REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

GENOMED
Está nos genes.

Attachment II - TEST LIST

(SNS code/Test Description)

CARDIOLOGY

34900	Dilated cardiomyopathy: *MYH7, TNNT2, TPM1, ACTC1* genes	O
34900	Dilated cardiomyopathy: *ACTC1* gene	O
34666	Dilated cardiomyopathy: *LMNA* gene	O
36352	Dilated cardiomyopathy: *MYH7* gene	O
36353	Dilated cardiomyopathy: *TNNT2* gene	O
34900	Dilated cardiomyopathy: *TPM1* gene	O
34900	Hypertrophic cardiomyopathy: *MYBPC3, MYH7, TNNT2, TNNI3, TPM1, ACTC1, MYL2, MYL3* genes	O
34900	Hypertrophic cardiomyopathy: *ACTC1* gene	O
34900	Hypertrophic cardiomyopathy: *CSRP3* gene	O
34900	Hypertrophic cardiomyopathy: *FHL1* gene	O
36351	Hypertrophic cardiomyopathy: *MYBPC3* gene	O
36352	Hypertrophic cardiomyopathy: *MYH7* gene	O
34900	Hypertrophic cardiomyopathy: *MYL2* gene	O
34900	Hypertrophic cardiomyopathy: *MYL3* gene	O
36354	Hypertrophic cardiomyopathy: *TNNI3* gene	O
36353	Hypertrophic cardiomyopathy: *TNNT2* gene	O
34900	Hypertrophic cardiomyopathy: *TPM1* gene	O
34900	Angiotensin converting enzyme (*ACE*) I/D polymorphism genotype	O
34900	Left ventricular noncompaction / Barth syndrome: *TAZ* gene	O
34900	Brugada syndrome: *SCN5A* gene	O
34900	Long QT syndrome: *KCNQ1, KCNH2, SCN5A* genes	O
34900	Long QT syndrome (LQT1): *KCNQ1* gene	O
34900	Long QT syndrome (LQT2): *KCNH2* gene	O
34900	Long QT syndrome (LQT3): *SCN5A* gene	O
34900	Long QT syndrome (LQT5): *KCNE1* gene	O
34727	Fabry disease: *GLA* gene - index case	O
34726	Fabry disease: *GLA* gene - familial case	O
34900	Hereditary lymphedema type I (Milroy disease): *FLT4* gene (*VEGFR-3*)	O
34900	Lymphedema-distichiasis syndrome: *FOXC2* gene	O
34900	Marfan syndrome: *FBN1* gene	O
34900	Aneurysm / thoracic aortic dissection: *ACTA2* gene	O
34900	Ehlers-Danlos syndrome, classic form: *COL5A1* gene	O
34900	Ehlers-Danlos syndrome, vascular type: *COL3A1* gene	O
34900	Loeys-Dietz syndrome: *TGFBR1* gene	O
34900	Loeys-Dietz syndrome: *TGFBR2* gene	O

DERMATOLOGY

36285	Basal cell nevus syndrome (Gorlin-Goltz syndrome): *PTCH1* gene	O
34900	Darier disease: *ATP2A2* gene	O
34900	Epidermolysis bullosa simplex: *KRT5* gene	O
34900	Epidermolysis bullosa: *COL7A1* gene	O
34900	Generalized pustular psoriasis: *IL36RN* gene	O
34900	Psoriasis type 2: *CARD14* gene	O
34900	Hailey-Hayley disease: *ATP2C1* gene	O
34900	Hypertrophic osteoarthropathy: *HPGD* gene	O
34900	Hypertrophic osteoarthropathy: *SLCO2A1* gene	O
34900	Ichthyosis: *FLG* gene (p.R501* and c.2282del4)	O
34900	Keratoderma punctate: *AAGAB* gene	O
34900	Lipoid proteinosis: *ECM1* gene	O
34900	Neurofibromatosis type 1: *NF1* gene	O
34900	Neurofibromatosis type 1: *NF1* gene (del/dup analysis)	O
34900	Neurofibromatosis type 1: *NF1* gene (seq + del/dup analysis)	O
34900	Neurofibromatosis type 2: *NF2* gene	O
34900	Neurofibromatosis type 2: *NF2* gene (del/dup analysis)	O
34900	Neurofibromatosis type 2: *NF2* gene (seq + del/dup analysis)	O
34900	Rothmund-Thomson syndrome: *RECQL4* gene	O

IMMUNE SYSTEM DISEASES

34900	Autoimmune interstitial lung, joint, and kidney disease: *COPA* gene	O
34900	Autoimmune lymphoproliferative syndrome (ALPS): *FAS* (*TNFRSF6*) gene	O
34900	Autoinflammatory syndrome, familial, Behcet-like 1: *TNFAIP3* gene	O
34900	Autosomal dominant hyper-IgE syndrome: *STAT3* gene	O
34900	Hiper-IgE syndrome: *DOCK8* gene	O
34900	Hiper-IgE syndrome: *DOCK8* gene (del/dup analysis)	O
34900	Autosomal dominant periodic fever syndrome: *TNFRSF1A* gene	O
34900	Blau syndrome: *NOD2* gene	O
34900	CAPS (cryopyrin-associated periodic syndrome): *NLRP3* gene	O
34900	Emberger syndrome: *GATA2* gene	O
34900	Muckle-Wells syndrome: *NLRP3* gene	O
34900	Familial congenital neutropenia: *ELANE* (*ELA2*) gene	O
34900	Familial mediterranean fever: *MEFV* gene	O
34900	Familial mediterranean fever: *MEFV* gene (exons 2, 3 and 10)	O
34900	IL1RN deficiency (DIRA): *IL1RN* gene	O
34900	IL36RN deficiency (DITRA): *IL36RN* gene	O
34900	Majeed syndrome: *LPIN2* gene	O
34900	Mevalonate kinase deficiency: *MVK* gene	O
34900	PAPA syndrome: *PSTPIP1* gene	O
34900	Severe combined immunodeficiency: *CD3D* gene	O
34900	Severe combined immunodeficiency: *IL2RG* gene	O
34900	Severe combined immunodeficiency: *IL7R* gene	O
34900	Severe combined immunodeficiency: *RAG1* gene	O
34900	Severe combined immunodeficiency: *RAG2* gene	O
34900	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome: *ADA2 (CECR1)* gene	O

METABOLIC DISEASES

34900	17-alpha-hydroxylase deficiency: *CYP17A1* gene	O
36113	21-hydroxylase deficiency: *CYP21A2* gene - index case	O
34577	21-hydroxylase deficiency: *CYP21A2* gene - familial case	O
34900	Abetalipoproteinemia: *MTTP* gene	O
34900	Alkaptonuria: *HGD* gene	O
34900	Alpha-L-iduronidase deficiency: *IDUA* gene	O
34900	Apolipoprotein B deficiency (hypobetalipoproteinemia): *APOB* gene	O
36169	Classic familial hypercholesterolemia: *LDLR* gene	O
34900	Classic homocystinuria: *CBS* gene	O
34900	Congenital disorder of glycosylation type Ia: *PMM2* gene	O
36332	Creatine transport deficiency: *SLC6A8* gene - index case	O
36535	Creatine transport deficiency: *SLC6A8* gene - familial case	O
34900	D-2-hydroxyglutaric aciduria: *D2HGDH* gene	O
34900	Extraoral halitosis: *SELENBP1* gene	O
34483	G6PD deficiency: *G6PD* gene - index case	O
34484	G6PD deficiency: *G6PD* gene - familial case	O
34900	GLUT1 deficiency syndrome 1: *SLC2A1* gene	O
34742	Glycogen storage disease type Ia: *G6PC* gene - index case	O
34743	Glycogen storage disease type Ia: *G6PC* gene - familial case	O
34744	Glycogen storage disease type Ib/Ic: *G6PT1* gene - index case	O
34745	Glycogen storage disease type Ib/Ic: *G6PT1* gene - familial case	O
36345	Glycogen storage disease type III: *AGL* gene - index case	O
34747	Glycogen storage disease type III: *AGL* gene - familial case	O
34746	Glycogen storage disease type III: *AGL* gene (exons 3, 4, 21, 24, 28, 31, 33 and 35) - index case	O
36331	Guanidinoacetate methyltransferase deficiency: *GAMT* gene	O
34900	HMG-CoA lyase deficiency: *HMGCL* gene	O
36205	Homocystinuria due to remethylation deficiency: *MTHFR* gene - index	O
36460	Homocystinuria due to remethylation deficiency: *MTHFR* gene - familial case	O
34900	Hypercholesterolemia: *APOB* gene (R3500Q and R3531C)	O
34900	Lecithin-cholesterol acyltransferase deficiency: *LCAT* gene	O
34900	Lysinuric protein intolerance: *SLC7A7* gene	O
34900	McArdle disease: *PYGM* gene	O
34900	Methylmalonic aciduria and homocystinuria type cblC: *MMACHC* gene	O
34900	Methylmalonic aciduria and homocystinuria type cblD: *MMADHC* gene	O
34900	Methylmalonic aciduria: *MUT* gene	O
34900	Ornithine transcarbamylase deficiency: *OTC* gene (seq + del/dup analysis)	O
34900	Phenylketonuria: *PAH* gene	O
34900	Pituitary adenoma 1, multiple types: *AIP* gene	O
36108	Primary carnitin deficiency: *SLC22A5* (*OCTN2*) gene	O
34900	Xanthinuria, type I: *XDH* gene	O

ENDOCRINOLOGY

34900	Barakat syndrome: *GATA3* gene	O
34900	Combined pituitary hormone deficiency: *PROP1* gene	O
36197	Familial hypocalciuric hypercalcemia / Neonatal severe hyperparathyroidism: *CASR* gene - index case	O
36198	Familial hypocalciuric hypercalcemia / Neonatal severe hyperparathyroidism: *CASR* gene - familial case	O
34864	Hyperparathyroidism-jaw tumor syndrome: *HRPT2* gene - index case	O
34865	Hyperparathyroidism-jaw tumor syndrome: *HRPT2* gene - familial case	O

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34900	Hypergonadotropic hypogonadism / Pseudohermaphroditism with Leydig cell hypoplasia: *LHCGR* gene	O
34563	Hypogonadotropic hypogonadism (Kallmann syndrome): *ANOS1* (*KAL1*) gene - index case	O
34565	Hypogonadotropic hypogonadism (Kallmann syndrome): *ANOS1* (*KAL1*) gene - familial case	O
34900	Mayer-Rokitansky-Küster-Hauser syndrome: *WNT4* gene	O
34900	Albright's hereditary osteodystrophy: *GNAS* gene (seq + del/dup analysis + methylation)	O
34900	McCune-Albright syndrome: *GNAS* gene (codons R201 and Q227)	O
34900	MODY1: *HNF4A* gene	O
34900	MODY2: *GCK* gene	O
34900	MODY3: *HNF1A* gene	O
34900	MODY5 or diabetes syndrome and renal cysts: *HNF1B* gene	O
34900	MODY5 or diabetes syndrome and renal cysts: *HNF1B* gene (del/dup analysis)	O
34900	Nonautoimmune hyperthyroidism: *TSHR* gene	O
34900	Pseudohypoaldosteronism type 1: *NR3C2* gene	O
34869	Thyroid hormone resistance: *THRB* gene - index case	O
34870	Thyroid hormone resistance: *THRB* gene - familial case	O
34900	Vitamin D receptor deficiency: *VDR* gene	O

PHARMACOGENETICS

34900	Dihydropyrimidine dehydrogenase deficiency: *DPYD* gene	O
34900	Dihydropyrimidine dehydrogenase deficiency: *DPYD* gene (c.1236G>A, c.1679T>G, c.1905+1G>A and c.2846A>T)	O
34900	P450 genotyping: *CYP1A2* gene (*1C, *1F)	O
34900	P450 genotyping: *CYP2C19* gene (*3)	O
34900	P450 genotyping: *CYP2C9* gene (*2, *3)	O
34900	P450 genotyping: *CYP2D6* gene (*3, *4, *5, *6, *9, *10, *41)	O
34900	P450 genotyping: *CYP3A4* gene (*1B)	O
34900	Pseudocholinesterase deficiency: *BCHE* gene	O
36269	Warfarin sensitivity/resistance: *CYP2C9* gene (*2, *3)	O
36270	Warfarin sensitivity/resistance: *VKORC1* gene (c.-1639G>A)	O

GASTROENTEROLOGY

34900	Alpha1-antitrypsin deficiency: *SERPINA1* gene	O
34900	Alpha1-antitrypsin deficiency: *SERPINA1* gene (alleles PI*S and PI*Z)	O
36277	Congenital generalized lipodystrophy type 1: *AGPAT2* gene	O
34900	Congenital generalized lipodystrophy type 2: *BSCL2* gene	O
34900	Familial intrahepatic cholestasis (BRIC1 and PFIC1): *ATP8B1* gene	O
34900	Familial intrahepatic cholestasis (BRIC2 and PFIC2): *ABCB11* gene	O
34900	Familial intrahepatic cholestasis (PFIC3): *ABCB4* gene	O
36224	Familial partial lipodystrophy type 2 (type Dunnigan): *LMNA* gene - index case	O
34900	Familial partial lipodystrophy type 3: *PPARG* gene	O
34835	Crigler-Najjar syndrome: *UGT1A1* gene	O
34838	Gilbert syndrome: *UGT1A1* gene	O
34837	Gilbert syndrome: *UGT1A1* gene (dupTA)	O
34900	Hemochromatosis: *HFE* gene	O
34493	Hemochromatosis: *HFE* gene (H63D and C282Y)	O
34494	Hemochromatosis: *HFE* gene (H63D and S65C)	O
36190	Hemochromatosis: *HFE* gene (H63D, C282Y and S65C)	O
34900	Interleukin 28B genotyping (*IFNL3* gene: SNP - rs12969860)	O
34900	Acute intermittent porphyria: *HMBS* gene	O
34900	Hereditary coproporphyria: *CPOX* gene	O
34900	Porphyria *cutanea tarda*: *UROD* gene	O
34900	Porphyria *variegata*: *PPOX* gene	O
34900	Wilson disease: *ATP7B* gene	O
34900	Wilson disease: *ATP7B* gene (del/dup analysis)	O

HEMATOLOGY

34301	Antithrombin III deficiency: *SERPINC1* gene	O
34325	Drepanocytosis (sickle cell anemia): *HBB* gene	O
34497	Thrombophilia study - Factor II, Factor V, *MTHFR* and *PAI1*	O
34370	Factor II deficiency (gene *F2*; Prothrombin): G20210A	O
34361	Factor V deficiency (gene *F5*): Leiden mutation	O
34367	Hyperhomocysteinemia: *MTHFR* gene (C677T and A1298C)	O
34364	Plasminogen activator inhibitor type 1 (*PAI1*) (4G)	O
34310	Factor VII deficiency: *F7* gene - index case	O
34311	Factor VII deficiency: *F7* gene - familial case	O
34900	Factor XII deficiency: *F12* gene (C46T)	O
34900	Factor XIII deficiency: *F13A1* gene	O
34900	Factor XIII deficiency: *F13B* gene	O
34305	Protein S deficiency: *PROS1* gene - index case	O
34306	Protein S deficiency: *PROS1* gene - familial case	O
34900	von Willebrand disease type 1, 2, 3: *VWF* gene	O
34900	von Willebrand disease type 2A, 2B or 2M: *VWF* gene (exon 28)	O
34900	Glanzmann thrombasthenia: *ITGA2B* and *ITGB3* genes	O
34900	Glanzmann thrombasthenia: *ITGA2B* gene	O
34900	Glanzmann thrombasthenia: *ITGB3* gene	O
34900	Hemophagocytic syndrome: *STXBP2*, *STX11*, *PRF1*, *UNC13D* genes	O
34900	Osler-Weber-Rendu disease: *ACVRL1* gene	O
34900	Osler-Weber-Rendu disease: *ENG* gene	O
34900	Osler-Weber-Rendu disease: *SMAD4* gene	O
34900	Osler-Weber-Rendu disease: *ACVRL1* and *ENG* genes (del/dup analysis)	O
34900	Thrombotic thrombocytopenic purpura: *ADAMTS13* gene	O

NEPHROLOGY

34900	Adenine phosphoribosyltransferase deficiency: *APRT* gene	O
34900	*APOL1* genotyping: haplotypes G1 and G2	O
34900	Autosomal dominant hypocalcemia, with Bartter syndrome: *CASR* gene	O
34900	Autosomal dominant hypocalcemia: *CASR* gene	O
34900	Autosomal dominant polycystic kidney disease: *PKD1* gene	O
34900	Autosomal dominant polycystic kidney disease: *PKD2* gene	O
34900	Autosomal dominant polycystic kidney disease: *PKHD1* gene	O
34900	Bartter syndrome type I: *SLC12A1* gene	O
34900	Bartter syndrome type II: *KCNJ1* gene	O
34900	Bartter syndrome type III: *CLCNKB* gene	O
34900	Gitelman syndrome: *SLC12A3* gene	O
36564	Cystinosis: *CTNS* gene	O
34900	Diabetes *insipidus*, renal form: *AVPR2* gene	O
34900	Familial hypocalciuric and hypercalcemia type II: *GNA11* gene	O
34900	Familial hypocalciuric and hypercalcemia type III: *AP2S1* gene	O
34900	Familial renal glucosuria: *SLC5A2* gene	O
34900	Hyperoxaluria type 1: *AGXT* gene	O
34900	Hyperoxaluria type 2: *GRHPR* gene	O
34900	Familial juvenile hyperuricemic nephropathy: *REN* gene	O
34900	Medullary cystic kidney disease type 1: *MUC1* gene (ins Citosine)	O
34900	Medullary cystic kidney disease type 2: *UMOD* gene	O
34900	Nephrogenic syndrome of inappropriate antidiuresis (NSIAD): *AVPR2* gene	O
34900	Nephrotic syndrome / Pierson syndrome: *LAMB2* gene	O
34900	Nephrotic syndrome: *NPHS1* gene	O
34900	Nephrotic syndrome: *NPHS2* gene	O
34900	Nephrotic syndrome: *PLCE1* gene	O
34900	Nephrotic syndrome: *WT1* gene	O
34900	Renal tubular acidosis: *SLC4A1* gene	O
34900	Renal tubular dysgenesis: *ACE* gene	O
34900	Renal tubular dysgenesis: *REN* gene	O
34900	X-linked Alport syndrome: *COL4A5* gene	O

NEUROLOGY

34900	Aicardi-Goutieres syndrome: *RNASEH2B* gene	O
34900	Ataxia with vitamin E deficiency: *TTPA* gene	O
34900	CADASIL: *NOTCH3* gene	O
34900	CADASIL: *NOTCH3* gene (exons 2 to 6 and 11)	O
34900	CARASIL: *HTRA1* gene	O
36330	Cerebrotendinous xanthomatosis: *CYP27A1* gene	O
34900	Charcot-Marie-Tooth disease type 1A: *PMP22* gene (17 dup)	O
34900	Charcot-Marie-Tooth disease type 1B: *MPZ* gene	O
34900	Charcot-Marie-Tooth disease type 1E: *PMP22* gene	O
34900	Charcot-Marie-Tooth disease type 2B: *RAB7A* gene	O
34900	Charcot-Marie-Tooth disease type 2B1: *LMNA* gene	O
34900	Charcot-Marie-Tooth disease X linked: *GJB1* gene (Cx32)	O
34900	Charcot-Marie-Tooth disease: *PMP22* gene (17 dup + seq)	O
34900	Hereditary neuropathy with liability to pressure palsies (HNPP): *PMP22* gene	O
34900	Choreoacanthocytosis: *VPS13A* gene	O
34900	Coffin-Lowry syndrome: *RPS6KA3* gene	O
34900	Congenital muscular dystrophy (CMD1A): *LAMA2* gene	O
36137	Congenital muscular dystrophy (CMD): *LMNA* gene	O
34329	Creutzfeldt-Jakob disease: *PRNP* gene	O
34900	Dementia with Lewy bodies: *SNCA* gene	O
34812	Emery-Dreifuss muscular dystrophy (EDMD2): gene *LMNA*	O
34900	Emery-Dreifuss muscular dystrophy (EDMD6): *FHL1* gene	O

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34900	Familial Alzheimer disease: *APOE* genotyping	O
34900	Familial Alzheimer disease: *APP* gene (exons 16 and 17)	O
34900	Familial Alzheimer disease: *PSEN1* gene	O
34900	Familial Alzheimer disease: *PSEN1, PSEN2, APP* genes (exons 16 and 17)	O
34900	Familial Alzheimer disease: *PSEN2* gene	O
34900	Quantification of total TAU, phosphorylated TAU and β-amyloid on CSF	O
34900	Familial amyotrophic lateral sclerosis: *C9ORF72* gene (GGGGCC expansion)	O
34900	Familial amyotrophic lateral sclerosis: *FUS* gene	O
34900	Familial amyotrophic lateral sclerosis: *SETX* gene	O
34900	Familial amyotrophic lateral sclerosis: *SOD1* gene	O
34900	Familial amyotrophic lateral sclerosis: *TARDBP* gene	O
34900	Familial amyotrophic lateral sclerosis: *VCP* gene	O
34900	Familial cerebral cavernous malformations: *KRIT1* gene	O
34900	Familial cerebral cavernous malformations: *KRIT1* gene (p.Gln455*)	O
34900	Familial glioma: *POT1* gene	O
34900	Episodic ataxia type 2: *CACNA1A* gene	O
34900	Familial hemiplegic migraine type 1: *CACNA1A* gene	O
34900	Familial hemiplegic migraine type 2: *ATP1A2* gene	O
34900	Familial hemiplegic migraine type 3: *SCN1A* gene	O
36158	Generalized epilepsy with febrile seizures “plus”: *SCN1A* gene	O
34900	Friedreich ataxia: *FXN* gene	O
34900	Frontotemporal dementia and/or familial amyotrophic lateral sclerosis: *TBK1* gene	O
34900	Frontotemporal dementia: *C9ORF72* gene (GGGGCC expansion)	O
34900	Frontotemporal dementia: *CHMP2B* gene	O
34900	Frontotemporal dementia: *GRN* gene	O
34900	Frontotemporal dementia: *MAPT* (exons 1 and 9 to 13) and *GRN* genes	O
34900	Frontotemporal dementia: *MAPT* gene (exons 1 and 9 to 13)	O
34900	Frontotemporal dementia: *TARDBP* gene	O
34900	Frontotemporal dementia: *VCP* gene	O
36528	GTP cyclohydrolase: *GCH1* gene - familial case	O
36096	GTP cyclohydrolase: *GCH1* gene - index case	O
34900	Hereditary motor neuropathy type VA: *BSCL2* gene	O
34900	Hereditary neuralgic amyotrophy: *SEPTIN9* gene	O
34900	Hereditary sensory neuropathy type 1A (HSAN1A): *SPTLC1* gene	O
34900	Hereditary sensory neuropathy type 1C (HSAN1C): *SPTLC2* gene	O
34900	Huntington disease: *HTT* gene	O
34900	Kennedy disease: *AR* gene	O
34721	Leigh/NARP syndrome - Neuropathy, Ataxia, and Retinitis: *MT-ATP6* gene (m.8993T>G, m.8993T>C) - index case	O
34720	Leigh/NARP syndrome - Neuropathy, Ataxia, and Retinitis: *MT-ATP6* gene (m.8993T>G, m.8993T>C) - familial case	O
34673	Limb-girdle muscular dystrophy type 2A: *CAPN3* gene - index case	O
34900	Machado-Joseph disease: *ATXN3* gene	O
34900	MELAS syndrome - Mitochondrial encephalomyopathy: *MT-TL1* (m.3243A>G, m.3244G>A, m.3252A>G, m.3256C>T, m.3271T>C, m.3291T>C) and *MT-ND5* genes (m.13513G>A)	O
34716	MERRF syndrome - Myoclonic epilepsy: *MT-TK* gene (m.8296A>G, m.8344A>G, m.8356T>C, m.8363G>A) - index case	O
34717	MERRF syndrome - Myoclonic epilepsy: *MT-TK* gene (m.8296A>G, m.8344A>G, m.8356T>C, m.8363G>A) - familial case	O
36343	Mitochondrial DNA depletion syndrome - Encephalomyopathic with or without mild methylmalonic aciduria: *SUCLA2* gene	O
34528	Myotonic dystrophy (DM1; Steinert disease): *DMPK* gene	O
34900	Myotonic dystrophy (DM2): *CNBP* gene	O
34900	Moyamoya disease: *ACTA2* gene	O
34900	Moyamoya disease: *RNF213* gene	O
34900	Myotonia *congenita*: *CLCN1* gene	O
34875	Hereditary transthyretin amyloidosis (Andrade type): *TTR* gene [p.V50M (V30M)]	O
34853	Hereditary transthyretin amyloidosis: *TTR* gene - index case	O
34900	Parkinson’s disease (PARK 1, 2 and 8) (del/dup analysis)	O
34900	Parkinson’s disease (PARK1): *SNCA* gene	O
34900	Parkinson’s disease (PARK2): *PRKN* gene	O
34900	Parkinson’s disease (PARK2): *PRKN* gene (del/dup analysis)	O
34900	Parkinson’s disease (PARK4): *SNCA* gene (del/dup analysis)	O
34900	Parkinson’s disease (PARK6): *PINK1* gene	O
34900	Parkinson’s disease (PARK8): *LRRK2* gene	O
34900	Parkinson’s disease (PARK8): *LRRK2* gene (exons 31, 34, 35, 41, 48)	O
34900	Rett syndrome: *CDKL5* gene	O
36161	Severe myoclonic epilepsy of infancy (Dravet syndrome): *SCN1A* gene	O
34900	Spinal muscular atrophy: *SMN1* and *SMN2* genes	O
34900	Spinocerebellar ataxia: SCA1, 2, 3, 6, 7, 8 and 12 genes	O
34900	Tuberous sclerosis: *TSC1* and *TSC2* genes	O
34900	X-linked lissencephaly: *DCX* gene	O

OPHTHALMOLOGY

34900	Early-onset glaucoma: *MYOC* gene	O
34900	Primary congenital glaucoma: *CYP1B1* gene	O
34900	Gyrate atrophy of choroid and retina w/ or w/o ornithinemia: *OAT* gene	O
34712	LHON syndrome - Leber hereditary optic neuropathy: *MT-ND1* (m.3460G>A), *MT-ND4* (m.11778G>A), *MT-ND6* (m.14484T>C) genes - index case	O
34713	LHON syndrome - Leber hereditary optic neuropathy: *MT-ND1* (m.3460G>A), *MT-ND4* (m.11778G>A), *MT-ND6* (m.14484T>C) genes - familial case	O
34320	Premature retinopathy (Norrie disease): *NDP* gene - index case	O
34321	Premature retinopathy (Norrie disease): *NDP* gene - familial case	O
34900	Retinitis *pigmentosa*: *RPE65* gene	O
34900	Stargardt disease: *ABCA4* gene	O
34900	Wolfram syndrome: *WFS1* gene	O

ONCOLOGY

Gastroenterology

34633	Colon adenomatous polyposis, level I: gene *MUTYH* (p.Y179C and p.G396D) - index case	O
36264	Colon adenomatous polyposis, level II: *MUTYH* gene - index case	O
34635	Colon adenomatous polyposis: *MUTYH* gene - familial case	O
34900	Colon adenomatous polyposis: *MUTYH* gene (del/dup analysis)	O
34394	Familial adenomatous polyposis: *APC* gene - index case	O
34395	Familial adenomatous polyposis: *APC* gene - familial case	O
36263	Familial adenomatous polyposis: *APC* gene (del/dup analysis)	O
36064	Hereditary diffuse gastric cancer (E-cadherin): *CDH1* gene	O
34900	Lynch syndrome: *MLH1* and *MSH2* genes	O
34844	Lynch syndrome: *MLH1* and *MSH2* genes (del/dup analysis)	O
34637	Lynch syndrome: *MLH1* gene - index case	O
34638	Lynch syndrome: *MLH1* gene - familial case	O
34398	Lynch syndrome: *MSH2* gene - index case	O
34399	Lynch syndrome: *MSH2* gene - familial case	O
36068	Lynch syndrome: *MSH6* gene - index case	O
36069	Lynch syndrome: *MSH6* gene - familial case	O
34900	Lynch syndrome: *MSH6* gene (del/dup analysis)	O
36070	Lynch syndrome: *PMS2* gene - index case	O
36290	Peutz-Jeghers syndrome: *STK11 (LKB1)* gene	O

Gynecology / Obstetrics / Others

34900	Pack 1: *BRCA1* and *BRCA2* testing - index case Portuguese founder mutation (blood) + NGS (blood or FFPE) + MLPA (blood) [1]	O
34900	Pack 2: *BRCA1* and *BRCA2* testing - index case Portuguese founder mutation (blood) + NGS (blood or FFPE) [1]	O
34900	*BRCA1* and *BRCA2* sequencing	O
36061	Portuguese founder mutation *BRCA2* (insAlu) - index case	O
36062	Portuguese founder mutation *BRCA2* (insAlu) - familial case	O
34543	*BRCA1* sequencing - index case	O
34544	*BRCA1* sequencing - familial case	O
34547	*BRCA2* sequencing - index case	O
34548	*BRCA2* sequencing - familial case	O
36059	*BRCA1*/*BRCA2* del/dup analysis - index case	O
36060	*BRCA1*/*BRCA2* del/dup analysis - familial case	O

Others

36272	Cowden syndrome: *PTEN* gene - index case	O
36273	Cowden syndrome: *PTEN* gene - familial case	O
34900	Familial medullary thyroid carcinoma: *RET* gene	O
34857	Multiple endocrine neoplasia type 1: *MEN1* gene - index case	O
34858	Multiple endocrine neoplasia type 1: *MEN1* gene - familial case	O
34900	Multiple endocrine neoplasia type 2 (MEN2): *RET* gene	O
34900	Multiple endocrine neoplasia type 2A (MEN2A): *RET* gene	O
34900	Multiple endocrine neoplasia type 2B (MEN2B): *RET* gene	O
36248	Multiple endocrine neoplasia type 4 (MEN4): *CDKN1B* gene	O
34900	Pheochromocytoma and paraganglioma type 1: *SDHD* gene	O
34900	Pheochromocytoma and paraganglioma type 3: *SDHC* gene	O
34900	Pheochromocytoma and paraganglioma type 4: *SDHB* gene	O
34900	Screening of the *TNFRSF10A* (*APO2*) gene	O
34862	von Hippel-Lindau syndrome: *VHL* gene - index case	O
34863	von Hippel-Lindau syndrome: *VHL* gene - familial case	O
36299	von Hippel-Lindau syndrome: *VHL* gene (del/dup analysis)	O

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REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

GENOMED
Está nos genes.

Pneumology

34900	Birt-Hogg-Dube syndrome: *FLCN* gene	O

OTORHINOLARYNGOLOGY

34900	Alström syndrome: *ALMS1* gene	O
36083	Connexin 26: *GJB2* gene	O
36537	Connexin 26: *GJB2* gene - familial case	O
36084	Connexin 30: *GJB6* gene	O
36539	Connexin 30: *GJB6* gene - familial case	O
34900	Maternally inherited diabetes mellitus and deafness (MIDD): *MT-TL1* (m.3243A>G), *MT-TE* (m.14692A>G, m.14709T>C) and *MT-TK* (8296A>G) genes	O
34900	Mitochondrial nonsyndromic hearing loss and deafness (NSHL): *MT-RNR1* gene (m.1555A>G)	O
34776	Mitochondrial nonsyndromic hearing loss and deafness: *MT-RNR1* (m.961T>G, m.961_962delTinsC(n), m.1095T>C, m.1494C>T, m.1555A>G), *MT-TL1* (m.3243A>G), *MT-TS1* (m.7443A>G, m.7444G>A, m.7445A>G/C/T, m.7462C>T, m.7472dupC, m.7505T>C, m.7510T>C, m.7511T>C) genes - index case	O
34777	Mitochondrial nonsyndromic hearing loss and deafness: *MT-RNR1* (m.961T>G, m.961_962delTinsC(n), m.1095T>C, m.1494C>T, m.1555A>G), *MT-TL1* (m.3243A>G), *MT-TS1* (m.7443A>G, m.7444G>A, m.7445A>G/C/T, m.7462C>T, m.7472dupC, m.7505T>C, m.7510T>C, m.7511T>C) genes - familial case	O
34900	Waardenburg syndrome: *PAX3* gene	O

PEDIATRICS

34900	Bone dysplasia: *FGFR3* gene	O
34509	Achondroplasia level I: *FGFR3* gene (p.G380R)	O
34512	Achondroplasia level II: *FGFR3* gene (p.G380R and p.G375C)	O
34900	Hypochondroplasia: *FGFR3* gene (exons 13 and 15)	O
34900	Muenke-type craniosynostosis: *FGFR3* gene (p.P250R)	O
34900	Thanatophoric dysplasia: *FGFR3* gene	O
34900	Acrodysostosis: *PRKAR1A* gene (exon 11)	O
34900	Acrodysostosis: *PRKAR1A* gene	O
34900	Carney complex: *PRKAR1A* gene	O
34900	Alagille syndrome: *JAG1* gene	O
34900	Apert syndrome: *FGFR2* gene	O
34900	Bardet-Biedl syndrome: *TMEM67* gene	O
34900	CHARGE syndrome: *CHD7* gene	O
34900	Cherubism disease: *SH3BP2* gene	O
34900	Cleidocranial dysplasia: *RUNX2* gene	O
34900	Costello syndrome: *HRAS* gene (exon 2)	O
34900	Crouzon syndrome: *FGFR2* gene	O
34900	Denys-Drash syndrome: *WT1* gene	O
34900	Facioaudiosymphalangism syndrome: *NOG* gene	O
34900	Frasier syndrome: *WT1* gene	O
34900	Greig syndrome: *GLI3* gene	O
36439	Hutchinson-Gilford syndrome (Progeria): *LMNA* gene	O
34900	Hypophosphatemic rickets: *FGF23* gene	O
34900	Jackson-Weiss syndrome: *FGFR2* gene	O
34900	Lowe syndrome: *OCRL* gene	O
34900	Macrocephaly/autism syndrome: *PTEN* gene	O
34900	*Osteogenesis Imperfecta*: *COL1A1* and *COL1A2* genes	O
34900	Osteopetrosis: *CLCN7* gene	O
34900	Osteoporosis: *LRP5* gene	O
34900	Pfeiffer syndrome: *FGFR2* gene	O
34900	Pseudoachondroplasia: *COMP* gene	O
34900	Saethre-Chotzen syndrome: *TWIST1* gene	O
34900	Smith-McCort syndrome: *DYM* gene	O
34900	Werner syndrome: *WRN* gene	O
34900	Sotos syndrome: *NSD1* gene	O
34900	Weill-Marchesani syndrome (AD): *FBN1* gene	O
34900	Screening for mutations in the *TP63* gene	O

PNEUMOLOGY

34335	Cystic fibrosis (mucoviscidosis): *CFTR* gene - index case, level 1	O
34341	Cystic fibrosis (mucoviscidosis): *CFTR* gene - familial case	O
36180	Cystic fibrosis: *CFTR* gene	O
34900	Pulmonary hypertension: *BMPR2* gene	O

OTHERS*

34201	DNA extraction	O
	_____	O
	_____	O

[1] If you requested the somatic mutation research trough NGS (tumor sample), please let us know the percentage of tumor cells. Pack 1 and 2 include testing for mutation origin (germline or somatic) if blood and tumor are sent.

***NOTES:**

For other tests or NGS panels please see attachment I, contact us or visit our website.

If you would like to order another genetic test that is not listed here, please contact us.

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