Molecular Diagnostic Laboratory
1600 Rockland Road, Wilmington, DE 19803
302.651.6775 email: MDL@nemours.org

	2016 CPT Code	2016 Price	2016 CPT Known Familial Variant	2016 Price Known Familial Variant	Sample Requirements: Whole Blood in EDTA tube
Acrodysostosis 1 (*PRKAR1A*)	81479	$900	81479	$200	2-4 cc in one or two tubes
Allan-Herndon-Dudley (*SLC16A2*)	81405	$800	81403	$200	2-4 cc in one or two tubes
APOL1 Genotyping (*APOL1*)	81479	$200	81479	$200	2-4 cc in one or two tubes
Autosomal Dominant Leukodystrophy (*LMNB1*)	81479	$350	81479	$200	2-4 cc in one or two tubes
Autosomal dominant torsion dystonia type 4; DYT4 (*TUBB4A*)	81479	$200	81479	$200	2-4 cc in one or two tubes
Barth Syndrome (*TAZ*)	81406	$650	81403	$185	2-4 cc in one or two tubes
Benign Hereditary Chorea (*NKX2.1*)	81479	$600	81479	$200	2-4 cc in one or two tubes
Calcium Homeostasis Disorders (*CASR*)	81405	$950	81403	$200	2-4 cc in one or two tubes
CFC Syndrome (*BRAF*)	81479	$600	81403	$200	2-4 cc in one or two tubes
CFC Syndrome (*MAP2K1; MAP2K2*)	81479 x 2	$525	81403	$200	2-4 cc in one or two tubes
CFC Syndrome/ Noonan Syndrome (*KRAS*)	81405	$425	81403	$200	2-4 cc in one or two tubes
Congenital Nongoitrous Hypothyroidism 2 (*PAX8*)	81479	$1,025	81479	$200	2-4 cc in one or two tubes
Costello Syndrome (ex2/3) (*HRAS*)	81479	$200	81403	$200	2-4 cc in one or two tubes
Costello Syndrome (full) (*HRAS*)	81404	$500	81403	$200	2-4 cc in one or two tubes
Deafness & Myopia Syndrome (*SLITRK6*)	81479	$600	81479	$200	2-4 cc in one or two tubes
DFNB59 Related Nonsyndromic Deafness (DFNB59)	81405	$600	81403	$200	2-4 cc in one or two tubes
Duchenne/Becker Muscular Dystrophy(*DMD*)	81161	$525	81403	$200	2-4 cc in one or two tubes
Emery-Dreifuss Muscular Dystrophy (*EMD*)	81405	$500	81403	$200	2-4 cc in one or two tubes
Escobar Syndrome (*CHRNG*)	81479	$700	81479	$200	2-4 cc in one or two tubes
Familial Hypercholestrolemia Tier 1 (common mutations *LDLR, APOB, PCSK9*)	81401, 81479x2	$370	81403	$200	2-4 cc in one or two tubes
Familial Hypercholestrolemia Tier 2 (additional exons *LDLR, APOB, PCSK9, & del/dup selected LDLR & PCSK9 exons)*	81401 | 81479x3 | 81405	$1,050	81403	$200	2-4 cc in one or two tubes
Familial Hypercholestrolemia full sequencing and del/dup (LDLR, APOB, PCSK9)	81401 | 81405| 81406 x2 | 81479	$1,420	81403		2-4 cc in one or two tubes
Familial Isolated Hypoparathyroidism (*GCM2*)	81479	$500	81479	$200	2-4 cc in one or two tubes
Fatal Infantile Cardioencephalomyopathy (*SCO2*)	81404	$200	81403	$200	2-4 cc in one or two tubes

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Feingold Syndrome 1 (*MYCN*)	81479	$550	81479	$200	2-4 cc in one or two tubes
Feingold Syndrome 2 (*MIR17HG*)	81479	$200	81479	$200	2-4 cc in one or two tubes
Glycogen Storage disease Type V (*PYGM ex1 and 5 only*)	81401	$250	81403	$200	2-4 cc in one or two tubes
Glycogen Storage disease Type V (*PYGM seq*)	81406	$750	81403	$200	2-4 cc in one or two tubes
Hypomyelinating Leukodystrophy 9 (*RARS*)	81479	$900	81479	$200	2-4 cc in one or two tubes
Hypomyelinating leukodystrophy w/ atrophy of basal ganglia,cerebellum ; H-ABC (*TUBB4A*)	81479	$500	81479	$200	2-4 cc in one or two tubes
Hypomyelination and Congenital Cataract (*HCC*)	81479	$1,180	81479	$200	2-4 cc in one or two tubes
Infantile Hypercalcemia (*CYP24A1* tier 1)	81479	$350	81479	$200	2-4 cc in one or two tubes
Infantile Hypercalcemia (CYP24A1 tier 2)	81479	$450	81479	$200	2-4 cc in one or two tubes
Infantile Hypercalcemia (CYP24A1 full)	81479	$800	81479	$200	2-4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* tier 1)	81479	$200	81479	$200	2-4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* tier 2)	81479	$725	81479	$200	2-4 cc in one or two tubes
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement (LBSL) (*DARS2* full gene sequencing)	81479	$925	81479	$200	2-4 cc in one or two tubes
LIG4 Related Disorders (*LIG4*)	81479	$350	81479	$200	2-4 cc in one or two tubes
Megalencephalic Leukoencephalopathy w/ Subcort cysts (*MLC1*)	81479	$750	81479	$200	2-4 cc in one or two tubes
Megalencephalic Leukoencephalopathy w/ Subcort cysts (*HEPACAM*)	81479	$600	81479	$200	2-4 cc in one or two tubes
Meier-Gorlin Syndrome 1 (*ORC1*)	81479	$925	81479	$200	2-4 cc in one or two tubes
Meier-Gorlin Syndrome 2 (*ORC4*)	81479	$775	81479	$200	2-4 cc in one or two tubes
Meier-Gorlin Syndrome 3 (*ORC6*)	81479	$500	81479	$200	2-4 cc in one or two tubes
Meier-Gorlin Syndrome 4 (*CDT1*)	81479	$550	81479	$200	2-4 cc in one or two tubes
Meier-Gorlin Syndrome 5 (*CDC6*)	81479	$725	81479	$200	2-4 cc in one or two tubes
Metatropic Dysplasia/Brachyomia/Spondylometaphyseal dysplasia (*TRPV4*)	81479	$1,450	81479	$200	2-4 cc in one or two tubes
Microcephalic Osteodysplastic Primordial Dwarfism type I (*RNU4ATAC*)	81479	$200	81479	$200	2-4 cc in one or two tubes
Microcephalic Osteodysplastic Primordial Dwarfism type II (*PCNT2*)	81479	$2,200	81479	$200	2-4 cc in one or two tubes
Nonsyndromic Hearing Loss/Deafness (*GJB2*)	81252	$300	81253	$200	2-4 cc in one or two tubes
Nonsyndromic Hearing Loss/Deafness (*GJB6 del + seq*)	81254	$300	81403	$200	2-4 cc in one or two tubes

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Noonan Syndrome (*PTPN11*)	81406	$1,400	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (*SOS1*)	81406	$1,500	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (*RAF1*)	81404	$250	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (*KRAS*)	81405	$350	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (*SHOC2*)	81400	$200	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (BRAF)	81406	$600	81403	$200	2-4 cc in one or two tubes
Noonan Syndrome (*MAP2K1*)	81479	$250	81403	$200	2-4 cc in one or two tubes
Pelizaeus-Merzbacher Disease (*PLP1 Duplication*)	81404	$375	81403	$200	2-4 cc in one or two tubes
PMD & Spastic Paraplegia 2 Sequencing (*PLP1* seq)	81405	$775	81403	$200	2-4 cc in one or two tubes
Pelizaeus-Merzbacher-Like Disease (*GJC2*)	81479	$575	81479	$200	2-4 cc in one or two tubes
Pendred Syndrome & DFNB4 (*SLC26A4*)	81406	$1,550	81403	$200	2-4 cc in one or two tubes
POL III leukodystrophies (*POLR3A*)	81479	$1,700	81479	$200	2-4 cc in one or two tubes
POL III leukodystrophies (*POLR3B*)	81479	$1,700	81479	$200	2-4 cc in one or two tubes
Renal Hypouricemia Type 1 (*SLC22A12*)	81479	$550	81479	$200	2-4 cc in one or two tubes
Renal Hypouricemia Type 2 (*SLC2A9 sequencing*)	81479	$800	81479	$200	2-4 cc in one or two tubes
Renal Hypouricemia Type 2 (*SLC2A9 dosage*)	81479	$350	81479	$200	2-4 cc in one or two tubes
Rett Syndrome (*MECP2* seq)	81302	$685	81303	$200	2-4 cc in one or two tubes
Rett Syndrome (*MECP2* deletion/duplication)	81304	$300	81303	$200	2-4 cc in one or two tubes
Smith-McCort dysplasia; SMC (*RAB33B*)	81479	$250	81479	$200	2-4 cc in one or two tubes
Spinal Muscular Atrophy (*SMN* exon 7 deletion)	81400	$425	81403	$200	2-4 cc in one or two tubes
Spinal Muscular Atrophy (*SMN1/SMN2* dosage)	81401	$475	81403	$200	2-4 cc in one or two tubes
Spinal Muscular Atrophy (*SMN* sequencing)	81405	$900	81403	$200	2-4 cc in one or two tubes
SMA w/ Respiratory Distress; SMARD (*IGHMBP2*)	81479	$1,500	81479	$200	2-4 cc in one or two tubes
TARP Syndrome (*RBM10*)	81479	$1,500	81479	$200	2-4 cc in one or two tubes
Timothy Syndrome (*CACNA1C* exons 8, 8a only)	81479	$250	81281	$200	2-4 cc in one or two tubes
TRPV4-Related Neuropathies (*TRPV4* seq)	81479	$1,450	81479	$200	2-4 cc in one or two tubes
TRPV4-Related Neuropathies (*TRPV4* only ex.5&6)	81479	$200	81479	$200	2-4 cc in one or two tubes
DNA Extraction	81479	$25			2-4 cc in one or two tubes
**updated Dec 2015*					