



## Molecular Diagnostic Laboratory

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	2019 CPT Code	2019 Price	2019 CPT Known Familial Variant	2019 Price Known Familial Variant	Sample Requirements: Whole Blood - EDTA tube
Acrodysostosis 1 ( <i>PRKAR1A</i> )	81479	\$900	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Allan-Herndon-Dudley ( <i>SLC16A2</i> )	81405	\$800	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
APOL1 Genotyping ( <i>APOL1</i> )	81479	\$225	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Autosomal Dominant Leukodystrophy ( <i>LMNB1</i> ) del/dup	81479	\$350	81479	<b>\$350</b>	2 - 4 cc in one or two tubes
Autosomal Dominant Leukodystrophy ( <i>LMNB1</i> ) upstream del	81479	\$450	81479	<b>\$350</b>	2 - 4 cc in one or two tubes
Autosomal dominant torsion dystonia type 4; DYT4 ( <i>TUBB4A</i> )	81479	\$225	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Barth Syndrome ( <i>TAZ</i> )	81406	\$650	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Benign Hereditary Chorea ( <i>NKX2.1</i> )	81479	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Calcium Homeostasis Disorders ( <i>CASR</i> )	81405	\$950	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
CFC Syndrome ( <i>BRAF</i> )	81479	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
CFC Syndrome ( <i>MAP2K1; MAP2K2</i> )	81479 x 2	\$525	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
CFC Syndrome/ Noonan Syndrome ( <i>KRAS</i> )	81405	\$425	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Congenital Nongoitrous Hypothyroidism ( <i>PAX8</i> )	81479	\$1,025	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Costello Syndrome (ex2/3) ( <i>HRAS</i> )	81403	\$225	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Costello Syndrome (full) ( <i>HRAS</i> )	81404	\$500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Deafness & Myopia Syndrome ( <i>SLITRK6</i> )	81479	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
<i>DFNB59</i> Related Nonsyndromic Deafness ( <i>DFNB59</i> )	81405	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Duchenne/Becker Muscular Dystrophy( <i>DMD</i> )	81161	\$525			2 - 4 cc in one or two tubes
Emery-Dreifuss Muscular Dystrophy ( <i>EMD</i> )	81405	\$500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Escobar Syndrome ( <i>CHRNA7</i> )	81479	\$700	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Familial Hypercholesterolemia Tier 1 (common mutations <i>LDLR, APOB, PCSK9</i> )	81401, 81406, 81479	\$370	81479	<b>\$225</b>	2 - 4 cc in one or two tubes

Familial Hypercholesterolemia Tier 2 (additional exons <i>LDLR</i> , <i>APOB</i> , <i>PCSK9</i> , & <i>del/dup selected LDLR &amp; PCSK9 exons</i> )	81401   81405 81406 81479x3	\$1,050	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Familial Hypercholesterolemia full sequencing and <i>del/dup</i> ( <i>LDLR</i> , <i>APOB</i> , <i>PCSK9</i> )	81401   81405  81406 x2   81479	\$1,420	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Familial Isolated Hypoparathyroidism ( <i>GCM2</i> )	81479	\$500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Fatal Infantile Cardioencephalomyopathy ( <i>SCO2</i> )	81404	\$225	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Feingold Syndrome 1 ( <i>MYCN</i> )	81479	\$550	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Feingold Syndrome 2 ( <i>MIR17HG</i> )	81479	\$250	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Glycogen Storage disease Type V ( <i>PYGM ex1 and 5 only</i> )	81401	\$250	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Glycogen Storage disease Type V ( <i>PYGM seq</i> )	81406	\$750	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Hypomyelinating Leukodystrophy 9 ( <i>RARS</i> )	81479	\$900	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Hypomyelinating Leukodystrophy 11 ( <i>POLR1C</i> )	81479	\$350	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Hypomyelinating leukodystrophy w/ atrophy of basal ganglia, cerebellum ; H-ABC ( <i>TUBB4A</i> )	81479	\$500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Hypomyelination and Congenital Cataract ( <i>FAM126A</i> )	81479	\$1,180	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Infantile Hypercalcemia ( <i>CYP24A1 tier 1</i> )	81479	\$350	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Infantile Hypercalcemia ( <i>CYP24A1 tier 2</i> )	81479	\$450	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Infantile Hypercalcemia ( <i>CYP24A1 full</i> )	81479	\$800	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) ( <i>DARS2 tier 1</i> )	81479	\$300	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) ( <i>DARS2 tier 2</i> )	81479	\$725	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) ( <i>DARS2 full gene sequencing</i> )	81479	\$925	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
LIG4 Related Disorders ( <i>LIG4</i> )	81479	\$350	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Megalencephalic Leukoencephalopathy w/ Subcort cysts ( <i>MLC1</i> )	81479	\$750	81479	<b>\$225</b>	2 - 4 cc in one or two tubes

Megalencephalic Leukoencephalopathy w/ Subcort cysts ( <i>HEPACAM</i> )	81479	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Meier-Gorlin Syndrome 1 ( <i>ORC1</i> )	81479	\$925	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Meier-Gorlin Syndrome 2 ( <i>ORC4</i> )	81479	\$775	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Meier-Gorlin Syndrome 3 ( <i>ORC6</i> )	81479	\$500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Meier-Gorlin Syndrome 4 ( <i>CDT1</i> )	81479	\$550	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Meier-Gorlin Syndrome 5 ( <i>CDC6</i> )	81479	\$725	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Metatropic Dysplasia /Brachyomia/ Spondylometaphyseal dysplasia ( <i>TRPV4</i> )	81479	\$1,450	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Microcephalic Osteodysplastic Primordial Dwarfism type I ( <i>RNU4ATAC</i> )	81479	\$225	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Microcephalic Osteodysplastic Primordial Dwarfism type II ( <i>PCNT2</i> )	81479	\$2,200	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Nonsyndromic Hearing Loss/Deafness ( <i>GJB2</i> )	81252	\$300	81253	<b>\$225</b>	2 - 4 cc in one or two tubes
Nonsyndromic Hearing Loss/Deafness ( <i>GJB6 del + seq</i> )	81254	\$300	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>PTPN11</i> )	81406	\$1,400	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>SOS1</i> )	81406	\$1,500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>RAF1</i> )	81404	\$250	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>KRAS</i> )	81405	\$350	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>SHOC2</i> )	81400	\$200	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>BRAF</i> )	81406	\$600	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Noonan Syndrome ( <i>MAP2K1</i> )	81479	\$250	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Pelizaeus-Merzbacher Disease ( <i>PLP1 Duplication</i> )	81404	\$375	81479	<b>\$375</b>	2 - 4 cc in one or two tubes
PMD & Spastic Paraplegia 2 Sequencing ( <i>PLP1 seq</i> )	81405	\$775	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Pelizaeus-Merzbacher-Like Disease ( <i>GJC2</i> )	81479	\$575	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Pendred Syndrome & DFNB4 ( <i>SLC26A4</i> )	81406	\$1,550	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
POL III leukodystrophies ( <i>POLR3A</i> )	81479	\$1,700	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
POL III leukodystrophies ( <i>POLR3B</i> )	81479	\$1,700	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Renal Hypouricemia Type 1 ( <i>SLC22A12</i> )	81479	\$559	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Renal Hypouricemia Type 2 ( <i>SLC2A9 sequencing</i> )	81479	\$800	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Renal Hypouricemia Type 2 ( <i>SLC2A9 dosage</i> )	81479	\$350	81479	<b>\$225</b>	2 - 4 cc in one or two tubes

Rett Syndrome ( <i>MECP2</i> seq)	81302	\$685	81303	<b>\$225</b>	2 - 4 cc in one or two tubes
Rett Syndrome ( <i>MECP2</i> deletion/duplication)	81304	\$300	81303	<b>\$225</b>	2 - 4 cc in one or two tubes
Smith-McCort dysplasia; SMC ( <i>RAB33B</i> )	81479	\$250	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Spinal Muscular Atrophy ( <i>SMN</i> exon 7 deletion)	81329	\$425	81329	<b>\$425</b>	2 - 4 cc in one or two tubes
Spinal Muscular Atrophy ( <i>SMN1/SMN2</i> dosage)	81329	\$475	81329	<b>\$475</b>	2 - 4 cc in one or two tubes
Spinal Muscular Atrophy ( <i>SMN</i> sequencing)	81336	\$900	81337	<b>\$225</b>	2 - 4 cc in one or two tubes
SMA w/ Respiratory Distress; SMARD ( <i>IGHMBP2</i> )	81479	\$1,500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Spondyloocular Syndrome; SOS ( <i>XYLT2</i> )	81479	\$700	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
TARP Syndrome ( <i>RBM10</i> )	81479	\$1,500	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
Timothy Syndrome ( <i>CACNA1C</i> exons 8, 8a only)	81479	\$250	81281	<b>\$225</b>	2 - 4 cc in one or two tubes
<i>TRPV4</i> -Related Neuropathies ( <i>TRPV4</i> seq)	81479	\$1,450	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
<i>TRPV4</i> -Related Neuropathies ( <i>TRPV4</i> only exons 3, 5 and 6)	81479	\$325	81479	<b>\$225</b>	2 - 4 cc in one or two tubes
DNA Extraction only	81479	\$50			2 - 4 cc in one or two tubes
<i>*updated January 2019</i>					