

DNA Diagnostic Laboratory at Johns Hopkins

Test, Fee and CPT Code List

*If targeted testing for a known mutation or prenatal testing is desired, please contact the lab for fees and CPT codes.

Includes Next Generation Sequencing (NGS) panels

Test / Gene Name	2016 CPT Full Test	Full Test Fee	
Acatlasemia (NGS; sequence and del/dup)	81479	\$2,450.00	
Alpha Methylacyl-CoA Racemase Deficiency (NGS; sequence and del/dup)	81479	\$2,450.00	
Achondroplasia/Hypochondroplasia FGFR3 panel	81404	\$542.00	
Achondroplasia/Thanatophoric Dysplasia FGFR3 panel	81404	\$662.00	
Antley-Bixler Like Syndrome FGFR2 panel	81404	\$422.00	
Apert Syndrome FGFR2 panel	81479	\$302.00	
Ataxia Telangiectasia (ATM; NGS; sequence and del/dup)	81408	\$2,750.00	
Campomelic Dysplasia SOX9 sequence	81479	\$902.00	
CF Linkage Analysis	81479	\$344.00	
CFTR Intron 8 T/TG tract typing	81224	\$302.00	
Cystic Fibrosis (CFTR; NGS; sequencing and del/dup)	81223; 81222	\$2,450.00	
Cystic Fibrosis and CF-Related Disorders NGS Panel (Includes CFTR) (sequencing and del/dup)	81223; 81222; 81406 x 3; 81479	\$2,750.00	
Cystic Fibrosis-Related Disorders NGS Panel (Excludes CFTR) (sequencing and del/dup)	81406 x 3; 81479	\$2,600.00	
Childhood Interstitial Lung Disease NGS Panel (sequencing and del/dup)	81479 x 3	\$3,252.00	
Coronal Craniosynostosis FGFR2, FGFR3 panel	81404; 81403	\$542.00	
Craniofrontonasal Syndrome EFNB1 sequence	81479	\$1,022.00	
Crouzon Syndrome FGFR2, FGFR3 panel	81404; 81403	\$542.00	
Crouzon Syndrome with Acanthosis Nigricans FGFR3 panel	81403	\$302.00	
Diffuse Lung Disease Comprehensive NGS Panel (sequencing and del/dup)	81479 x 14	\$4,634.00	
Huntington Disease	81401	\$355.00	
Huntington Disease-Like HDL2	81479	\$437.00	
Idiopathic Pulmonary Fibrosis NGS Panel (sequencing and del/dup)	81479 x 9	\$3,807.00	
Liddle Syndrome SCNN1B and SCNN1G panel	81479	\$662.00	
Loeys-Dietz 1C, SMAD3	81479	\$994.00	
Loeys-Dietz Syndrome TGFBR1 sequence	81405	\$773.00	* This is a reflexed test - see web site for details
Loeys-Dietz Syndrome TGFBR2 sequence	81405	\$937.00	* This is a reflexed test - see web site for details
Marfan Syndrome Type 2 TGFBR2 sequence	81405	\$937.00	
Mulibrey Nanism (NGS)	81479	\$2,450.00	
Neonatal Respiratory Distress NGS Panel (sequencing and del/dup)	81479 x 5	\$3,250.00	
NKX2-1 Gene sequence	81479	\$882.00	* This is a reflexed test - see web site for details
NKX2-1 MLPA	81479	\$429.00	* This is a reflexed test - see web site for details
Oculodentodigital Dysplasia GJA1 sequence	81479	\$662.00	

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Test / Gene Name	2016 CPT Full Test	Full Test Fee	
Peroxisomal β -Oxidation Defects NGS Panel (sequencing and del/dup)	81479 x 3	\$2,601.00	
Peroxisomal Genes NGS Panel, Comprehensive (sequencing and del/dup)	81479 x 24	\$4,104.00	
Primary Hyperoxaluria (NGS; sequencing and del/dup)	81479	\$2,450.00	
Pfeiffer Syndrome FGFR2, FGFR3, FGFR1 panel	81404; 81403; 81479	\$662.00	
Pseudohypoadosteronism Type 1 NGS Panel (sequencing and del/dup)	81406 x 3; 81479	\$2,600.00	
Pseudohypoparathyroidism 1A: GNAS methylation	81479	\$482.00	* This is a reflexed test - see web site for details
Pseudohypoparathyroidism 1A: GNAS sequence	81479	\$1,478.00	* This is a reflexed test - see web site for details
Pseudohypoparathyroidism 1B: GNAS methylation	81479	\$482.00	
Pulmonary Alveolar Proteinosis NGS Panel (sequencing and del/dup)	81479 x 8	\$3,528.00	
Refsum Disease NGS Panel (sequencing and del/dup)	81479 x 2	\$2,450.00	
Rhizomelic Chondrodysplasia Punctata NGS Panel (sequencing and del/dup)	81479	\$2,601.00	
SADDAN FGFR3 panel	81479	\$302.00	
Saethre-Chotzen Syndrome FGFR2, FGFR3, TWIST1 panel	81403; 81404; 81479	\$662.00	* This is a reflexed test - see web site for details
Saethre-Chotzen Syndrome TWIST MLPA	81479	\$429.00	* This is a reflexed test - see web site for details
Telomere Shortening Disorders Spectrum NGS Panel (sequencing and del/dup)	81479 x 8	\$2,752.00	
Treacher Collins Syndrome NGS Panel (sequencing and del/dup)	81479 x 3	\$2,601.00	
von Hippel-Lindau Syndrome VHL sequence	81404	\$542.00	* This is a reflexed test - see web site for details
von Hippel-Lindau Syndrome VHL MLPA	81403	\$429.00	* This is a reflexed test - see web site for details
X-Adrenoleukodystrophy ABCD1 MLPA	81479	\$541.00	* This is a reflexed test - see web site for details
X-Adrenoleukodystrophy ABCD1 sequence	81405	\$1,158.00	* This is a reflexed test - see web site for details
Zellweger Spectrum Disorder NGS Panel (sequencing and del/dup)	81479 x 13	\$3,211.00	* This is a reflexed test - see web site for details
Zellweger Spectrum Disorder + Peroxisomal β -Oxidation Defects NGS Panel (sequencing and del/dup)	81479 x 16	\$3,664.00	* This is a reflexed test - see web site for details