremic Syndromes Panel**		2178	81443	6 weeks	$2,000
Rett syndrome testing					
Rett/Angelman Syndrome Panel**		1112	81443	6 weeks	$2,500
Rickets testing					
Hypophosphatemic Rickets Panel**		2173	81404, 81406, 81479	4 weeks	$2,500
Roberts syndrome testing					
ESCO2 Mutation Analysis		1282	81479	4 weeks	$1,000
Robinow syndrome testing					
Robinow Syndrome Panel**		1319	81479	6 weeks	$2,000
Rubinstein-Taybi syndrome testing					
Rubenstein-Taybi Syndrome Panel**		1320	81406, 81407, 81479	6 weeks	$2,000
Schinzel-Giedion syndrome testing					
SETBP1 Mutation Analysis		1292	81479	4 weeks	$1,000
SHORT syndrome testing					
PIK3R1 Mutation Analysis		1294	81479	4 weeks	$1,000
Sotos syndrome testing					
NSD1 Mutation Analysis		1317	81405, 81406	4 weeks	$1,000
Thrombocytopenia testing					
Thrombocytopenia Panel**		5106	81443	6 weeks	$4,000
Temple-Baraitser syndrome testing					
KCNH1 Mutation Analysis		1146	81479	4 weeks	$900
Thyroid Disorders testing					
Hypothyroidism Panel**		2170	81443	4 weeks	$3,000
Hyperparathyroidism Panel**		2171	81404, 81405x2, 81406, 81479	4 weeks	$2,000
Hypoparathyroidism Panel**		2172	81443	4 weeks	$2,500
Type A Insulin Resistant Diabetes with Acanthosis Nigricans testing					
INSR Mutation Analysis	Type A Insulin Resistant Diabetes with Acanthosis Nigricans	2163	81479	4 weeks	$1,000

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***UGT1A1* testing**					
UGT1A1 genotyping for Gilbert syndrome		1295	81350	2 weeks	$450
UGT1A1 genotyping for irinotecan dosing		1296	81350	1 week	$450
UGT1A1 Mutation Analysis	Crigler-Najjar syndrome	1297	81404	4 weeks	$875
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	$540
UPD7 testing	Russell-Silver syndrome	1300	81402	4 weeks	$540
UPD14 testing	UPD14	1301	81402	4 weeks	$540
UPD15 testing	Angelman and Prader-Willi syndromes	1105	81402	4 weeks	$540
Warburg Micro syndrome testing					
Warburg Micro Syndrome Panel**		1316	81479	6 weeks	$2,000
Weaver syndrome testing					
EZH2 Mutation Analysis		1318	81236	4 weeks	$1,000
Wiedemann-Steiner syndrome testing					
KMT2A (*MLL*) Mutation Analysis	Wiedemann-Steiner syndrome	1305	81479	4 weeks	$1,000
Wilson Disease testing					
ATP7B Mutation Analysis	Wilson disease	1307	81406	4 weeks	$1,000
Wolfram syndrome testing					
Wolfram Syndrome Panel**		2167	81479	4 weeks	$2,000
Targeted Testing					
Targeted mutation analysis (known familial mutation)		7102	81403 (unless gene specific)	3 weeks	$500
Testing for a known familial mutation by deletion/duplication analysis (MLPA)		gene specific	81402 (unless gene specific)	4 weeks	$1,000
Single Gene Testing					
Single Gene Mutation Analysis		gene specific	gene specific	4 weeks	$1,000
Prenatal Testing					
Prenatal testing for known mutation by sequence analysis		7104	81403 (unless gene specific)	1 week	$1,800
MISCELLANEOUS FEES					
Cell culture service (skin fibroblasts)		1147	88233	2-3 weeks	$350
STAT fee for known mutations, MS-MLPA or UPD***		n/a	99060	MAY CUT TAT IN HALF	$200
STAT fee for full gene mutation analysis (single gene, requires prior approval)***		n/a	99060	MAY CUT TAT IN HALF	$500
STAT fee for Next-Generation Sequencing panel (requires prior approval)***		n/a	99060	MAY CUT TAT IN HALF	$1,000
Technical lab charge for initial processing of failed sample		n/a	99199	n/a	$150
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 top tube					
***Prices listed are applicable for institutional billing only. Please contact us for discounted patient self-pay prices.**					
****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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