

	MOLECULAR GENETIC ANALYSIS	MIM	ANALYSIS	GENE	TIME since sample reception*	SAMPLE
MUT-PUNT	Pointed Mutational Analysis (any gene from this list)		Any locus		2 weeks	5 ml blood EDTA/Saliva on swab
SRY-MUT	46,XX True Hermaphroditism, SRY-positive (Gonadal Dysgenesis)	400045	Detection	SRY	2 weeks	5 ml blood EDTA/Saliva on swab
ACH	Achondroplasia	100800	p.G380R and p.G375C	FGFR3	2 weeks	5 ml blood EDTA/Saliva on swab
PML-RARA	Acute Promyelocytic Leukemia	102578	Fusion PML/RARA	PML/RARA	4 weeks	5 ml blood EDTA on ice
MCADD	ACYL-COA Dehydrogenase Medium-Chain, Deficiency of	201450	c.985A>G (p.Lys304Glu)	ACADM	3 weeks	5 ml blood EDTA/Saliva on swab
MCADD-SEQ	ACYL-COA Dehydrogenase Medium-Chain, Deficiency of	201450	cds	ACADM	2 months	5 ml blood EDTA
ADIPOQ	Adiponectin (Hipopadiponectinemia)	612556	c.276G>T	ADIPOQ	1 week	5 ml blood EDTA/Saliva on swab
CYP21A2-2MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	201910	IVS2-13A/C>G and c.G110del8nt	CYP21A2	2 weeks	5 ml blood EDTA/Saliva on swab
CYP21A2-8MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	201910	p.Pro30Leu, c.293-13A/C>G, p.Val281Leu, p.Leu307PhefsX6,	CYP21A2	4 weeks	5 ml blood EDTA
CYP21A2-1MUT	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	201910	p.Val281Leu	CYP21A2	2 weeks	5 ml blood EDTA/Saliva on swab
CYP21A2	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	201910	cds	CYP21A2	5 weeks	5 ml blood EDTA
CYP21A2-DEL	Adrenal hyperplasia congenital (due to 21-hydroxylase deficiency)	201910	Deletions	CYP21A2	5 weeks	5 ml blood EDTA
ABCD1-SEQ	Adrenoleukodystrophy (X-linked)	300100	cds	ABCD1	4 weeks	5 ml blood EDTA/Saliva on swab
ABCD1-DEL	Adrenoleukodystrophy (X-linked)	300100	Deletions	ABCD1	5 weeks	5 ml blood EDTA
FGA	Afibrinogenemia, congenital	202400	cds	FGA	4 weeks	5 ml blood EDTA/Saliva on swab
FGB	Afibrinogenemia, congenital	202400	cds	FGB	4 weeks	5 ml blood EDTA/Saliva on swab
FGG	Afibrinogenemia, congenital	202400	cds	FGG	4 weeks	5 ml blood EDTA/Saliva on swab
OCA1	Albinism oculocutaneous Type IA (OCA1A)	203100 and 606952	cds	TYR	3 weeks	5 ml blood EDTA/Saliva on swab
OCA2	Albinism oculocutaneous Type II (OCA2)	203200	cds	OCA2	2 months	5 ml blood EDTA
AAT	Alpha-1-Antitrypsin Deficiency (AAT)	613490	Variants Z, S and M	SERPINA1	2 weeks	5 ml blood EDTA/Saliva on swab
HBA	Alpha-Thalassaemia	604131	α 3.7, α 4.2, α 20.5, α SEA, α FIL and α MED	HBA	3 weeks	5 ml blood EDTA
HBA1y2	Alpha-Thalassaemia	604131	cds	HBA1 and HBA2	4 weeks	5 ml blood EDTA/Saliva on swab
ALPORT-D	Alport Syndrome (Autosomal Dominant)	104200	cds	COL4A3	4 months	5 ml blood EDTA
ALPORT-R	Alport Syndrome (Autosomal Recessive)	203780	cds	COL4A3	4 months	5 ml blood EDTA
COL4A4	Alport Syndrome (Autosomal Recessive)	203780	cds	COL4A4	4 months	5 ml blood EDTA
COL4A5	Alport Syndrome (X-linked, ATS)	301050	cds	COL4A5	4 months	5 ml blood EDTA
COL4A5-DEL	Alport Syndrome (X-linked, ATS)	301050	Deletions	COL4A5	7 weeks	5 ml blood EDTA
AD1	Alzheimer Disease Familial Type 1 (AD1)	104300	cds	APP	7 weeks	5 ml blood EDTA/Saliva on swab
AD1-EX	Alzheimer Disease Familial Type 1 (AD1)	104300	Exons 16 and 17	APP	3 weeks	5 ml blood EDTA/Saliva on swab

AD3	Alzheimer Disease Familial Type 3 (AD3)	607822	cds	PSEN1	4 weeks	5 ml blood EDTA/Saliva on swab
AD4	Alzheimer Disease Familial Type 4 (AD4)	606889	cds	PSEN2	4 weeks	5 ml blood EDTA/Saliva on swab
APOE	Alzheimer Disease Type 2 (AD2), Late Onset	104310	Genotyping E2/E3/E4	APOE	2 weeks	5 ml blood EDTA/Saliva on swab
TTR	Amyloidosis hereditary, transthyretin-related	105210	cds	TTR	3 weeks	5 ml blood EDTA/Saliva on swab
AS	Angelman Syndrome	105830	cds	UBE3A	2 months	5 ml blood EDTA/Saliva on swab
AS-UPD	Angelman Syndrome	105830	Uniparental disomy STRs	15q11-q13	5 weeks	5 ml blood EDTA
AS-MET	Angelman Syndrome	105830	Methylation	AS	3 weeks	5 ml blood EDTA
AS-DEL	Angelman Syndrome	105830	Deletions, methylation and uniparental disomy	AS	4 weeks	5 ml blood EDTA
C1NH	Angioedema hereditary types I and II (have C1 esterase inhibitor, deficiency of).	106100	cds	SERPING1 (C1NH)	5 weeks	5 ml blood EDTA/Saliva on swab
ECA	Angiotensin Converting Enzyme (ACE)	106180	I/D	ACE	1 week	5 ml blood EDTA/Saliva on swab
HLA-B27	Ankylosing Spondylitis	106300	HLA-B27	HLA-B	2 weeks	5 ml blood EDTA/Saliva on swab
APERT-MUT	Apert Syndrome	101200	p.S252W and p.P253R	FGFR2	2 weeks	5 ml blood EDTA/Saliva on swab
APERT	Apert Syndrome	101200	cds	FGFR2	2 months	5 ml blood EDTA
APOA5	Apolipoprotein A-V (APOA5)	606368	c.-1131T>C	APOA5	1 week	5 ml blood EDTA/Saliva on swab
ARVD10	Arrhythmogenic Right Ventricular Dysplasia Type 10 (ARVD10)	610193	cds	DSG2	2 months	5 ml blood EDTA
ARVD8	Arrhythmogenic Right Ventricular Dysplasia Type 8 (ARVD8)	607450	cds	DSP	3 months	5 ml blood EDTA
ARVD9	Arrhythmogenic Right Ventricular Dysplasia Type 9 (ARVD9)	609040	cds	PKP2	2 months	5 ml blood EDTA
ATM	Ataxia-telangiectasia (Louis-Bar, Syndrome)	208900	cds	ATM	5 months	5 ml blood EDTA
ATM-DEL	Ataxia-telangiectasia (Louis-Bar, Syndrome)	208900	Deletions/Duplication	ATM	5 weeks	5 ml blood EDTA
HBB-B	Beta-Thalassaemia	604131	cds + introns	HBB	2 weeks	5 ml blood EDTA/Saliva on swab
HBB-DB	Beta-Thalassaemia	604131	Deletion Spanish (delta-beta)	HBB	4 weeks	5 ml blood EDTA
HBB-B-DEL	Beta-Thalassaemia	604131	Deletions	HBB	5 weeks	5 ml blood EDTA
BRUG	Brugada Syndrome	601144	cds	SCN5A	3 months	5 ml blood EDTA
CADAS-2EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	125310	Exons 3 and 4	NOTCH3	2 weeks	5 ml blood EDTA/Saliva on swab
CADAS-4EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	125310	Exons 2, 5, 6 and 11	NOTCH3	4 weeks	5 ml blood EDTA/Saliva on swab
CADAS-6EX	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	125310	Exons 2, 3, 4, 5, 6 and 11	NOTCH3	4 weeks	5 ml blood EDTA/Saliva on swab
CADAS	CADASIL (Cerebral arteriopathy, Autosomal Dominant)	125310	cds	NOTCH3	3 months	5 ml blood EDTA
SOX9	Campomelic Dysplasia	114290	cds	SOX9	4 weeks	5 ml blood EDTA/Saliva on swab
CANAV	Canavan Disease	271900	cds	ASPA	4 weeks	5 ml blood EDTA/Saliva on swab
CMD1A	Cardiomyopathy Dilated (CMD1A)	115200	cds	LMNA	3 months	5 ml blood EDTA
CMD1D	Cardiomyopathy Dilated (CMD1D)	601494	cds	TNNT2	2 months	5 ml blood EDTA
CMH2	Cardiomyopathy Hypertrophic familial (CMH2)	115195	cds	TNNT2	2 months	5 ml blood EDTA
CMH4	Cardiomyopathy Hypertrophic familial (CMH4)	115197	cds	MYBPC	3 months	5 ml blood EDTA

CEL	Celiac disease	212750	Complete genotyping	HLA-DQA1 and HLA-DQB1	2 weeks	5 ml blood EDTA/Saliva on swab
RYR1-MUT	Central Core Disease Myopathy	117000	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
RYR1-EX	Central Core Disease Myopathy	117000	Exons 1-17, 39-48 and 90-104	RYR1	3 months	5 ml blood EDTA/Saliva on swab
CCM-DEL	Cerebral Cavernous Malformations		Deletions	KRIT1, CCM2 and PDCD10	2 months	5 ml blood EDTA
CCM1	Cerebral Cavernous Malformations 1 (CCM1)	116860	cds	KRIT1	6 weeks	5 ml blood EDTA/Saliva on swab
CCM2	Cerebral Cavernous Malformations 2 (CCM2)	603248	cds	CCM2	2 months	5 ml blood EDTA/Saliva on swab
CCM3	Cerebral Cavernous Malformations 3 (CCM3)	603285	cds	PDCD10	4 weeks	5 ml blood EDTA/Saliva on swab
CMT1A	Charcot-Marie-Tooth 1A (CMT1A)	118220	STRs	PMP22	4 weeks	5 ml blood EDTA/Saliva on swab
CMT1B	Charcot-Marie-Tooth 1B (CMT1B)	118200	cds	MPZ	4 weeks	5 ml blood EDTA/Saliva on swab
CMT1E	Charcot-Marie-Tooth 1E (CMT1E)	118300	cds	PMP22	4 weeks	5 ml blood EDTA/Saliva on swab
CMT2A2	Charcot-Marie-Tooth 2A2 (CMT2A2)	609260	cds	MFN2	3 months	5 ml blood EDTA
CMT2B-MUT	Charcot-Marie-Tooth 2B (CMT2B)	600882	p.Leu129Phe, p.Lys157Asn, p.Asn161Thr and p.Val162Met	RAB7A	2 weeks	5 ml blood EDTA/Saliva on swab
CMT2B	Charcot-Marie-Tooth 2B (CMT2B)	600882	cds	RAB7A	4 weeks	5 ml blood EDTA/Saliva on swab
CHARGE	Charge syndrome	214800	cds	CHD7	4 months	5 ml blood EDTA
CONDRO	Chondrodysplasia Punctata, X-linked dominant (Conradi-Hunermann syndrome)	302960	cds	EBP	4 weeks	5 ml blood EDTA/Saliva on swab
RUNX2	Cleidocranial Dysplasia	119600	cds	RUNX2	4 weeks	5 ml blood EDTA/Saliva on swab
RUNX2-DEL	Cleidocranial Dysplasia	119600	Deletions	RUNX2	5 weeks	5 ml blood EDTA
CBAVD	Congenital bilateral absence of vas deferens (CBAVD)	277180	36 mutations + Tn	CFTR	2 weeks	5 ml blood EDTA/Saliva on swab
EFNB1	Craniofrontonasal Dysplasia	304110	cds	EFNB1	6 weeks	5 ml blood EDTA
CCA-FGFR2	Craniosynostosis FGFR2 related		cds	FGFR2	2 months	5 ml blood EDTA
CCA-FGFR3	Craniosynostosis FGFR3 related		cds	FGFR3	2 months	5 ml blood EDTA/Saliva on swab
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
CROHN	Crohn disease, susceptibility to	266600	cds	NOD2 (CARD15)	4 weeks	5 ml blood EDTA/Saliva on swab
CROHN-MUT	Crohn disease, susceptibility to	266600	p.R720W, p.G908R and p.1007fs	NOD2 (CARD15)	2 weeks	5 ml blood EDTA/Saliva on swab
CROUZON-MUT	Crouzon syndrome	123500	p.C342Y and p.C342R	FGFR2	2 weeks	5 ml blood EDTA/Saliva on swab
CROUZON	Crouzon syndrome	123500	cds	FGFR2	2 months	5 ml blood EDTA
CROUZON-AC	Crouzon syndrome with acanthosis nigricans	612247	cds	FGFR3	2 months	5 ml blood EDTA/Saliva on swab

CURR	Currarino Syndrome (Sacral Agenesis Hereditary)	176450	cds	MXN1	3 weeks	5 ml blood EDTA/Saliva on swab
CFTR-MUT	Cystic Fibrosis	219700	36 mutations	CFTR	1 week	5 ml blood EDTA/Saliva on swab
CFTR	Cystic Fibrosis	219700	cds	CFTR	2 months	5 ml blood EDTA
SN-MYO	Deafness Type 3 (Autosomal Recessive)	600316	cds	MYO15A	4 months	5 ml blood EDTA
SN-SLC	Deafness Type 4 (Autosomal Recessive)	600791	cds	SLC26A4	2 months	5 ml blood EDTA
SORDERA	Deafness, Aminoglycoside Induced	580000	m.1555A>G, m.961_962delTinsC(n), m.7443A>G, m.7444G>A and m.7445A>G	MT-RNR1 and MT-CO1	3 weeks	5 ml blood EDTA/Saliva on swab
DRPLA	Dentatorubro-pallidoluysian Atrophy (Naito-Oyanagi disease) (DRPLA)	125370	Expansion CAG	ATN1	3 weeks	5 ml blood EDTA
DMN	Diabetes Mellitus Neonatal	610582	cds	KCNJ11	3 weeks	5 ml blood EDTA/Saliva on swab
DBA1	Diamond-Blackfan Anemia (DBA1)	105650	cds	RPS19	4 weeks	5 ml blood EDTA/Saliva on swab
DBA1-DEL	Diamond-Blackfan Anemia (DBA1)	105650	Deletions/Duplications	RPS19	5 weeks	5 ml blood EDTA
DBA10	Diamond-Blackfan Anemia (DBA10)	613309	cds	RPS26	3 weeks	5 ml blood EDTA
DBA3	Diamond-Blackfan Anemia (DBA3)	610629	cds	RPS24	6 weeks	5 ml blood EDTA
DBA4	Diamond-Blackfan Anemia (DBA4)	612527	cds	RPS17	4 weeks	5 ml blood EDTA
DBA5	Diamond-Blackfan Anemia (DBA5)	612528	cds	RPL35A	4 weeks	5 ml blood EDTA
DBA6	Diamond-Blackfan Anemia (DBA6)	612561	cds	RPL5	2 months	5 ml blood EDTA
DBA7	Diamond-Blackfan Anemia (DBA7)	612562	cds	RPL11	2 months	5 ml blood EDTA
DBA8	Diamond-Blackfan Anemia (DBA8)	612563	cds	RPS7	2 months	5 ml blood EDTA
DBA9	Diamond-Blackfan Anemia (DBA9)	613608	cds	RPS10	4 weeks	5 ml blood EDTA
CATCH22	DiGeorge syndrome, included CATCH22	188400	Deletion 22q11.2	(TBX1)	3 weeks	10 ml LA
DUCHENNE-DEL	Duchenne/Becker Muscular Dystrophy	310200	Deletions	DMD	5 weeks	5 ml blood EDTA
DUCHENNE	Duchenne/Becker Muscular Dystrophy	310200	cds	DMD	5 months	5 ml blood EDTA
DISFIBRIN	Dysfibrinogenemia	134820	cds	FGA	2 months	5 ml blood EDTA/Saliva on swab
DYT1	Early-onset Torsion Dystonia (DYT1) (Autosomal dominant)	128100	del(GAG)	TOR1A	2 weeks	5 ml blood EDTA/Saliva on swab
COL5A1	Ehlers-Danlos Syndrome, Type I	130000	cds	COL5A1	5 months	5 ml blood EDTA
COL5A2	Ehlers-Danlos Syndrome, Type I	130000	cds	COL5A2	4 months	5 ml blood EDTA
COL3A1	Ehlers-Danlos Syndrome, Type III	130020	cds	COL3A1	4 months	5 ml blood EDTA
TNXB	Ehlers-Danlos Syndrome, Type III	130020	cds	TNXB	4 months	5 ml blood EDTA
COL3A1-IV	Ehlers-Danlos Syndrome, Type IV	130050	cds	COL3A1	4 months	5 ml blood EDTA
PLOD1	Ehlers-Danlos Syndrome, Type VI	225400	cds	PLOD1	3 months	5 ml blood EDTA
EHLERS-VII	Ehlers-Danlos Syndrome, Type VII	130060	cds	COL1A1	4 months	5 ml blood EDTA
EHLERS-VII2	Ehlers-Danlos Syndrome, Type VII	130060	cds	COL1A2	4 months	5 ml blood EDTA
ADAMTS2	Ehlers-Danlos Syndrome, Type VII	225410	cds	ADAMTS2	3 months	5 ml blood EDTA
EDMD-AD	Emery-Dreifuss Muscular Dystrophy (Autosomal Dominant) (AD-EDMD)	181350	cds	LMNA	3 months	5 ml blood EDTA
EDMD-XL	Emery-Dreifuss Muscular Dystrophy, X-linked (XL-EDMD)	310300	cds	EMD	6 weeks	5 ml blood EDTA/Saliva on swab

COL7A1-AD-EX	Epidermolysis Bullosa Dystrophica (Autosomal Dominant)	131750	Exons 73, 74 and 75	COL7A1	3 weeks	5 ml blood EDTA
COL7A1-AD	Epidermolysis Bullosa Dystrophica (Autosomal Dominant)	131750	cds	COL7A1	3 months	5 ml blood EDTA
COL7A1-AR	Epidermolysis Bullosa Dystrophica (Autosomal Recessive)	226600	cds	COL7A1	3 months	5 ml blood EDTA
EBS-MUT	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	131800 (EBS-WC), 131900 (EBS-K), 131760 (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
KRT5	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	131800 (EBS-WC), 131900 (EBS-K), 131760 (EBS-DM)	cds	KRT5	3 months	5 ml blood EDTA/Saliva on swab
KRT14	Epidermolysis Bullosa Simplex, Weber-Cockayne Type (EBS-WC), Koebner Type (EBS-K) and Dowling-Meara (EBS-DM)	131800 (EBS-WC), 131900 (EBS-K), 131760 (EBS-DM)	cds	KRT14	2 months	5 ml blood EDTA/Saliva on swab
ALDH7A1	Epilepsy Pyridoxine-Dependent (AASA Dehydrogenase deficiency)	266100	cds	ALDH7A1	2 months	5 ml blood EDTA/Saliva on swab
FABRY	Fabry Disease (Galactosidase deficiency)	301500	cds	GLA	4 weeks	5 ml blood EDTA/Saliva on swab
FABRY-DEL	Fabry Disease (Galactosidase deficiency)	301500	Deletions	GLA	5 weeks	5 ml blood EDTA
FGD1	Facio-genital Dysplasia (Aarskog-Scott syndrome)	305400	cds	FGD1	3 months	5 ml blood EDTA/Saliva on swab
FSHD	Facioscapulohumeral Muscular Dystrophy	158900	Deletion D4Z4	FSHD	4 months	15 ml blood EDTA
FSHD-HAPL	Facioscapulohumeral Muscular Dystrophy	158900	Haplotype 4qA161	FSHD	2 weeks	5 ml blood EDTA/Saliva on swab
FSHD-FRG1	Facioscapulohumeral Muscular Dystrophy	158900	cds	FRG1	7 weeks	5 ml blood EDTA
FV	Factor V Leiden Thrombophilia	188055	p.Arg506Gln (p.R506Q)	F5	3 days	5 ml blood EDTA/Saliva on swab
FXII-MUT	Factor XII Deficiency	234000	c.C46T	F12	2 weeks	5 ml blood EDTA/Saliva on swab
FXII	Factor XII Deficiency	234000	cds	F12	2 months	5 ml blood EDTA/Saliva on swab
MEFV-EX	Familial Mediterranean Fever (MEFV)	608107	Exons 2, 3, 5 and 10	MEFV	3 weeks	5 ml blood EDTA/Saliva on swab
MEFV	Familial Mediterranean Fever (MEFV)	608107	cds	MEFV	2 months	5 ml blood EDTA/Saliva on swab
PNPLA3	Fatty Liver Disease Nonalcoholic, susceptibility to	613282	p.I148M	PNPLA3	2 weeks	5 ml blood EDTA/Saliva on swab
SEX	Fetal gender prediction on peripheral maternal blood		DYS14	Yq	3 days	5 ml blood EDTA
FRAXA	Fragile X Syndrome (FRAXA)	300624	Expansion CGG-Screening	FMR1	3 weeks	5 ml blood EDTA
FRDA	Friedrich's Ataxia	229300	Expansion GAA	FRDA	3 weeks	5 ml blood EDTA
ALX4	Frontonasal Dysplasia	613451	cds	ALX4	4 weeks	5 ml blood EDTA
WNT7A	Fuhrmann Syndrome	228930	cds	WNT7A	4 weeks	5 ml blood EDTA/Saliva on swab
GBA-MUT	Gaucher Disease (Types 1, 2 and 3)	230800, 230900 and 231000	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

GBA	Gaucher Disease (Types 1, 2 and 3)	230800, 230900 and 231000	cds	GBA	2 months	5 ml blood EDTA/Saliva on swab
UGT1A1-MUT	Gilbert Syndrome	143500	(TA)7	UGT1A1	3 weeks	5 ml blood EDTA/Saliva on swab
UGT1A1	Gilbert Syndrome	143500	cds	UGT1A1	2 months	5 ml blood EDTA/Saliva on swab
HDC	Gilles de la Tourette Syndrome	137580	p.Trp317Ter	HDC	3 weeks	5 ml blood EDTA/Saliva on swab
MYOC	Glaucoma 1 Open Angle (POAG)	137750	cds	MYOC	4 weeks	5 ml blood EDTA/Saliva on swab
CYP1B1	Glaucoma Primary Congenital	231300	cds	CYP1B1	4 weeks	5 ml blood EDTA/Saliva on swab
CYP11B	Glucocorticoid-Suppressible Hyperaldosteronism	103900	Chimerism	CYP11B1 – CYP11B2	4 weeks	5 ml blood EDTA
G6PD	Glucosa 6-fosfato deshidrogenasa, Deficiency	305900	cds	G6PD	2 months	5 ml blood EDTA
GLUT1	GLUT1, Deficiency Syndrome	612126 and 606777	cds	SLC2A1	3 weeks	5 ml blood EDTA/Saliva on swab
GCDH	Glutaric Acidemia Type I	231670	cds	GCDH	4 weeks	5 ml blood EDTA
PYGM-MUT	Glycogen Storage disease V (McArdle disease)	232600	p.Arg50X, p.Gly205Ser, p.Trp798Arg, p.Tyr85X	PYGM	3 weeks	5 ml blood EDTA/Saliva on swab
PYGM	Glycogen Storage disease V (McArdle disease)	232600	cds	PYGM	3 months	5 ml blood EDTA
GHRL-METAB	Grelina (Ghrelin, Obestatin) (Metabolic Syndrome,susceptibility to)	605353	p.Arg51Gln and p.Leu72Met	GHRL	2 weeks	5 ml blood EDTA/Saliva on swab
GHRL-OBES	Grelina (Ghrelin, Obestatin) (Obesity, susceptibility to)	605353	p.Gln90Leu	GHRL	2 weeks	5 ml blood EDTA/Saliva on swab
GHRL	Grelina (Ghrelin, Obestatin) (Obesity/Metabolic Syndrome, susceptibility to)	605353	cds	GHRL	4 weeks	5 ml blood EDTA/Saliva on swab
HFE	Hemochromatosis	235200	p.C282Y, p.H63D and p.S65C	HFE	4 days	5 ml blood EDTA/Saliva on swab
HJV	Hemochromatosis juvenile (Type 2A)	602390	p.G320V	HJV	2 weeks	5 ml blood EDTA/Saliva on swab
FVIII-MUT	Hemophilia A	306700	Intron inversion 22A / Intron inversion 1	F8	3 months	5 ml blood EDTA
FVIII	Hemophilia A	306700	cds	F8	3 months	5 ml blood EDTA
FIX	Hemophilia B	306900	cds	F9	4 weeks	5 ml blood EDTA/Saliva on swab
IL28B	Hepatitis C virus infection, response to therapy of		g.120070005C>T	IL28B	1 week	5 ml blood EDTA/Saliva on swab
ZIC3	Heterotaxy Visceral 1, X-linked (Congenital heart defects, nonsyndromic)	306955	cds	ZIC3	2 months	5 ml blood EDTA
CCR5	HIV infection, Resistance to	601373.0001	Deletion	CCR5	15 days	5 ml blood EDTA/Saliva on swab
HLA-DQ	HLA-DQ, Genotyping		Complete genotyping High Resolution	HLA-DQA1 and HLA-DQB1	2 weeks	5 ml blood EDTA/Saliva on swab
HLA-DRB1	HLA-DR, Genotyping		Complete genotyping High Resolution	HLA-DRB1	2 weeks	5 ml blood EDTA/Saliva on swab
HPV	Human Papiloma Virus (HPV)		Complete genotyping		4 days	Lesion on swab/Liquid cytology
HTT	Huntington Disease	143100	Expansion CAG	HTT	4 weeks	5 ml blood EDTA
LDLR	Hypercholesterolemia (Apo B)	143890	cds	LDLR	2 months	5 ml blood EDTA
LDLR-DEL	Hypercholesterolemia (Apo B)	143890	Deletions	LDLR	5 weeks	5 ml blood EDTA

PCSK9	Hypercholesterolemia (Apo B)	143890	cds	PCSK9	2 months	5 ml blood EDTA
APOB	Hypercholesterolemia Familiar (Apo B)	144010	p.Arg3527Gln, p.Arg3527Trp, p.Arg3558Cys	APOB	2 weeks	5 ml blood EDTA/Saliva on swab
CYP11B2	Hyperreninemic Hypoaldosteronism, Familial (Corticosterone Methyloxidase Type I)	203400	cds	CYP11B2	7 weeks	5 ml blood EDTA/Saliva on swab
NR3C2	Hypertension Early-onset, Autosomal Dominant	605115	cds	NR3C2	2 months	5 ml blood EDTA/Saliva on swab
FHH	Hypocalciuric Hypercalcemia Familiar Type I	145980	cds	CASR	2 months	5 ml blood EDTA
FGFR3-MUT	Hypochondroplasia	146000	p.N540K	FGFR3	2 weeks	5 ml blood EDTA/Saliva on swab
FGFR3-EX	Hypochondroplasia	146000	Exons 9 and 15	FGFR3	2 weeks	5 ml blood EDTA/Saliva on swab
TGM1	Ichthyosis Lamellar 1 (LI1)	242300	cds	TGM1	2 months	5 ml blood EDTA/Saliva on swab
ABCA12	Ichthyosis Lamellar 2 (LI2)	601277	cds	ABCA12	4 months	5 ml blood EDTA
IKBKG-DEL	Incontinentia Pigmenti	308300	Deletion exons 4-10	IKBKG	5 weeks	5 ml blood EDTA
IKBKG	Incontinentia Pigmenti	308300	cds	IKBKG	2 months	5 ml blood EDTA/Saliva on swab
FGFR2	Jackson-Weiss Syndrome	123150	cds	FGFR2	2 months	5 ml blood EDTA
KAL1	Kallmann Syndrome - Type 1 (X-Linked)	308700	cds	KAL1	4 weeks	5 ml blood EDTA/Saliva on swab
FGFR1	Kallmann Syndrome - Type 2	147950	cds	FGFR1	3 months	5 ml blood EDTA/Saliva on swab
PROKR2	Kallmann Syndrome - Type 3	244200	cds	PROKR2	3 weeks	5 ml blood EDTA/Saliva on swab
PROK2	Kallmann Syndrome - Type 4	610628	cds	PROK2	3 weeks	5 ml blood EDTA/Saliva on swab
KEARNS	Kearns-Sayre Syndrome (KSS)	530000	Deletions	mtDNA	5 weeks	5 ml blood EDTA
TBCE	Kenny-Caffey Syndrome (KCS1)	244460	c.155del12nt	TBCE	2 weeks	5 ml blood EDTA/Saliva on swab
KFSDX	Keratosis Follicularis Spinulosa Decalvans	308800	cds	MBTPS2	2 months	5 ml blood EDTA
KIR	KIR Genotyping (Repetitive Abortions)	605305	Genotyping	KIR	2 weeks	5 ml blood EDTA/Saliva on swab
MCM6	Lactose Intolerance (LCT)	223100	c.-13910C>T and c.-22018G>A	MCM6	2 weeks	5 ml blood EDTA/Saliva on swab
LCA6	Leber congenital amaurosis (LCA 6)	204000	cds	RPGRIP1	2 months	5 ml blood EDTA
LCA14	Leber congenital amaurosis 14 (LCA 14)	613341	cds	LRAT	3 weeks	5 ml blood EDTA/Saliva on swab
LCA2	Leber congenital amaurosis 2 (LCA 2)	204100	cds	RPE65	6 weeks	5 ml blood EDTA
LHON-1MUT	Leber Hereditary Optic Neuropathy (LHON) (Leber Optic Atrophy)	535000	m.11778G>A	mtDNA	2 weeks	5 ml blood EDTA/Saliva on swab
LHON-3MUT	Leber Hereditary Optic Neuropathy (LHON) (Leber Optic Atrophy)	535000	m.11778G>A, m.14484T>C and m.3460G>A	mtDNA	2 weeks	5 ml blood EDTA/Saliva on swab
LERI	Leri-Weill, Dyschondrosteosis	127300	cds	SHOX	4 weeks	5 ml blood EDTA
LERI-DEL	Leri-Weill, Dyschondrosteosis	127300	Deletions/Duplications	SHOX	5 weeks	5 ml blood EDTA
BCR-ABL	Leucemia Mieloide Crónica	608232	Fusion BCR/ABL	BCR/ABL	2 weeks	5 ml blood EDTA on ice
PML-RARA	Leukemia Acute Promyelocytic, PML/RARA type	102578	Fusion PML/RARA	PML/RARA	4 weeks	5 ml blood EDTA on ice
BCR-ABL	Leukemia Chronic Myeloid	608232	Fusion BCR/ABL	BCR/ABL	2 weeks	5 ml blood EDTA on ice
LAD	Leukocyte adhesion deficiency (LAD)	116920	cds	ITGB2	2 months	5 ml blood EDTA/Saliva on swab
EIF2B5	Leukoencephalopathy with vanishing white matter	603896	cds	EIF2B5	3 months	5 ml blood EDTA
EIF2B2	Leukoencephalopathy with vanishing white matter	603896	cds	EIF2B2	2 months	5 ml blood EDTA/Saliva on swab
EIF2B4	Leukoencephalopathy with vanishing white matter	603896	cds	EIF2B4	3 months	5 ml blood EDTA

EIF2B3	Leukoencephalopathy with vanishing white matter	603896	cds	EIF2B3	3 months	5 ml blood EDTA
EIF2B1	Leukoencephalopathy with vanishing white matter	603896	cds	EIF2B1	2 months	5 ml blood EDTA
SCNN1B	Liddle Syndrome	177200	cds	SCNN1B	2 months	5 ml blood EDTA
SCNN1G	Liddle Syndrome	177200	cds	SCNN1G	2 months	5 ml blood EDTA
LPL-MUT	Lipoprotein Lipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	238600	p.G188E	LPL	2 weeks	5 ml blood EDTA/Saliva on swab
LPL	Lipoprotein Lipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	238600	cds	LPL	4 weeks	5 ml blood EDTA
LPL-DEL	Lipoprotein Lipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	238600	Deletions/Duplications	LPL	5 weeks	5 ml blood EDTA
HNPCC-INES	Lipoprotein Lipase Deficiency (Hyperlipoproteinemia, Hyperlipemia Essential Familial)	120435	Microsatellites inestability studies		4 weeks	5 ml blood EDTA
LQT1	Long QT Syndrome (LQT1)	192500	cds	KCNQ1	2 months	5 ml blood EDTA
LQT2	Long QT Syndrome (LQT2)	613688	cds	KCNH2	2 months	5 ml blood EDTA
LQT3	Long QT Syndrome (LQT3)	603830	cds	SCN5A	3 months	5 ml blood EDTA
PMS2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	PMS2	3 months	5 ml blood EDTA
SCA3	Machado-Joseph Disease (MJD) (SCA3)	109150	Expansion CAG	ATXN3	4 weeks	5 ml blood EDTA
HIPERTERM	Malignant Hyperthermia, Susceptibility to	145600	p.R614C/L and p.G2434R	RYR1	2 weeks	5 ml blood EDTA/Saliva on swab
FBN1	Marfan Syndrome	154700	cds	FBN1	2 months	5 ml blood EDTA
FBN1-DEL	Marfan Syndrome	154700	Deletions	FBN1	5 weeks	5 ml blood EDTA
LDS2A	Marfan Type 2A Syndrome (Loeys-Dietz, Syndrome) (LDS2A)	608967	cds	TGFBR1	7 weeks	5 ml blood EDTA/Saliva on swab
LDS2B	Marfan Type 2B Syndrome (Loeys-Dietz, Syndrome) (LDS2B)	610380	cds	TGFBR2	7 weeks	5 ml blood EDTA/Saliva on swab
SIL1	Marinesco-Sjögren Syndrome	248800	cds	SIL1	2 months	5 ml blood EDTA
KIT-MUT	Mastocytosis	164920	p.Lys509Ile	KIT	2 weeks	5 ml blood EDTA/Saliva on swab
KIT	Mastocytosis	164920	cds	KIT	3 months	5 ml blood EDTA
KIT-DIFUS	Mastocytosis cutaneous diffuse		p.Ala533Asp	KIT	2 weeks	5 ml blood EDTA/Saliva on swab
KIT-HEMAT	Mastocytosis with associated hematologic disorder	164920.0009	p.Asp816Val	KIT	2 weeks	5 ml blood EDTA/Saliva on swab
CONT-MAT	Maternal Cell Contamination, on prenatal samples		STRs			
PYGM-MUT	McArdle Disease (Glycogen Storage Disease V, Myophosphorylase Deficiency)	232600	p.Arg50X, p.Gly205Ser, p.Trp798Arg and p.Tyr85X	PYGM	3 weeks	5 ml blood EDTA/Saliva on swab
PYGM	McArdle Disease (Glycogen Storage Disease V, Myophosphorylase Deficiency)	232600	cds	PYGM	3 months	5 ml blood EDTA
MELAS	MELAS Syndrome (Encephalopathy mitochondrial)	540000	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
ATP7A	Menkes Disease	309400	cds	ATP7A	3 weeks	5 ml blood EDTA
ATP7A-DEL	Menkes Disease	309400	Big Deletions	ATP7A	5 weeks	5 ml blood EDTA
CYB5R3	Methemoglobinemia	250800	cds	CYB5R3	4 weeks	5 ml blood EDTA/Saliva on swab
AZF	Microdeletions Y chromosome (Spermatogenic Failure non-obstructive, Azoospermia)	415000	AZFa, AZFb and AZFc	Yq	2 weeks	5 ml blood EDTA/Saliva on swab

MODY2	MODY 2	125851	cds	GCK	4 weeks	5 ml blood EDTA/Saliva on swab
MODY3	MODY 3	600496	cds	HNF1A	4 weeks	5 ml blood EDTA/Saliva on swab
MTHFR-MUT	MTHFR Metylenetetrahydrofolate reductase (Homocystinuria)	188050	c.677C>T and c.1298A>C	MTHFR	3 days	5 ml blood EDTA/Saliva on swab
MTHFR-SEQ	MTHFR Metylenetetrahydrofolate reductase (Homocystinuria)	188050	cds	MTHFR	2 months	5 ml blood EDTA/Saliva on swab
ARSB	Mucopolysaccharidosis Type VI (Maroteaux-Lamy Syndrome)	253200	cds	ARSB	4 weeks	5 ml blood EDTA/Saliva on swab
GALNS	Mucopolysaccharidosis Type IV A (Morquio Syndrome A, Galactosamine-6-Sulfatase Deficiency)	253000	cds	GALNS	2 months	5 ml blood EDTA
GLB1	Mucopolysaccharidosis Type IV B (Morquio Syndrome B)	253010	cds	GLB1	2 months	5 ml blood EDTA
MUENKE	Muenke Syndrome	602849	cds	FGFR3	2 months	5 ml blood EDTA/Saliva on swab
RYR1-SEPN1	Multiminicore Disease (MmD)	255320	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
RYR1	Multiminicore Disease (MmD)	255320	cds	RYR1	5 months	5 ml blood EDTA
RYR1-EX1	Multiminicore Disease (MmD)	255320	Exons 2 and 6-18	RYR1	7 weeks	5 ml blood EDTA
RYR1-EX2	Multiminicore Disease (MmD)	255320	Exons 39-48	RYR1	7 weeks	5 ml blood EDTA
RYR1-EX3	Multiminicore Disease (MmD)	255320	Exons 85-104	RYR1	2 months	5 ml blood EDTA
SEPN1	Multiminicore Disease (MmD)	255320	cds	SEPN1	2 months	5 ml blood EDTA/Saliva on swab
JAK2-MI	Myelofibrosis	254450	p.V617F	JAK2	2 weeks	5 ml blood EDTA/Saliva on swab
JAK2	Myeloproliferative disorder with erythrocytosis JAK2 related (Polycythemia Vera, Thrombocythemia Essential and Myelofibrosis)	147796	p.V617F	JAK2	2 weeks	5 ml blood EDTA/Saliva on swab
DNM2	Myopathy Centronuclear	160150	cds	DNM2	3 months	5 ml blood EDTA
CLCN1	Myotonia Congenita Dominant (Thomsen Disease; Becker Disease)	160800 and 255700	cds	CLCN1	2 months	5 ml blood EDTA/Saliva on swab
DM-STEINERT	Myotonic Dystrophy Type 1 (DM1) (Steinert disease)	160900	Expansion CTG	DMPK	3 weeks	5 ml blood EDTA
NBLAST1	Neuroblastoma, Susceptibility to (NBLST1)	256700	cds	KIF1B	5 months	5 ml blood EDTA
NBLAST2	Neuroblastoma, Susceptibility to (NBLST2)	603851	cds	PHOX2B	4 weeks	5 ml blood EDTA/Saliva on swab
NBLAST3	Neuroblastoma, Susceptibility to (NBLST3)	613014	cds	ALK	4 months	5 ml blood EDTA
PMP22-STRS	Neuropathy Hereditary With Liability to Pressure Palsies (HNPP)	162500	STRs	PMP22	4 weeks	5 ml blood EDTA/Saliva on swab
PMP22	Neuropathy Hereditary With Liability to Pressure Palsies (HNPP)	162500	cds	PMP22	4 weeks	5 ml blood EDTA/Saliva on swab

OTOF-MUT	Neurosensory Nonsyndromic Recessive Deafness	601071	p.Gln829Ter and p.Pro1825Ala	OTOF	3 weeks	5 ml blood EDTA/Saliva on swab
OTOF	Neurosensory Nonsyndromic Recessive Deafness	601071	cds	OTOF	4 months	5 ml blood EDTA
GJB2-MUT	Neurosensory Nonsyndromic Recessive Deafness	220290	c.35delG	GJB2 (Conexina 26)	2 weeks	5 ml blood EDTA/Saliva on swab
GJB2	Neurosensory Nonsyndromic Recessive Deafness	220290	cds	GJB2 (Conexina 26)	3 weeks	5 ml blood EDTA/Saliva on swab
NCS	Neutropenia Severe Congenital	202700	cds	ELANE (ELA2)	4 weeks	5 ml blood EDTA/Saliva on swab
SMPD1	Niemann-Pick Disease (Types A and B)	257200 and 607616	cds	SMPD1	4 weeks	5 ml blood EDTA/Saliva on swab
PTPN11	Noonan Syndrome	163950	cds	PTPN11	4 weeks	5 ml blood EDTA
MC4R	Obesity Morbid	601665	cds	MC4R	2 weeks	5 ml blood EDTA/Saliva on swab
DMO	Oculopharyngeal Muscular Dystrophy	164300	Expansion GCG	PABPN1	3 weeks	5 ml blood EDTA/Saliva on swab
OPA1-EX	Optic Atrophy Dominant Type 1 (ADOA)	165500	Exons 8-16 and 27-28	OPA1	4 weeks	5 ml blood EDTA/Saliva on swab
OPA1	Optic Atrophy Dominant Type 1 (ADOA)	165500	cds	OPA1	2 months	5 ml blood EDTA
OPA1-DEL	Optic Atrophy Dominant Type 1 (ADOA)	165500	Deletions	OPA1	5 weeks	5 ml blood EDTA
OPA3-MUT	Optic Atrophy Type 3 with cataract	165300	p.G93S and p.Q105E	OPA3	2 weeks	5 ml blood EDTA/Saliva on swab
OPA3	Optic Atrophy Type 3 with cataract	165300	cds	OPA3	3 weeks	5 ml blood EDTA/Saliva on swab
OTC	Ornithine transcarbamylase deficiency	311250	cds	OTC	2 months	5 ml blood EDTA
OTC-DEL	Ornithine transcarbamylase deficiency	311250	Deletions	OTC	5 weeks	5 ml blood EDTA
COL1A1	Osteogenesis Imperfecta	166200	cds	COL1A1	4 months	5 ml blood EDTA
COL1A2	Osteogenesis Imperfecta	166200	cds	COL1A2	4 months	5 ml blood EDTA
COL1A1-2	Osteogenesis Imperfecta	166200	cds	COL1A1 and COL1A2	5 months	5 ml blood EDTA
PRSS1	Pancreatitis Hereditary	167800	cds	PRSS1	4 weeks	5 ml blood EDTA/Saliva on swab
PRSS1-EX	Pancreatitis Hereditary	167800	Exons 2 and 3 (variant p.R122H included)	PRSS1	3 weeks	5 ml blood EDTA/Saliva on swab
SPINK1	Pancreatitis Hereditary	167800	cds	SPINK1	4 weeks	5 ml blood EDTA/Saliva on swab
SPINK1-EX	Pancreatitis Hereditary	167800	Exon 3 (variant N34S included)	SPINK1	4 weeks	5 ml blood EDTA/Saliva on swab
PANCR	Pancreatitis Hereditary	167800	cds	CFTR	2 months	5 ml blood EDTA
LRRK2-MUT	Parkinson Disease 8 (Autosomal Dominant)	607060	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
LRRK2	Parkinson Disease 8 (Autosomal Dominant)	607060	cds	LRRK2	4 months	5 ml blood EDTA

PARK2	Parkinson Juvenile Disease	600116	cds	PARK2	4 weeks	5 ml blood EDTA/Saliva on swab
PARK2-DEL	Parkinson Juvenile Disease	600116	Deletions/Duplications	PARK2	4 weeks	5 ml blood EDTA
PFEIFFER-MUT	Pfeiffer Syndrome	101600	p.C278F and p.C342G	FGFR2	2 weeks	5 ml blood EDTA/Saliva on swab
PFEIFFER-2	Pfeiffer Syndrome	101600	cds	FGFR2	2 months	5 ml blood EDTA
PFEIFFER-EX	Pfeiffer Syndrome	101600	Exon 7 FGFR1; exon 7,8,13,14 and 15 FGFR2	FGFR1 and FGFR2	4 weeks	5 ml blood EDTA/Saliva on swab
PFEIFFER-1	Pfeiffer Syndrome	101600	cds	FGFR1	3 months	5 ml blood EDTA/Saliva on swab
PKU-EX	Phenylketonuria (PAH)	261600	Exons 7,9,11 and 12	PAH	2 weeks	5 ml blood EDTA/Saliva on swab
PKU	Phenylketonuria (PAH)	261600	cds	PAH	4 weeks	5 ml blood EDTA/Saliva on swab
SERPINE1	Plasminogen Activator Inhibitor 1 Deficiency	613329	c.-675_4G/5G	SERPINE1	2 weeks	5 ml blood EDTA/Saliva on swab
PKHD1	Polycystic Kidney Autosomal Recessive (ARPKD)	263200	cds	PKHD1	5 months	5 ml blood EDTA
PKD1	Polycystic Kidney Syndrome, Autosomal Dominant	173900	cds	PKD1	5 months	5 ml blood EDTA
PKD2	Polycystic Kidney Syndrome, Autosomal Dominant	173900	cds	PKD2	2 months	5 ml blood EDTA/Saliva on swab
JAK2-PV	Polycythemia Vera	263300	p.V617F	JAK2	2 weeks	5 ml blood EDTA/Saliva on swab
PPOX	Porphyria Variegata	176200	cds	PPOX	4 weeks	5 ml blood EDTA/Saliva on swab
PW-UPD	Prader Willi Syndrome	176270	Uniparental Disomy STRs	15q11-q13	5 weeks	5 ml blood EDTA
PW-MET	Prader Willi Syndrome	105830	Metilation	PW	3 weeks	5 ml blood EDTA
PW-DEL	Prader Willi Syndrome	176270	Deletions, metilation and uniparental disomy	PW	4 weeks	5 ml blood EDTA
FII	Prothrombin Thrombophilia (Factor II)	176930	c.20210G>A	F2	3 days	5 ml blood EDTA/Saliva on swab
ARSA	Pseudoarylsulfatase A Deficiency (Metachromatic leukodystrophy)	250100	cds	ARSA	2 months	5 ml blood EDTA/Saliva on swab
NR3C2	Pseudohypoaldosteronism Type I, Autosomal Dominant	177735	cds	NR3C2	2 months	5 ml blood EDTA
BMPR2	Pulmonary Hypertension Primary (PPH1)	178600	cds	BMPR2	3 months	5 ml blood EDTA
BMPR2-DEL	Pulmonary Hypertension Primary (PPH1)	178600	Deletions	BMPR2	5 weeks	5 ml blood EDTA
RHO	Retinitis Pigmentosa Autosomal Dominant	268000	cds	RHO	4 weeks	5 ml blood EDTA/Saliva on swab
RPAR	Retinitis Pigmentosa Autosomal Recessive	268000	cds	LRAT and RPE65	2 months	5 ml blood EDTA
LRAT	Retinitis Pigmentosa Autosomal Recessive	268000	cds	LRAT	3 weeks	5 ml blood EDTA/Saliva on swab
MERTK	Retinitis Pigmentosa Autosomal Recessive	268000	cds	MERTK	2 months	5 ml blood EDTA
RPE65	Retinitis Pigmentosa Autosomal Recessive	268000	cds	RPE65	4 weeks	5 ml blood EDTA
RPLX	Retinitis Pigmentosa X-linked	268000	cds	RP2 and RPGR	3 months	5 ml blood EDTA
RPGR	Retinitis Pigmentosa X-linked	268000	cds	RPGR	2 months	5 ml blood EDTA
RP2	Retinitis Pigmentosa X-linked	268000	cds	RP2	4 weeks	5 ml blood EDTA/Saliva on swab
RS1	Retinoschisis 1 Juvenile, X-Linked	312700	cds	RS1	4 weeks	5 ml blood EDTA/Saliva on swab
RETT	Rett Syndrome	312750	cds	MECP2	2 weeks	5 ml blood EDTA/Saliva on swab
RETT-DEL	Rett Syndrome	312750	Big Deletions	MECP2	5 weeks	5 ml blood EDTA
CREBBP	Rubinstein-Taybi Syndrome	180849	cds	CREBBP	5 months	5 ml blood EDTA
CREBBP-DEL	Rubinstein-Taybi Syndrome	180849	Deletions	CREBBP	5 weeks	5 ml blood EDTA

HRD	Sanjad-Sakati Syndrome (HRD)	241410	c.66delAG (p.V23fs48X) and c.1113T>A (p.C371X)	TBCE	2 weeks	5 ml blood EDTA/Saliva on swab
SLC6A4	Serotonin Transporter SERT (Obsessive-compulsive disorder)	182138	44bp ins/del	SLC6A4 (5HTTLPR)	3 weeks	5 ml blood EDTA/Saliva on swab
SCN1A	Severe Myoclonic epilepsy of infancy (Dravet Syndrome) (SMEI)	607208	cds	SCN1A	2 months	5 ml blood EDTA
SCN1A-DEL	Severe Myoclonic epilepsy of infancy (Dravet Syndrome) (SMEI)	607208	Big Deletions	SCN1A	4 weeks	5 ml blood EDTA
SHOX	Short Stature, idiopathic	300582	cds	SHOX	4 weeks	5 ml blood EDTA/Saliva on swab
SHOX-DEL	Short Stature, idiopathic	300582	Deletions/Duplications	SHOX	5 weeks	5 ml blood EDTA
HBB-MUT	Sickle cell anemia	603903	p.E6V	HBB	2 weeks	5 ml blood EDTA/Saliva on swab
HBB	Sickle cell anemia	603903	cds	HBB	4 weeks	5 ml blood EDTA/Saliva on swab
SLOS	Smith-Lemli-Opitz Syndrome (SLOS)	270400	cds	DHCR7	4 weeks	5 ml blood EDTA/Saliva on swab
SLOS-MUT	Smith-Lemli-Opitz Syndrome (SLOS)	270400	p.Phe302Leu and p.Arg446Gln	DHCR7	3 weeks	5 ml blood EDTA/Saliva on swab
PLP1	Spastic Paraplegia (Pelizaeus-Merzbacher Disease)	312080 and 312920	cds	PLP1	4 weeks	5 ml blood EDTA/Saliva on swab
PLP1-DUP	Spastic Paraplegia (Pelizaeus-Merzbacher Disease)	312080 and 312920	Duplication	PLP1	4 weeks	5 ml blood EDTA
SPAST	Spastic Paraplegia Familiar (Autosomal Dominant)	182601	cds	SPAST (SPG4)	2 months	5 ml blood EDTA
SPAST	Spastic Paraplegia Familiar (Autosomal Dominant)	182601	Deleciones	SPAST (SPG4) and SPG3A	2 months	5 ml blood EDTA
SPH1	Spherocytosis Type 1 (SPH1)	182900	cds	ANK1	5 months	5 ml blood EDTA/Saliva on swab
SPH2	Spherocytosis Type 2 (SPH2)	182870	cds	SPTB	4 months	5 ml blood EDTA/Saliva on swab
SPH3	Spherocytosis Type 3 (SPH3)	270970	cds	SPTA1	4 months	5 ml blood EDTA/Saliva on swab
SPH4	Spherocytosis Type 4 (SPH4)	109270	cds	SLC4A1	3 months	5 ml blood EDTA/Saliva on swab
SPH5	Spherocytosis Type 5 (SPH5)	612690	cds	EPB42	2 months	5 ml blood EDTA/Saliva on swab
SBMA	Spinal and Bulbar Muscular Atrophy of Kennedy (SBMA)	313200	Expansion CAG	AR	3 weeks	5 ml blood EDTA/Saliva on swab
SMA-LED	Spinal Muscular Atrophy (SMA-LED) (Kugelberg-Welander Syndrome)	158600	cds	DYNC1H1	5 months	5 ml blood EDTA
SMA-AF	Spinal Muscular Atrophy (SMA)	253300	Deletion Exons 7 and 8 (patients)	SMN1	2 weeks	5 ml blood EDTA/Saliva on swab
SMA-PORT	Spinal Muscular Atrophy (SMA)	253300	Deletion Exons 7 and 8 (carriers)	SMN1	4 weeks	5 ml blood EDTA
SCA12	Spinocerebellar Ataxia 12, SCA12	604326	Expansion CAG	PPP2R2B	4 weeks	5 ml blood EDTA
SCA1	Spinocerebellar Ataxia Dominant SCA1	164400	Expansion CAG	ATXN1	4 weeks	5 ml blood EDTA
SCA17	Spinocerebellar Ataxia Dominant SCA17	607136	Expansion CAG/CAA	TBP	4 weeks	5 ml blood EDTA
SCA2	Spinocerebellar Ataxia Dominant SCA2	183090	Expansion CAG	ATXN2	4 weeks	5 ml blood EDTA
SCA3	Spinocerebellar Ataxia Dominant SCA3 (Machado-Joseph Disease)	109150	Expansion CAG	ATXN3	4 weeks	5 ml blood EDTA

SCA5	Spinocerebellar Ataxia Dominant SCA5	600224	cds	SPTBN2	3 months	5 ml blood EDTA
SCA6	Spinocerebellar Ataxia Dominant SCA6	183086	Expansion CAG	CACNA1A	4 weeks	5 ml blood EDTA
SCA7	Spinocerebellar Ataxia Dominant SCA7	164500	Expansion CAG	ATXN7	4 weeks	5 ml blood EDTA
SCA8	Spinocerebellar Ataxia Dominant SCA8	608768	Expansion CTG	ATXN8OS	4 weeks	5 ml blood EDTA
TNFRSF	Squamous Cell Carcinoma, Head and Neck	275355	cds	TNFRSF10B	7 weeks	5 ml blood EDTA/Saliva on swab
ING1	Squamous Cell Carcinoma, Head and Neck	275355	cds	ING1	4 weeks	5 ml blood EDTA/Saliva on swab
STICKLER1	Stickler Syndrome Type 1	108300	cds	COL2A1	2 months	5 ml blood EDTA
			Exons			
STICKLER1-EX	Stickler Syndrome Type 1 (Arthroophthalmopathy)	108300	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
STICKLER2	Stickler Syndrome Type 2	604841	cds	COL11A1	4 months	5 ml blood EDTA
SVAS	Supravalvar Aortic Stenosis (SVAS)	185500	cds	ELN	3 months	5 ml blood EDTA
SVAS	Supravalvular Aortica Stenosis (SVAS)	185500	Deletions/Duplications	ELN	5 weeks	5 ml blood EDTA
HEXA	Tay-Sachs Disease	272800	cds	HEXA	2 months	5 ml blood EDTA
HHT	Telangiectasia Hereditary Hemorrhagic (Osler-Weber-Rendu Disease) (HHT)		cds	ENG and ACVRL1	2 months	5 ml blood EDTA
HHT-DEL	Telangiectasia Hereditary Hemorrhagic (Osler-Weber-Rendu Disease) (HHT)		Big Deletions	ENG and ACVRL1	5 weeks	5 ml blood EDTA
HHT1	Telangiectasia Hereditary Hemorrhagic (Osler-Weber-Rendu Disease) (HHT1)	187300	cds	ENG	2 months	5 ml blood EDTA/Saliva on swab
HHT2	Telangiectasia Hereditary Hemorrhagic (Osler-Weber-Rendu Disease) (HHT2)	600376	cds	ACVRL1	5 weeks	5 ml blood EDTA/Saliva on swab
JPHT	Telangiectasia Hereditary Hemorrhagic (Osler-Weber-Rendu Disease) (JPHT, HHT, Juvenile polyposis)	175050	cds	SMAD4	2 months	5 ml blood EDTA/Saliva on swab
TANATOF-1	Thanatophoric Dysplasia Type I	187600	p.R248C and p.Y373C	FGFR3	2 weeks	5 ml blood EDTA/Saliva on swab
TANATOF-1y2	Thanatophoric Dysplasia Type I and Type II		cds	FGFR3	2 months	5 ml blood EDTA/Saliva on swab
TANATOF-2	Thanatophoric Dysplasia Type II	187601	p.K650E	FGFR3	2 weeks	5 ml blood EDTA/Saliva on swab
JAK2-TE	Thrombocythemia Essential	187950	p.V617F	JAK2	2 weeks	5 ml blood EDTA/Saliva on swab
THRB	Thyroid Hormone Resistance	188570	cds	THRB	7 weeks	5 ml blood EDTA/Saliva on swab
TCOF1	Treacher Collins Syndrome	154500	cds	TCOF1	2 months	5 ml blood EDTA
TRPS1	Trichorhinophalangeal Syndrome	190350	cds	TRPS1	2 months	5 ml blood EDTA
FMO3	Trimethylaminuria	602079	cds	FMO3	4 weeks	5 ml blood EDTA/Saliva on swab
ET-SEQ	Tuberous Sclerosis	191100 and 613254	cds	TSC1 and TSC2	4 weeks	5 ml blood EDTA
TSC1	Tuberous Sclerosis	191100	cds	TSC1	3 months	5 ml blood EDTA
TSC2	Tuberous Sclerosis	613254	cds	TSC2	3 months	5 ml blood EDTA
TSC1-DEL	Tuberous Sclerosis	191100	Deletions	TSC1	5 weeks	5 ml blood EDTA
TSC2-DEL	Tuberous Sclerosis	613254	Deletions	TSC2	5 weeks	5 ml blood EDTA
BEST1	Vitelliform Macular Distrophy (Best disease)	153700	cds	BEST1	3 months	5 ml blood EDTA
VHL	Von Hippel Lindau Disease	193300	cds	VHL	2 weeks	5 ml blood EDTA/Saliva on swab

VHL-DEL	Von Hippel Lindau Disease	193300	Deletions/Duplications	VHL	5 weeks	5 ml blood EDTA
VWF1	Von Willebrand Disease Type 1 (VWD1)	193400	cds	VWF	4 months	5 ml blood EDTA
VWF1-EX	Von Willebrand Disease Type 1 (VWD1)	193400	Exons 18-28	VWF	2 months	5 ml blood EDTA/Saliva on swab
VWF2-AD	Von Willebrand Disease Type 2A (VWD2)	613554	Exon 28	VWF	2 weeks	5 ml blood EDTA/Saliva on swab
VWF2-AR	Von Willebrand Disease Type 2A (VWD2)	613554	Exons 11-16, 22, 25-27 and 52	VWF	2 months	5 ml blood EDTA/Saliva on swab
VW2N	Von Willebrand Disease Type 2N (VWDN)	613554	Exons 18-20	VWF	2 weeks	5 ml blood EDTA
VWF3	Von Willebrand Disease Type 3 (VWD3)	277480	cds	VWF	4 months	5 ml blood EDTA
SOX10-2E	Waardenburg Syndrome (Type 2E)	611584	cds	SOX10	3 weeks	5 ml blood EDTA/Saliva on swab
SOX10-4C	Waardenburg Syndrome (Type 4C)	613266	cds	SOX10	3 weeks	5 ml blood EDTA/Saliva on swab
ELN-DEL	Williams Syndrome (Williams-Beuren Syndrome)	194050	Deletion	ELN	5 weeks	5 ml blood EDTA
ELN	Williams Syndrome (Williams-Beuren Syndrome)	194050	cds	ELN	3 months	5 ml blood EDTA
ATP7B	Wilson Disease	277900	cds	ATP7B	2 months	5 ml blood EDTA
ATP7B-MUT	Wilson Disease	277900	p.H1069Q	ATP7B	2 weeks	5 ml blood EDTA/Saliva on swab
WAS	Wiskott-Aldrich Syndrome	301000	cds	WAS	2 months	5 ml blood EDTA/Saliva on swab
XPC	Xeroderma Pigmentosum Complementation Group C (XPC)	278720	cds	XPC	3 months	5 ml blood EDTA
XPD	Xeroderma Pigmentosum Complementation Group D (XPD)	278730	cds	ERCC2	3 months	5 ml blood EDTA
XPV	Xeroderma Pigmentosum Variant Type (XPV)	278750	cds	POLH	3 months	5 ml blood EDTA

CYTOGENETICS ANALYSIS

QF-PCR	Aneuploidy crom 13, 18, 21, X and Y	QF-PCR	3 days	2 ml AF/5 ml blood EDTA
QF-RA	Aneuploidy crom 15 and 22		3 days	Tissues -abortions-
CGH-60	CGH-array 60K - PRENATAL	Deletions/Duplications with 60k resolution (60.000 probes)	Complete genome	2 weeks 10 ml AF
CGH-60	CGH-array 60K	Deletions/Duplications with 60k resolution (60.000 probes)	Complete genome	4 weeks 5 ml blood EDTA
CGH-180	CGH-array 180K	Deletions/Duplications with 180k resolution (180.000 probes)	Complete genome	4 weeks 5 ml blood EDTA
CGH-400	CGH-array 400K	Deletions/Duplications with 400k resolution (400.000 probes)	Complete genome	4 weeks 5 ml blood EDTA
FISH-BCR	FISH BCR-ABL		3 weeks	5 ml blood EDTA
FISH-TELOM	Integrity telomeric analysis		3 weeks	15 ml blood HEPARIN/10 ml AF
CAR-VC	Karyotype chorionic villus		3 weeks	Chorionic villus
CAR-SP	Karyotype peripheral blood		3 weeks	15 ml blood HEPARIN
CAR-RA	Karyotype others tissues -abortions-		3 weeks	Consult
CAR-LA	Karyotype amniotic fluid		3 weeks	10 ml AF

PREDICTIVE MEDICINE ANALYSIS

DIABES	Diabesity (genetic predisposition)		7 genes		2 weeks	5 ml blood EDTA/Saliva on swab
OBES-EI	Obesity (genetic predisposition) - Initial study		6 genes		2 weeks	5 ml blood EDTA/Saliva on swab
OBES-EC	Obesity (genetic predisposition) - Complete study		18 genes		2 weeks	5 ml blood EDTA/Saliva on swab
OSTEO	Osteoporosis	166710	IVS1-440G>T (Sp1), IVS1-397T>C and IVS10+283G>A	COL1A1, ESR1 and VDR	2 weeks	5 ml blood EDTA/Saliva on swab
OSTEO-COL1A1	Osteoporosis	166710	IVS1-440G>T (Sp1)	COL1A1	2 weeks	5 ml blood EDTA/Saliva on swab
OSTEO-ESR1	Osteoporosis	166710	IVS1-397T>C	ESR1	2 weeks	5 ml blood EDTA/Saliva on swab
OSTEO-VDR	Osteoporosis	166710	IVS10+283G>A	VDR	2 weeks	5 ml blood EDTA/Saliva on swab
HTA	Cardiovascular risk: Hypertension		I/D (ECA), E298D (NOS3), G16R (ADRB2) and 825C>T (GNB3)	ECA, NOS3, ADRB2 and GNB3	2 weeks	5 ml blood EDTA/Saliva on swab
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
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

PHARMACOGENETICS

DRUGS

GENE

CYP2C9	CYP2C9		Warfarina, Ibuprofeno, Aceclofenaco, Celecoxib, Acenocumarol, Glibenclamida, Gluclazida, Glimeperida, Fenprocumon, Fenitoina	CYP2C9	2 weeks	5 ml blood EDTA/Saliva on swab
CYP2C19	CYP2C19		Citalopram, Clopidogrel, Esomeprazol, Imipramina, Lansoprazol, Moclobemida, Omeprazol, Pantoprazol, Sertarlina, Voriconazol	CYP2C19	2 weeks	5 ml blood EDTA/Saliva on swab
CYP2D6*	CYP2D6*		Amitriptilina, Aripiprazol, Atomoxetina, Clomipramina, Codeina, Doxepina, Flecaínida, Haloperidol, Imipramina, Metoprolol, Nortriptilina, Oxícodona, Profafenona, Risperidona, Tamoxifeno, Tramadol, Velafaxina, Zuclopentixol	CYP2D6*	2 weeks	5 ml blood EDTA
CYP2C8	CYP2C8		Bifosfonatos	CYP2C8	2 weeks	5 ml blood EDTA/Saliva on swab

CYP3A4	CYP3A4	Efavirenz, Docetaxel, Tacrolimus, Ciclofosfamida, Midazolam, Ciclosporina, Indinavir, Sirolimus, Tenipósido, Paclitaxel, Doxorubicina, Agentes Antineoplásicos, Nelfinavir, Tamoxifeno, Etopósido, Nevirapina, Fluorouracilo, Tipifarnib	CYP3A4	2 weeks	5 ml blood EDTA/Saliva on swab
CYP3A5	CYP3A5	Tacrolimus	CYP3A5	2 weeks	5 ml blood EDTA/Saliva on swab
CYP1A2	CYP1A2	Clopidogrel, Warfarina, Cafeina, Teofilina Inhibidor Fluvoxamina	CYP1A2	2 weeks	5 ml blood EDTA/Saliva on swab
COMT	COMT	Nicotina, Antipsicóticos, Morfina, Litio, Fluvoxamina	COMT	2 weeks	5 ml blood EDTA/Saliva on swab
TPMT	TPMT	Azatioprina, Mercaptopurina, Tioguanina	TPMT	2 weeks	5 ml blood EDTA/Saliva on swab
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
UGT1A1	UGT1A1	Irinotecan	UGT1A1	2 weeks	5 ml blood EDTA/Saliva on swab
NAT2	NAT2	Isoniazida, Rifamicina, Pirazinamida, Isosorbida, Hidrazalina	NAT2	2 weeks	5 ml blood EDTA/Saliva on swab
SLC01B1	SLC01B1	Estatinas-Simvastatina	SLC01B1	2 weeks	5 ml blood EDTA/Saliva on swab
SLC6A4	SLC6A4	Antidepresivos, Ondansetron, Risperidona, Berberine, Inhibidores Selectivos Recaptación Serotonina, Clomipramina, Evodiamina, Escitalopram, Nortriptylina	SLC6A4	2 weeks	5 ml blood EDTA/Saliva on swab
SCNA5	SCNA5	Tioridazina, Claritromicina	SCNA5	2 weeks	5 ml blood EDTA/Saliva on swab
HTR2A	HTR2A	Antidepresivos, citalopram, venlafaxina, Inhibidores Selectivos recaptación serotonina, Olanzapina, Risperidona, Paroxetina, Escitalopram	HTR2A	2 weeks	5 ml blood EDTA/Saliva on swab
DRD2	DRD2	Risperidona, Nicotina, Antipsicóticos, Clorpromazina, Clozapina, Nemonaprida, Olanzapina, Bromperidol, Aripiprazole, Bupropion	DRD2	2 weeks	5 ml blood EDTA/Saliva on swab
Factor V	Factor V	Terapia Hormonal-Estrógenos	F5	2 weeks	5 ml blood EDTA/Saliva on swab
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
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

VKORC1	VKORC1	Acenocumarol, Fenprocumon, Warfarina	VKORC1	2 weeks	5 ml blood EDTA/Saliva on swab
DPYD	DPYD	Cetuximab, Oxaliplatino, Bevacizumab, Raltitrexed, Leucovorin, Tegafur Fluorouracilo, Capecitabina, Análogos Pirimidina	DPYD	2 weeks	5 ml blood EDTA/Saliva on swab
IL-28	IL-28	Boceprevir, Telaprevir, PegInterferon	IL-28	2 weeks	5 ml blood EDTA/Saliva on swab
HLA-B*5701	HLA-B*5701	Abacavir	HLA-B*5701	2 weeks	5 ml blood EDTA/Saliva on swab
HLA-A*1502	HLA-A*1502	Carbamazepina, Fenitoina	HLA-A*1502	2 weeks	5 ml blood EDTA/Saliva on swab
MTHFR	MTHFR	Azatioprina, Fluorouracilo, Acido Fólico, Leucovorin, Mercaptopurina, Metotrexato	MTHFR	2 weeks	5 ml blood EDTA/Saliva on swab
VDR	VDR	A. Etidrónico, Zoledronato, Medroxiprogesterona Calcio, Alendronato, Midazolam, Clodronato, Bifosfonatos, Estrógenos conjugados, Raloxifeno	VDR	2 weeks	5 ml blood EDTA/Saliva on swab
ALOX5	ALOX5	Montelukast	ALOX5	2 weeks	5 ml blood EDTA/Saliva on swab
PTGS2	PTGS2	AINES, Rofecoxib, Coxibs, Aspirina Ibuprofeno, A Grasos poliinsaturados omega-3	PTGS2	2 weeks	5 ml blood EDTA/Saliva on swab
HMGCoA	HMGCoA	Estatinas	HMGCoA	2 weeks	5 ml blood EDTA/Saliva on swab
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
FGEN-HORM	Hormone Therapy Pharmacogenomics			2 weeks	5 ml blood EDTA/Saliva on swab
FGEN-DEPRE	SSRI Antidepressants Pharmacogenomics			2 weeks	5 ml blood EDTA/Saliva on swab

PATERNITY TEST

PAT	Paternity Test (father-child) with legal value	16 markers		1 week	Saliva on swab
PAT-ANON	Paternity Test (father-child) informative value	16 markers		1 week	Saliva on swab
PARENT	Biological relationship with relatives (2 persons) with legal value	16 markers		1 week	Saliva on swab
PAT-13STRS	Nuclear STRs markers extended	13 markers		1 week	Saliva on swab
Y-PARENT	Paternal familial relationship via Chromosome Y	Chromosome and STRs		2 weeks	Saliva on swab
M-ADICIONAL	Additional sample			1-2 weeks	Saliva on swab
M-NOSALIVA	Additional price for other samples than blood or saliva (except bone/tooth)			Consult	Consult
M-HUESO	Additional price for bone/tooth			Consult	Consult
MYT-PARENT	Maternal familial relationship via mtDNA	mtDNA		2 weeks	Saliva on swab
MYT-MUESTRA	Additional sample mtDNA	mtDNA		2 weeks	Saliva on swab
PERFIL	Genetic Profile	16 markers		1 week	Saliva on swab

INF-COMP Genetic Profile Comparative Report

ONCOLOGY ANALYSIS

PML-RARA	Acute Promyelocytic Leukemia	102578	Fusion PML/RARA	PML/RARA	4 weeks	5 ml blood EDTA
BRAF	BRAF	164757	cds	BRAF	2 months	5 ml blood EDTA/Saliva on swab
BRAF-MUT	BRAF, Mutation	164757	p.V600E	BRAF	2 weeks	5 ml blood EDTA/Saliva on swab
MAMA	Breast-ovarian cancer familial (BRCA1 and BRCA2)	604370	cds	BRCA1 and BRCA2	3 months	10 ml blood EDTA
MAMA-LOH	Breast-ovarian cancer familial (BRCA1 and BRCA2)	604370	Loss of heterozygosity (LOH)	BRCA1 and BRCA2	4 weeks	Consult
BRCA1-DEL	Breast-ovarian cancer familial (BRCA1 and BRCA2)	604370	Deletions	BRCA1 and BRCA2	5 weeks	5 ml blood EDTA
BRCA1	Breast-ovarian cancer familial (BRCA1)	604370	cds	BRCA1	6 weeks	5 ml blood EDTA
BRCA1-EX	Breast-ovarian cancer familial (BRCA1)	604370	Exon 10	BRCA1	3 weeks	5 ml blood EDTA/Saliva on swab
BRCA2	Breast-ovarian cancer familial (BRCA2)	604370	cds	BRCA2	2 months	5 ml blood EDTA
BRCA2-EX	Breast-ovarian cancer familial (BRCA2)	604370	Exon 11	BRCA2	3 weeks	5 ml blood EDTA/Saliva on swab
MUTYH-MUT	Colorectal Adenomatous Polyptosis, Autosomal Recessive	608456	p.Y165C and p.G382C	MUTYH (MYH)	2 weeks	5 ml blood EDTA/Saliva on swab
MUTYH	Colorectal Adenomatous Polyptosis, Autosomal Recessive	608456	cds	MUTYH (MYH)	2 months	5 ml blood EDTA
COWDEN-PTEN	Cowden disease	158350	cds	PTEN	4 weeks	5 ml blood EDTA/Saliva on swab
COWDEN-BMPR1A	Cowden disease	158350	cds	BMPR1A	3 months	5 ml blood EDTA
COWDEN	Cowden disease	158350	Big Deletions	PTEN and BMPR1A	4 weeks	5 ml blood EDTA
APC	Familial Adenomatous Polyposis (APC)	175100	cds	APC	2 months	10 ml blood EDTA
APC-MUT	Familial Adenomatous Polyposis (APC)	175100	Exon 15 region (p.Ile1307Lys)	APC	3 weeks	5 ml blood EDTA/Saliva on swab
APC-EX	Familial Adenomatous Polyposis (APC)	175100	Exon 15	APC	4 weeks	10 ml blood EDTA
APC-DEL	Familial Adenomatous Polyposis (APC)	175100	Big Deletions	APC	5 weeks	5 ml blood EDTA
CDH1	Gastric Cancer Hereditary Diffuse	137215	cds	CDH1	2 months	5 ml blood EDTA/Saliva on swab
PTCH1	Gorlin Syndrome	109400	cds	PTCH1	2 months	5 ml blood EDTA
KRAS	KRAS, Mutations		cds	KRAS	5 weeks	5 ml blood EDTA/Saliva on swab
KRAS-EX	KRAS, Mutations		Codons 12, 13 and 61	KRAS	3 weeks	5 ml blood EDTA/Saliva on swab
BCR-ABL	Leucemia Mieloiide Crónica	608232	Fusion BCR/ABL	BCR/ABL	2 months	5 ml blood EDTA
TP53	Li-Fraumeni, Syndrome	151623	cds	p53 (TP53)	4 weeks	5 ml blood EDTA/Saliva on swab
HNPCC	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	MLH1, MSH2 and MSH6	4 months	10 ml blood EDTA
MLH1	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	MLH1	4 weeks	10 ml blood EDTA
MSH2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	MSH2	4 weeks	10 ml blood EDTA
MSH6	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	MSH6	6 weeks	10 ml blood EDTA

MLH1-MSH2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	Big Deletions	MLH1 and MSH2	4 weeks	5 ml blood EDTA
HNPCC-INES	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	Microsatellite instability (MSI)		4 weeks	Tumour sample and 5 ml blood EDTA
PMS2	Lynch Syndrome (Colorectal Cancer, Hereditary Nonpolyposis) (HNPCC)	120435	cds	PMS2	3 months	5 ml blood EDTA
FMTC	Medullary thyroid carcinoma (FMTC)	155240	cds	RET	3 months	5 ml blood EDTA
FMTC-EX	Medullary thyroid carcinoma (FMTC)	155240	Exons 10, 11, 13, 14, 15 and 16	RET	3 weeks	5 ml blood EDTA/Saliva on swab
P16	Melanoma Hereditary	600160	cds	p16	4 weeks	5 ml blood EDTA/Saliva on swab
MEN1	MEN 1 - Multiple Endocrine Neoplasia Type I	131100	cds	MEN1	4 weeks	5 ml blood EDTA
MEN1-DEL	MEN 1 - Multiple Endocrine Neoplasia Type I	131100	Big Deletions	MEN1	5 weeks	5 ml blood EDTA
MEN2A	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	171400	cds	RET	3 months	5 ml blood EDTA
MEN2A-2EX	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	171400	Exons 10 and 11	RET	3 weeks	5 ml blood EDTA/Saliva on swab
MEN2A-4EX	MEN 2A - Multiple Endocrine Neoplasia Type 2A (Sipple Syndrome)	171400	Exons 10, 11, 13, 14, 15 and 16	RET	4 weeks	5 ml blood EDTA/Saliva on swab
MEN2B	MEN 2B -Multiple Endocrine Neoplasia Type IIB	162300	cds	RET	3 months	5 ml blood EDTA
MEN2B-EX	MEN 2B -Multiple Endocrine Neoplasia Type IIB	162300	Exons 15 and 16 (p.M918T and	RET	3 weeks	5 ml blood EDTA/Saliva on swab
NBLAST1	Neuroblastoma, Susceptibility to (NBLST1)	256700	cds	KIF1B	5 months	5 ml blood EDTA
NBLAST2	Neuroblastoma, Susceptibility to (NBLST2)	603851	cds	PHOX2B	4 weeks	5 ml blood EDTA/Saliva on swab
NBLAST3	Neuroblastoma, Susceptibility to (NBLST3)	613014	cds	ALK	4 months	5 ml blood EDTA
NF2	Neurofibromatosis Type II (NF2)	101000	cds	NF2	2 months	5 ml blood EDTA
NF2-DEL	Neurofibromatosis Type II (NF2)	101000	Big Deletions	NF2	5 weeks	5 ml blood EDTA
NF1	Neurofibromatosis Type I (NF1)	162200	cds	NF1	5 months	5 ml blood EDTA
NF1-DEL	Neurofibromatosis Type I (NF1)	162200	Big Deletions	NF1	4 weeks	5 ml blood EDTA
NSCLC	Non Small Cell Lung Cancer (NSCLC)	211980	Exons 18-21	EGFR	4 weeks	5 ml blood EDTA/Saliva on swab
STK11	Peutz-Jeghers Syndrome	175200	cds	STK11	2 months	5 ml blood EDTA/Saliva on swab
SDHD	Pheochromocytoma and/or Paraganglioma	171300 and/or 168000	cds	SDHD	4 weeks	5 ml blood EDTA/Saliva on swab
SDHB	Pheochromocytoma and/or Paraganglioma	171300 and/or 168000	cds	SDHB	4 weeks	5 ml blood EDTA/Saliva on swab
SDHC	Pheochromocytoma and/or Paraganglioma	171300 and/or 168000	cds	SDHC	4 weeks	5 ml blood EDTA/Saliva on swab
RB1	Retinoblastoma	180200	cds	RB1	3 months	5 ml blood EDTA
RB1-DEL	Retinoblastoma	180200	Big Deletions	RB1	5 weeks	5 ml blood EDTA
TNFRSF	Squamous cell carcinoma of the head and neck	275355	cds	TNFRSF10B	7 weeks	5 ml blood EDTA/Saliva on swab
ING1	Squamous cell carcinoma of the head and neck	275355	cds	ING1	4 weeks	5 ml blood EDTA/Saliva on swab
VHL	Von Hippel Lindau Disease	193300	cds	VHL	2 weeks	5 ml blood EDTA/Saliva on swab

VHL-DEL	Von Hippel Lindau Disease	193300	Deletions/Duplications	VHL	5 weeks	5 ml blood EDTA
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* Result´s deadline starts at the arrival of the sample in our laboratory.

** The payment will be done in EUROS.

All analyses can be done from prenatal samples. Please consult.

If you want to make a specific test that is not included in this list, do not hesitate to ask to GENYCA´s Technical Management (info@genyca.es). This list will be updated periodically.

These prices do not include the delivery of the samples neither the sampling kits (tubes, swabs,...).