

	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
	Hem B (Factor IX): Full Sequencing	4-6 weeks	\$840	81405
Hemophilia B	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
Heredity 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, KCNJ11, GLUD1, GCK</i>	7-10 days	\$2,800	81407, 81403, 81406x2
	Expanded Level 2 CHI Panel: <i>ABCC8, KCNJ11, GLUD1, GCK, SLC16A1, UCP2, HNF1A, HNF4A, 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)

	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 & Vascular Malformations Panel (v3)	4-6 weeks	\$3,000	81479
	Somatic Overgrowth & Vascular Malformations Panel (v3) (Prenatal)	2-4 weeks	\$3,100	81479, 81265
LYM	Lymphedema Panel (v1)	4-6 weeks	\$3,000	81479
	Lymphedema Panel (v1) (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