University of Eastern Finlan

GENOME CENTER OF EASTERN FINLAND

Gene Diagnostics

Pricelist

Genetic tests

Prices until 30.4.2023

21-hydroxylase deficiency	Price (vat 0%)
Screening of the coding region of the CYP21A2 gene and deletion analysis	890 €
Screening of the coding region of the CYP21A2 gene and deletion analysis including parents	1300 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Amyloidosis	Price (vat 0%)
Amyloidosis: Screening of the coding region of the TTR gene	700 €
A previously identified gene variant	220 €
Androgen insensitivity syndrome	Price (vat 0%)
Screening of the coding region of the AR gene and deletion analysis	890 €
Deletion analysis of the AR gene	600 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
APECED	Price (vat 0%)
Screening of the coding region of the AIRE gene	700 €
Screening of the AIRE p.Arg257Term gene variant	220 €
A previously identified gene variant	220 €
Dilated cardiomyopathy	Price (vat 0%)
Screening of the LMNA p.Ser143Pro (rs61661343) mutation	220 €
Screening of the coding region of the Lamin A/C (LMNA) gene and deletion analysis	890 €
Deletion analysis of the LMNA gene	600 €
Hyperthrophic cardiomyopatho and dilated cardiomyopathy related genes (screening of the coding regions of the 62 genes)	1300 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Fabry disease	Price (vat 0%)
Screening of the coding region of the GLA gene and deletion analysis	890 €
Screening of the introns of the GLA gene	700 €
A previously identified deletion or insertion	400 €
A previously identified gene variant	220 €
Familial hypercholesterolemia	Price (vat 0%)
Screening of the FH-Helsinki, FH-Pohjois-Karjala, FH-Turku, FH-Pori and FH-Pogosta mutations in the LDL receptor gene	300 €
Screening of the FH-Helsinki, FH-Pohjois-Karjala, FH-Turku, FH-Pori and FH-Pogosta mutations in the LDL receptor gene if result negative then Screening of the coding region of the LDL-receptor (LDL) gene and deletion analysis	1000 €
Screening of the FH-Helsinki, FH-Pohjois-Karjala, FH-Turku, FH-Pori and FH-Pogosta mutations in the LDL receptor gene if result negative then deletion analysis and screening of the coding region and promoter area (approximately 1000 bases before exon 1) of the LDL-receptor (LDL) gene	1000 €
Deletion analysis and screening of the coding region and promoter area (approximately 1000 bases before exon 1) of the LDL-receptor (LDL) gene	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €

Pheochromocytoma	Price (vat 0%)
Screening of the coding regions of the NF1, RET, VHL, SDHA, SDHB, SDHC, SDHD, SDHAF2, TMEM127, MAX, FH, EPAS1, EGLN1, KIF1B, IDH1, HRAS genes and deletion analysis of SDHB, SDHC and ADHD genes	1400 €
Deletion analysis of the SHDB, SDHC and SDHD genes	600 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Gaucher's disease	**Price (vat 0%)**
Screening of the coding region of the GBA gene and deletion analysis	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Gitelman syndrome	**Price (vat 0%)**
Screening of the coding regions of the SLC12A3 gene	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Hemochromatosis	**Price (vat 0%)**
Screening of the HFE Cys282Tyr and His63Asp mutations	440 €
Screening of the coding regions of the HFE, HFE2, HAMP, TFR2 and SLC40A1 genes	990 €
A previously identified gene variant	220 €
Hypercalsemia	**Price (vat 0%)**
All Hypercalcemias: Screening of the coding regions of the CASR, AP2S1, GNA11, CDC73 (HRPT2), MEN1, RET, CDKN1B and SLC34A3 genes	1400 €
Familial hypocalciuric hypercalcemia (FHH): Screening of the coding regions of the CASR, AP2S1 and GNA11 genes	990 €
Familial hypocalciuric hypercalcemia (FHH): Screening of the coding regions of the CASR, AP2S1 and GNA11 genes (990€) if result negative then deletion analysis of CASR gene	1200 €
Familial isolated primary hyperparathyroidism (FIPH): Screening of the coding regions of the CASR, MEN1 and CDC73 (HRPT2) genes	990 €
Jaw tumor-associated hyperparathyroidism: Screening of the coding regions of the CDC73 (HRPT2) gene and deletion analysis	890 €
MEN1, MEN2A and MEN4 related hyperparathyroidism: Screening of the coding regions of the MEN1, RET and CDKN1B genes	990 €
Neonatal severe hyperparathyroidism: Screening of the coding regions of the CASR gene and deletion analysis	890 €
Hereditary hypophosphatemic rickets with hypercalciuria (HHRH): Screening of the coding regions of the SLC34A3 gene	700 €
Jaw tumor-associated hyperparathyroidism: Deletion analysis of CDC73 (HRPT2) gene	600 €
Deletion analysis of the CASR gene	600 €
Deletion analysis of the MEN1 gene	600 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
hypertriglyceridemia	**Price (vat 0%)**
Screening of the coding regions of the LPL, APOC2 and APOC3 genes	990 €
A previously identified gene variant	220 €
Hypertrophic cardiomyopathy	**Price (vat 0%)**
Screening of the MYBPC Gln1061Term (rs397516005) mutation	220 €
Screening of the TPM1 Asp175Asn (rs104894503) mutation	220 €
Screening of the MYH7 Arg1053Gln (rs587782962) mutation	220 €
Screening of the MYBPC Gln1061Term, TPM1 Asp175Asn and MYH7 Arg1053Gln mutations	660 €
Hyperthrophic cardiomyopatho and dilated cardiomyopathy related genes (screening of the coding regions of the 62 genes)	1300 €
A previously identified gene variant	220 €

Hypocalsemia	Price (vat 0%)
Screening of the coding regions of the GCMB-, PTH-, GATA3-, TBCE-, AIRE-, GNAS-, CASR-, GNA11-, TRPM6-, CYP27A1, VDR-, FAM111A and TBX1 genes	1300 €
Familial isolated hypoparathyroidism: Screening of the coding regions of the GCMB- and PTH genes	990 €
Deletion analysis of the CASR gene	600 €
Autosomal dominant hypocalcemia: Screening of the coding regions of the CASR and GNA11 genes	990 €
Bartter's syndrome type 5: Screening of the coding regions of the CASR gene	700 €
Hypomagnesemia With Secondary Hypocalcemia: Screening of the coding regions of the TRPM6 gene	700 €
Vitamin D-dependent rickets type 1: Screening of the coding regions of the CYP27A1- and VDR genes	990 €
Kenny-Caffey syndrome type 2: : Screening of the coding regions of the FAM111A gene	700 €
DiGeorge syndrome: Screening of the coding regions of the TBX1 gene	700 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Familial hypothyroidism	Price (vat 0%)
Screening of the coding regions of the GNAS, SLC5A5, SLC26A4, TG, TPO, DUOX1, DUOX2, DUOXA2, NCOR1, NKX2-1, SECISBP2, TRH, TRHR, TSHR, USP47, PAX8, TSHB, THRA, THRB, NKX2-5, FOXE1 and DEHAL1 genes	1300 €
A previously identified gene variant	220 €
Type III hyperlipidemia	Price (vat 0%)
Genotyping of the apolipoprotein E gene: A previously identified gene variant	440 €
Thyroid hormone resistance	Price (vat 0%)
Screening of the coding region of the THRA and THRB genes	990 €
A previously identified gene variant	220 €
Evaluation of the efficacy of clopidogrel treatment	Price (vat 0%)
Screening of the variant c.681G>A (rs4244285) in the CYP2C19 gene	220 €
Liddle syndrome	Price (vat 0%)
Screening of the coding regions of the SCNN1B and SCNN1G genes	990 €
A previously identified gene variant	220 €
Aldosterone producing adenoma	Price (vat 0%)
Screening of the coding region of the KCNJ5 gene	700 €
A previously identified gene variant	220 €
Marfan syndrome	Price (vat 0%)
Screening of the coding region of the FBN1 gene and deletion analysis	890 €
Deletion analysis of the FBN1 gene	600 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Obesity caused by melanocortin-4 receptor mutations	Price (vat 0%)
Screening of the coding region of the MC4R gene and deletion analysis	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Mitochondrial diabetes	Price (vat 0%)
Screening of the mtDNA - tRNALeu(UUR) A3243G mutation	220 €

MODY diabetes	Price (vat 0%)
Screening of the coding regions of the HNF4A, GCK and HNF1A genes and deletion analysis (MODY1-3)	1300 €
Screening of the coding region of the HNF4A gene and deletion analysis (MODY 1)	890 €
Screening of the coding region of the GCK gene and deletion analysis (MODY 2)	890 €
Screening of the coding region of the HNF1A gene and deletion analysis (MODY 3)	890 €
Screening of the coding region of the HNF1B gene and deletion analysis (MODY 5)	890 €
Screening of the coding regions of the PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS and BLK genes and deletion analysis (exceluding BLK gene) (MODY 4-10)	1300 €
Screening of the coding regions of the UCP2, SLC2A2, SLC16A1, PTF1A, PDX1, PAX4, NEUROD1, KLF11, KCNJ11, INS, HNF4A, HNF1B, HNF1A, HADH, GLUD1, GCK, FOXP3, EIF2AK3, CEL, BLK and ABCC8	1300 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Monogenic diabetes	Price (vat 0%)
Screening of the coding regions of the ABCC8- (SUR1), KCNJ11- (Kir6.2), FOXP3-, HNF4A-, GCK-, HNF1A-, PDX1-, HNF1B-, NEUROD1-, KLF11-, CEL-, PAX4-, INS- and BLK genes; deletion analysis of the ABCC8- (SUR1), HNF4A-, GCK-, HNF1A-, PDX1-, HNF1B-, NEUROD1-, KLF11-, CEL-, PAX4-, INS-genes; Screening of the mtDNA - MTTL1 gene tRNALeu(UUR) m.3243A>G variant	1500 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Multiple endocrine neoplasia type 1	Price (vat 0%)
Screening of the coding region of the MEN1 gene and deletion analysis	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Multiple endocrine neoplasia type 2	Price (vat 0%)
Screening of the coding regions of the RET gene	700 €
A previously identified gene variant	220 €
Other familial hypercholesterolemia	Price (vat 0%)
Screening of the coding regions of the LDLR, LDLRAP1, APOB, APOE, PCSK9, ABCG5 and ABCG8 genes and deletion analysis of the LDLR gene	1300 €
A previously identified gene variant	220 €
Statin-induced myopathy	Price (vat 0%)
Screening of the variant c.1498-1331T>C (rs4363657) in the SLCO1B1 gene	220 €
Nephrotic syndrome	Price (vat 0%)
Screening of the coding region of the NPHS2 gene	700 €
A previously identified gene variant	220 €
Polycystic kidney disease	Price (vat 0%)
Screening of the coding region of the PKD1 and PKD2 genes and deletion analysis	1300 €
Screening of the coding regions of the PKHD1 gene and deletion analysis	890 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €
Sitosterolemia	Price (vat 0%)
Screening of the coding regions of the ABCG5 and ABCG8 genes	990 €
A previously identified gene variant	220 €
Congenital hyperinsulinemia	Price (vat 0%)
Screening of the coding regions of the ABCC8 (SUR1) and KCNJ11 (Kir6.2) genes and deletion analysis of ABCC8 (SUR1) gene	990 €
Screening of the coding region of the GCK gene and deletion analysis	890 €
Screening of the coding regions of the other genes causing congenital hyperinsulinemia (GLUD1, HADH, HNF4A, UCP2, SLC16A1, SLC2A2, PTF1A, EIF2AK3)	1300 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €

Evaluation of the efficacy of warfarin treatment	Price (vat 0%)
Screening of the variant c.-1639G>A (rs9923231) in the VKORC1 gene and variant c.1075A>C (rs1057910) in the CYP2C9 gene	440 €

Permanent neonatal diabetes mellitus	Price (vat 0%)
Screening of the coding regions of the ABCC8 (SUR1), KCNJ11 (Kir6.2), GCK, INS and FOXP3 genes and deletion analysis of ABCC8 (SUR1) gene	1300 €
Screening of the coding regions of the ABCC8 (SUR1) and KCNJ11 (Kir6.2) genes and deletio analysis of ABCC8 (SUR1) gene	990 €
Screening of the coding region of the ABCC8 gene and deletion analysis	890 €
Screening of the coding region of the KCNJ11 (Kir6.2) gene	700 €
Screening of the coding region of the GCK gene and deletion analysis	890 €
Screening of the coding region of the INS gene	700 €
Screening of the coding region of the FOXP3 gene	700 €
A previously identified gene variant	220 €
A previously identified deletion or insertion	400 €