Endocrine Genetic Test List

Test No.	Test Name	Genes Included	Associated conditions and phenotypes
Bone			
630292	Hypophosphatasia and Hypophosphatemic Rickets Panel	*ALPL, CLCN5, CYP2R1, CYP27B1, DMP1, ENPP1, FGF23, PHEX, SLC34A3, VDR*	HPP, X-Linked Hypophosphatemia, X-Linked Hypophosphatemic Rickets, XLH, Hypophosphatemic Rickets, X-Linked Dominant Hypophosphatemic Rickets (XLHR), X-Linked Rickets (XLR), Vitamin D-Resistant Rickets, X-Linked Vitamin D-Resistant Rickets (VDRR), Hypophosphatemic Vitamin D-Resistant Rickets (HPDR), Phosphate Diabetes, Familial Hypophosphatemic Rickets
630543	Osteogenesis Imperfecta Genetic Panel	*BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1*	OI, Juvenile Primary Osteoporosis
Cancer			
481374	VistaSeq® Endocrine Cancer Panel*	*CDC73, MAX, MEN1, NF1, PRKAR1A, PTEN, RET, SDHB, SDHC, SDHD, TMEM127, TP53 and VHL*	Familial Isolated Hyperparathyroidism, Multiple Endocrine Neoplasia, Paraganglioma, Parathyroid Cancer, Pheochromocytoma
Diabetes and Insulin			
504603	Maturity-Onset Diabetes of the Young (MODY) 4-gene Panel	*GCK, HNF1A, HNF4A, HNF1B*	
630513	Maturity-Onset Diabetes of the Young (MODY) Expanded Genetic Panel	*ABCC8, APPL1, BLK, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1*	
630500	Congenital Hyperinsulinism Genetic Panel	*ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, KCNJ11, PGM1, PMM2, SLC16A1, UCP2*	Hypoglycemia
Growth and Development			
630534	Combined Pituitary Hormone Deficiency Genetic Panel	*GLI1, HESX1, LHX3, LHX4, OTX2, POU1F1, PROKR2, PROP1, SOX2, SOX3*	
630520	Comprehensive Short Stature Genetic Panel	*ACAN, BRAF, BTK, CBL, CCDC8, COL10A1, COL11A1, COL11A2, COL1A1, COL2A1, COL9A1, COL9A2, COL9A3, CUL7, EVC, FBN1, FGFR3, GH1, GHR, GHSR, GHRHR, GLI2, GPC3, H19, HESX1, HRAS, IGF1, IGF2, IGF1R, IGFALS, IHH, KRAS, LHX3, LHX4, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NPPC, NPR2, NRAS, OBSL1, OTX2, POU1F1, PPP1CB, PROKR2, PROP1, PTPN11, RAF1, RIT1, RRAS, SHOC2, SHOX, SOS1, SOS2, SOX3, SPRED1, SRCAP, STAT5B*	Idiopathic Short Stature
630527	Growth Hormone Deficiency Genetic Panel	*BTK, GH1, GHR, GHRHR, GHSR, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1*	GHD, Isolated Growth Hormone Deficiency
630542	Kallmann Syndrome Genetic Panel	*ANOS1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF8, FGF17, FGFR1, FLRT3, GNRH1, HS6ST1, IL17RD, KISS1R, NSMF, PROK2, PROKR2, SEMA3A, SEMA7A, SOX10, SPRY4, TAC3, TACR3, WDR11*	Hypogonadotropic Hypogonadism
Hypertension			
630258	Monogenic Hypertension Genetic Panel	*CUL3, CYP11B1, CYP11B2, HSD11B2, KCNJ5, KLHL3, NR3C2, SCNN1B, SCNN1G, WNK1, WNK4*	Pseudohypoaldosteronism, Familial Hyperaldosteronism, Liddle syndrome, Gordon syndrome
Thyroid			
630264	Congenital Hypothyroidism Genetic Panel	*DUOX2, DUOXA2, FOXE1, IYD, NKX2-5, PAX8, SLC5A5, SLC26A4, TG, THRA, THRB, TPO, TRHR, TSHB, TSHR*	Congenital Hypothyroidism, Thyroid Resistance, Bamforth-Lazarus Syndrome

*GeneSeq® Plus testing not available with this panel.

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Endocrine Genetic Test List

Single Gene Assays

Test No.	Test Name	Gene Sequenced
504513	Calcium-Sensing Receptor (*CASR*) Gene Sequencing Analysis	CASR
504576	*COMT* Genetic Test	COMT
500768	Congenital Adrenal Hyperplasia (*CAH*) 21-Hydroxylase (*CYP21*) Mutation (CAHDetx™)	CYP21A2
504010	*MEN1* Gene Sequencing Analysis	MEN1
504008	MEN2: *RET* Gene Sequencing	RET
500110	*SHOX*, DHPLC (Endocrine Sciences); SHOX-DNA-Dx ™	SHOX
504142	Thiopurine Methyltransferase (*TPMT*) Gentyping	TPMT
630540	Thyroid Hormone Resistance Beta (*THRB*) Gene Sequencing	THRB
630494	*PHEX* Gene Sequencing	PHEX

GeneSeq Plus® [630068]

Labcorp's GeneSeq® Plus test allows providers to request full gene sequencing on a single gene from the panels above or from our Inheritest® 500 Plus panel. Some common endocrine-related genes are below. Additional genes are available.

To utilize this option, simply order the GeneSeq® Plus test number and then specify on the order form what gene you would like sequenced.

Genes available for sequencing by GeneSeq® Plus
Bardet-Biedl Syndrome (*ARL6*)
Bardet-Biedl Syndrome (*BBS1*)
Bardet-Biedl Syndrome (*BBS12*)
Bardet-Biedl Syndrome (*BBS2*)
Bardet-Biedl Syndrome (*BBS4*)
Bardet-Biedl Syndrome (*BBS5*)
Bardet-Biedl Syndrome (*BBS7*)
Bardet-Biedl Syndrome (*BBS9*)
Bardet-Biedl Syndrome (*MKKS*)
Bardet-Biedl Syndrome (*SDCCAG8*)
Bardet-Biedl Syndrome (*TTC8*)
Cohen Syndrome (*VPS13B*)
Congenital Adrenal Hyperplasia (*CYP11B1*)
Congenital Adrenal Hyperplasia (*CYP17A1*)
Congenital Adrenal Hyperplasia (*HSD3B2*)
Congenital Adrenal Hyperplasia (*POR*)
Congenital Adrenal Hyperplasia (*StAR*)
Congenital Adrenal Hyperplasia, X-Linked (*NROB1*)
Nephrogenic Diabetes Insipidus Evaluation (*AVPR2*)

Genetic counselors available to answer testing or result questions: **800-345-GENE (4363)**

Visit **Labcorp.com** for full test information, including CPT coding, RUO/IUO status, and current specimen collection requirements.

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