THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Quick Guide To Genetic Testing

TEST	DISORDER	TEST CODE	CPT	TAT	COST*
Aceruloplasminemia testing					
CP sequencing	Aceruloplasminemia	2101	81406	4 weeks	$1,000
CP deletion/duplication		2102	81405	4 weeks	$1,000
Albinism testing					
Albinism Sequencing Panel**		1101	81407	8 weeks	$2,500
Albinism Deletion/Duplication Panel		1102	81407	6 weeks	$2,500
Alstrom syndrome testing					
ALMS1 sequencing	Alstrom syndrome	2103	81406	4 weeks	$1,000
ALMS1 deletion/duplication		2104	81405	4 weeks	$1,000
Alternating Hemiplegia of Childhood testing					
ATP1A3 sequencing	Alternating hemiplegia of childhood	1103	81406	4 weeks	$1,000
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	81403	4 weeks	$450
UBE3A sequencing		1107	81406	4 weeks	$1,000
UBE3A deletion/duplication		1108	81405	4 weeks	$1,000
SLC9A6 sequencing	X-linked Angelman-like syndrome	1109	81406	4 weeks	$1,000
SLC9A6 deletion/duplication		1110	81405	4 weeks	$1,000
Angelman Syndrome Tier 2 Panel**		1111	81407	4 weeks	$2,500
Rett/Angelman Syndrome Sequencing Panel**		1112	81407	8 weeks	$2,500
Rett/Angelman Syndrome Deletion/Duplication Panel		1113	81407	6 weeks	$2,500
Aniridia testing					
*PAX6*sequencing		2105	81406	4 weeks	$1,000
PAX6 deletion/duplication		2106	81405	4 weeks	$1,000
Baraitser Winter syndrome testing					
Baraitser Winter syndrome sequencing panel**		1114	81406	8 weeks	$1,500
Baraitser Winter syndrome deletion/duplication panel		1115	81405	4 weeks	$1,545
Beckwith-Wiedemann syndrome/IMAGe syndrome testing					
CDKN1C sequencing	Beckwith-Wiedemann syndrome	1116	81404	4 weeks	$840
Bernard-Soulier syndrome testing					
GPIbβ sequencing	Bernard-Soulier syndrome	1117	81403	4 weeks	$400
GPIbβ deletion/duplication		1118	81402	4 weeks	$1,000
Brain Malformation testing					
Cerebellar/Pontocerebellar Hypoplasia Sequencing Panel**	Cerebellar/Pontocerebellar Hypoplasia (PCH)	1119	81407	8 weeks	$2,800
Cerebellar/Pontocerebellar Hypoplasia Deletion/Duplication Panel		1120	81407	6 weeks	$2,500
TSEN54 sequencing		1121	81406	4 weeks	$1,000
TSEN54 deletion/duplication		1122	81405	4 weeks	$1,000
CASK sequencing		1123	81407	4 weeks	$1,000
CASK deletion/duplication		1124	81406	4 weeks	$1,000
OPHN1 sequencing		1125	81406	4 weeks	$1,000
OPHN1 deletion/duplication		1126	81405	4 weeks	$1,000
Cerebral Cortical Malformations Sequencing Panel**	Cerebral Cortical Malformations	1127	81407	8 weeks	$3,500
Cerebral Cortical Malformations Deletion/Duplication Panel		1128	81407	6 weeks	$2,500
Holoprosencephaly Sequencing Panel**	Holoprosencephaly	1129	81407	8 weeks	$2,800
Holoprosencephaly Deletion/Duplication Panel		1130	81407	6 weeks	$1,545
L1CAM sequencing	L1 syndrome	1131	81407	4 weeks	$1,000
L1CAM deletion/duplication		1132	81406	4 weeks	$1,000
Comprehensive Hydrocephalus Panel**	Hydrocephalus	1321	81406, 81407	6 weeks	$3,500
Autosomal Recessive Non-Syndromic Hydrocephalus Sequencing Panel**		1133	81407	4 weeks	$2,000
Autosomal Recessive Non-Syndromic Hydrocephalus Deletion/Duplication Panel		1134	81407	4 weeks	$1,545
Lissencephaly Sequencing Panel**	Lissencephaly	1322	81407	8 weeks	$3,000
Lissencephaly Deletion/Duplication Panel		1323	81407	6 weeks	$2,500
Comprehensive Lissencephaly panel**		1135	81406, 81407	8 weeks	$4,000
Cobblestone Lissencephaly Sequencing Panel**		1136	81407	8 weeks	$2,500
Cobblestone Lissencephaly Deletion/Duplication Panel		1137	81407	6 weeks	$1,545
DCX (XLIS) sequencing		1138	81405	4 weeks	$660
DCX deletion/duplication		1139	81404	4 weeks	$1,000
PAFAH1B1 (LIS1) sequencing		1140	81405	4 weeks	$1,000
PAFAH1B1 (LIS1) deletion/duplication		1141	81404	4 weeks	$1,000
TUBA1A sequencing		1142	81404	4 weeks	$900
TUBA1A deletion/duplication		1143	81403	4 weeks	$1,000
ARX sequencing		1144	81404	4 weeks	$900
ARX deletion/duplication		1145	81403	4 weeks	$1,000
Polymicrogyria Sequencing Panel**	Polymicrogyria	1148	81407	8 weeks	$2,500
Polymicrogyria Deletion/Duplication Panel		1149	81407	6 weeks	$2,500
GPR56 sequencing		1150	81406	4 weeks	$1,000
GPR56 deletion/duplication		1151	81405	4 weeks	$1,000
OCLN sequencing (exons 2 - 5)		1152	81405	4 weeks	$875
OCLN deletion/duplication (exons 2 - 5)		1153	81404	4 weeks	$1,000

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TUBB2B sequencing	Polymicrogyria (cont)	1154	81404	4 weeks	$1,000
*TUBB2B*deletion/duplication		1155	81403	4 weeks	$1,000
TUBB3 sequencing		1156	81404	4 weeks	$1,000
TUBB3 deletion/duplication		1157	81403	4 weeks	$1,000
CHARGE syndrome testing					
CHD7 sequencing	CHARGE syndrome	1158	81407	4 weeks	$1,000
CHD7 deletion/duplication		1159	81406	4 weeks	$1,000
CHILD syndrome testing					
NSDHL sequencing	CHILD syndrome	1160	81405	4 weeks	$1,000
NSDHL deletion/duplication		1161	81404	4 weeks	$1,000
CHIME syndrome testing					
PIGL sequencing	CHIME syndrome	1162	81405	4 weeks	$1,000
PIGL deletion/duplication		1163	81404	4 weeks	$1,000
Chondrodysplasia punctata testing					
ARSE sequencing	X-linked recessive (CDPX1)	1164	81405	4 weeks	$900
ARSE deletion/duplication		1165	81404	4 weeks	$1,000
EBP sequencing	X-linked dominant (CDPX2)	1166	81404	4 weeks	$540
EBP deletion/duplication		1167	81403	4 weeks	$1,000
Rhizomelic Chondrodysplasia Punctata Sequencing Panel**		1170	81407	4 weeks	$1,500
Ciliopathy testing					
Bardet-Biedl Syndrome Sequencing Panel**	Bardet-Biedl syndrome	2107	81407	8 weeks	$2,000
Bardet-Biedl Syndrome Deletion/Duplication Panel		2108	81407	6 weeks	$1,545
Joubert/Meckel Gruber Syndrome Sequencing Panel**	Joubert syndrome and Meckel Gruber syndrome	1171	81407	8 weeks	$3,000
Joubert/Meckel Gruber Syndrome Deletion/Duplication Panel		1172	81407	6 weeks	$2,500
Meckel Gruber Syndrome Sequencing Panel**	Meckel Gruber syndrome	1173	81407	8 weeks	$2,500
Meckel Gruber Syndrome Deletion/Duplication Panel		1174	81407	6 weeks	$2,500
Nephronophthisis Sequencing Panel**	Nephronophthisis	1175	81407	8 weeks	$2,000
Nephronophthisis Deletion/Duplication Panel		1176	81407	6 weeks	$1,545
Coffin-Siris syndrome testing					
Coffin-Siris Syndrome Sequencing Panel**		1178	81407	8 weeks	$2,500
Coffin-Siris Syndrome Deletion/Duplication Panel		1179	81407	6 weeks	$1,545
Congenital heart defects (isolated) testing					
NKX2.5 sequencing	Isolated congenital heart defects	1181	81404	4 weeks	$430
NKX2.5 deletion/duplication		1182	81403	4 weeks	$1,000
Congenital Muscle Disease testing					
Bethlem Myopathy and Ullrich Muscular Dystrophy Sequencing Panel**		3121	81407	8 weeks	$2,000
Centronuclear Myopathy Sequencing Panel**		3122	81407	8 weeks	$2,000
Congenital Myopathy Sequencing Panel**		3100	81407	8 weeks	$2,000
Congenital Myopathy Deletion/Duplication Panel		3101	81407	6 weeks	$1,545
Congenital Muscular Dystrophy Sequencing Panel**		3102	81407	8 weeks	$2,000
Congenital Muscular Dystrophy Deletion/Duplication Panel		3103	81407	6 weeks	$1,545
Congenital Myopathy with Prominent Contractures Sequencing Panel**		3104	81407	8 weeks	$2,000
Congenital Myopathy with Prominent Contractures Deletion/Duplication Panel		3105	81407	6 weeks	$1,545
Congenital Myopathy with Fiber-Type Disproportion Sequencing Panel**		3123	81407	8 weeks	$2,000
Congenital Muscular Dystrophy-Dystroglycanopathy Sequencing Panel**		3119	81407	8 weeks	$2,000
Emery-Dreifuss Muscular Dystrophy Sequencing Panel**		3125	81407	8 weeks	$2,000
Limb Girdle Muscular Dystrophy Sequencing Panel**		3106	81407	8 weeks	$2,000
Limb Girdle Muscular Dystrophy Deletion/Duplication Panel		3107	81407	6 weeks	$1,545
Congenital Myasthenic Syndrome Sequencing Panel**		3108	81407	8 weeks	$2,000
Congenital Myasthenic Syndrome Deletion/Duplication Panel		3109	81407	6 weeks	$1,545
Multiminicore Disease Sequencing Panel**		3123	81407	8 weeks	$2,000
Myopathy with Tubular Aggregates Sequencing Panel**		3124	81407	8 weeks	$2,000
Neuromuscular Disorders Sequencing Panel**		3118	81407	8 weeks	$2,000
Nemaline Myopathy Sequencing Panel		3120	81407	8 weeks	$2,000
BIN1 sequencing	Autosomal recessive centronuclear myopathy	3110	81406	4 weeks	$1,000
BIN1 deletion/duplication		3111	81405	4 weeks	$1,000
RYR1 sequencing		3112	81408	4 weeks	$1,000
*RYR1*deletion/duplication		3113	81407	4 weeks	$1,000
DNM2 sequencing	Autosomal dominant centronuclear myopathy, CMT	3114	81406	4 weeks	$1,000
DNM2 deletion/duplication		3115	81405	4 weeks	$1,000
MTM1 sequencing	X-linked myotubular myopathy	3116	81406	4 weeks	$1,000
MTM1 deletion/duplication		3117	81405	4 weeks	$1,000
Cornelia de Lange syndrome testing					
Cornelia de Lange Syndrome Panel**	Cornelia de Lange Syndrome	1324	81406, 81407	4 weeks	$2,500
NIPBL sequencing		1186	81407	4 weeks	$1,000
NIPBL deletion/duplication		1187	81406	4 weeks	$1,000
SMC1A sequencing		1188	81406	4 weeks	$1,000
SMC1A deletion/duplication		1189	81405	4 weeks	$1,000
SMC3,RAD21, HDAC8 sequencing		1190	81407	4 weeks	$1,500
SMC3, RAD21, HDAC8 deletion/duplication		1191	81407	4 weeks	$1,545
Cornelia de Lange PLUS Sequencing Panel**		1192	81407	8 weeks	$3,500
Craniofacial testing					
Craniofacial Sequencing Panel**		1193	81407	8 weeks	$3,000
Craniofacial Deletion/Duplication Panel		1194	81407	6 weeks	$2,500
Facial Dysostosis Sequencing Panel**		1195	81407	8 weeks	$2,500
Facial Dysostosis Deletion/Duplication Panel		1196	81407	6 weeks	$1,545
Currarino syndrome					
MNX1 sequencing		2175	81404	4 weeks	$1,000
MNX1 deletion/duplication analysis		2176	81403	4 weeks	$1,000

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Custom mutation analysis (requires approval by UCGS Lab staff)					
Custom mutation analysis (targeted analysis for known sequence change for first family member)		7101	81403	4 weeks	$540
Custom targeted analysis (for known sequence change for additional family members)		7102	81403	3 weeks	$500
Custom deletion/duplication analysis for known familial deletion/duplication by real-time q-pcr		7103	81402	6 weeks	$1,000
Disorders of Sex Development testing					
Abnormal/Ambiguous Genitalia Sequencing Panel **		2109	81407	8 weeks	$4,000
Hypogonadotropic Hypogonadism Sequencing Panel**		2110	81407	8 weeks	$3,500
Kallmann Syndrome Sequencing Panel**		2174	81407	8 weeks	$3,000
Hypospadias Sequencing Panel**		2111	81407	8 weeks	$2,000
46,XX Disorders of Sex Development/Complete Gonadal Dysgenesis Sequencing Panel**		2112	81407	8 weeks	$2,000
46,XY Disorders of Sex Development/Complete Gonadal Dysgenesis Sequencing Panel**		2113	81407	8 weeks	$3,000
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Distal arthrogryposes testing					
Distal Arthrogryposes Sequencing Panel**		1198	81407	8 weeks	$2,500
Distal Arthrogryposes Deletion/Duplication Panel		1199	81407	6 weeks	$1,545
Donnai-Barrow syndrome testing					
LRP2 sequencing	Donnai-Barrow syndrome	1200	81406	4 weeks	$1,000
LRP2 deletion/duplication		1201	81405	4 weeks	$1,000
Epilepsy testing					
Early Infantile Epileptic Encephalopathy Sequencing Panel**		1326	81407	8 weeks	$2,000
Epilepsy Exome Panel (includes exome sequencing of proband and both parents)**		6102	81415	6 weeks	$5,800
Epilepsy Exome Panel (proband only)**		6103	81415	6 weeks	$4,000
ARX sequencing		1144	81404	4 weeks	$900
ARX deletion/duplication		1145	81403	4 weeks	$1,000
STXBP1 sequencing		4103	81406	4 weeks	$1,000
STXBP1 deletion/duplication		4104	81405	4 weeks	$1,000
SLC25A22 sequencing		4105	81406	4 weeks	$1,000
SLC25A22 deletion/duplication		4106	81405	4 weeks	$1,000
SPTAN1 sequencing		4107	81408	4 weeks	$1,000
SPTAN1 deletion/duplication		4108	81407	4 weeks	$1,000
PCDH19 sequencing		4109	81404	4 weeks	$1,000
PCDH19 deletion/duplication		4110	81403	4 weeks	$1,000
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Exome Sequencing					
Exome Sequencing (Proband only)**		6103	81415	6 weeks	$4,000
Exome Sequencing (Trio)**		6104	81415, 81416	6 weeks	$5,800
STAT Exome Sequencing (Proband only)**		6107	81415	Prelim result in 2 weeks	$8,000
STAT Exome Sequencing (Trio)**		6108	81415, 81416	Prelim result in 2 weeks	$11,600
Exome Select**		6105	81415	6 weeks	$3,000
Fanconi-Bickel syndrome testing					
SLC2A2 sequencing	Fanconi-Bickel syndrome	2114	81406	4 weeks	$1,000
SLC2A2 deletion/duplication		2115	81405	4 weeks	$1,000
Floating Harbor syndrome testing					
SRCAP sequencing	Floating Harbor syndrome	1202	81407	4 weeks	$1,000
SRCAP deletion/duplication		1203	81406	4 weeks	$1,000
Glucose transporter type 1 deficiency testing					
SLC2A1 sequencing	Glucose transporter type 1 deficiency	4111	81405	4 weeks	$1,000
SLC2A1 deletion/duplication		4112	81404	4 weeks	$1,000
Goldberg-Shprintzen megacolon syndrome testing					
KIAA1279 sequencing	Goldberg-Shprintzen megacolon	1204	81405	4 weeks	$1,000
KIAA1279 deletion/duplication		1205	81404	4 weeks	$1,000
Hearing loss testing					
GJB2 sequencing (Connexin 26)	Hearing loss	1206	81252	4 weeks	$500
GJB2 deletion/duplication		1207	81402	4 weeks	$1,000
Targeted analysis for a known familial *GJB2* mutation		7107	81253	3 weeks	$500
Hereditary Cancer testing					
Comprehensive Hereditary Cancer panel**		5144	81406, 81407	6 weeks	$5,000
BRCA1 and BRCA2 Mutation Analysis (sequence and deletion/duplication analysis)**		5146	81211, 81213	4 weeks	$2,500
BRCA1, BRCA2 and PALB2 Mutation Analysis (sequence and deletion/duplication analysis)**		5147	81211, 81213	4 weeks	$2,500
BRCA1, BRCA2 and TP53 Mutation Analysis (sequence and deletion/duplication analysis)**		5148	81211, 81213	4 weeks	$2,500
Hereditary Breast and Ovarian Cancer High Risk Panel**		5149	81211, 81213	6 weeks	$3,500
Comprehensive Hereditary Breast and Ovarian Cancer Panel**		5150	81211, 81213	6 weeks	$3,500
Lynch Syndrome Panel**		5151	81292, 81294, 81295, 81297, 81317, 81319, 81298, 81300	6 weeks	$3,500
Hereditary Colorectal Cancer High Risk Panel**		5152	81292, 81294, 81295, 81297, 81317, 81319, 81298, 81300, 81201, 81203	6 weeks	$3,500
Colorectal Polyposis Panel**		5153	81321, 81323, 81201, 81203	6 weeks	$3,500
Comprehensive Hereditary Colorectal Cancer Panel**		5154	81435, 81436	6 weeks	$4,000
Hereditary Prostate Cancer Panel**		5155	81406, 81407	6 weeks	$3,500

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Familial *BRCA1* mutation	Hereditary Breast and Ovarian Cancer	5101	81215	3 weeks	$500
Familial *BRCA2* mutation		5102	81217	3 weeks	$500
3 Ashkenazi *BRCA1* and *BRCA2* mutations		5103	81212	3 weeks	$500
Comprehensive Familial Myelodysplastic Syndrome/Acute Leukemia Panel**		5143	81406, 81407	6 weeks	$4,000
Tier 1: Familial Myelodysplastic Syndrome / Acute Leukemia Panel**		5104	81406, 81407	6 weeks	$3,500
Tier 2: Familial Myelodysplastic Syndrome / Acute Leukemia Panel**		5107	81406, 81407	6 weeks	$2,000
Hereditary Leukemia and Breast Cancer Panel**		5142	81406, 81407	6 weeks	$4,000
Tier 1: Hereditary Lymphoma and Immunodeficiency Panel**		5108	81406, 81407	6 weeks	$3,300
Tier 2: Hereditary Lymphoma and Immunodeficiency Panel**		5109	81406, 81407	6 weeks	$2,000
Diamond-Blackfan Anemia Sequencing Panel**		5112	81407	8 weeks	$2,000
Diamond-Blackfan Anemia Deletion/Duplication panel		5113	81406	6 weeks	$1,545
Comprehensive Telomere Biology Disorder/Dyskeratosis Congenita Panel**		5114	81406, 81407	6 weeks	$3,500
Telomere Biology Disorder/Dyskeratosis Congenita Sequencing Panel**		5115	81407	6 weeks	$2,500
Dyskeratosis Congenita Deletion/Duplication Panel		5116	81406	6 weeks	$1,545
Fanconi Anemia Sequencing Panel**		5117	81407	8 weeks	$2,000
Fanconi Anemia Deletion/Duplication Panel		5118	81406	6 weeks	$1,545
Hereditary Melanoma Sequencing panel**		5119	81445	6 weeks	$2,000
Hereditary Melanoma Deletion/Duplication panel		5120	81407	6 weeks	$1,545
Hereditary Gastric Cancer Panel**		5121	81445, 81406	6 weeks	$3,500
Hereditary Pheochromocytoma and Paraganglioma Panel**		5122	81445, 81406	6 weeks	$3,000
Hereditary Thyroid Cancer Panel**		5145	81406, 81407	4 weeks	$3,500
Comprehensive Bone Marrow Failure Panel**	Inherited Bone Marrow Failure Disorderes	5133	81406, 81407	6 weeks	$3,000
Inherited Bone Marrow Failure Sequencing Panel**		5134	81407	6 weeks	$2,000
Inherited Bone Marrow Failure Deletion/Duplication Panel		5135	81406	6 weeks	$1,545
SCG5/GREM1 targeted duplication testing (founder mutation)	Hereditary mixed polyposis syndrome	5136	81402	3 weeks	$500
Severe Congenital Neutropenia Sequencing Panel **		5137	81407	8 weeks	$2,000
Severe Congenital Neutropenia Deletion/Duplication Panel		5138	81406	6 weeks	$1,545
SMARCA4 sequencing	Rhabdoid Tumor Predisposition Syndrome	5139	81407	4 weeks	$1,000
SMARCA4 deletion/duplication		5140	81406	4 weeks	$1,000
Hereditary Hemorrhagic Telangictasia (HHT)					
Hereditary Hemorrhagic Telangectasia (HHT) Sequencing Panel**		1314	81407	8 weeks	$1,500
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum					
SLC12A6 sequencing	Hereditary Motor and Sensory Neuropathy with ACC	1208	81406	4 weeks	$1,000
SLC12A6 deletion/duplication		1209	81405	4 weeks	$1,000
Hydroxyglutaric Aciduria (D2 and L2)					
D-2-Hydroxyglutaric Aciduria Sequencing Panel**		1197	81406	4 weeks	$1,500
L2HGDH sequencing		1217	81405	4 weeks	$1,000
SLC25A1 sequencing		1180	81405	4 weeks	$1,000
D-2 and L2-Hydroxyglutaric Aciduria Sequencing Panel**		1325	81407	8 weeks	$1,500
Hyperinsulinism, Congenital testing					
Tier 1 Panel: Diazoxide Unresponsive Hyperinsulinism	Familial hyperinsulinism	2116	81407	7 days	$3,000
Congenital Hyperinsulinism Sequencing Panel		2117	81407	4 weeks	$1,400
Comprehensive Congenital Hyperinsulinism Panel		2118	81406, 81407	4 weeks	$3,000
Hypoinsulinemic Hypoglycemia with Hemihypertrophy testing					
AKT2 sequencing	Hypoinsulinemic Hypoglycemia with Hemihypertrophy	2119	81406	4 weeks	$1,000
AKT2 deletion/duplication		2120	81405	4 weeks	$1,000
Intellectual Disability (ID) testing					
Autosomal Recessive Non-Specific ID Sequencing Panel**		1210	81407	8 weeks	$2,000
X-Linked Non-Specific ID Sequencing Panel**		1211	81470	8 weeks	$2,000
Non-Specific ID Sequencing Panel**		1212	81407, 81470	8 weeks	$3,500
Intellectual Disability Exome Panel (trio)**		6106	81415, 81416	6 weeks	$5,500
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
IPEX syndrome testing					
FOXP3 sequencing	IPEX syndrome	2121	81406	4 weeks	$1,000
FOXP3 deletion/duplication		2122	81405	4 weeks	$1,000
Kabuki syndrome testing					
KMT2D (MLL2) sequencing	Kabuki syndrome	1213	81408	4 weeks	$1,000
KMT2D (MLL2) deletion/duplication		1214	81407	4 weeks	$1,000
KDM6A sequencing		1215	81407	4 weeks	$1,000
KDM6A deletion/duplication		1216	81406	4 weeks	$1,000
Comprehensive Kabuki syndrome panel**		1327	81406, 81407	8 weeks	$2,500
Lipodystrophy testing					
Comprehensive Lipodystrophy Sequencing Panel**		2123	81407	8 weeks	$3,000
Comprehensive Lipodystrophy Deletion/Duplication Panel		2124	81407	6 weeks	$2,500
Congenital Generalized Lipodystrophy Sequencing panel**		2125	81407	8 weeks	$2,000
Congenital Generalized Lipodystrophy Deletion/Duplication panel		2126	81405	6 weeks	$1,545
Partial Lipodystrophy Sequencing Panel**		2127	81407	8 weeks	$3,000
Partial Lipodystrophy Deletion/Duplication Panel		2128	81407	6 weeks	$1,545

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Laminopathy testing					
LMNA sequencing (Lamin A/C)	Laminopathies	1218	81406	4 weeks	$1,000
LMNA deletion/duplication		1219	81405	4 weeks	$1,000
Macrocephaly testing					
Macrocephaly Sequencing Panel**		1220	81407	8 weeks	$3,000
Macrocephaly Deletion/Duplication Panel		1221	81407	6 weeks	$2,500
NSD1 sequencing	Sotos syndrome	1222	81406	4 weeks	$1,000
NSD1 deletion/duplication		1223	81405	4 weeks	$1,000
NSD1 mutation analysis (sequencing + deletion/duplication)		1317	81405, 81406	4 weeks	$1,700
EZH2 sequencing	Weaver syndrome	1224	81406	4 weeks	$1,000
EZH2 deletion/duplication		1225	81405	4 weeks	$1,000
EZH2 mutation analysis (sequencing + deletion/duplication)		1318	81405, 81406	4 weeks	$1,700
NFIX sequencing	Sotos-like syndrome	1226	81405	4 weeks	$1,000
***MCT8* testing: (NOTE: 3-10cc of blood in a red top tube AND 3-10cc of blood in a purple top EDTA tube)**					
Thyroid testing only	Allan-Herndon-Dudley syndrome/*MCT8*-related thyroid hormone cell transporter deficiency	2129	84436, 84481, 84443, 84482	2 weeks	$350
SLC16A2 (MCT8) sequencing (performed after abnormal thyroid testing)		2130	81405	4 weeks	$1,000
SLC16A2 (MCT8) deletion/duplication		2131	81404	4 weeks	$1,000
Menkes disease testing					
ATP7A sequencing	Menkes disease	1227	81406	4 weeks	$1,000
ATP7A deletion/duplication		1228	81405	4 weeks	$1,000
Microcephalic Osteodysplastic Primordial Dwarfism					
Seckel Syndrome Sequencing Panel**		1229	81407	8 weeks	$2,000
Seckel Syndrome Deletion/Duplication Panel		1230	81407	6 weeks	$1,545
Meier-Gorlin Syndrome Sequencing Panel**		1231	81407	8 weeks	$2,000
Meier-Gorlin Syndrome Deletion/Duplication Panel		1232	81407	6 weeks	$1,545
Comprehensive Primordial Dwarfism Sequencing Panel**		1233	81407	8 weeks	$2,000
Comprehensive Primordial Dwarfism Deletion/Duplication Panel		1234	81407	6 weeks	$1,545
3-M Syndrome Sequencing Panel**		1328	81407	8 weeks	$1,500
Microcephaly testing					
Microcephaly Sequencing Panel**		1237	81407	8 weeks	$4,000
Comprehensive Autosomal Recessive Primary Microcephaly Panel**	Autosomal Recessive Primary Microcephaly	1315	81406, 81407	8 weeks	$3,000
ASPM sequencing		1240	81407	4 weeks	$1,000
ASPM deletion/duplication		1241	81406	4 weeks	$1,000
IER3IP1 sequencing	Microcephaly, Epilepsy and Diabetes Syndrome	2132	81404	4 weeks	$1,000
IER3IP1 deletion/duplication		2133	81403	4 weeks	$1,000
NDE1 sequencing		1244	81405	4 weeks	$1,000
NDE1 deletion/duplication		1245	81404	4 weeks	$1,000
PNKP sequencing		1246	81406	4 weeks	$1,000
PNKP deletion/duplication		1247	81405	4 weeks	$1,000
STAMBP sequencing	Microcephaly-Capillary Malformation Syndrome	1248	81405	4 weeks	$1,000
STAMBP deletion/duplication		1249	81404	4 weeks	$1,000
*WDR62*sequencing		1250	81407	4 weeks	$1,000
WDR62 deletion/duplication		1251	81406	4 weeks	$1,000
Mitchell-Riley syndrome testing					
RFX6 sequencing	Mitchell-Riley syndrome	2134	81406	4 weeks	$1,000
RFX6 deletion/duplication		2135	81405	4 weeks	$1,000
Mowat Wilson syndrome testing					
ZEB2 sequencing	Mowat-Wilson syndrome	1252	81405	4 weeks	$1,000
ZEB2 deletion/duplication		1253	81404	4 weeks	$1,000
Multiple Congenital Anomaly testing					
Cytogenomic SNP array (postnatal)		8100	81229	4 weeks	$1,500
Movement Disorder testing					
Ataxia Exome Panel**		6101	81415	6 weeks	$4,000
Dystonia Exome Panel**		6109	81415	6 weeks	$3,500
Hereditary Spastic Paraplegia Exome Panel**		6110	81415	6 weeks	$3,000
Neonatal Diabetes Mellitus, Maturity-Onset Diabetes of the Young and Monogenic Obesity testing					
Comprehensive Neonatal Diabetes Mutation Analysis		2136	81407	4 weeks	$3,000
6q24 Methylation-Specific MLPA	Transient neonatal diabetes	2137	81402	4 weeks	$600
Neonatal Diabetes Mellitus (NDM) Sequencing Panel		2138	81407	4 weeks	$2,500
Neonatal Diabetes Mellitus (NDM) Deletion/Duplication Panel		2139	81407	6 weeks	$1,545
Neonatal Diabetes/MODY Sequencing Panel		2140	81407	8 weeks	$3,000
MODY Panel		2141	81406, 81407	6 weeks	$3,000
GCK sequencing	MODY type 2	2142	81405	4 weeks	$850
*GCK*deletion/duplication		2143	81404	4 weeks	$1,000
GLIS3 sequencing	NDM with Congenital Hypothyroidism	2144	81405	4 weeks	$1,000
GLIS3 deletion/duplication		2145	81404	4 weeks	$1,000
KCNJ11 sequencing	Neonatal diabetes	2146	81403	4 weeks	$400
KCNJ11 deletion/duplication		2147	81402	4 weeks	$1,000
NEUROG3 sequencing	NDM with Severe Malabsorptive Diarrhea	2148	81403	4 weeks	$1,000
NEUROG3 deletion/duplication		2149	81403	4 weeks	$1,000
Bardet-Biedl Syndrome Sequencing Panel**		2107	81407	8 weeks	$2,000
Bardet-Biedl Syndrome Deletion/Duplication Panel		2108	81407	6 weeks	$1,545
Non-Syndromic Monogenic Obesity Sequencing Panel**	Monogenic Obesity	2169	81407	8 weeks	$2,500
Monogenic Obesity Sequencing Panel**		2151	81407	8 weeks	$3,000

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Quick Guide To Genetic Testing

TEST	DISORDER	TEST CODE	CPT	TAT	COST*
Neurodegeneration with brain iron accumulation (NBIA) testing					
NBIA Sequencing Panel**		1254	81407	8 weeks	$2,500
NBIA Deletion/Duplication Panel		1255	81407	6 weeks	$1,545
PANK2 sequencing	PKAN/Hallervorden-Spatz syndrome	1258	81405	4 weeks	$900
PANK2 deletion/duplication		1259	81404	4 weeks	$1,000
PLA2G6 sequencing	Infantile neuroaxonal dystrophy	1260	81406	4 weeks	$1,000
PLA2G6 deletion/duplication		1261	81405	4 weeks	$1,000
Neuronal Ceroid- Lipofuscinoses testing					
Neuronal Ceroid-Lipofuscinoses Panel**		4113	81406, 81407	8 weeks	$2,000
Nicolaides-Baraitser syndrome testing					
SMARCA2 sequencing	Nicolaides-Baraitser syndrome	1262	81407	4 weeks	$1,000
SMARCA2 deletion/duplication		1263	81406	4 weeks	$1,000
Noonan syndrome testing					
Noonan Syndrome Sequencing panel**		1312	81407	8 weeks	$2,000
Noonan Syndrome Deletion/Duplication panel		1313	81407	6 weeks	$1,545
Oculodentodigital dysplasia testing					
GJA1 sequencing		1264	81404	4 weeks	$540
Oral-facial-digital syndrome testing					
OFD1 sequencing	Oral-facial-digital syndrome	1265	81406	4 weeks	$1,000
OFD1 deletion/duplication		1266	81405	4 weeks	$1,000
Pancreatic Agenesis					
*PDX1*sequencing	Pancreatic Agenesis	2152	81404	4 weeks	$1,000
PDX1 deletion/duplication		2153	81403	4 weeks	$1,000
GATA6 sequencing	Pancreatic Agenesis and Congenital Heart Defects	2154	81405	4 weeks	$1,000
GATA6 deletion/duplication		2155	81404	4 weeks	$1,000
*PTF1A*sequencing	Pancreatic and Cerebellar Agenesis	2156	81404	4 weeks	$1,000
PTF1A deletion/duplication		2157	81403	4 weeks	$1,000
Pigmented hypertrichotic dermatosis with insulin-dependent diabetes mellitus testing					
SLC29A3 sequencing		2158	81405	4 weeks	$1,000
SLC29A3 deletion/duplication		2159	81404	4 weeks	$1,000
Pitt-Hopkins syndrome testing					
TCF4 sequencing	Pitt-Hopkins syndrome	1267	81406	4 weeks	$1,000
TCF4 deletion/duplication		1268	81405	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	81403	4 weeks	$450
MAGEL2 sequencing		1272	81403	4 weeks	$1,000
Premature Ovarian Failure testing					
Premature Ovarian Failure Panel**		2160	81407	8 weeks	$2,000
Renal Cystic Disorders testing					
Renal Cystic Disorders Sequencing Panel**		1311	81407	8 weeks	$2,000
Rett/Atypical Rett syndrome testing					
Rett/Atypical Rett Syndrome Panel**		1273	81406, 81407	4 weeks	$2,500
Rett/Angelman Syndrome Sequencing Panel**		1112	81407	8 weeks	$2,500
Rett/Angelman Syndrome Deletion/Duplication Panel		1113	81407	6 weeks	$2,500
MECP2 sequencing	Rett syndrome	1274	81302	4 weeks	$900
MECP2 deletion/duplication		1275	81304	4 weeks	$1,000
CDKL5 sequencing		1276	81406	4 weeks	$1,000
CDKL5 deletion/duplication		1277	81405	4 weeks	$1,000
FOXG1 sequencing	Rett syndrome, congenital variant	1278	81404	4 weeks	$550
FOXG1 deletion/duplication		1279	81402	4 weeks	$1,000
MEF2C sequencing		1280	81405	4 weeks	$1,000
MEF2C deletion/duplication		1281	81404	4 weeks	$1,000
Rickets testing					
Rickets Sequencing Panel**		2173	81407	4 weeks	$2,500
Roberts syndrome testing					
ESCO2 sequencing		1282	81405	4 weeks	$1,000
ESCO2 deletion/duplication		1283	81404	4 weeks	$1,000
Robinow syndrome testing					
ROR2 sequencing	Autosomal Recessive Robinow syndrome, brachydactyly type B1	1284	81405	4 weeks	$1,000
ROR2 deletion/duplication		1285	81404	4 weeks	$1,000
WNT5A sequencing	Autosomal Dominant Robinow syndrome	1286	81404	4 weeks	$1,000
WNT5A deletion/duplication		1287	81403	4 weeks	$1,000
Comprehensive Robinow Syndrome panel**		1319	81406, 81407	8 weeks	$2,500
Rubinstein-Taybi syndrome testing					
CREBBP sequencing		1288	81407	4 weeks	$1,000
CREBBP deletion/duplication		1289	81406	4 weeks	$1,000
EP300 sequencing		1290	81407	4 weeks	$1,000
EP300 deletion/duplication		1291	81406	4 weeks	$1,000
Comprehensive Rubenstein-Taybi Syndrome panel**		1320	81406, 81407	8 weeks	$2,500
Schinzel-Giedion syndrome testing					
SETBP1 sequencing		1292	81404	4 weeks	$1,000
SETBP1 deletion/duplication		1293	81403	4 weeks	$1,000

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TEST	DISORDER	TEST CODE	CPT	TAT	COST*
SHORT syndrome testing					
*PIK3R1*sequencing		1294	81406	4 weeks	$1,000
Thrombocytopenia testing					
Thrombocytopenia Sequencing panel**		5141	81407	6 weeks	$3,000
Temple-Baraitser syndrome testing					
*KCNH1*sequencing		1146	81406	4 weeks	$900
Thiamine Responsive Megaloblastic Anemia syndrome testing					
SLC19A2 sequencing		2161	81405	4 weeks	$1,000
SLC19A2 deletion/duplication		2162	81404	4 weeks	$1,000
Thyroid Disorders testing					
Congenital Hypothyroidism Sequencing panel**		2170	81407	4 weeks	$2,500
Hyperparathyroidism Sequencing panel**		2171	81407	4 weeks	$2,500
Hypoparathyroidism Sequencing panel**		2172	81407	4 weeks	$2,500
Type A Insulin Resistant Diabetes with Acanthosis Nigricans testing					
INSR sequencing	Type A Insulin Resistant Diabetes with Acanthosis Nigricans	2163	81406	4 weeks	$1,000
INSR deletion/duplication		2164	81405	4 weeks	$1,000
***UGT1A1* testing**					
UGT1A1 genotyping for Gilbert syndrome		1295	81350	2 weeks	$390
UGT1A1 genotyping for irinotecan dosing		1296	81350	1 week	$390
UGT1A1 sequencing	Crigler-Najjar syndrome	1297	81404	4 weeks	$875
UGT1A1 deletion/duplication		1298	81403	4 weeks	$1,000
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 Sequencing Panel**		1303	81407	8 weeks	$1,500
Warburg Micro Syndrome Deletion/Duplication Panel		1304	81407	6 weeks	$1,545
Warburg Micro Syndrome Comprehensive Panel**		1316	81406, 81407	8 weeks	$2,500
Wiedemann-Steiner syndrome testing					
KMT2A (MLL) sequencing	Wiedemann-Steiner syndrome	1305	81407	4 weeks	$1,000
KMT2A (MLL) deletion/duplication		1306	81406	4 weeks	$1,000
Wilson Disease testing					
ATP7B sequencing	Wilson disease	1307	81406	4 weeks	$1,000
ATP7B deletion/duplication		1308	81405	4 weeks	$1,000
Wolcott-Rallison syndrome					
EIF2AK3 sequencing	Wolcott-Rallison syndrome	2165	81406	4 weeks	$1,000
EIF2AK3 deletion/duplication		2166	81405	4 weeks	$1,000
Wolfram syndrome testing					
Wolfram Syndrome Sequencing panel**		2167	81406	4 weeks	$1,500
Wolfram Syndrome Deletion/Duplication panel		2168	81405	4 weeks	$1,545
Woodhouse-Sakati syndrome testing					
DCAF17 sequencing		1309	81406	4 weeks	$1,000
DCAF17 deletion/duplication		1310	81405	4 weeks	$1,000
Targeted Testing					
Testing for a known mutation in additional family member by sequence analysis		7102	81403	3 weeks	$500
Testing for a known mutation in additional family member by deletion/duplication analysis (MLPA or array-CGH)		gene specific	gene specific	4 weeks	$1,000
Prenatal Testing					
Prenatal testing for known mutation by sequence analysis		7104	81403	1 week	$1,800
Prenatal testing for known deletion/duplication by MLPA		7105	81402	2 weeks	$1,800
Prenatal testing for known deletion/duplication by real-time q-pcr		7106	81402	3 weeks	$1,800
MISCELLANEOUS FEES					
STAT fee for known mutations, deletion/duplication testing, MS-MLPA or UPD***		n/a	99060	MAY CUT TAT IN HALF	$200
STAT fee for full gene sequencing (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 options only. Please contact us for prices for direct insurance billing for applicable tests.**

****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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6/2017