THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

# Quick Guide To Genetic Testing

| TEST | DISORDER | TEST CODE | CPT | TAT | INSTITUTIONAL COST* |
|---|---|---|---|---|---|
| **Aceruloplasminemia testing** | | | | | |
| CP Mutation Analysis | Aceruloplasminemia | 2101 | 81405, 81406 | 4 weeks | $1,000 |
| **Albinism testing** | | | | | |
| Albinism Panel** | | 1101 | 81406, 81407 | 8 weeks | $2,500 |
| **Alstrom syndrome testing** | | | | | |
| ALMS1 Mutation Analysis | Alstrom syndrome | 2103 | 81405, 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 |
| Angelman Syndrome Tier 2 Panel** | | 1111 | 81407 | 4 weeks | $2,500 |
| Rett/Angelman Syndrome Panel** | | 1112 | 81406, 81407 | 8 weeks | $2,500 |
| **Aniridia testing** | | | | | |
| *PAX6* Mutation Analysis | | 2105 | 81405, 81406 | 4 weeks | $1,000 |
| **Baraitser Winter syndrome testing** | | | | | |
| Baraitser Winter syndrome Panel** | | 1114 | 81405, 81406 | 8 weeks | $1,500 |
| **IMAGe syndrome/Beckwith-Wiedemann syndrome testing** | | | | | |
| CDKN1C Mutation Analysis | Beckwith-Wiedemann syndrome | 1116 | 81404 | 4 weeks | $1,000 |
| **Bernard-Soulier syndrome testing** | | | | | |
| GPIbβ Mutation Analysis | Bernard-Soulier syndrome | 1117 | 81402, 81403 | 4 weeks | $1,000 |
| **Brain Malformation testing** | | | | | |
| Cerebellar/Pontocerebellar Hypoplasia Panel** | Cerebellar/Pontocerebellar Hypoplasia (PCH) | 1119 | 81407 | 8 weeks | $2,800 |
| Cerebral Cortical Malformations Panel** | Cerebral Cortical Malformations | 1127 | 81406, 81407 | 8 weeks | $3,500 |
| Holoprosencephaly Panel** | Holoprosencephaly | 1129 | 81406, 81407 | 8 weeks | $2,800 |
| Hydrocephalus Panel** | Hydrocephalus | 1321 | 81406, 81407 | 6 weeks | $3,500 |
| Autosomal Recessive Non-Syndromic Hydrocephalus Panel** | | 1133 | 81406, 81407 | 4 weeks | $2,000 |
| Lissencephaly Panel** | | 1135 | 81406, 81407 | 8 weeks | $4,000 |
| Cobblestone Lissencephaly Panel** | | 1136 | 81406, 81407 | 8 weeks | $2,500 |
| Polymicrogyria Panel** | Polymicrogyria | 1148 | 81406, 81407 | 8 weeks | $2,500 |
| **CHARGE syndrome testing** | | | | | |
| CHD7 Mutation Analysis | CHARGE syndrome | 1158 | 81406, 81407 | 4 weeks | $1,000 |
| **CHILD syndrome testing** | | | | | |
| NSDHL Mutation Analysis | CHILD syndrome | 1160 | 81404, 81405 | 4 weeks | $1,000 |
| **CHIME syndrome testing** | | | | | |
| PIGL Mutation Analysis | CHIME syndrome | 1162 | 81404, 81405 | 4 weeks | $1,000 |
| **Chondrodysplasia punctata testing** | | | | | |
| ARSE Mutation Analysis | X-linked recessive (CDPX1) | 1164 | 81404, 81405 | 4 weeks | $1,000 |
| EBP Mutation Analysis | X-linked dominant (CDPX2) | 1166 | 81403, 81404 | 4 weeks | $1,000 |
| Rhizomelic Chondrodysplasia Punctata Panel** | | 1170 | 81406, 81407 | 4 weeks | $1,500 |
| **Ciliopathy testing** | | | | | |
| Bardet-Biedl Syndrome Panel** | Bardet-Biedl syndrome | 2107 | 81406, 81407 | 8 weeks | $2,000 |
| Joubert/Meckel Gruber Syndrome Panel** | Joubert syndrome and Meckel Gruber syndrome | 1171 | 81406, 81407 | 8 weeks | $3,000 |
| Meckel Gruber Syndrome Panel** | Meckel Gruber syndrome | 1173 | 81406, 81407 | 8 weeks | $2,500 |
| Nephronophthisis Panel** | Nephronophthisis | 1175 | 81406, 81407 | 8 weeks | $2,000 |
| **Coffin-Siris syndrome testing** | | | | | |
| Coffin-Siris Syndrome Panel** | | 1178 | 81406, 81407 | 8 weeks | $2,500 |
| **Congenital Muscle Disease testing** | | | | | |
| Bethlem Myopathy and Ullrich Muscular Dystrophy Panel** | | 3121 | 81406, 81407 | 8 weeks | $2,000 |
| Centronuclear Myopathy Panel** | | 3122 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Myopathy Panel** | | 3100 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Muscular Dystrophy Panel** | | 3102 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Myopathy with Prominent Contractures Panel** | | 3104 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Myopathy with Fiber-Type Disproportion Panel** | | 3126 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Muscular Dystrophy-Dystroglycanopathy Panel** | | 3119 | 81406, 81407 | 8 weeks | $2,000 |
| Emery-Dreifuss Muscular Dystrophy Panel** | | 3125 | 81406, 81407 | 8 weeks | $2,000 |
| Limb Girdle Muscular Dystrophy Panel** | | 3106 | 81406, 81407 | 8 weeks | $2,000 |
| Congenital Myasthenic Syndrome Panel** | | 3108 | 81406, 81407 | 8 weeks | $2,000 |
| Multiminicore Disease Panel** | | 3123 | 81406, 81407 | 8 weeks | $2,000 |
| Myopathy with Tubular Aggregates Panel** | | 3124 | 81406, 81407 | 8 weeks | $2,000 |
| Neuromuscular Disorders Exome Panel** | | 6112 | 81415 | 6 weeks | $4,000 |
| Neuromuscular Disorders Panel** | | 3118 | 81406, 81407 | 8 weeks | $2,000 |
| Nemaline Myopathy Panel | | 3120 | 81406, 81407 | 8 weeks | $2,000 |
| **Cornelia de Lange syndrome testing** | | | | | |
| Cornelia de Lange Syndrome Mosaicism Panel (Sequencing Only)** | Cornelia de Lange Syndrome | 1324 | 81407 | 4 weeks | $2,000 |
| Cornelia de Lange PLUS Panel** | | 1192 | 81406, 81407 | 8 weeks | $3,500 |
| **Craniofacial testing** | | | | | |
| Craniofacial Panel** | | 1193 | 81406, 81407 | 8 weeks | $3,000 |
| Facial Dysostosis Panel** | | 1195 | 81406, 81407 | 8 weeks | $2,500 |
| **Currarino syndrome** | | | | | |
| MNX1 Mutation Analysis | | 2175 | 81403, 81404 | 4 weeks | $1,000 |
| **Custom Mutation Analysis (requires approval by UCGS Lab staff)** | | | | | |
| Custom Mutation Analysis for known sequence change in 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 Panel ** | | 2109 | 81406, 81407 | 8 weeks | $4,000 |
| Hypogonadotropic Hypogonadism Panel** | | 2110 | 81406, 81407 | 8 weeks | $3,500 |
| Kallmann Syndrome Panel** | | 2174 | 81406, 81407 | 8 weeks | $3,000 |
| 46,XX Disorders of Sex Development/Complete Gonadal Dysgenesis Panel** | | 2112 | 81406, 81407 | 8 weeks | $2,000 |
| 46,XY Disorders of Sex Development/Complete Gonadal Dysgenesis Panel** | | 2113 | 81406, 81407 | 8 weeks | $3,000 |
| Cytogenomic SNP array (postnatal) | | 8100 | 81229 | 4 weeks | $1,500 |

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| **Distal arthrogryposes testing** | | | | | |
| Distal Arthrogryposes Panel** | | 1198 | 81406, 81407 | 8 weeks | $2,500 |
| **Donnai-Barrow syndrome testing** | | | | | |
| LRP2 Mutation Analysis | Donnai-Barrow syndrome | 1200 | 81405, 81406 | 4 weeks | $1,000 |
| **Dyslipidemia testing** | | | | | |
| Dyslipidemia Panel | | 2186 | 81406, 81407 | 8 weeks | $3,000 |
| Hypercholesterolemia Panel | | 2188 | 81406, 81407 | 8 weeks | $2,500 |
| **Epilepsy testing** | | | | | |
| Early Infantile Epileptic Encephalopathy Panel** | | 1326 | 81406, 81407 | 8 weeks | $3,000 |
| Epilepsy Exome (includes exome sequencing of proband and both parents)** | | 6102 | 81415 | 6 weeks | $5,800 |
| Epilepsy Exome (proband only)** | | 6103 | 81415 | 6 weeks | $4,000 |
| 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 Mutation Analysis | Fanconi-Bickel syndrome | 2114 | 81405, 81406 | 4 weeks | $1,000 |
| **Floating Harbor syndrome testing** | | | | | |
| SRCAP Mutation Analysis | Floating Harbor syndrome | 1202 | 81406, 81407 | 4 weeks | $1,000 |
| **Glucose transporter type 1 deficiency testing** | | | | | |
| SLC2A1 Mutation Analysis | Glucose transporter type 1 deficiency | 4111 | 81404, 81405 | 4 weeks | $1,000 |
| **Goldberg-Shprintzen megacolon syndrome testing** | | | | | |
| KIAA1279 Mutation Analysis | Goldberg-Shprintzen megacolon | 1204 | 81404, 81405 | 4 weeks | $1,000 |
| **Hereditary Cancer testing** | | | | | |
| Comprehensive Hereditary Cancer Panel** | | 5144 | 81406, 81407 | 6 weeks | $4,000 |
| BRCA1, BRCA2 and PALB2 Mutation Analysis** | | 5147 | 81163, 81164 | 4 weeks | $2,500 |
| BRCA1, BRCA2 and TP53 Mutation Analysis** | | 5148 | 81163, 81164 | 4 weeks | $2,500 |
| Hereditary Breast and Ovarian Cancer High Risk Panel** | | 5149 | 81163, 81164 | 6 weeks | $3,500 |
| Hereditary Breast and Ovarian Cancer Panel** | | 5150 | 81163, 81164 | 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 |
| Hereditary Colorectal Cancer Panel** | | 5154 | 81435, 81436 | 6 weeks | $4,000 |
| Hereditary Prostate Cancer Panel** | | 5155 | 81407 | 6 weeks | $3,500 |
| 3 Ashkenazi BRCA1 and BRCA2 mutations | | 5103 | 81212 | 3 weeks | $500 |
| Hereditary Myeloid Malignancy and Inherited Bone Marrow Failure Panel** | | 5156 | 81406, 81407 | 6 weeks | $4,000 |
| Familial Myelodysplastic Syndrome/Acute Leukemia Panel** | | 5143 | 81406, 81407 | 6 weeks | $4,000 |
| Hereditary Leukemia and Breast Cancer Panel** | | 5142 | 81406, 81407 | 6 weeks | $4,000 |
| Lymphoma and Immunodeficiency Panel** | | 5158 | 81406, 81407 | 6 weeks | $4,000 |
| Diamond-Blackfan Anemia Panel** | | 5159 | 81406, 81407 | 6 weeks | $3,000 |
| Telomere Biology Disorder/Dyskeratosis Congenita Panel** | | 5114 | 81406, 81407 | 6 weeks | $3,500 |
| Fanconi Anemia Panel** | | 5160 | 81406, 81407 | 6 weeks | $3,000 |
| Hereditary Melanoma Panel** | | 5161 | 81445, 81407 | 6 weeks | $3,000 |
| Hereditary Gastric Cancer Panel** | | 5121 | 81445 | 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 |
| Inherited Bone Marrow Failure Panel** | | 5133 | 81406, 81407 | 6 weeks | $3,000 |
| SCG5/GREM1 targeted duplication testing (founder mutation) | Hereditary mixed polyposis syndrome | 5136 | 81402 | 3 weeks | $500 |
| Severe Congenital Neutropenia Panel** | | 5162 | 81406, 81407 | 6 weeks | $3,000 |
| SMARCA4 Mutation Analysis | Rhabdoid tumor predisposition syndrome testing | 5163 | 81406, 81407 | 4 weeks | $1,000 |
| **Hereditary Hemorrhagic Telangictasia (HHT)** | | | | | |
| Hereditary Hemorrhagic Telangectasia (HHT) Panel** | | 1314 | 81406, 81407 | 8 weeks | $1,500 |
| **Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum** | | | | | |
| SLC12A6 Mutation Analysis | Hereditary Motor and Sensory Neuropathy with ACC | 1208 | 81405, 81406 | 4 weeks | $1,000 |
| **Hydroxyglutaric Aciduria (D2 and L2)** | | | | | |
| D-2-Hydroxyglutaric Aciduria Panel** | | 1197 | 81406, 81407 | 4 weeks | $1,500 |
| D-2 and L2-Hydroxyglutaric Aciduria Panel** | | 1325 | 81406, 81407 | 8 weeks | $1,500 |
| **Hyperinsulinism, Congenital testing** | | | | | |
| Tier 1 Panel: Diazoxide Unresponsive Hyperinsulinism | Familial hyperinsulinism | 2116 | 81406, 81407 | 7 days | $3,000 |
| Congenital Hyperinsulinism Panel | | 2118 | 81406, 81407 | 4 weeks | $3,000 |
| **Hypoinsulinemic Hypoglycemia with Hemihypertrophy testing** | | | | | |
| AKT2 Mutation Analysis | Hypoinsulinemic Hypoglycemia with Hemihypertrophy | 2119 | 81405, 81406 | 4 weeks | $1,000 |
| **Intellectual Disability (ID) testing** | | | | | |
| Autosomal Recessive Non-Specific ID Panel** | | 1210 | 81406, 81407 | 8 weeks | $2,000 |
| X-Linked Non-Specific ID Panel** | | 1211 | 81406, 81407 | 8 weeks | $2,000 |
| Non-Specific ID Panel** | | 1212 | 81406, 81407 | 8 weeks | $3,500 |
| Intellectual Disability Exome (Trio)** | | 6106 | 81415, 81416 | 6 weeks | $5,500 |
| Cytogenomic SNP array (postnatal) | | 8100 | 81229 | 4 weeks | $1,500 |
| **IPEX syndrome testing** | | | | | |
| FOXP3 Mutation Analysis | IPEX syndrome | 2121 | 81405, 81406 | 4 weeks | $1,000 |
| **Kabuki syndrome testing** | | | | | |
| Kabuki Syndrome Panel** | | 1327 | 81406, 81407 | 8 weeks | $2,500 |
| **Lipodystrophy testing** | | | | | |
| Comprehensive Lipodystrophy Panel** | | 2123 | 81406, 81407 | 8 weeks | $3,000 |
| Congenital Generalized Lipodystrophy Panel** | | 2125 | 81406, 81407 | 8 weeks | $2,000 |
| Partial Lipodystrophy Panel** | | 2127 | 81406, 81407 | 8 weeks | $3,000 |
| **Laminopathy testing** | | | | | |
| LMNA Mutation Analysis (Lamin A/C) | Laminopathies | 1218 | 81405, 81406 | 4 weeks | $1,000 |

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| **Macrocephaly testing** | | | | | |
| Macrocephaly Panel** | | 1220 | 81406, 81407 | 8 weeks | $3,000 |
| **Marshall-Smith syndrome testing** | | | | | |
| NFIX Mutation Analysis | | 1226 | 81405, 81406 | 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)** | | | | | |
| Tier 1: Thyroid testing only | Allan-Herndon-Dudley syndrome/*MCT8*-related thyroid hormone cell transporter deficiency | 2129 | 84436, 84481, 84443, 84482 | 2 weeks | $350 |
| Tier 2: SLC16A2 (MCT8) Mutation Analysis (performed after abnormal thyroid testing) | | 2130 | 81405 | 4 weeks | $1,000 |
| **Menkes disease testing** | | | | | |
| ATP7A Mutation Analysis | Menkes disease | 1227 | 81405, 81406 | 4 weeks | $1,000 |
| **Microcephalic Osteodysplastic Primordial Dwarfism** | | | | | |
| Seckel Syndrome Panel** | | 1229 | 81406, 81407 | 8 weeks | $2,000 |
| Meier-Gorlin Syndrome Panel** | | 1231 | 81406, 81407 | 8 weeks | $2,000 |
| Primordial Dwarfism Panel** | | 1233 | 81406, 81407 | 8 weeks | $2,000 |
| 3-M Syndrome Panel** | | 1328 | 81406, 81407 | 8 weeks | $1,500 |
| **Microcephaly testing** | | | | | |
| Microcephaly Panel** | | 1237 | 81406, 81407 | 8 weeks | $4,000 |
| Autosomal Recessive Primary Microcephaly Panel** | Autosomal Recessive Primary Microcephaly | 1315 | 81406, 81407 | 8 weeks | $3,000 |
| **Mitchell-Riley syndrome testing** | | | | | |
| RFX6 Mutation Analysis | Mitchell-Riley syndrome | 2134 | 81405, 81406 | 4 weeks | $1,000 |
| **Mowat Wilson syndrome testing** | | | | | |
| ZEB2 Mutation Analysis | Mowat-Wilson syndrome | 1252 | 81404, 81405 | 4 weeks | $1,000 |
| **Multiple Congenital Anomaly testing** | | | | | |
| Cytogenomic SNP array (postnatal) | | 8100 | 81229 | 4 weeks | $1,500 |
| **Movement Disorder testing** | | | | | |
| Ataxia Exome** | | 6101 | 81415 | 6 weeks | $4,000 |
| Ataxia Repeat Expansion Panel | | 4101 | 81401 | 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 | 81401, 81415 | 6 weeks | $5,000 |
| Comprehensive Ataxia Testing (reflex testing option)** | | 6111 | 81401, 81415 | 6-8 weeks | $1,250 (repeat exapansion) $3,750 (exome) |
| Dystonia Exome** | | 6109 | 81415 | 6 weeks | $3,500 |
| Hereditary Spastic Paraplegia Exome** | | 6110 | 81415 | 6 weeks | $3,000 |
| **Neonatal Diabetes and Hyperinsulinism of Infancy testing** | | | | | |
| KCNJ11 Mutation Analysis | | 2146 | 81402, 81403 | 4 weeks | $1,000 |
| **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) Panel | | 2138 | 81406, 81407 | 4 weeks | $2,500 |
| Neonatal Diabetes/MODY Panel | | 2140 | 81406, 81407 | 8 weeks | $3,000 |
| MODY Panel | | 2141 | 81406, 81407 | 6 weeks | $3,000 |
| GCK Mutation Analysis | MODY type 2 | 2142 | 81404, 81405 | 4 weeks | $850 |
| GLIS3 Mutation Analysis | NDM with Congenital Hypothyroidism | 2144 | 81404, 81405 | 4 weeks | $1,000 |
| Bardet-Biedl Syndrome Panel** | | 2107 | 81406, 81407 | 8 weeks | $2,000 |
| Non-Syndromic Monogenic Obesity Panel** | Monogenic Obesity | 2169 | 81406, 81407 | 8 weeks | $2,500 |
| Monogenic Obesity Panel** | | 2151 | 81406, 81407 | 8 weeks | $3,000 |
| **Neurodegeneration with brain iron accumulation (NBIA) testing** | | | | | |
| NBIA Panel** | | 1254 | 81406, 1407 | 8 weeks | $2,500 |
| **Neuronal Ceroid- Lipofuscinoses testing** | | | | | |
| Neuronal Ceroid-Lipofuscinoses Panel** | | 4113 | 81406, 81407 | 8 weeks | $2,000 |
| **Nicolaides-Baraitser syndrome testing** | | | | | |
| SMARCA2 Mutation Analysis | Nicolaides-Baraitser syndrome | 1262 | 81406, 81407 | 4 weeks | $1,000 |
| **Noonan syndrome testing** | | | | | |
| Noonan Syndrome Panel** | | 1312 | 81406, 81407 | 8 weeks | $2,000 |
| **Oculodentodigital dysplasia testing** | | | | | |
| GJA1 Mutation Analysis | | 1264 | 81404 | 4 weeks | $1,000 |
| **Oral-facial-digital syndrome testing** | | | | | |
| OFD1 Mutation Analysis | Oral-facial-digital syndrome | 1265 | 81405, 81406 | 4 weeks | $1,000 |
| **Pancreatic Agenesis** | | | | | |
| PDX1 Mutation Analysis | Pancreatic Agenesis | 2152 | 81403, 81404 | 4 weeks | $1,000 |
| GATA6 Mutation Analysis | Pancreatic Agenesis and Congenital Heart Defects | 2154 | 81404, 81405 | 4 weeks | $1,000 |
| PTF1A Mutation Analysis | Pancreatic and Cerebellar Agenesis | 2156 | 81403, 81404 | 4 weeks | $1,000 |
| **Pigmented hypertrichotic dermatosis with insulin-dependent diabetes mellitus testing** | | | | | |
| SLC29A3 Mutation Analysis | | 2158 | 81404, 81405 | 4 weeks | $1,000 |
| **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 | 81403 | 4 weeks | $450 |
| MAGEL2 Mutation Analysis | Shaaf-Yang syndrome | 1272 | 81403 | 4 weeks | $1,000 |
| **Premature Ovarian Failure testing** | | | | | |
| Premature Ovarian Failure Panel** | | 2160 | 81406, 81407 | 8 weeks | $2,000 |
| **Renal Cystic Disorders testing** | | | | | |
| Renal Cystic Disorders Panel** | | 1311 | 81406, 81407 | 8 weeks | $2,000 |
| **Rett/Atypical Rett syndrome testing** | | | | | |
| Rett/Atypical Rett Syndrome Panel** | | 1273 | 81407 | 4 weeks | $2,500 |
| Rett/Angelman Syndrome Panel** | | 1112 | 81406, 81407 | 8 weeks | $2,500 |
| **Rickets testing** | | | | | |
| Rickets Panel** | | 2173 | 81406, 81407 | 4 weeks | $2,500 |

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| **Roberts syndrome testing** | | | | | |
| ESCO2 Mutation Analysis | | 1282 | 81404, 81405 | 4 weeks | $1,000 |
| **Robinow syndrome testing** | | | | | |
| Robinow Syndrome Panel** | | 1319 | 81406, 81407 | 8 weeks | $2,500 |
| **Rubinstein-Taybi syndrome testing** | | | | | |
| Rubenstein-Taybi Syndrome Panel** | | 1320 | 81406, 81407 | 8 weeks | $2,500 |
| **Schinzel-Giedion syndrome testing** | | | | | |
| SETBP1 Mutation Analysis | | 1292 | 81403, 81404 | 4 weeks | $1,000 |
| **SHORT syndrome testing** | | | | | |
| PIK3R1 Mutation Analysis | | 1294 | 81406 | 4 weeks | $1,000 |
| **Sotos syndrome testing** | | | | | |
| NSD1 Mutation Analysis | | 1317 | 81405, 81406 | 4 weeks | $1,000 |
| **Thrombocytopenia testing** | | | | | |
| **Thrombocytopenia Panel**** | | 5106 | 81406, 81407 | 6 weeks | $4,000 |
| **Temple-Baraitser syndrome testing** | | | | | |
| KCNH1 Mutation Analysis | | 1146 | 81406 | 4 weeks | $1,000 |
| **Thiamine Responsive Megaloblastic Anemia syndrome testing** | | | | | |
| SLC19A2 Mutation Analysis | | 2161 | 81404, 81405 | 4 weeks | $1,000 |
| **Thyroid Disorders testing** | | | | | |
| Congenital Hypothyroidism Panel** | | 2170 | 81406, 81407 | 4 weeks | $2,500 |
| Hyperparathyroidism Panel** | | 2171 | 81406, 81407 | 4 weeks | $2,500 |
| Hypoparathyroidism Panel** | | 2172 | 81406, 81407 | 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 | 81405, 81406 | 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 Mutation Analysis | Crigler-Najjar syndrome | 1297 | 81403, 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 | 81407 | 8 weeks | $2,500 |
| **Weaver syndrome testing** | | | | | |
| EZH2 Mutation Analysis | | 1318 | 81405, 81406 | 4 weeks | $1,000 |
| **Wiedemann-Steiner syndrome testing** | | | | | |
| KMT2A (MLL) Mutation Analysis | Wiedemann-Steiner syndrome | 1305 | 81406, 81407 | 4 weeks | $1,000 |
| **Wilson Disease testing** | | | | | |
| ATP7B Mutation Analysis | Wilson disease | 1307 | 81405, 81406 | 4 weeks | $1,000 |
| **Wolcott-Rallison syndrome** | | | | | |
| EIF2AK3 Mutation Analysis | Wolcott-Rallison syndrome | 2165 | 81405, 81406 | 4 weeks | $1,000 |
| **Wolfram syndrome testing** | | | | | |
| Wolfram Syndrome Panel** | | 2167 | 81405, 81406 | 4 weeks | $1,500 |
| **Woodhouse-Sakati syndrome testing** | | | | | |
| DCAF17 Mutation Analysis | | 1309 | 81405, 81406 | 4 weeks | $1,000 |
| **Targeted Testing** | | | | | |
| Targeted mutation analysis (known familial mutation) | | 7102 | 81403 | 3 weeks | $500 |
| Testing for a known familial mutation by deletion/duplication analysis (MLPA) | | gene specific | 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 | 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** | | | | | |
| 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 options only. Please contact us for prices for patient self-pay and 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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