

	Name of Test	Turnaround Time	Cost	CPT codes
Beckwith-Wiedemann Syndrome	Methylation and high resolution copy number analysis of 11p15.5 with automatic reflex to CDKN1C if negative	4-6 weeks	\$1,950*	81401x2, 81479x2
	Methylation analysis of 11p15.5 with automatic reflex to CDKN1C if negative	4-6 weeks	\$1,200*	81401x2, 81479
	BWS: Methylation analysis of 11p15.5 only	3-4 weeks	\$600	81401x2
	BWS: 11p15.5 high resolution copy number analysis only (aCGH)	3-4 weeks	\$750	81479
	BWS: CDKN1C sequencing analysis only	3 weeks	\$600	81479
	BWS: Site specific analysis (familial)	2-3 weeks	\$600 (methylation) \$360 (CDKN1C)	81401x2 for methylation 81403
	Methylation analysis of 11p15.5 with automatic reflex to CDKN1C if negative (Prenatal)	1-3 weeks	\$1,300*	81401x2, 81479, 81265
	BWS: CDKN1C sequencing analysis only (Prenatal)	1-3 weeks	\$700	81479, 81265
	BWS: Site specific analysis (familial) (Prenatal)	1-3 weeks	\$700 (methylation) \$460 (CDKN1C)	81401x2, 81265 for methylation 81403, 81265 for CDKN1C
FOP	Fibrodysplasia Ossificans Progressiva (FOP)	3 weeks	\$420	81403
	Fibrodysplasia Ossificans Progressiva (FOP) (Prenatal)	1-3 weeks	\$520	81403, 81265
Hemophilia A	Hem A (Factor VIII): Inversion Analysis	3-4 weeks	\$360	81403
	HemA (Factor VIII): Full Sequencing	4-6 weeks	\$1,350	81407
	Hem A (Factor VIII): Deletion/Duplication	2-3 weeks	\$750	81406
	Hem A (Factor VIII): Site specific analysis (familial)	2-3 weeks	\$360	81403
	Hem A (Factor VIII): Site Specific - Inversion, Seq. or del/dup (Prenatal)	7-10 days	\$460	81403, 81265 for prenatal inversion 81403, 81265 for prenatal sequencing
	Hem A (Factor VIII): Linkage Analysis	3-4 weeks	\$740	81479
Hemophilia B	Hem B (Factor IX): Full Sequencing	4-6 weeks	\$840	81405
	Hem B: Promoter Analysis only for Factor IX Leyden	3-4 weeks	\$360	81404
	Hem B (Factor IX): Site specific analysis (familial)	2-3 weeks	\$360	81403
	Hem B (Factor IX): Site specific analysis (familial) (Prenatal)	7-10 days	\$460	81403, 81265

	Name of Test	Turnaround Time	Cost	CPT codes
Hereditary Hemorrhagic Telangiectasia	HHT: <i>ENG/ACRVL1</i> Sequencing with automatic reflex to Deletion/Duplication Analysis	6-8 weeks	\$2,050*	81406, 81479x2, 81405
	HHT: <i>ENG/ACRVL1</i> Deletion/Duplication Analysis	3-4 weeks	\$750	81479, 81405
	HHT: <i>SMAD4</i> Deletion/Duplication Analysis	3-4 weeks	\$750	81405
	HHT: <i>ENG/ACRVL1</i> Site specific analysis (familial)	2-3 weeks	\$360	81403
	HHT: <i>ENG/ACRVL1</i> Site specific analysis (familial) (Prenatal)	7-10 days	\$460	81403, 81265
	HHT: <i>SMAD4</i> Full Sequencing	8-10 weeks	\$600	81406
	HHT: <i>SMAD4</i> Site specific analysis (familial)	2-3 weeks	\$360	81403
	HHT: <i>SMAD4</i> Site specific analysis (familial) (Prenatal)	7-10 days	\$460	81403, 81265
Hyperinsulinism	Expedited Level 1 CHI panel: <i>ABCC8</i> , <i>KCNJ11</i> , <i>GLUD1</i> , <i>GCK</i>	7-10 days	\$2,800	81407, 81403, 81406x2
	Expanded Level 2 CHI Panel: <i>ABCC8</i> , <i>KCNJ11</i> , <i>GLUD1</i> , <i>GCK</i> , <i>SLC16A1</i> , <i>UCP2</i> , <i>HNF1A</i> , <i>HNF4A</i> , <i>HADH</i>	4-6 weeks	\$3,000	81479
	HI: <i>ABCC8</i> Sequencing	2-3 weeks	\$1,000	81407
	HI: <i>KCNJ11</i> Sequencing	2-3 weeks	\$600	81403
	HI: <i>GLUD1</i> Sequencing	2-3 weeks	\$700	81406
	HI: <i>GCK</i> Sequencing	2-3 weeks	\$700	81406
	HI: Deletion/Duplication analysis	3 weeks	\$750	81479
	HI: Site specific analysis (familial)	7-10 days	\$360	81403
HI: Site specific analysis (familial) (Prenatal)	1-3 weeks	\$460	81403, 81265	
IMAGe	IMAGe: <i>CDKN1C</i> sequencing	3 weeks	\$600	81479
	IMAGe: Site specific analysis (familial)	2-3 weeks	\$360	81403
	IMAGe: <i>CDKN1C</i> sequencing (Prenatal)	1-3 weeks	\$700 \$460	81479, 81265 (full sequencing) 81403, 81265 (site specific, known familial)

UNIVERSITY OF PENNSYLVANIA
GENETIC DIAGNOSTIC LABORATORY

*REFLEX TESTING OPTIONS: The maximum cost is listed, although our lab only bills for the services performed. Final charges may be less than what is listed.

	Name of Test	Turnaround Time	Cost	CPT codes
MODY	MODY Comprehensive Panel (Sequencing AND Deletion/Duplication analysis)	4-6 weeks	\$2,500	81403 x 2, 81404 x 2, 81405 x 2 81406 x 2, 81407 X 1, 81479 x 7
	MODY Sequencing analysis ONLY	4-6 weeks	\$1,750	81403 x 2, 81404 x 2, 81405 x 2 81406 x 2, 81407 X 1, 81479 x 6
	MODY Deletion/Duplication analysis ONLY	3-4 weeks	\$750	81479
	MODY: Site specific analysis (familial)	2-3 weeks	\$360	81403
	MODY: Site specific analysis (familial) (Prenatal)	1-3 weeks	\$460	81403, 81265
OVG	Somatic Overgrowth Panel (v2)	6-8 weeks	\$3,000	81479
	Somatic Overgrowth Panel (v2) (Prenatal)	2-4 weeks	\$3,100	81479, 81265
Retinoblastoma	Retinoblastoma: Sequence analysis with reflex to deletion/duplication [Blood]	6-8 weeks	\$2,070*	81479x2
	<u>Frozen</u> tumor comprehensive analysis with reflex to site specific testing on constitutional DNA	8-10 weeks	\$2,630	81479x2, 81401, 81402
	Retinoblastoma: <u>Paraffin-embedded</u> Tumor comprehensive analysis with reflex to site specific testing on constitutional DNA	8-12 weeks	\$2,740	81479x2, 81401, 81402, 88381
	Retinoblastoma: Deletion/duplication (blood)	4-6 weeks	\$750	81479
	Retinoblastoma: Site specific analysis (familial)	2-3 weeks	\$360	81403 Call for code for deletion/ duplication >5 exons of RB1
	Retinoblastoma: Site specific analysis (familial) (Prenatal)	7-10 days	\$460	81403, 81265 Call for code for deletion/ duplication >5 exons of RB1
	Retinoblastoma: MYCN analysis	2-3 weeks	\$360	81402
	Retinoblastoma: Methylation only (tumor &/or blood)	2-3 weeks	\$400	81401x2

	Name of Test	Turnaround Time	Cost	CPT codes
Russell- Silver Syndrome	Methylation and high resolution copy number analysis of 11p15.5; if negative, reflex to methylation analysis of GRB10/PEG1 genes	4-6 weeks	\$1,980*	81401x2, 81402, 81479
	Methylation and high resolution copy number analysis of 11p15.5 with automatic reflex to methylation analysis of GRB10/PEG1 genes if negative with automatic reflex to CDKN1C if negative	4-8 weeks	\$2,580*	81401x2, 81402, 81479x2
	Methylation analysis of 11p15.5 with automatic reflex to methylation analysis of GRB10/PEG1 genes if negative	4-6 weeks	\$1,230*	81401x2, 81402
	RSS: Methylation analysis of 11p15.5 only	3-4 weeks	\$600	81401x2
	RSS: 11p15.5 high resolution copy number analysis only (aCGH)	3-4 weeks	\$750	81479
	RSS: methylation analysis of GRB10/PEG1 genes only	3 weeks	\$630	81402
	RSS: Site specific analysis (familial)	2-3 weeks	\$600 (methylation) \$360 (CDKN1C)	81401x2 for methylation 81403 for CDKN1C familial mutation
	Methylation analysis of 11p15.5 with automatic reflex to methylation analysis of GRB10/PEG1 genes if negative (Prenatal)	1-3 weeks	\$1,330*	81401x2, 81402, 81265
	RSS: Site specific analysis (familial) (Prenatal)	2-3 weeks	\$700 (methylation) \$460 (CDKN1C)	81401x2, 81265 for methylation 81403, 81265 for CDKN1C familial mutation
Uveal melanoma	Uveal Melanoma: SNP-array analysis of Chromosomes 1p, 3, 6, and 8	8-10 weeks	\$1,000	81406
	BAP1 sequencing with a reflex to deletion/duplication analysis (Germline only)	4-6 weeks	\$1,550*	81479x2
	BAP1: Sequencing only	4-6 weeks	\$800	81479
	BAP1: Deletion/Duplication analysis only	3-4 weeks	\$750	81479
	BAP1: Site specific analysis (familial)	2-3 weeks	\$360	81403
X inactivation	X-Inactivation Studies: Individual	3-4 weeks	\$500	81401
	X-Inactivation Studies: Individual and Parent(s)	3-4 weeks	\$720	81401x3