UNIVERSITY OF PENNSYLVANIA
GENETIC DIAGNOSTIC LABORATORY

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	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 for CDKN1C familial mutation
	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
Familial Adenomatous Polyposis	FAP: APC or MYH Site specific analysis (familial)	2-3 weeks	$360	81202 for APC known mutation 81401 for MYH known mutation
	FAP: APC or MYH Site specific analysis (familial) ***(Prenatal)***	7-10 days	$460	81202, 81265 for APC prenatal 81401, 81265 for MYH prenatal
FOP	Fibrodysplasia Ossificans Progressiva (FOP)	3 weeks	$420	81403
	Fibrodysplasia Ossificans Progressiva (FOP) ***(Prenatal)***	1-3 weeks	$520	81403, 81265
GNA	GNAQ/GNA11 Somatic analysis	4-5 weeks	$800	81401x2

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	Name of Test	Turnaround Time	Cost	CPT codes
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
Hereditary Hemorrhagic Telangiectasia	HHT: *ENG/ACRVL1* Sequencing with automatic reflex to Deletion/Duplication Analysis	6-8 weeks	$2,050*	81406, 81479x2, 81405
	HHT: *ENG/ACRVL1* Deletion/Duplication Analysis	3-4 weeks	$750	81479, 81405
	HHT: SMAD4 Deletion/Duplication Analysis	3-4 weeks	$750	81405
	HHT: *ENG/ACRVL1* Site specific analysis (familial)	2-3 weeks	$360	81403
	HHT: *ENG/ACRVL1* Site specific analysis (familial) ***(Prenatal)***	7-10 days	$460	81403, 81265
	HHT: *SMAD4* Full Sequencing	8-10 weeks	$600	81406
	HHT: *SMAD4* Site specific analysis (familial)	2-3 weeks	$360	81403
	HHT: *SMAD4* Site specific analysis (familial) ***(Prenatal)***	7-10 days	$460	81403, 81265

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Hyperinsulinism	Level 1 CHI panel: ABCC8, KCNJ11, GLUD1, GCK	7-10 days	$2,800	81407, 81403, 81406x2
	Level 2 CHI Panel: *ABCC8*, *KCNJ11*, *GLUD1*, *GCK*, *SLC16A1*, *UCP2*, *HNF1A*, *HNF4A*, *HADH*	4-6 weeks	$3,000	81479
	HI: ABCC8 Sequencing	2-3 weeks	$1,000	81407
	HI: KCNJ11 Sequencing	2-3 weeks	$600	81403
	HI: GLUD1 Sequencing	2-3 weeks	$700	81406
	HI: GCK 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: *CDKN1C* sequencing	3 weeks	$600	81479
	IMAGe: Site specific analysis (familial)	2-3 weeks	$360	81403
	IMAGe: *CDKN1C* sequencing ***(Prenatal)***	1-3 weeks	$700 $460	81479, 81265 (full sequencing) 81403, 81265 (site specific, known familial)
Li Fraumeni	Li-Fraumeni Syndrome: Site specific analysis (familial)	2-3 weeks	$360	81403
	Li-Fraumeni Syndrome: Site specific analysis (familial) ***(Prenatal)***	7-10 days	$460	81403, 81265
Lynch Sydnrome	HNPCC: MLH1, MSH2, MSH6 Site specific analysis (familial)	2-3 weeks	$360	81293 for MLH1 81296 for MSH2 81298 for MSH 6
	HNPCC: MLH1, MSH2, MSH6 Site specific analysis (familial) ***(Prenatal)***	1-3 weeks	$460	81293, 81265 for MLH1 81296, 81265 for MSH2 81298, 81265 for MSH 6

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	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	PIK3CA Sequence analysis	4-6 weeks	$1000	81479
	Somatic Overgrowth Panel (v2)	6-8 weeks	$2,500	81479
	PIK3CA Sequence analysis ***(Prenatal)***	2-4 weeks	$1,100	81479, 81265
	Somatic Overgrowth Panel (v2) ***(Prenatal)***	2-4 weeks	$2,600	81479, 81265
Retinoblastoma	Retinoblastoma: Sequence analysis with reflex to deletion/duplication *[Blood]*	6-8 weeks	$2,070*	81479x2
	Frozen tumor comprehensive analysis with reflex to site specific testing on constitutional DNA	8-10 weeks	$2,630	81479x2, 81401, 81402
	Retinoblastoma: Paraffin-embedded 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

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	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
	UM Related NGS Panel: *GNAQ, GNA11, BAP1*	4-6 weeks	$1,200	81479
	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

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