NEMOURS CHILDREN'S HEALTH

Molecular Diagnostics Laboratory
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CLIA # 08D0706140

	2023 CPT Code	2023 Price	2023 CPT Known Familial Variant	2023 Price Known Familial Variant	Sample Requirements: Whole Blood - EDTA tube
Acrodysostosis 1 (*PRKAR1A*)	81479	$900	81479	$225	2ml - 4ml
Allan-Herndon-Dudley (*SLC16A2*)	81405	$800	81479	$225	2ml - 4ml
APOL1 Genotyping (*APOL1*)	81479	$225	81479	$225	2ml - 4ml
Autosomal Dominant Leukodystrophy (*LMNB1*) del/dup	81479	$350	81479	$350	2ml - 4ml
Autosomal Dominant Leukodystrophy (*LMNB1*) upstream del	81479	$450	81479	$350	2ml - 4ml
Autosomal dominant torsion dystonia type 4; DYT4 (*TUBB4A*)	81479	$225	81479	$225	2ml - 4ml
Barth Syndrome (*TAZ*)	81406	$650	81479	$225	2ml - 4ml
Benign Hereditary Chorea (*NKX2.1*)	81479	$600	81479	$225	2ml - 4ml
Calcium Homeostasis Disorders (*CASR*)	81405	$950	81479	$225	2ml - 4ml
CFC Syndrome (*BRAF*)	81479	$600	81479	$225	2ml - 4ml
CFC Syndrome (*MAP2K1; MAP2K2*)	81479 x 2	$525	81479	$225	2ml - 4ml
CFC Syndrome/ Noonan Syndrome (*KRAS*)	81405	$425	81479	$225	2ml - 4ml
Congenital Nongoitrous Hypothyroidism (*PAX8*)	81479	$1,025	81479	$225	2ml - 4ml
Costello Syndrome (ex2/3) (*HRAS*)	81403	$225	81479	$225	2ml - 4ml
Costello Syndrome (full) (*HRAS*)	81404	$500	81479	$225	2ml - 4ml
Deafness & Myopia Syndrome (*SLITRK6*)	81479	$600	81479	$225	2ml - 4ml
DFNB59 Related Nonsyndromic Deafness (*DFNB59)*	81405	$600	81479	$225	2ml - 4ml
Duchenne/Becker Muscular Dystrophy (*DMD*)	81161	$525			2ml - 4ml
Emery-Dreifuss Muscular Dystrophy (*EMD*)	81405	$500	81479	$225	2ml - 4ml
Escobar Syndrome (*CHRNG*)	81479	$700	81479	$225	2ml - 4ml
Familial Hypercholestrolemia Tier 1 (common mutations *LDLR, APOB, PCSK9*)	81401 | 81406 | 81479	$370	81479	$225	2ml - 4ml
Familial Hypercholestrolemia Tier 2 (additional exons *LDLR, APOB, PCSK9, & del/dup selected LDLR & PCSK9 exons)*	81401 | 81405 81406 81479x3	$1,050	81479	$225	2ml - 4ml
Familial Hypercholestrolemia full sequencing and del/dup (*LDLR, APOB, PCSK9*)	81401 | 81405 | 81406 x2 | 81479	$1,420	81479	$225	2ml - 4ml
Familial Isolated Hypoparathyroidism (*GCM2*)	81479	$500	81479	$225	2ml - 4ml
Fatal Infantile Cardioencephalomyopathy (*SCO2*)	81404	$225	81479	$225	2ml - 4ml
Feingold Syndrome 1 (*MYCN*)	81479	$550	81479	$225	2ml - 4ml
Feingold Syndrome 2 (*MIR17HG*)	81479	$300	81479	$225	2ml - 4ml
GJB2 related disorders	81252	$300	81253	$225	2ml - 4ml

GJB6 related disorders del + seq	81254	$300	81479	$225	2ml - 4ml
Glycogen Storage disease Type V (*PYGM ex1 and 5 only*)	81401	$250	81479	$225	2ml - 4ml
Glycogen Storage disease Type V (*PYGM seq*)	81406	$750	81479	$225	2ml - 4ml
Hypomyelination and Congenital Cataract, HLD5 (*FAM126A*)	81479	$1,180	81479	$225	2ml - 4ml
Hypomyelinating leukodystrophy w/ atrophy of basal ganglia,cerebellum ; H-ABC; HLD6 (*TUBB4A*)	81479	$500	81479	$225	2ml - 4ml
Hypomyelinating Leukodystrophy 9 (*RARS*)	81479	$900	81479	$225	2ml - 4ml
Hypomyelinating Leukodystrophy 11 (*POLR1C*)	81479	$350	81479	$225	2ml - 4ml
Infantile Hypercalcemia (*CYP24A1* tier 1)	81479	$350	81479	$225	2ml - 4ml
Infantile Hypercalcemia (*CYP24A1* tier 2)	81479	$450	81479	$225	2ml - 4ml
Infantile Hypercalcemia (*CYP24A1* full)	81479	$800	81479	$225	2ml - 4ml
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* tier 1)	81479	$300	81479	$225	2ml - 4ml
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* tier 2)	81479	$725	81479	$225	2ml - 4ml
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* full gene sequencing)	81479	$1,025	81479	$225	2ml - 4ml
LIG4 Related Disorders (*LIG4*)	81479	$350	81479	$225	2ml - 4ml
Megalencephalic Leukoencephalopathy w/ Subcort cysts (*MLC1*)	81479	$750	81479	$225	2ml - 4ml
Megalencephalic Leukoencephalopathy w/ Subcort cysts (*HEPACAM*)	81479	$600	81479	$225	2ml - 4ml
Meier-Gorlin Syndrome 1 (*ORC1*)	81479	$925	81479	$225	2ml - 4ml
Meier-Gorlin Syndrome 2 (*ORC4*)	81479	$775	81479	$225	2ml - 4ml
Meier-Gorlin Syndrome 3 (*ORC6*)	81479	$500	81479	$225	2ml - 4ml
Meier-Gorlin Syndrome 4 (*CDT1*)	81479	$550	81479	$225	2ml - 4ml
Meier-Gorlin Syndrome 5 (*CDC6*)	81479	$725	81479	$225	2ml - 4ml
Metatropic Dysplasia /Brachyomia/ Spondylometaphyseal dysplasia (*TRPV4*)	81479	$1,450	81479	$225	2ml - 4ml
Microcephalic Osteodysplastic Primordial Dwarfism type I (*RNU4ATAC*)	81479	$225	81479	$225	2ml - 4ml
Microcephalic Osteodysplastic Primordial Dwarfism type II (*PCNT2*)	81479	$2,200	81479	$225	2ml - 4ml
Noonan Syndrome (*PTPN11*)	81406	$1,400	81479	$225	2ml - 4ml
Noonan Syndrome (*SOS1*)	81406	$1,500	81479	$225	2ml - 4ml
Noonan Syndrome (*RAF1*)	81404	$250	81479	$225	2ml - 4ml
Noonan Syndrome (*KRAS*)	81405	$425	81479	$225	2ml - 4ml
Noonan Syndrome (*SHOC2*)	81400	$225	81479	$225	2ml - 4ml
Noonan Syndrome (BRAF)	81406	$600	81479	$225	2ml - 4ml

Noonan Syndrome (*MAP2K1*)	81479	$250	81479	$225	2ml - 4ml
Pelizaeus-Merzbacher Disease (*PLP1 Deletion/Duplication*)	81404	$375	81479	$375	2ml - 4ml
PMD & Spastic Paraplegia 2 Sequencing (*PLP1* seq)	81405	$775	81479	$225	2ml - 4ml
Pelizaeus-Merzbacher-Like Disease (*GJC2*)	81479	$575	81479	$225	2ml - 4ml
Pendred Syndrome & DFNB4 (*SLC26A4*)	81406	$1,550	81479	$225	2ml - 4ml
POL III leukodystrophies (*POLR3A*)	81479	$1,700	81479	$225	2ml - 4ml
POL III leukodystrophies (*POLR3B*)	81479	$1,700	81479	$225	2ml - 4ml
Renal Hypouricemia Type 1 (*SLC22A12*)	81479	$550	81479	$225	2ml - 4ml
Renal Hypouricemia Type 2 (*SLC2A9 sequencing*)	81479	$800	81479	$225	2ml - 4ml
Renal Hypouricemia Type 2 (*SLC2A9 dosage*)	81479	$350	81479	$350	2ml - 4ml
Rett Syndrome (*MECP2* seq)	81302	$685	81303	$225	2ml - 4ml
Rett Syndrome (*MECP2* deletion/duplication)	81304	$300	81303	$300	2ml - 4ml
Smith-McCort dysplasia; SMC (*RAB33B*)	81479	$250	81479	$225	2ml - 4ml
Spinal Muscular Atrophy (*SMN* exon 7 deletion)	81329	$425	81329	$425	2ml - 4ml
Spinal Muscular Atrophy (*SMN1/SMN2* dosage)	81329	$475	81329	$475	2ml - 4ml
Spinal Muscular Atrophy (*SMN* sequencing)	81336	$900	81337	$225	2ml - 4ml
SMA w/ Respiratory Distress; SMARD (*IGHMBP2*)	81479	$1,500	81479	$225	2ml - 4ml
Spondyloocular Syndrome; SOS (*XYLT2)*	81479	$700	81479	$225	2ml - 4ml
TARP Syndrome (*RBM10*)	81479	$1,500	81479	$225	2ml - 4ml
Timothy Syndrome (*CACNA1C* exons 8, 8a only)	81479	$250	81479	$225	2ml - 4ml
TRPV4 -Related Neuropathies *(TRPV4* full seq)	81479	$1,450	81479	$225	2ml - 4ml
TRPV4 -Related Neuropathies *(TRPV4* only exons 3, 5 and 6)	81479	$325	81479	$225	2ml - 4ml
TRPV4 Related Skeletal Dysplasias (*TRPV4*)	81479	$1,450	81479	$225	2ml - 4ml
DNA Extraction and Banking DNA ***only*** (no testing)	81479	$100			2ml - 4ml
updated January 2023					

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