	MOLECULAR GENETIC ANALYSIS	ANALYSIS	GENE	TIME since sample reception*	SAMPLE	VND
MUT-PUNT	Pointed Mutational Analysis (any gene from this list)	Any locus		**2 weeks**	5 ml blood EDTA/Saliva on swab	9 000 000
SRY-MUT	46,XX True Hermaphroditism, SRY-positive (Gonadal Dysgenesis)	Detection	SRY	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 000 000
ACH	Achondroplasia	p.G380R and p.G375C	FGFR3	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
PML-RARA	Acute Promyelocytic Leukemia	Fusion PML/RARA	PML/RARA	**4 weeks**	5 ml blood EDTA on ice	8 300 000
MCADD	ACYL-COA Dehydrogenase Medium-Chain, Deficiency of	c.985A>G (p.Lys304Glu)	ACADM	**3 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
MCADD-SEQ	ACYL-COA Dehydrogenase Medium-Chain, Deficiency of	cds	ACADM	**2 months**	5 ml blood EDTA	37 400 000
ADIPOQ	Adiponectin (Hipoadiponectinemia)	c.276G>T	ADIPOQ	**1 week**	5 ml blood EDTA/Saliva on swab	5 700 000
CYP21A2-2MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	IVS2-13A/C>G and c.G110del8nt	CYP21A2	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
CYP21A2-8MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	p.Pro30Leu, c.293-13A/C>G, p.Val281Leu, p.Leu307PhefsX6, p.Gln318X, p.Arg356Trp, p.Arg444X and p.Pro453Ser	CYP21A2	**4 weeks**	5 ml blood EDTA	17 300 000
CYP21A2-1MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	p.Val281Leu	CYP21A2	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
CYP21A2	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	cds	CYP21A2	**5 weeks**	5 ml blood EDTA	24 800 000
CYP21A2-DEL	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	Deletions	CYP21A2	**5 weeks**	5 ml blood EDTA	20 800 000
ABCD1-SEQ	Adrenoleukodystrophy (X-linked)	cds	ABCD1	**4 weeks**	5 ml blood EDTA/Saliva on swab	37 400 000
ABCD1-DEL	Adrenoleukodystrophy (X-linked)	Deletions	ABCD1	**5 weeks**	5 ml blood EDTA	23 100 000
FGA	Afibrinogenemia, congenital	cds	FGA	**4 weeks**	5 ml blood EDTA/Saliva on swab	30 800 000
FGB	Afibrinogenemia, congenital	cds	FGB	**4 weeks**	5 ml blood EDTA/Saliva on swab	37 400 000
FGG	Afibrinogenemia, congenital	cds	FGG	**4 weeks**	5 ml blood EDTA/Saliva on swab	47 100 000
OCA1	Albinism oculocutaneous Type IA (OCA1A)	cds	TYR	**3 weeks**	5 ml blood EDTA/Saliva on swab	21 500 000
OCA2	Albinism oculocutaneous Type II (OCA2)	cds	OCA2	**2 months**	5 ml blood EDTA	60 100 000
AAT	Alpha-1-Antitrypsin Deficiency (AAT)	Variants Z, S and M	SERPINA1	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 200 000
HBA	Alpha-Thalassaemia	α3.7, α4.2, α20.5, αSEA, αFIL and αMED	HBA	**3 weeks**	5 ml blood EDTA	10 200 000
HBA1y2	Alpha-Thalassaemia	cds	HBA1 and HBA2	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 000 000
ALPORT-D	Alport Syndrome (Autosomal Dominant)	cds	COL4A3	**4 months**	5 ml blood EDTA	94 700 000
ALPORT-R	Alport Syndrome (Autosomal Recessive)	cds	COL4A3	**4 months**	5 ml blood EDTA	94 700 000
COL4A4	Alport Syndrome (Autosomal Recessive)	cds	COL4A4	**4 months**	5 ml blood EDTA	94 700 000
COL4A5	Alport Syndrome (X-linked, ATS)	cds	COL4A5	**4 months**	5 ml blood EDTA	94 700 000
COL4A5-DEL	Alport Syndrome (X-linked, ATS)	Deletions	COL4A5	**7 weeks**	5 ml blood EDTA	36 500 000
AD1	Alzheimer Disease Familial Type 1 (AD1)	cds	APP	**7 weeks**	5 ml blood EDTA/Saliva on swab	38 300 000

AD1-EX	Alzheimer Disease Familial Type 1 (AD1)	Exons 16 and 17	APP	**3 weeks**	5 ml blood EDTA/Saliva on swab	9 200 000
AD3	Alzheimer Disease Familial Type 3 (AD3)	cds	PSEN1	**4 weeks**	5 ml blood EDTA/Saliva on swab	24 600 000
AD4	Alzheimer Disease Familial Type 4 (AD4)	cds	PSEN2	**4 weeks**	5 ml blood EDTA/Saliva on swab	22 800 000
APOE	Alzheimer Disease Type 2 (AD2), Late Onset	Genotyping E2/E3/E4	APOE	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
TTR	Amyloidosis hereditary, transthyretin-related	cds	TTR	**3 weeks**	5 ml blood EDTA/Saliva on swab	17 800 000
AS	Angelman Syndrome	cds	UBE3A	**2 months**	5 ml blood EDTA/Saliva on swab	38 300 000
AS-UPD	Angelman Syndrome	Uniparental disomy STRs	15q11-q13	**5 weeks**	5 ml blood EDTA	10 700 000
AS-MET	Angelman Syndrome	Metilation	AS	**3 weeks**	5 ml blood EDTA	10 000 000
AS-DEL	Angelman Syndrome	Deletions, metilation and uniparental disomy	AS	**4 weeks**	5 ml blood EDTA	20 800 000
C1NH	Angioedema hereditary types I and II (hane C1 esterase inhibitor, deficiency of).	cds	SERPING1 (C1NH)	**5 weeks**	5 ml blood EDTA/Saliva on swab	30 800 000
ECA	Angiotensin Converting Enzyme (ACE)	I/D	ACE	**1 week**	5 ml blood EDTA/Saliva on swab	4 700 000
HLA-B27	Ankylosing Spondylitis	HLA-B27	HLA-B	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 600 000
APERT-MUT	Apert Syndrome	p.S252W and p.P253R	FGFR2	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
APERT	Apert Syndrome	cds	FGFR2	**2 months**	5 ml blood EDTA	33 000 000
APOA5	Apolipoprotein A-V (APOA5)	c.-1131T>C	APOA5	**1 week**	5 ml blood EDTA/Saliva on swab	4 700 000
ARVD10	Arrhythmogenic Right Ventricular Dysplasia Type 10 (ARVD10)	cds	DSG2	**2 months**	5 ml blood EDTA	48 400 000
ARVD8	Arrhythmogenic Right Ventricular Dysplasia Type 8 (ARVD8)	cds	DSP	**3 months**	5 ml blood EDTA	70 500 000
ARVD9	Arrhythmogenic Right Ventricular Dysplasia Type 9 (ARVD9)	cds	PKP2	**2 months**	5 ml blood EDTA	41 800 000
ATM	Ataxia-telangiectasia (Louis-Bar, Syndrome)	cds	ATM	**5 months**	5 ml blood EDTA	156 500 000
ATM-DEL	Ataxia-telangiectasia (Louis-Bar, Syndrome)	Deletions/Duplication	ATM	**5 weeks**	5 ml blood EDTA	34 100 000
HBB-B	Beta-Thalassaemia	cds + introns	HBB	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 000 000
HBB-DB	Beta-Thalassaemia	Deletion Spanish (delta-beta)	HBB	**4 weeks**	5 ml blood EDTA	22 000 000
HBB-B-DEL	Beta-Thalassaemia	Deletions	HBB	**5 weeks**	5 ml blood EDTA	23 100 000
BRUG	Brugada Syndrome	cds	SCN5A	**3 months**	5 ml blood EDTA	63 800 000
CADAS-2EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	Exons 3 and 4	NOTCH3	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 000 000
CADAS-4EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	Exons 2, 5, 6 and 11	NOTCH3	**4 weeks**	5 ml blood EDTA/Saliva on swab	13 400 000
CADAS-6EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	Exons 2, 3, 4, 5, 6 and 11	NOTCH3	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 700 000
CADAS	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	cds	NOTCH3	**3 months**	5 ml blood EDTA	85 500 000
SOX9	Campomelic Dysplasia	cds	SOX9	**4 weeks**	5 ml blood EDTA/Saliva on swab	23 500 000
CANAV	Canavan Disease	cds	ASPA	**4 weeks**	5 ml blood EDTA/Saliva on swab	20 200 000
CMD1A	Cardiomyopathy Dilated (CMD1A)	cds	LMNA	**3 months**	5 ml blood EDTA	35 200 000
CMD1D	Cardiomyopathy Dilated (CMD1D)	cds	TNNT2	**2 months**	5 ml blood EDTA	34 300 000
CMH2	Cardiomyopathy Hypertrophic familial (CMH2)	cds	TNNT2	**2 months**	5 ml blood EDTA	34 300 000
CMH4	Cardiomyopathy Hypertrophic familial (CMH4)	cds	MYBPC	**3 months**	5 ml blood EDTA	68 300 000

CEL	Celiac disease	Complete genotyping	HLA-DQA1 and HLA-DQB1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
RYR1-MUT	Central Core Disease Myopathy	p.L4793P, p.R4825C, p.R4861H, p.R4861C, p.R4893W, p.R4893G, p.G4899E and p.R4914G	RYR1	**3 weeks**	5 ml blood EDTA/Saliva on swab	14 000 000
RYR1-EX	Central Core Disease Myopathy	Exons 1-17, 39-48 and 90-104	RYR1	**3 months**	5 ml blood EDTA/Saliva on swab	84 600 000
CCM-DEL	Cerebral Cavernous Malformations	Deletions	KRIT1, CCM2 and PDCD10	**2 months**	5 ml blood EDTA	36 500 000
CCM1	Cerebral Cavernous Malformations 1 (CCM1)	cds	KRIT1	**6 weeks**	5 ml blood EDTA/Saliva on swab	38 300 000
CCM2	Cerebral Cavernous Malformations 2 (CCM2)	cds	CCM2	**2 months**	5 ml blood EDTA/Saliva on swab	29 200 000
CCM3	Cerebral Cavernous Malformations 3 (CCM3)	cds	PDCD10	**4 weeks**	5 ml blood EDTA/Saliva on swab	23 700 000
CMT1A	Charcot-Marie-Tooth 1A (CMT1A)	STRs	PMP22	**4 weeks**	5 ml blood EDTA/Saliva on swab	11 800 000
CMT1B	Charcot-Marie-Tooth 1B (CMT1B)	cds	MPZ	**4 weeks**	5 ml blood EDTA/Saliva on swab	23 100 000
CMT1E	Charcot-Marie-Tooth 1E (CMT1E)	cds	PMP22	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 100 000
CMT2A2	Charcot-Marie-Tooth 2A2 (CMT2A2)	cds	MFN2	**3 months**	5 ml blood EDTA	46 200 000
CMT2B-MUT	Charcot-Marie-Tooth 2B (CMT2B)	p.Leu129Phe, p.Lys157Asn, p.Asn161Thr and p.Val162Met	RAB7A	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
CMT2B	Charcot-Marie-Tooth 2B (CMT2B)	cds	RAB7A	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 300 000
CHARGE	Charge syndrome	cds	CHD7	**4 months**	5 ml blood EDTA	118 500 000
CONDRO	Chondrodysplasia Punctata, X-linked dominant (Conradi-Hunermann syndrome)	cds	EBP	**4 weeks**	5 ml blood EDTA/Saliva on swab	25 000 000
RUNX2	Cleidocranial Dysplasia	cds	RUNX2	**4 weeks**	5 ml blood EDTA/Saliva on swab	33 000 000
RUNX2-DEL	Cleidocranial Dysplasia	Deletions	RUNX2	**5 weeks**	5 ml blood EDTA	23 100 000
CBAVD	Congenital bilateral absence of vas deferens (CBAVD)	36 mutations + Tn	CFTR	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 200 000
EFNB1	Craniofrontonasal Dysplasia	cds	EFNB1	**6 weeks**	5 ml blood EDTA	29 900 000
CCA-FGFR2	Craniosynostosis FGFR2 related	cds	FGFR2	**2 months**	5 ml blood EDTA	33 000 000
CCA-FGFR3	Craniosynostosis FGFR3 related	cds	FGFR3	**2 months**	5 ml blood EDTA/Saliva on swab	36 500 000
CNS	Craniosynostosis non syndromic	Exon 7 (FGFR1); exon 7 (FGFR2); exons 6 and 8 (FGFR3)	FGFR1, FGFR2 and FGFR3	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
CROHN	Crohn disease, susceptibility to	cds	NOD2 (CARD15)	**4 weeks**	5 ml blood EDTA/Saliva on swab	45 500 000
CROHN-MUT	Crohn disease, susceptibility to	p.R720W, p.G908R and p.1007fs	NOD2 (CARD15)	**2 weeks**	5 ml blood EDTA/Saliva on swab	11 800 000
CROUZON-MUT	Crouzon syndrome	p.C342Y and p.C342R	FGFR2	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
CROUZON	Crouzon syndrome	cds	FGFR2	**2 months**	5 ml blood EDTA	33 000 000
CROUZON-AC	Crouzon syndrome with acanthosis nigricans	cds	FGFR3	**2 months**	5 ml blood EDTA/Saliva on swab	36 500 000
CURR	Currarino Syndrome (Sacral Agenesis Hereditary)	cds	MNX1 (HLXB9)	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000

CFTR-MUT	Cystic Fibrosis	36 mutations	CFTR	**1 week**	5 ml blood EDTA/Saliva on swab	7 300 000
CFTR	Cystic Fibrosis	cds	CFTR	**2 months**	5 ml blood EDTA	51 700 000
SN-MYO	Deafness Type 3 (Autosomal Recessive)	cds	MYO15A	**4 months**	5 ml blood EDTA	178 100 000
SN-SLC	Deafness Type 4 (Autosomal Recessive)	cds	SLC26A4	**2 months**	5 ml blood EDTA	46 900 000
SORDERA	Deafness, Aminoglycoside Induced	m.1555A>G, m.961_962delTinsC(n), m.7443A>G, m.74444G>A and m.7445A>G	MT-RNR1 and MT-CO1	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 300 000
DRPLA	Dentatorubro-pallidoluysian Atrophy (Naito-Oyanagi disease) (DRPLA)	Expansion CAG	ATN1	**3 weeks**	5 ml blood EDTA	9 400 000
DMN	Diabetes Mellitus Neonatal	cds	KCNJ11	**3 weeks**	5 ml blood EDTA/Saliva on swab	15 300 000
DBA1	Diamond-Blackfan Anemia (DBA1)	cds	RPS19	**4 weeks**	5 ml blood EDTA/Saliva on swab	25 700 000
DBA1-DEL	Diamond-Blackfan Anemia (DBA1)	Deletions/Duplications	RPS19	**5 weeks**	5 ml blood EDTA	20 800 000
DBA10	Diamond-Blackfan Anemia (DBA10)	cds	RPS26	**3 weeks**	5 ml blood EDTA	24 200 000
DBA3	Diamond-Blackfan Anemia (DBA3)	cds	RPS24	**6 weeks**	5 ml blood EDTA	31 200 000
DBA4	Diamond-Blackfan Anemia (DBA4)	cds	RPS17	**4 weeks**	5 ml blood EDTA	28 100 000
DBA5	Diamond-Blackfan Anemia (DBA5)	cds	RPL35A	**4 weeks**	5 ml blood EDTA	24 200 000
DBA6	Diamond-Blackfan Anemia (DBA6)	cds	RPL5	**2 months**	5 ml blood EDTA	40 500 000
DBA7	Diamond-Blackfan Anemia (DBA7)	cds	RPL11	**2 months**	5 ml blood EDTA	31 200 000
DBA8	Diamond-Blackfan Anemia (DBA8)	cds	RPS7	**2 months**	5 ml blood EDTA	31 200 000
DBA9	Diamond-Blackfan Anemia (DBA9)	cds	RPS10	**4 weeks**	5 ml blood EDTA	31 200 000
CATCH22	DiGeorge syndrome, included CATCH22	Deletion 22q11.2	(TBX1)	**3 weeks**	10 ml LA	9 800 000
DUCHENNE-DEL	Duchenne/Becker Muscular Dystrophy	Deletions	DMD	**5 weeks**	5 ml blood EDTA	23 100 000
DUCHENNE	Duchenne/Becker Muscular Dystrophy	cds	DMD	**5 months**	5 ml blood EDTA	147 200 000
DISFIBRIN	Dysfibrinogenemia	cds	FGA	**2 months**	5 ml blood EDTA/Saliva on swab	36 300 000
DYT1	Early-onset Torsion Dystonia (DYT1) (Autosomal dominant)	del(GAG)	TOR1A	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
COL5A1	Ehlers-Danlos Syndrome, Type I	cds	COL5A1	**5 months**	5 ml blood EDTA	157 500 000
COL5A2	Ehlers-Danlos Syndrome, Type I	cds	COL5A2	**4 months**	5 ml blood EDTA	147 200 000
COL3A1	Ehlers-Danlos Syndrome, Type III	cds	COL3A1	**4 months**	5 ml blood EDTA	112 400 000
TNXB	Ehlers-Danlos Syndrome, Type III	cds	TNXB	**4 months**	5 ml blood EDTA	112 400 000
COL3A1-IV	Ehlers-Danlos Syndrome, Type IV	cds	COL3A1	**4 months**	5 ml blood EDTA	112 400 000
PLOD1	Ehlers-Danlos Syndrome, Type VI	cds	PLOD1	**3 months**	5 ml blood EDTA	63 800 000
EHLERS-VII	Ehlers-Danlos Syndrome, Type VII	cds	COL1A1	**4 months**	5 ml blood EDTA	59 000 000
EHLERS-VII2	Ehlers-Danlos Syndrome, Type VII	cds	COL1A2	**4 months**	5 ml blood EDTA	64 700 000
ADAMTS2	Ehlers-Danlos Syndrome, Type VII	cds	ADAMTS2	**3 months**	5 ml blood EDTA	92 500 000
EDMD-AD	Emery-Dreifuss Muscular Dystrophy (Autosomal Dominant) (AD-EDMD)	cds	LMNA	**3 months**	5 ml blood EDTA	35 200 000
EDMD-XL	Emery-Dreifuss Muscular Dystrophy, X-linked (XL-EDMD)	cds	EMD	**6 weeks**	5 ml blood EDTA/Saliva on swab	25 900 000
COL7A1-AD-EX	Epidermolysis Bullosa Dystrophica (Autosomal Dominant)	Exons 73, 74 and 75	COL7A1	**3 weeks**	5 ml blood EDTA	10 900 000

COL7A1-AD	Epidermolysis Bullosa Dystrophica (Autosomal Dominant)	cds	COL7A1	**3 months**	5 ml blood EDTA	182 900 000
COL7A1-AR	Epidermolysis Bullosa Dystrophica (Autosomal Recessive)	cds	COL7A1	**3 months**	5 ml blood EDTA	182 900 000
EBS-MUT	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	[p.Pro25Leu, p.Gly550Alafs*77 and p.Glu477Lys] and [p.Met119Thr, p.Asn123Ser, p.Arg125Cys and p.Arg125His]	KRT5 and KRT14	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000
KRT5	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	cds	KRT5	**3 months**	5 ml blood EDTA/Saliva on swab	41 100 000
KRT14	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	cds	KRT14	**2 months**	5 ml blood EDTA/Saliva on swab	41 100 000
ALDH7A1	Epilepsy Pyridoxine-Dependent (AASA Dehydrogenase deficiency)	cds	ALDH7A1	**2 months**	5 ml blood EDTA/Saliva on swab	59 400 000
FABRY	Fabry Disease (Galactosidase defiency)	cds	GLA	**4 weeks**	5 ml blood EDTA/Saliva on swab	28 100 000
FABRY-DEL	Fabry Disease (Galactosidase defiency)	Deletions	GLA	**5 weeks**	5 ml blood EDTA	20 800 000
FGD1	Faciogenital Dysplasia (Aarskog-Scott syndrome)	cds	FGD1	**3 months**	5 ml blood EDTA/Saliva on swab	63 800 000
FSHD	Facioscapulohumeral Muscular Dystrophy	Deletion D4Z4	FSHD	**4 months**	15 ml blood EDTA	40 900 000
FSHD-HAPL	Facioscapulohumeral Muscular Dystrophy	Haplotype 4qA161	FSHD	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
FSHD-FRG1	Facioscapulohumeral Muscular Dystrophy	cds	FRG1	**7 weeks**	5 ml blood EDTA	36 700 000
FV	Factor V Leiden Thrombophilia	p.Arg506Gln (p.R506Q)	F5	**3 days**	5 ml blood EDTA/Saliva on swab	4 400 000
FXII-MUT	Factor XII Deficiency	c.C46T	F12	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 500 000
FXII	Factor XII Deficiency	cds	F12	**2 months**	5 ml blood EDTA/Saliva on swab	31 900 000
MEFV-EX	Familial Mediterranean Fever (MEFV)	Exons 2, 3, 5 and 10	MEFV	**3 weeks**	5 ml blood EDTA/Saliva on swab	15 100 000
MEFV	Familial Mediterranean Fever (MEFV)	cds	MEFV	**2 months**	5 ml blood EDTA/Saliva on swab	35 800 000
PNPLA3	Fatty Liver Disease Nonalcoholic, susceptibility to	p.I148M	PNPLA3	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
SEX	Fetal gender prediction on peripheral maternal blood	DYS14	Yq	**3 days**	5 ml blood EDTA	8 400 000
FRAXA	Fragile X Syndrome (FRAXA)	Expansion CGG-Screening	FMR1	**3 weeks**	5 ml blood EDTA	8 700 000
FRDA	Friedrich´s Ataxia	Expansion GAA	FRDA	**3 weeks**	5 ml blood EDTA	13 100 000
ALX4	Frontonasal Dysplasia	cds	ALX4	**4 weeks**	5 ml blood EDTA	24 800 000
WNT7A	Fuhrmann Syndrome	cds	WNT7A	**4 weeks**	5 ml blood EDTA/Saliva on swab	17 600 000

GBA-MUT	Gaucher Disease (Types 1, 2 and 3)	c.27+1G>A, c.93_94insG, N409S and L483P (IVS2+1G>A, 84GG, N370S and L444P)	GBA	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
GBA	Gaucher Disease (Types 1, 2 and 3)	cds	GBA	**2 months**	5 ml blood EDTA/Saliva on swab	36 100 000
UGT1A1-MUT	Gilbert Syndrome	(TA)7	UGT1A1	**3 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
UGT1A1	Gilbert Syndrome	cds	UGT1A1	**2 months**	5 ml blood EDTA/Saliva on swab	35 400 000
HDC	Gilles de la Tourette Syndrome	p.Trp317Ter	HDC	**3 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
MYOC	Glaucoma 1 Open Angle (POAG)	cds	MYOC	**4 weeks**	5 ml blood EDTA/Saliva on swab	17 300 000
CYP1B1	Glaucoma Primary Congenital	cds	CYP1B1	**4 weeks**	5 ml blood EDTA/Saliva on swab	16 700 000
CYP11B	Glucocorticoid-Supressible Hyperaldosteronism	Chimerism	CYP11B1 – CYP11B2	**4 weeks**	5 ml blood EDTA	15 300 000
G6PD	Glucosa 6-fosfato deshidrogenasa, Deficiency	cds	G6PD	**2 months**	5 ml blood EDTA	33 000 000
GLUT1	GLUT1, Deficiency Syndrome	cds	SLC2A1	**3 weeks**	5 ml blood EDTA/Saliva on swab	29 000 000
GCDH	Glutaric Acidemia Type I	cds	GCDH	**4 weeks**	5 ml blood EDTA	26 400 000
PYGM-MUT	Glycogen Storage disease V (McArdle disease)	p.Arg50X, p.Gly205Ser, p.Trp798Arg, p.Tyr85X	PYGM	**3 weeks**	5 ml blood EDTA/Saliva on swab	14 400 000
PYGM	Glycogen Storage disease V (McArdle disease)	cds	PYGM	**3 months**	5 ml blood EDTA	69 800 000
GHRL-METAB	Grelina (Ghrelin, Obestatin) (Metabolic Syndrome,susceptibility to)	p.Arg51Gln and p.Leu72Met	GHRL	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
GHRL-OBES	Grelina (Ghrelin, Obestatin) (Obesity, susceptibility to)	p.Gln90Leu	GHRL	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
GHRL	Grelina (Ghrelin, Obestatin) (Obesity/Metabolic Syndrome, susceptibility to)	cds	GHRL	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 900 000
HFE	Hemochromatosis	p.C282Y, p.H63D and p.S65C	HFE	**4 days**	5 ml blood EDTA/Saliva on swab	4 400 000
HJV	Hemochromatosis juvenile (Type 2A)	p.G320V	HJV	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
FVIII-MUT	Hemophilia A	Intron inversion 22A / Intron inversion 1	F8	**3 months**	5 ml blood EDTA	22 000 000
FVIII	Hemophilia A	cds	F8	**3 months**	5 ml blood EDTA	74 900 000
FIX	Hemophilia B	cds	F9	**4 weeks**	5 ml blood EDTA/Saliva on swab	26 800 000
IL28B	Hepatitis C virus infection, response to therapy of	g.120070005C>T	IL28B	**1 week**	5 ml blood EDTA/Saliva on swab	4 700 000
ZIC3	Heterotaxy Visceral 1, X-linked (Congenital heart defects, nonsyndromic)	cds	ZIC3	**2 months**	5 ml blood EDTA	34 900 000
CCR5	HIV infection, Resistance to	Deletion	CCR5	**15 days**	5 ml blood EDTA/Saliva on swab	5 300 000
HLA-DQ	HLA-DQ, Genotyping	Complete genotyping High Resolution	HLA-DQA1 and HLA-DQB1	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
HLA-DRB1	HLA-DR, Genotyping	Complete genotyping High Resolution	HLA-DRB1	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 300 000
HPV	Human Papiloma Virus (HPV)	Complete genotyping		**4 days**	Lesion on swab/Liquid cytology	5 000 000

HTT	Huntington Disease	Expansion CAG	HTT	**4 weeks**	5 ml blood EDTA	9 400 000
LDLR	Hypercholesterolemia (Apo B)	cds	LDLR	**2 months**	5 ml blood EDTA	46 200 000
LDLR-DEL	Hypercholesterolemia (Apo B)	Deletions	LDLR	**5 weeks**	5 ml blood EDTA	23 100 000
PCSK9	Hypercholesterolemia (Apo B)	cds	PCSK9	**2 months**	5 ml blood EDTA	38 500 000
APOB	Hypercholesterolemia Familial (Apo B)	p.Arg3527Gln, p.Arg3527Trp, p.Arg3558Cys	APOB	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 400 000
CYP11B2	Hyperreninemic Hypoaldosteronism, Familial (Corticosterone Methyloxidase Type I)	cds	CYP11B2	**7 weeks**	5 ml blood EDTA/Saliva on swab	36 700 000
NR3C2	Hypertension Early-onset, Autosomal Dominant	cds	NR3C2	**2 months**	5 ml blood EDTA/Saliva on swab	33 000 000
FHH	Hypocalciuric Hypercalcemia Familiar Type I	cds	CASR	**2 months**	5 ml blood EDTA	30 100 000
FGFR3-MUT	Hypochondroplasia	p.N540K	FGFR3	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
FGFR3-EX	Hypochondroplasia	Exons 9 and 15	FGFR3	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
TGM1	Ichthyosis Lamellar 1 (LI1)	cds	TGM1	**2 months**	5 ml blood EDTA/Saliva on swab	46 200 000
ABCA12	Ichthyosis Lamellar 2 (LI2)	cds	ABCA12	**4 months**	5 ml blood EDTA	143 200 000
IKBKG-DEL	Incontinentia Pigmenti	Deletion exons 4-10	IKBKG (NEMO)	**5 weeks**	5 ml blood EDTA	20 800 000
IKBKG	Incontinentia Pigmenti	cds	IKBKG (NEMO)	**2 months**	5 ml blood EDTA/Saliva on swab	24 600 000
FGFR2	Jackson-Weiss Syndrome	cds	FGFR2	**2 months**	5 ml blood EDTA	33 000 000
KAL1	Kallmann Syndrome - Type 1 (X-Linked)	cds	KAL1	**4 weeks**	5 ml blood EDTA/Saliva on swab	33 000 000
FGFR1	Kallmann Syndrome - Type 2	cds	FGFR1	**3 months**	5 ml blood EDTA/Saliva on swab	5 900 000
PROKR2	Kallmann Syndrome - Type 3	cds	PROKR2	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000
PROK2	Kallmann Syndrome - Type 4	cds	PROK2	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000
KEARNS	Kearns-Sayre Syndrome (KSS)	Deletions	mtDNA	**5 weeks**	5 ml blood EDTA	20 800 000
TBCE	Kenny-Caffey Syndrome (KCS1)	c.155del12nt	TBCE	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
KFSDX	Keratosis Follicularis Spinulosa Decalvans	cds	MBTPS2	**2 months**	5 ml blood EDTA	37 400 000
KIR	KIR Genotyping (Repetitive Abortions)	Genotyping	KIR	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 300 000
MCM6	Lactose Intolerance (LCT)	c.-13910C>T and c.-22018G>A	MCM6	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
LCA6	Leber congenital amaurosis (LCA 6)	cds	RPGRIP1	**2 months**	5 ml blood EDTA	68 300 000
LCA14	Leber congenital amaurosis 14 (LCA 14)	cds	LRAT	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 500 000
LCA2	Leber congenital amaurosis 2 (LCA 2)	cds	RPE65	**6 weeks**	5 ml blood EDTA	29 700 000
LHON-1MUT	Leber Hereditary Optic Neuropathy (LHON) (Leber Optic Atrophy)	m.11778G>A	mtDNA	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
LHON-3MUT	Leber Hereditary Optic Neuropathy (LHON) (Leber Optic Atrophy)	m.11778G>A, m.14484T>C and m.3460G>A	mtDNA	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 500 000
LERI	Leri-Weill, Dyschondrosteosis	cds	SHOX	**4 weeks**	5 ml blood EDTA	26 400 000
LERI-DEL	Leri-Weill, Dyschondrosteosis	Deletions/Duplications	SHOX	**5 weeks**	5 ml blood EDTA	20 800 000
BCR-ABL	Leucemia Mieloide Crónica	Fusion BCR/ABL	BCR/ABL	**2 weeks**	5 ml blood EDTA on ice	8 300 000
PML-RARA	Leukemia Acute Promyelocytic, PML/RARA type	Fusion PML/RARA	PML/RARA	**4 weeks**	5 ml blood EDTA on ice	8 300 000
BCR-ABL	Leukemia Chronic Myeloid	Fusion BCR/ABL	BCR/ABL	**2 weeks**	5 ml blood EDTA on ice	8 300 000

LAD	Leukocyte adhesion deficiency (LAD)	cds	ITGB2	**2 months**	5 ml blood EDTA/Saliva on swab	41 400 000
EIF2B5	Leukoencephalopathy with vanishing white matter	cds	EIF2B5	**3 months**	5 ml blood EDTA	34 300 000
EIF2B2	Leukoencephalopathy with vanishing white matter	cds	EIF2B2	**2 months**	5 ml blood EDTA/Saliva on swab	28 600 000
EIF2B4	Leukoencephalopathy with vanishing white matter	cds	EIF2B4	**3 months**	5 ml blood EDTA	30 300 000
EIF2B3	Leukoencephalopathy with vanishing white matter	cds	EIF2B3	**3 months**	5 ml blood EDTA	34 900 000
EIF2B1	Leukoencephalopathy with vanishing white matter	cds	EIF2B1	**2 months**	5 ml blood EDTA	28 600 000
SCNN1B	Liddle Syndrome	cds	SCNN1B	**2 months**	5 ml blood EDTA	38 500 000
SCNN1G	Liddle Syndrome	cds	SCNN1G	**2 months**	5 ml blood EDTA	39 600 000
LPL-MUT	Lipoprotein Llipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	p.G188E	LPL	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
LPL	Lipoprotein Llipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	cds	LPL	**4 weeks**	5 ml blood EDTA	37 800 000
LPL-DEL	Lipoprotein Llipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	Deletions/Duplications	LPL	**5 weeks**	5 ml blood EDTA	23 100 000
HNPCC-INES	Lipoprotein Llipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	Microsatellites inestability studies		**4 weeks**	5 ml blood EDTA	19 700 000
LQT1	Long QT Syndrome (LQT1)	cds	KCNQ1	**2 months**	5 ml blood EDTA	43 800 000
LQT2	Long QT Syndrome (LQT2)	cds	KCNH2	**2 months**	5 ml blood EDTA	42 700 000
LQT3	Long QT Syndrome (LQT3)	cds	SCN5A	**3 months**	5 ml blood EDTA	63 800 000
PMS2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	PMS2	**3 months**	5 ml blood EDTA	52 400 000
SCA3	Machado-Joseph Disease (MJD) (SCA3)	Expansion CAG	ATXN3	**4 weeks**	5 ml blood EDTA	10 200 000
HIPERTERM	Malignant Hyperthermia, Susceptibility to	p.R614C/L and p.G2434R	RYR1	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
FBN1	Marfan Syndrome	cds	FBN1	**2 months**	5 ml blood EDTA	99 100 000
FBN1-DEL	Marfan Syndrome	Deletions	FBN1	**5 weeks**	5 ml blood EDTA	30 800 000
LDS2A	Marfan Type 2A Syndrome (Loeys-Dietz, Syndrome) (LDS2A)	cds	TGFBR1	**7 weeks**	5 ml blood EDTA/Saliva on swab	24 600 000
LDS2B	Marfan Type 2B Syndrome (Loeys-Dietz, Syndrome) (LDS2B)	cds	TGFBR2	**7 weeks**	5 ml blood EDTA/Saliva on swab	24 600 000
SIL1	Marinesco-Sjögren Syndrome	cds	SIL1	**2 months**	5 ml blood EDTA	38 300 000
KIT-MUT	Mastocytosis	p.Lys509Ile	KIT	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
KIT	Mastocytosis	cds	KIT	**3 months**	5 ml blood EDTA	69 600 000
KIT-DIFUS	Mastocytosis cutaneous diffuse	p.Ala533Asp	KIT	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
KIT-HEMAT	Mastocytosys with associated hematologic disorder	p.Asp816Val	KIT	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
CONT-MAT	Maternal Cell Contamination, on prenatal samples	STRs				7 600 000

PYGM-MUT	McArdle Disease (Glycogen Storage Disease V, Myophosphorylase Deficiency)	p.Arg50X, p.Gly205Ser, p.Trp798Arg and p.Tyr85X	PYGM	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 800 000
PYGM	McArdle Disease (Glycogen Storage Disease V, Myophosphorylase Deficiency)	cds	PYGM	**3 months**	5 ml blood EDTA	63 800 000
MELAS	MELAS Syndrome (Encephalopaty mitochondrial)	m.3243A>G, m.3252A>G, m.3256C>T, m.3271T>C and m.3291T>C	MT-TL1 (TRNL1)	**3 weeks**	5 ml blood EDTA/Saliva on swab	8 900 000
ATP7A	Menkes Disease	cds	ATP7A	**3 weeks**	5 ml blood EDTA	72 700 000
ATP7A-DEL	Menkes Disease	Big Deletions	ATP7A	**5 weeks**	5 ml blood EDTA	20 800 000
CYB5R3	Methemoglobinemia	cds	CYB5R3	**4 weeks**	5 ml blood EDTA/Saliva on swab	24 800 000
AZF	Microdeletions Y chromosome (Spermatogenic Failure non-obstructive, Azoospermia)	AZFa, AZFb and AZFc	Yq	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 900 000
MODY2	MODY 2	cds	GCK	**4 weeks**	5 ml blood EDTA/Saliva on swab	26 400 000
MODY3	MODY 3	cds	HNF1A	**4 weeks**	5 ml blood EDTA/Saliva on swab	26 400 000
MTHFR-MUT	MTHFR Methylenetetrahydrofolate reductase (Homocystinuria)	c.677C>T and c.1298A>C	MTHFR	**3 days**	5 ml blood EDTA/Saliva on swab	4 700 000
MTHFR-SEQ	MTHFR Methylenetetrahydrofolate reductase (Homocystinuria)	cds	MTHFR	**2 months**	5 ml blood EDTA/Saliva on swab	46 200 000
ARSB	Mucopolysaccharidosis Type VI (Maroteaux-Lamy Syndrome)	cds	ARSB	**4 weeks**	5 ml blood EDTA/Saliva on swab	33 000 000
GALNS	Mucopolysacharidosis Type IV A (Morquio Syndrome A, Galactosamine-6-Sulfatase Deficiency)	cds	GALNS	**2 months**	5 ml blood EDTA	50 600 000
GLB1	Mucopolysacharidosis Type IV B (Morquio Syndrome B)	cds	GLB1	**2 months**	5 ml blood EDTA	63 800 000
MUENKE	Muenke Syndrome	cds	FGFR3	**2 months**	5 ml blood EDTA/Saliva on swab	36 500 000
RYR1-SEPN1	Multiminicore Disease (MmD)	Muts in SEPN1: M1V, G273E, H293R, G315S, N340I, R439X, W453S, U462G, U462X, R466Q, c.22dup10bp, c.713-714insA, c.1446delC; Muts en RYR1: R109W, A1577T, N2283H, P3527S, IVS101+2990A>G	SEPN1 and RYR1	**4 weeks**	5 ml blood EDTA/Saliva on swab	20 400 000
RYR1	Multiminicore Disease (MmD)	cds	RYR1	**5 months**	5 ml blood EDTA	222 600 000
RYR1-EX1	Multiminicore Disease (MmD)	Exons 2 and 6-18	RYR1	**7 weeks**	5 ml blood EDTA	30 800 000
RYR1-EX2	Multiminicore Disease (MmD)	Exons 39-48	RYR1	**7 weeks**	5 ml blood EDTA	31 800 000
RYR1-EX3	Multiminicore Disease (MmD)	Exons 85-104	RYR1	**2 months**	5 ml blood EDTA	50 600 000
SEPN1	Multiminicore Disease (MmD)	cds	SEPN1	**2 months**	5 ml blood EDTA/Saliva on swab	46 200 000
JAK2-MI	Myelofibrosis	p.V617F	JAK2	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 600 000
JAK2	Myeloproliferative disorder with erythrocytosis JAK2 related (Polycythemia Vera, Thrombocythemia Essential and Myelofibrosis)	p.V617F	JAK2	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 600 000

DNM2	Myopathy Centronuclear	cds	DNM2	**3 months**	5 ml blood EDTA	57 200 000
CLCN1	Myotonia Congenita Dominant (Thomsen Disease; Becker Disease)	cds	CLCN1	**2 months**	5 ml blood EDTA/Saliva on swab	30 800 000
DM-STEINERT	Myotonic Dystrophy Type 1 (DM1) (Steinert disease)	Expansion CTG	DMPK	**3 weeks**	5 ml blood EDTA	11 400 000
NBLAST1	Neuroblastoma, Susceptibility to (NBLST1)	cds	KIF1B	**5 months**	5 ml blood EDTA	134 400 000
NBLAST2	Neuroblastoma, Susceptibility to (NBLST2)	cds	PHOX2B	**4 weeks**	5 ml blood EDTA/Saliva on swab	15 300 000
NBLAST3	Neuroblastoma, Susceptibility to (NBLST3)	cds	ALK	**4 months**	5 ml blood EDTA	107 900 000
PMP22-STRS	Neuropathy Hereditary With Liability to Pressure Palsies (HNPP)	STRs	PMP22	**4 weeks**	5 ml blood EDTA/Saliva on swab	11 800 000
PMP22	Neuropathy Hereditary With Liability to Pressure Palsies (HNPP)	cds	PMP22	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 100 000
OTOF-MUT	Neurosensory Nonsyndromic Recessive Deafness	p.Gln829Ter and p.Pro1825Ala	OTOF	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
OTOF	Neurosensory Nonsyndromic Recessive Deafness	cds	OTOF	**4 months**	5 ml blood EDTA	107 900 000
GJB2-MUT	Neurosensory Nonsyndromic Recessive Deafness	c.35delG	GJB2 (Conexina 26)	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
GJB2	Neurosensory Nonsyndromic Recessive Deafness	cds	GJB2 (Conexina 26)	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
NCS	Neutropenia Severe Congenital	cds	ELANE (ELA2)	**4 weeks**	5 ml blood EDTA/Saliva on swab	20 600 000
SMPD1	Niemann-Pick Disease (Types A and B)	cds	SMPD1	**4 weeks**	5 ml blood EDTA/Saliva on swab	29 700 000
PTPN11	Noonan Syndrome	cds	PTPN11	**4 weeks**	5 ml blood EDTA	33 600 000
MC4R	Obesity Morbid	cds	MC4R	**2 weeks**	5 ml blood EDTA/Saliva on swab	12 300 000
DMO	Oculopharyngeal Muscular Dystrophy	Expansion GCG	PABPN1	**3 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
OPA1-EX	Optic Atrophy Dominant Type 1 (ADOA)	Exons 8-16 and 27-28	OPA1	**4 weeks**	5 ml blood EDTA/Saliva on swab	27 900 000
OPA1	Optic Atrophy Dominant Type 1 (ADOA)	cds	OPA1	**2 months**	5 ml blood EDTA	66 100 000
OPA1-DEL	Optic Atrophy Dominant Type 1 (ADOA)	Deletions	OPA1	**5 weeks**	5 ml blood EDTA	23 100 000
OPA3-MUT	Optic Atrophy Type 3 with cataract	p.G93S and p.Q105E	OPA3	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
OPA3	Optic Atrophy Type 3 with cataract	cds	OPA3	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 900 000
OTC	Ornithine transcarbamylase deficiency	cds	OTC	**2 months**	5 ml blood EDTA	32 300 000
OTC-DEL	Ornithine transcarbamylase deficiency	Deletions	OTC	**5 weeks**	5 ml blood EDTA	23 100 000
COL1A1	Osteogenesis Imperfecta	cds	COL1A1	**4 months**	5 ml blood EDTA	59 000 000
COL1A2	Osteogenesis Imperfecta	cds	COL1A2	**4 months**	5 ml blood EDTA	64 700 000
COL1A1-2	Osteogenesis Imperfecta	cds	COL1A1 and COL1A2	**5 months**	5 ml blood EDTA	121 200 000
PRSS1	Pancreatitis Hereditary	cds	PRSS1	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 200 000
PRSS1-EX	Pancreatitis Hereditary	Exons 2 and 3 (variant p.R122H included)	PRSS1	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 500 000
SPINK1	Pancreatitis Hereditary	cds	SPINK1	**4 weeks**	5 ml blood EDTA/Saliva on swab	17 100 000
SPINK1-EX	Pancreatitis Hereditary	Exon 3 (variant N34S included)	SPINK1	**4 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000

PANCR	Pancreatitis Hereditary	cds	CFTR	**2 months**	5 ml blood EDTA	51 700 000
LRRK2-MUT	Parkinson Disease 8 (Autosomal Dominant)	p.Asn1437His, p.Arg1441His, p.Arg1441Cys, p.Arg1441Gly, p.Arg1628Pro, p.Tyr1699Cys, p.Gly2019Ser, p.Ile2020Thr and p.Gly2385Arg	LRRK2	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 700 000
LRRK2	Parkinson Disease 8 (Autosomal Dominant)	cds	LRRK2	**4 months**	5 ml blood EDTA	107 900 000
PARK2	Parkinson Juvenile Disease	cds	PARK2	**4 weeks**	5 ml blood EDTA/Saliva on swab	23 500 000
PARK2-DEL	Parkinson Juvenile Disease	Deletions/Duplications	PARK2	**4 weeks**	5 ml blood EDTA	20 800 000
PFEIFFER-MUT	Pfeiffer Syndrome	p.C278F and p.C342G	FGFR2	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
PFEIFFER-2	Pfeiffer Syndrome	cds	FGFR2	**2 months**	5 ml blood EDTA	33 000 000
PFEIFFER-EX	Pfeiffer Syndrome	Exon 7 FGFR1; exon 7,8,13,14 and 15 FGFR2	FGFR1 and FGFR2	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 700 000
PFEIFFER-1	Pfeiffer Syndrome	cds	FGFR1	**3 months**	5 ml blood EDTA/Saliva on swab	37 200 000
PKU-EX	Phenylketonuria (PAH)	Exons 7,9,11 and 12	PAH	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 900 000
PKU	Phenylketonuria (PAH)	cds	PAH	**4 weeks**	5 ml blood EDTA/Saliva on swab	32 600 000
SERPINE1	Plasminogen Activator Inhibitor 1 Deficiency	c.-675_4G/5G	SERPINE1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 300 000
PKHD1	Polycystic Kidney Autosomal Recessive (ARPKD)	cds	PKHD1	**5 months**	5 ml blood EDTA	165 300 000
PKD1	Polycystic Kidney Syndrome, Autosomal Dominant	cds	PKD1	**5 months**	5 ml blood EDTA	130 000 000
PKD2	Polycystic Kidney Syndrome, Autosomal Dominant	cds	PKD2	**2 months**	5 ml blood EDTA/Saliva on swab	38 500 000
JAK2-PV	Polycythemia Vera	p.V617F	JAK2	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 600 000
PPOX	Porphyria Variegata	cds	PPOX	**4 weeks**	5 ml blood EDTA/Saliva on swab	31 700 000
PW-UPD	Prader Willi Syndrome	Uniparental Disomy STRs	15q11-q13	**5 weeks**	5 ml blood EDTA	10 700 000
PW-MET	Prader Willi Syndrome	Metilation	PW	**3 weeks**	5 ml blood EDTA	10 000 000
PW-DEL	Prader Willi Syndrome	Deletions, metilation and uniparental disomy	PW	**4 weeks**	5 ml blood EDTA	20 800 000
FII	Prothrombin Thrombophilia (Factor II)	c.20210G>A	F2	**3 days**	5 ml blood EDTA/Saliva on swab	4 400 000
ARSA	Pseudoarylsulfatase A Deficiency (Metachromatic leukodystrophy)	cds	ARSA	**2 months**	5 ml blood EDTA/Saliva on swab	25 000 000
NR3C2	Pseudohypoaldosteronism Type I, Autosomal Dominant	cds	NR3C2	**2 months**	5 ml blood EDTA	33 000 000
BMPR2	Pulmonary Hypertension Primary (PPH1)	cds	BMPR2	**3 months**	5 ml blood EDTA	55 700 000
BMPR2-DEL	Pulmonary Hypertension Primary (PPH1)	Deletions	BMPR2	**5 weeks**	5 ml blood EDTA	28 400 000
RHO	Retinitis Pigmentosa Autosomal Dominant	cds	RHO	**4 weeks**	5 ml blood EDTA/Saliva on swab	23 100 000
RPAR	Retinitis Pigmentosa Autosomal Recessive	cds	LRAT and RPE65	**2 months**	5 ml blood EDTA	38 500 000
LRAT	Retinitis Pigmentosa Autosomal Recessive	cds	LRAT	**3 weeks**	5 ml blood EDTA/Saliva on swab	12 300 000
MERTK	Retinitis Pigmentosa Autosomal Recessive	cds	MERTK	**2 months**	5 ml blood EDTA	49 900 000
RPE65	Retinitis Pigmentosa Autosomal Recessive	cds	RPE65	**4 weeks**	5 ml blood EDTA	29 700 000

RPLX	Retinitis Pigmentosa X-linked	cds	RP2 and RPGR	**3 months**	5 ml blood EDTA	64 900 000
RPGR	Retinitis Pigmentosa X-linked	cds	RPGR	**2 months**	5 ml blood EDTA	50 600 000
RP2	Retinitis Pigmentosa X-linked	cds	RP2	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 700 000
RS1	Retinoschisis 1 Juvenile, X-Linked	cds	RS1	**4 weeks**	5 ml blood EDTA/Saliva on swab	28 600 000
RETT	Rett Syndrome	cds	MECP2	**2 weeks**	5 ml blood EDTA/Saliva on swab	17 100 000
RETT-DEL	Rett Syndrome	Big Deletions	MECP2	**5 weeks**	5 ml blood EDTA	17 600 000
CREBBP	Rubinstein-Taybi Syndrome	cds	CREBBP	**5 months**	5 ml blood EDTA	79 300 000
CREBBP-DEL	Rubinstein-Taybi Syndrome	Deletions	CREBBP	**5 weeks**	5 ml blood EDTA	20 800 000
HRD	Sanjad-Sakati Syndrome (HRD)	c.66delAG (p.V23fs48X) and c.1113T>A (p.C371X)	TBCE	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 200 000
SLC6A4	Serotonin Transporter SERT (Obsessive-compulsive disorder)	44bp ins/del	SLC6A4 (5HTTLPR)	**3 weeks**	5 ml blood EDTA/Saliva on swab	10 200 000
SCN1A	Severe Myoclonic epilepsy of infancy (Dravet Syndrome) (SMEI)	cds	SCN1A	**2 months**	5 ml blood EDTA	82 400 000
SCN1A-DEL	Severe Myoclonic epilepsy of infancy (Dravet Syndrome) (SMEI)	Big Deletions	SCN1A	**4 weeks**	5 ml blood EDTA	20 800 000
SHOX	Short Stature, idiopathic	cds	SHOX	**4 weeks**	5 ml blood EDTA/Saliva on swab	24 200 000
SHOX-DEL	Short Stature, idiopathic	Deletions/Duplications	SHOX	**5 weeks**	5 ml blood EDTA	20 800 000
HBB-MUT	Sickle cell anemia	p.E6V	HBB	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
HBB	Sickle cell anemia	cds	HBB	**4 weeks**	5 ml blood EDTA/Saliva on swab	11 600 000
SLOS	Smith-Lemli-Opitz Syndrome (SLOS)	cds	DHCR7	**4 weeks**	5 ml blood EDTA/Saliva on swab	29 200 000
SLOS-MUT	Smith-Lemli-Opitz Syndrome (SLOS)	p.Phe302Leu and p.Arg446Gln	DHCR7	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
PLP1	Spastic Paraplegia (Pelizaeus-Merzbacher Disease)	cds	PLP1	**4 weeks**	5 ml blood EDTA/Saliva on swab	21 500 000
PLP1-DUP	Spastic Paraplegia (Pelizaeus-Merzbacher Disease)	Duplication	PLP1	**4 weeks**	5 ml blood EDTA	20 800 000
SPAST	Spastic Paraplegia Familiar (Autosomal Dominant)	cds	SPAST (SPG4)	**2 months**	5 ml blood EDTA	33 900 000
SPAST	Spastic Paraplegia Familiar (Autosomal Dominant)	Deleciones	SPAST (SPG4) and SPG3A	**2 months**	5 ml blood EDTA	23 100 000
SPH1	Spherocytosis Type 1 (SPH1)	cds	ANK1	**5 months**	5 ml blood EDTA/Saliva on swab	112 400 000
SPH2	Spherocytosis Type 2 (SPH2)	cds	SPTB	**4 months**	5 ml blood EDTA/Saliva on swab	101 100 000
SPH3	Spherocytosis Type 3 (SPH3)	cds	SPTA1	**4 months**	5 ml blood EDTA/Saliva on swab	132 800 000
SPH4	Spherocytosis Type 4 (SPH4)	cds	SLC4A1	**3 months**	5 ml blood EDTA/Saliva on swab	48 400 000
SPH5	Spherocytosis Type 5 (SPH5)	cds	EPB42	**2 months**	5 ml blood EDTA/Saliva on swab	45 800 000
SBMA	Spinal and Bulbar Muscular Atrophy of Kennedy (SBMA)	Expansion CAG	AR	**3 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
SMA-LED	Spinal Muscular Atrophy (SMA-LED) (Kugelberg-Welander Syndrome)	cds	DYNC1H1	**5 months**	5 ml blood EDTA	200 600 000
SMA-AF	Spinal Muscular Atrophy (SMA)	Deletion Exons 7 and 8 (patients)	SMN1	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000

SMA-PORT	Spinal Muscular Atrophy (SMA)	Deletion Exons 7 and 8 (carriers)	SMN1	**4 weeks**	5 ml blood EDTA	11 800 000
SCA12	Spinocerebellar Ataxia 12, SCA12	Expansion CAG	PPP2R2B	**4 weeks**	5 ml blood EDTA	10 700 000
SCA1	Spinocerebellar Ataxia Dominant SCA1	Expansion CAG	ATXN1	**4 weeks**	5 ml blood EDTA	10 200 000
SCA17	Spinocerebellar Ataxia Dominant SCA17	Expansion CAG/CAA	TBP	**4 weeks**	5 ml blood EDTA	10 200 000
SCA2	Spinocerebellar Ataxia Dominant SCA2	Expansion CAG	ATXN2	**4 weeks**	5 ml blood EDTA	10 200 000
SCA3	Spinocerebellar Ataxia Dominant SCA3 (Machado-Joseph Disease)	Expansion CAG	ATXN3	**4 weeks**	5 ml blood EDTA	10 200 000
SCA5	Spinocerebellar Ataxia Dominant SCA5	cds	SPTBN2	**3 months**	5 ml blood EDTA	125 600 000
SCA6	Spinocerebellar Ataxia Dominant SCA6	Expansion CAG	CACNA1A	**4 weeks**	5 ml blood EDTA	10 200 000
SCA7	Spinocerebellar Ataxia Dominant SCA7	Expansion CAG	ATXN7	**4 weeks**	5 ml blood EDTA	10 200 000
SCA8	Spinocerebellar Ataxia Dominant SCA8	Expansion CTG	ATXN8OS	**4 weeks**	5 ml blood EDTA	10 200 000
TNFRSF	Squamous Cell Carcinoma, Head and Neck	cds	TNFRSF10B	**7 weeks**	5 ml blood EDTA/Saliva on swab	36 700 000
ING1	Squamous Cell Carcinoma, Head and Neck	cds	ING1	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 900 000
STICKLER1	Stickler Syndrome Type 1	cds	COL2A1	**2 months**	5 ml blood EDTA	77 100 000
STICKLER1-EX	Stickler Syndrome Type 1 (Arthroophthalmopathy)	Exons 1,2,3,4,5,7,9,10,11,12,13,15 ,17,18,19,20,21,24,25,26,32 ,35,36,37,40,41,44,45,46,47 ,50,51,52,53 and 54	COL2A1	**2 months**	5 ml blood EDTA	67 800 000
STICKLER2	Stickler Syndrome Type 2	cds	COL11A1	**4 months**	5 ml blood EDTA	160 900 000
SVAS	Supravalvar Aortic Stenosis (SVAS)	cds	ELN	**3 months**	5 ml blood EDTA	69 100 000
SVAS	Supravalvular Aortica Stenosis (SVAS)	Deletions/Duplications	ELN	**5 weeks**	5 ml blood EDTA	17 600 000
HEXA	Tay-Sachs Disease	cds	HEXA	**2 months**	5 ml blood EDTA	44 000 000
HHT	Telangiectasia Hereditary Hemorragic (Osler-Weber-Rendu Disease) (HHT)	cds	ENG and ACVRL1	**2 months**	5 ml blood EDTA	55 000 000
HHT-DEL	Telangiectasia Hereditary Hemorragic (Osler-Weber-Rendu Disease) (HHT)	Big Deletions	ENG and ACVRL1	**5 weeks**	5 ml blood EDTA	23 100 000
HHT1	Telangiectasia Hereditary Hemorragic (Osler-Weber-Rendu Disease) (HHT1)	cds	ENG	**2 months**	5 ml blood EDTA/Saliva on swab	37 400 000
HHT2	Telangiectasia Hereditary Hemorragic (Osler-Weber-Rendu Disease) (HHT2)	cds	ACVRL1	**5 weeks**	5 ml blood EDTA/Saliva on swab	27 700 000
JPHT	Telangiectasia Hereditary Hemorragic (Osler-Weber-Rendu Disease) (JPHT, HHT, Juvenile polyposis)	cds	SMAD4	**2 months**	5 ml blood EDTA/Saliva on swab	34 700 000
TANATOF-1	Thanatophoric Dysplasia Type I	p.R248C and p.Y373C	FGFR3	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
TANATOF-1y2	Thanatophoric Dysplasia Type I and Type II	cds	FGFR3	**2 months**	5 ml blood EDTA/Saliva on swab	36 500 000
TANATOF-2	Thanatophoric Dysplasia Type II	p.K650E	FGFR3	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 700 000
JAK2-TE	Thrombocythemia Essential	p.V617F	JAK2	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 600 000
THRB	Thyroid Hormone Resistance	cds	THRB	**7 weeks**	5 ml blood EDTA/Saliva on swab	24 200 000
TCOF1	Treacher Collins Syndrome	cds	TCOF1	**2 months**	5 ml blood EDTA	58 500 000
TRPS1	Trichorhinophalangeal Syndrome	cds	TRPS1	**2 months**	5 ml blood EDTA	48 400 000
FMO3	Trimethylaminuria	cds	FMO3	**4 weeks**	5 ml blood EDTA/Saliva on swab	25 200 000

ET-SEQ	Tuberous Sclerosis	cds	TSC1 and TSC2	**4 weeks**	5 ml blood EDTA	98 400 000
TSC1	Tuberous Sclerosis	cds	TSC1	**3 months**	5 ml blood EDTA	50 600 000
TSC2	Tuberous Sclerosis	cds	TSC2	**3 months**	5 ml blood EDTA	66 600 000
TSC1-DEL	Tuberous Sclerosis	Deletions	TSC1	**5 weeks**	5 ml blood EDTA	20 800 000
TSC2-DEL	Tuberous Sclerosis	Deletions	TSC2	**5 weeks**	5 ml blood EDTA	20 800 000
BEST1	Vitelliform Macular Distrophy (Best disease)	cds	BEST1 (VMD2)	**3 months**	5 ml blood EDTA	50 600 000
VHL	Von Hippel Lindau Disease	cds	VHL	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 900 000
VHL-DEL	Von Hippel Lindau Disease	Deletions/Duplications	VHL	**5 weeks**	5 ml blood EDTA	20 800 000
VWF1	Von Willebrand Disease Type 1 (VWD1)	cds	VWF	**4 months**	5 ml blood EDTA	119 000 000
VWF1-EX	Von Willebrand Disease Type 1 (VWD1)	Exons 18-28	VWF	**2 months**	5 ml blood EDTA/Saliva on swab	28 600 000
VWF2-AD	Von Willebrand Disease Type 2A (VWD2)	Exon 28	VWF	**2 weeks**	5 ml blood EDTA/Saliva on swab	18 900 000
VWF2-AR	Von Willebrand Disease Type 2A (VWD2)	Exons 11-16, 22, 25-27 and 52	VWF	**2 months**	5 ml blood EDTA/Saliva on swab	28 600 000
VW2N	Von Willebrand Disease Type 2N (VWDN)	Exons 18-20	VWF	**2 weeks**	5 ml blood EDTA	10 900 000
VWF3	Von Willebrand Disease Type 3 (VWD3)	cds	VWF	**4 months**	5 ml blood EDTA	119 000 000
SOX10-2E	Waardenburg Syndrome (Type 2E)	cds	SOX10	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000
SOX10-4C	Waardenburg Syndrome (Type 4C)	cds	SOX10	**3 weeks**	5 ml blood EDTA/Saliva on swab	16 400 000
ELN-DEL	Williams Syndrome (Williams-Beuren Syndrome)	Deletion	ELN	**5 weeks**	5 ml blood EDTA	20 800 000
ELN	Williams Syndrome (Williams-Beuren Syndrome)	cds	ELN	**3 months**	5 ml blood EDTA	69 100 000
ATP7B	Wilson Disease	cds	ATP7B	**2 months**	5 ml blood EDTA	50 600 000
ATP7B-MUT	Wilson Disease	p.H1069Q	ATP7B	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
WAS	Wiskott-Aldrich Syndrome	cds	WAS	**2 months**	5 ml blood EDTA/Saliva on swab	30 100 000
XPC	Xeroderma Pigmentosum Complementation Group C (XPC)	cds	XPC	**3 months**	5 ml blood EDTA	63 800 000
XPD	Xeroderma Pigmentosum Complementation Group D (XPD)	cds	ERCC2	**3 months**	5 ml blood EDTA	72 700 000
XPV	Xeroderma Pigmentosum Variant Type (XPV)	cds	POLH	**3 months**	5 ml blood EDTA	48 400 000
						2 100 000
						2 100 000
	CYTOGENETICS ANALYSIS					2 100 000
QF-PCR	Aneuploidy crom 13, 18, 21, X and Y	QF-PCR		**3 days**	2 ml AF/5 ml blood EDTA	5 900 000
QF-RA	Aneuploidy crom 15 and 22			**3 days**	Tissues -abortions-	12 500 000
CGH-60	CGH-array 60K - PRENATAL	Deletions/Duplications with 60k resolution (60.000 probes)	Complete genome	**2 weeks**	10 ml AF	34 300 000
CGH-60	CGH-array 60K	Deletions/Duplications with 60k resolution (60.000 probes)	Complete genome	**4 weeks**	5 ml blood EDTA	34 300 000
CGH-180	CGH-array 180K	Deletions/Duplications with 180k resolution (180.000 probes)	Complete genome	**4 weeks**	5 ml blood EDTA	40 000 000

CGH-400	CGH-array 400K	Deletions/Duplications with 400k resolution (400.000 probes)	Complete genome	**4 weeks**	5 ml blood EDTA	51 700 000
FISH-BCR	FISH BCR-ABL			**3 weeks**	5 ml blood EDTA	8 900 000
FISH-TELOM	Integrity telomeric analysis			**3 weeks**	15 ml blood HEPARIN/10 ml AF	8 900 000
CAR-VC	Karyotype chorionic villus			**3 weeks**	Chorionic villus	7 900 000
CAR-SP	Karyotype peripheral blood			**3 weeks**	15 ml blood HEPARIN	5 200 000
CAR-RA	Karyotype others tissues -abortions-			**3 weeks**	Consult	8 700 000
CAR-LA	Karyotype amniotic fluid			**3 weeks**	10 ml AF	6 500 000
						2 100 000
						2 100 000
	PREDICTIVE MEDICINE ANALYSIS					2 100 000
DIABES	Diabesity (genetic predisposition)	7 genes		**2 weeks**	5 ml blood EDTA/Saliva on swab	8 500 000
OBES-EI	Obesity (genetic predisposition) - Initial study	6 genes		**2 weeks**	5 ml blood EDTA/Saliva on swab	7 900 000
OBES-EC	Obesity (genetic predisposition) - Complete study	18 genes		**2 weeks**	5 ml blood EDTA/Saliva on swab	17 100 000
OSTEO	Osteoporosis	IVS1-440G>T (Sp1), IVS1-397T>C and IVS10+283G>A	COL1A1, ESR1 and VDR	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 100 000
OSTEO-COL1A1	Osteoporosis	IVS1-440G>T (Sp1)	COL1A1	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 800 000
OSTEO-ESR1	Osteoporosis	IVS1-397T>C	ESR1	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 800 000
OSTEO-VDR	Osteoporosis	IVS10+283G>A	VDR	**2 weeks**	5 ml blood EDTA/Saliva on swab	5 800 000
HTA	Cardiovacular risk: Hypertension	I/D (ECA), E298D (NOS3), G16R (ADRB2) anad 825C>T (GNB3)	ECA, NOS3, ADRB2 and GNB3	**2 weeks**	5 ml blood EDTA/Saliva on swab	12 500 000
TROM-EI	Cardiovascular risk: Venous thrombosis - Initial Study	1691G>A (F5), 20210G>A (F2), 677C>T (MTHFR) and 1298A>C /MTHFR)	F5, F2 and MTHFR	**1 weeks**	5 ml blood EDTA/Saliva on swab	9 300 000
TROM-EC	Cardiovascular risk: Venous thrombosis - Complete Study	1691G>A (F5), 20210G>A (F2), 677C>T (MTHFR) and 1298A>C (MTHFR)	F5, F2, MTHFR, PAI1 and FGB	**2 weeks**	5 ml blood EDTA/Saliva on swab	17 700 000
						2 100 000
						2 100 000
	PHARMACOGENETICS	**DRUGS**	**GENE**			2 100 000
CYP2C9	CYP2C9	Warfarina, Ibuprofeno, Aceclofenaco, Celecoxib	CYP2C9	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
CYP2C19	CYP2C19	Citalopram, Clopidogrel, Esomeprazol, Imipramina	CYP2C19	**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
CYP2D6*	CYP2D6*	Amitriptilina, Aripiprazol, Atomoxetina, Clomipramina	CYP2D6*	**2 weeks**	5 ml blood EDTA	9 800 000
CYP2C8	CYP2C8	Bifosfonatos	CYP2C8	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
CYP3A4	CYP3A4	Efavirenz, Docetaxel, Tacrolimus, Ciclofosfamida	CYP3A4	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
CYP3A5	CYP3A5	Tacrolimus	CYP3A5	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
CYP1A2	CYP1A2	Clopidogrel, Warfarina, Cafeina, Teofilina	CYP1A2	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
COMT	COMT	Nicotina, Antipsicoticos, Morfina, Litio, Fluvoxamina	COMT	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000

TPMT	TPMT	Azatioprina, Mercaptopurina, Tioguanina	TPMT	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
GSTP1	GSTP1	Difenhidramina, Ciclofosfamida, Dexametasona, Leucovorin, Epirubicina, Paclitaxel, Doxorubicina, Cisplatino, Ranitidina, Taxanos, Oxaliplatino, Compuestos de Platino, Fluorouracilo, Mercaptopurina, Metotrexato	GSTP1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
UGT1A1	UGT1A1	Irinotecan	UGT1A1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
NAT2	NAT2	Isoniazida, Rifamicina, Pirazinamida, Isosorbida, Hidrazalina	NAT2	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
SLC01B1	SLC01B1	Estatinas-Simvastatina	SLC01B1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
SLC6A4	SLC6A4	Antidepresivos, Ondansetron, Risperidona, Berberine, Inhibidores Selectivos Recaptación Serotonina, Clomipramina, Evodiamina, Escitalopram, Nortriptylina	SLC6A4	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
SCNA5	SCNA5	Tioridazina, Claritromicina	SCNA5	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
HTR2A	HTR2A	Antidepresivos, citalopram, venlafaxina, Inhibidores Selectivos recaptación serotonina, Olanzapina, Risperidona, Paroxetina, Escitalopram	HTR2A	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
DRD2	DRD2	Risperidona, Nicotina, Antipsicóticos, Clorpromazina, Clozapina, Nemonaprida, Olanzapina, Bromperidol, Aripiprazole, Bupropion	DRD2	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
Factor V	Factor V	Terapia Hormonal-Estrógenos	F5	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
ADRB2	ADRB2	Hidroclorotiazida, Agentes Beta Bloqueantes Corticosteroides, Formoterol, Losartan, Risperidona, Budesonida, Indacaterol, Isoproterenol, Salbutamol, Salmeterol, Atenolol	ADRB2	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000

ADRB1	ADRB1	Hidroclorotiazida, Agentes Beta Bloqueantes Verapamilo, Losartan, Bucindolol, Carvedilol, Metoprolol, Diltiazem, Bisoprolol, Muraglitazar, Atenolol	ADRB1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
VKORC1	VKORC1	Acenocumarol, Fenprocumon, Warfarina	VKORC1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
DPYD	DPYD	Cetuximab, Oxaliplatino, Bevacizumab, Raltitrexed, Leucovorin, Tegafur Fluorouracilo, Capecitabina, Análogos Pirimidina	DPYD	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
IL-28	IL-28	Boceprevir, Telaprevir, PegInterferon	IL-28	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
HLA-B*5701	HLA-B*5701	Abacavir	HLA-B*5701	**2 weeks**	5 ml blood EDTA/Saliva on swab	11 400 000
HLA-A*1502	HLA-A*1502	Carbamazepina, Fenitoina	HLA-A*1502	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
MTHFR	MTHFR	Azatioprina, Fluorouracilo, Acido Fólico, Leucovorin, Mercaptopurina, Metotrexato	MTHFR	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
VDR	VDR	A. Etidrónico, Zoledronato, Medroxiprogesterona Calcio, Alendronato, Midazolam, Clodronato, Bifosfonatos, Estrógenos conjugados, Raloxifeno	VDR	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
ALOX5	ALOX5	Montelukast	ALOX5	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
PTGS2	PTGS2	AINES, Rofecoxib, Coxibs, Aspirina Ibuprofeno, A Grasos poliinsaturados omega-3	PTGS2	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
HMGCoA	HMGCoA	Estatinas	HMGCoA	**2 weeks**	5 ml blood EDTA/Saliva on swab	9 800 000
ABCB1	ABCB1	Actinomicina, Aldosterona, Amitriptilina, Atorvastatina, Bromperidol, Carbamazepina, Clorpromacina, Citalopram, Clopidogrel, Cortocosteroides, Ciclosporina, Fenitoina, Dexametasona, Digoxina, Diltiazem, Eritromicina, Indinavir, Lansoprazol, Tetraciclina, etc	ABCB1	**2 weeks**	5 ml blood EDTA/Saliva on swab	6 700 000
FGEN-HORM	Hormone Therapy Pharmacogenomics			**2 weeks**	5 ml blood EDTA/Saliva on swab	11 400 000
FGEN-DEPRE	SSRI Antidepressants Pharmacogenomics			**2 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000

						2 100 000
						2 100 000
	ONCOLOGY ANALYSIS					0
PML-RARA	Acute Promyelocytic Leukemia	Fusion PML/RARA	PML/RARA	**4 weeks**	5 ml blood EDTA	8 300 000
BRAF	BRAF	cds	BRAF	**2 months**	5 ml blood EDTA/Saliva on swab	52 800 000
BRAF-MUT	BRAF, Mutation	p.V600E	BRAF	**2 weeks**	5 ml blood EDTA/Saliva on swab	7 000 000
MAMA	Breast-ovarian cancer familial (BRCA1 and BRCA2)	cds	BRCA1 and BRCA2	**3 months**	10 ml blood EDTA	116 800 000
MAMA-LOH	Breast-ovarian cancer familial (BRCA1 and BRCA2)	Loss of heterozygosity (LOH)	BRCA1 and BRCA2	**4 weeks**	Consult	12 300 000
BRCA1-DEL	Breast-ovarian cancer familial (BRCA1 and BRCA2)	Deletions	BRCA1 and BRCA2	**5 weeks**	5 ml blood EDTA	30 800 000
BRCA1	Breast-ovarian cancer familial (BRCA1)	cds	BRCA1	**6 weeks**	5 ml blood EDTA	53 900 000
BRCA1-EX	Breast-ovarian cancer familial (BRCA1)	Exon 10	BRCA1	**3 weeks**	5 ml blood EDTA/Saliva on swab	22 000 000
BRCA2	Breast-ovarian cancer familial (BRCA2)	cds	BRCA2	**2 months**	5 ml blood EDTA	72 700 000
BRCA2-EX	Breast-ovarian cancer familial (BRCA2)	Exon 11	BRCA2	**3 weeks**	5 ml blood EDTA/Saliva on swab	24 200 000
MUTYH-MUT	Colorectal Adenomatus Polyptosis, Autosomal Recessive	p.Y165C and p.G382C	MUTYH (MYH)	**2 weeks**	5 ml blood EDTA/Saliva on swab	11 400 000
MUTYH	Colorectal Adenomatus Polyptosis, Autosomal Recessive	cds	MUTYH (MYH)	**2 months**	5 ml blood EDTA	41 800 000
COWDEN-PTEN	Cowden disease	cds	PTEN	**4 weeks**	5 ml blood EDTA/Saliva on swab	27 700 000
COWDEN-BMPR1A	Cowden disease	cds	BMPR1A	**3 months**	5 ml blood EDTA	41 800 000
COWDEN	Cowden disease	Big Deletions	PTEN and BMPR1A	**4 weeks**	5 ml blood EDTA	28 400 000
APC	Familial Adenomatous Polyposis (APC)	cds	APC	**2 months**	10 ml blood EDTA	59 400 000
APC-MUT	Familial Adenomatous Polyposis (APC)	Exon 15 region (p.Ile1307Lys and p.Glu1309AspfsX4 included)	APC	**3 weeks**	5 ml blood EDTA/Saliva on swab	8 300 000
APC-EX	Familial Adenomatous Polyposis (APC)	Exon 15	APC	**4 weeks**	10 ml blood EDTA	37 800 000
APC-DEL	Familial Adenomatous Polyposis (APC)	Big Deletions	APC	**5 weeks**	5 ml blood EDTA	23 100 000
CDH1	Gastric Cancer Hereditary Diffuse	cds	CDH1	**2 months**	5 ml blood EDTA/Saliva on swab	52 800 000
PTCH1	Gorlin Syndrome	cds	PTCH1	**2 months**	5 ml blood EDTA	59 400 000
KRAS	KRAS, Mutations	cds	KRAS	**5 weeks**	5 ml blood EDTA/Saliva on swab	23 700 000
KRAS-EX	KRAS, Mutations	Codons 12, 13 and 61	KRAS	**3 weeks**	5 ml blood EDTA/Saliva on swab	9 400 000
BCR-ABL	Leucemia Mieloide Crónica	Fusion BCR/ABL	BCR/ABL	**2 months**	5 ml blood EDTA	8 300 000
TP53	Li-Fraumeni, Syndrome	cds	p53 (TP53)	**4 weeks**	5 ml blood EDTA/Saliva on swab	22 000 000
HNPCC	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	MLH1, MSH2 and MSH6	**4 months**	10 ml blood EDTA	92 900 000
MLH1	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	MLH1	**4 weeks**	10 ml blood EDTA	36 500 000
MSH2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	MSH2	**4 weeks**	10 ml blood EDTA	31 200 000
MSH6	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	MSH6	**6 weeks**	10 ml blood EDTA	29 400 000

MLH1-MSH2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	Big Deletions	MLH1 and MSH2	**4 weeks**	5 ml blood EDTA	20 800 000
HNPCC-INES	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	Microsatellite instability (MSI)		**4 weeks**	Tumour sample and 5 ml blood EDTA	19 700 000
PMS2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	cds	PMS2	**3 months**	5 ml blood EDTA	52 400 000
FMTC	Medullary thyroid carcinoma (FMTC)	cds	RET	**3 months**	5 ml blood EDTA	53 900 000
FMTC-EX	Medullary thyroid carcinoma (FMTC)	Exons 10, 11, 13, 14, 15 and 16	RET	**3 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
P16	Melanoma Hereditary	cds	p16 (CDKN2A)	**4 weeks**	5 ml blood EDTA/Saliva on swab	16 200 000
MEN1	MEN 1 - Multiple Endocrine Neoplasia Type I	cds	MEN1	**4 weeks**	5 ml blood EDTA	20 400 000
MEN1-DEL	MEN 1 - Multiple Endocrine Neoplasia Type I	Big Deletions	MEN1	**5 weeks**	5 ml blood EDTA	20 800 000
MEN2A	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	cds	RET	**3 months**	5 ml blood EDTA	53 900 000
MEN2A-2EX	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	Exons 10 and 11	RET	**3 weeks**	5 ml blood EDTA/Saliva on swab	8 700 000
MEN2A-4EX	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	Exons 10, 11, 13, 14, 15 and 16	RET	**4 weeks**	5 ml blood EDTA/Saliva on swab	13 100 000
MEN2B	MEN 2B -Multiple Endocrine Neoplasia Type IIB	cds	RET	**3 months**	5 ml blood EDTA	53 900 000
MEN2B-EX	MEN 2B -Multiple Endocrine Neoplasia Type IIB	Exons 15 and 16 (p.M918T and p.A883F)	RET	**3 weeks**	5 ml blood EDTA/Saliva on swab	8 700 000
NBLAST1	Neuroblastoma, Susceptibility to (NBLST1)	cds	KIF1B	**5 months**	5 ml blood EDTA	134 400 000
NBLAST2	Neuroblastoma, Susceptibility to (NBLST2)	cds	PHOX2B	**4 weeks**	5 ml blood EDTA/Saliva on swab	15 300 000
NBLAST3	Neuroblastoma, Susceptibility to (NBLST3)	cds	ALK	**4 months**	5 ml blood EDTA	107 900 000
NF2	Neurofibromatosis Type II (NF2)	cds	NF2	**2 months**	5 ml blood EDTA	37 400 000
NF2-DEL	Neurofibromatosis Type II (NF2)	Big Deletions	NF2	**5 weeks**	5 ml blood EDTA	20 800 000
NF1	Neurofribromatosis Type I (NF1)	cds	NF1	**5 months**	5 ml blood EDTA	69 300 000
NF1-DEL	Neurofribromatosis Type I (NF1)	Big Deletions	NF1	**4 weeks**	5 ml blood EDTA	20 800 000
NSCLC	Non Small Cell Lung Cancer (NSCLC)	Exons 18-21	EGFR	**4 weeks**	5 ml blood EDTA/Saliva on swab	19 700 000
STK11	Peutz-Jeghers Syndrome	cds	STK11	**2 months**	5 ml blood EDTA/Saliva on swab	27 700 000
SDHD	Pheochromocytoma and/or Paragaglioma	cds	SDHD	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 900 000
SDHB	Pheochromocytoma and/or Paragaglioma	cds	SDHB	**4 weeks**	5 ml blood EDTA/Saliva on swab	27 700 000
SDHC	Pheochromocytoma and/or Paragaglioma	cds	SDHC	**4 weeks**	5 ml blood EDTA/Saliva on swab	22 600 000
RB1	Retinoblastoma	cds	RB1	**3 months**	5 ml blood EDTA	50 600 000
RB1-DEL	Retinoblastoma	Big Deletions	RB1	**5 weeks**	5 ml blood EDTA	20 800 000
TNFRSF	Squamous cell carcinoma of the head and neck	cds	TNFRSF10B	**7 weeks**	5 ml blood EDTA/Saliva on swab	36 700 000
ING1	Squamous cell carcinoma of the head and neck	cds	ING1	**4 weeks**	5 ml blood EDTA/Saliva on swab	18 900 000
VHL	Von Hippel Lindau Disease	cds	VHL	**2 weeks**	5 ml blood EDTA/Saliva on swab	10 900 000
VHL-DEL	Von Hippel Lindau Disease	Deletions/Duplications	VHL	**5 weeks**	5 ml blood EDTA	20 800 000