

ENDOCRINOLOGY PANELS

PANEL NAME	SYNDROMES PHENOTYPES CONDITIONS	GENES
Hypo-parathyroidism, standard	Hypoparathyroidism, Autosomal Dominant, Autoimmune Polyendocrine Syndrome, type I, with or without reversible metaphyseal dysplasia, Hypoparathyroidism, familial isolated, Hypoparathyroidism, sensorial deafness, and renal disease, Pseudohypoparathyroidism, type IB, Pseudohypoparathyroidism, type IA, Pseudopseudohypoparathyroidism	PTH AIRE CaSR GNA11
Hypo-parathyroidism, extended		PTH AIRE GCM2 GATA3 CaSR STX16 GNA11 GCM1
Pseudohypo-parathyroidism	Pseudohypoparathyroidism, Pseudopseudohypoparathyroidism	GNAS
Hyper-parathyroidism, standard	Pituitary adenoma, prolactin secreting, Multiple endocrine neoplasia, type I + IV, Thyroid carcinoma, papillary, Parathyroid carcinoma, Hypoparathyroidism, familial isolated, Hypocalcemia, autosomal dominant 2, Hypocalciuric hypercalcemia, familial, type III	MENIN RET CDC73 CaSR AIP

Hyper-parathyroidism, extended		MENIN RET CDC73 CaSR AIP CDKN1b GCM2 AP2S1
Osteogenesis Imperfecta, standard		COL1A1 COL1A2
Osteogenesis Imperfecta, extended		COL1A1 COL1A2 PPIB CRTAP LEPRE1 FKBP10 PLOD2 SERPINF1 PLS3 SERPINH1 SP7 WNT1 TMEM38B, BMP1, IFITM5
Osteopetrosis		CA2 CLCN7 LRP5 OSTM1 PLEKHM1 SNX10 TCIRG1 TNFRSF11A TNFSF11

Adipositas	Obesity: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [MIM:601665]	AGRP CARTPT FTO LEP LEPR MC4R NR0B2 POMC SIM1 UCP1 UCP3
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