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GENERAL INFORMATION

- **TEST-MATERIAL**
  - Purified DNA is preferred (3-5µg DNA, min concentration 40ng/ml).
  - Can also use blood (5ml, EDTA stabilized).
- **TURN-AROUND-TIME** (TAT) is 5-6 weeks from receipt of sample.
  - *IMPORTANT:* waiting time caused of lacking parent samples for segregation analysis will cause the TAT-clock to be reset. Therefore, if possible, send parents together with the patient sample.
- **REPORT:** in pdf. Hard-copy if requested.
- The **price includes** validation of suspected pathological mutation(s) by Sanger *in the proband only*.
- The **price does <u>not</u> include** segregation analysis (parental follow-up). The price of segregation/parental analysis depends on the number of mutations to investigate. The number of mutations to be investigated is not known until the proband has been analyses and evaluated.
  - €200/mutation/parent.

ENDOCRINOLOGY PANELS

PANEL NAME (# of genes)	**GENES**
Adiposity (8)	POMC, MC4R, NR0B2, UCP1, CARTPT, SIM1, UCP3, AGRP
Familial Hypercholesterolaemia (11)	APOA2, ITIH4, GHR, PPP1R17, EPHX2, ABCA1, LDLR, LPL, PCSK9, DLRAP1, APOB
Hyperinsulinism (9)	ABCC8, KCNJ11, HADH, GLUD1, GCK, INSR, INS, UCP2, SLC16A1
Hyperparathyroidism (12)	CaSR, MEN1, GCM1, GCM2, SOX3, GATA3, TBX1, CLCNKB, CDC73, CDKN1B, AIP
Hypocalcicuric Hypercalcemia (3)	CaSR, GNA11, AP2S1
Hypophosphatasia (1)	ALPL
Idiopathic hypoparathyroidism (8)	PTH, AIRE, HADHA, HADHB, PTH1R, ATP7B , MICA, MICB
Maturity Onset Diabetes of the Young, MODY (11)	HNF1A, HBF1B, HNF4A, GCK, ABCC8, KCNJ11, BLK1, PAX4, NEUROD, PDX1, INS
Osteogenesis Imperfecta (15)	Col1A1, Col1A2, PPIB, CRTAP, LEPRE1, FKBP10, PLOD2, SERPINF1, SERPINH1, SP7, WNT1, TMEM38B, BMP1, IFITM5, PLS3
Osteopetrosis (14)	CLCN7, TCIRG1, IKBKG, CA2, CLCN7, IKBKG, ITGB3, OSTM1, PLEKHM1, TCIRG1, TNFRSF11A, TNFSF11, LRP5, SNX10
Porphyria (7)	UROD, UROS, FECH, PPOX, CPOX, HMBS, ALAD
Pseudohypoparathyroidism / Pseudopseudohypoparathyroidism (3)	GNAS, STX16, TSHR
Rickets (9)	PHEX, FGF23, ClCN5, DPM1, SLC34A3, ENPP1, CYP2R1, VDR
Thrombosis (18)	PROS1, SERPINC1, PROC, F2, F5, MTHFR, THBD, ADAMTS13, F13A1, HABP2, CBS, FGA, HABP2, MMACHC, HRG, PLG, PROZ, SERPIND1