KITGEN
Laboratory for molecular diagnostics
V Hurkach 3
158 00 Prague
Czech Republic

# Requisition form for human molecular syndromology (human molecular genetics) and predictive genetics

| **Patient:** | **Referring clinician:** |
|---|---|
| Name of the patient:<br>Address:<br>ID of the patient:<br>Sex:<br>Insurance type:<br>Diagnosis: | ID of the clinician:<br>ID of the referring institute:<br>Address:<br>Signature, Stamp: |

**Biological material (please specify):**

☐ Peripheral blood

☐ Buccal swab

☐ Other …….

Date and time of sampling:

**Requested tests** (please specify):
*The laboratory performs sequencing of whole coding regions and exon/intron boundaries of the genes listed below. The only exception is NGS multiplexes for predictive genetics, listed in separate section at the end of the requisition form.*

| | **Genetical syndrome** | **Associated gene / genes** |
|---|---|---|
| | α1-antitrypsin deficiency | SERPINA1 |
| | Alagille syndrome | JAG1 |
| | Aniridia | PAX6 |
| | Ataxia teleangiectasia | ATM |
| | Bardet-Biedl syndrome | BBS1 |
| | Bartter syndrome | SLC12A1 |
| | Bartter syndrome, type II | KCNJ1 |
| | Barttter syndrome, type III | CLCNKB |
| | Brachydactylia | ROR2 |
| | C3 Glomerulonephritis | C3, CD46, CFB, CFH, CFHR5, CFI |
| | CADASIL syndrome | NOTCH3 |

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|---|---|---|
| | Carnitinepalmytoyltransferase-1 deficiency | CPT1A |
| | Cockayne syndrome / Xeroderma pigmentosum | ERCC1,2,5,6,8 |
| | Coffin-Lowry syndrome | RPS6KA3 |
| | Congenital angioedema | SERPING1 |
| | Congenital pachyonychia, steatocystoma multiplex | KRT17 |
| | Costello syndrome | HRAS |
| | Cystic fibrosis | CFTR |
| | D-2-hydroxyglutaric aciduria | D2HGDH |
| | Dent's disease, X-linked nephrolithiasis, X-linked hypophosphatemic rachitis | CLCN5 |
| | Ehlers-Danlos syndrome | COL3A1 |
| | Ellis-van Creveld syndrome | EVC, EVC2 |
| | Fabry disease | GLA |
| | Factor VII deficiency | F7 |
| | Fibrodysplasia ossificans progressiva | ACVR1 |
| | Goldberg-Shprintzen syndrome | KIF1BP |
| | Hereditary pancreatitis | SPINK1, CLDN2, CPA1, CTRC, PRSS1 |
| | Hypercholesterolemia | LDLR |
| | Hypophosphatemia | ALPL |
| | CHARGE syndrome | CHD7 |
| | Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked | FLNA |
| | Iuvenile myoclonic epilepsy | EFHC1 |
| | Kallmann syndrome | FGFR1, KAL1 |
| | Kartagener syndrome | DNAI1 |
| | Kidney polycystosis | PKHD1 |
| | L-2-hydroxyglutaric aciduria | L2HGDH |
| | Leber congenital amaurosis | CEP290, GUCY2D |
| | LEOPARD syndrome | RAF1 |
| | Leucinosis | BCKDHA, BCKDHB, DBT |
| | Lisencephaly Subcortical laminar heterotopy | DCX |
| | Marfan syndrome | FBN1 |
| | MASA syndrome | L1CAM |
| | Microcephaly | MCPH1 |
| | Migraine, hemiplegic, familial | CACNA1A |
| | MOPD II | PCNT |
| | Mowat-Wilson syndrome | ZEB2 |
| | Multiple endocrine neoplasia IIB | RET, CDC73 (aka HRPT2) |

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|---|---|---|
| | Multiple exostosis | EXT1, EXT2 |
| | N. opticus hypoplasia, micropthalmia | SOX2 |
| | Nail-patella syndrome | LMX1B |
| | Neurofibromatosis type I | NF1 |
| | Neutropenia, cyclic | ELANE |
| | Noonan syndrome | PTPN11, SOS1, KRAS, NRAS, BRAF, RAF1 |
| | Ocular albinism type I | GPR143 |
| | Ollier disease | PTH1R |
| | Opitz GBBB syndrome, type I | MID1 |
| | Opitz GBBB syndrome, type II | SPECC1L |
| | Osteogenesis imperfecta | ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, P3H1, LRP5, PLOD2, PLS3, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1 |
| | Peters-plus syndrome | B3GALT1 |
| | Pickardt syndrome, Morsier syndrome | HESX1 |
| | Progressive supranuclear paralysis | MAPT |
| | Proteus syndrome | AKT1 |
| | Retinopathy, autosomal dominant | FZD4, C1QTNF5, GNAT1, CAPN5, NR2E3, RHO |
| | Robinow syndrome | DVL1, DVL3 |
| | Rubinstein-Taybi syndrome | CREBBP |
| | Sotos syndrome | NSD1, NFIX, EZH2, PTEN, DNMT3A, SETD2, IGF2, GPC3, APC2 |
| | Short stature, idiopathic familial | SHOX |
| | Stickler syndrome | COL2A1, COL11A1 |
| | Surfactant dysfunction | ABCA3, SFTPC |
| | Syndromic myastenia | RAPSN |
| | Testicular feminization | DHH |
| | Three-M syndrome (3M) | CUL7 |
| | Tricho-rhino-phalangeal syndrome | TRPS1 |
| | Usher syndrome | USH2A |
| | Waardenburg syndrome | PAX3, SOX10, SNAI2, EDN3, EDNRB, MITF, TYR |
| | Werner syndrome | RECQL2 |
| | Wilms tumor syndrome | WT1 |
| | X-linked Alport syndrome | COL4A5 |
| | X-linked hypophosphatemia | FGF23, PHEX |
| | X-linked mental retardation | MED12, SMS, UPF3B, KDM5C (aka JARID1C), SLC16A2, PQBP1, SLC6A8 |
| | X-linked mental retardation, Aarskog-Scott syndrome | FGD1 |
| | X-linked retinitis pigmentosa | RPGR, RP2 |

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|---|---|---|
| | Solid tumors (somatic mutations); NGS panel | HRAS, KRAS, NRAS, BRAF, EGFR, RET, CDC73 |
| | Breast and ovarian cancer (germline mutations); NGS panel | BRCA-1, BRCA-2 |
| | Breast and ovarian cancer (germline mutations); NGS panel; extended | BRCA-1, BRCA-2, TP53, MLH1, MSH2 |
| | Colon cancer (germline mutations); NGS panel | APC, PTEN, STK11, MLH1, MRE11A, MSH2, MSH6, MUTYH, PMS1, PMS2 |
| | Prostate cancer (germline mutations); NGS panel | BRCA-1, BRCA-2 |
| | Pancreatic cancer (germline mutations); NGS panel | PALB2, TP53, STK11, BRCA-1, BRCA-2 |
| | Gastric cancer (germline mutations); NGS panel | CDH1, MLH1, MSH2, TP53, STK1 |
| | Lung cancer (germline mutations); NGS panel | TP53, EGFR, KRAS |
| | Other types of cancer (germline mutations); NGS panel | BARD1, BRIP1, CHEK2, MEN1, NBN, RAD50, RAD51C, RAD51D, XRCC2 |
| | Female infertility NGS panel | CYP21A2, FSHR, LHCGR, BMP15, LHB, ZP1, FMR1 |
| | Male infertility NGS panel | AR, CATSPER1, CFTR, FSHR, LHCGR |
| | Familial hypercholesterolemia NGS panel | APOB, LDLR, PCSK9 |
| | Dementia NGS panel | APOE, APP, PRNP, PSEN1, PSEN2, SORL1, TREM2 |

☐ Other gene / panel (please specify) …….

**Panels for predictive genetics (multiplex NGS genotyping of selected SNPs)**

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|---|---|
| DetoxiGenePlex - NGS panel | CYP1A1*2A, CYP1A1*2C, CYP1B1, CYP1B1, CYP2A6*2, CYP2C9*2, CYP2C9*3, CYP2C19*2, CYP2C19*3, CYP2D6*3, CYP3A4*17, CYP3A4*1B, COMT, NAT1, NAT1, NAT2, NAT2, NAT2, NAT2, NAT2, GSTM1, GSTP1, GSTP1, SOD1, SOD1, SOD2 |
| EstroGenePlex - NGS panel | APOE, APOE, APOE, CYP1B1, CYP1B1, MTHFR, MTHFR, COMT, TNFa, IL-6, VDR, CYP1A1*2A, CYP1A1*2C, GSTM1, GSTP1, GSTP1, GP3A (ITGB3), PAI-1, FII Prothrombin |
| CardioGenePlex - NGS panel | APOE, APOE, APOE, CETP, CETP, CETP, SELE, MTHFR, MTHFR, GNB3, AGT, AGT, AGTR1, CYBA, GP3A (ITGB3), PAI-1, FII Prothrombin, FV Leiden |

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|---|---|
| NeuroGenePlex - NGS panel | MTHFR, MTHFR, COMT, GSTM1, GSTP1, GSTP1, SOD2 |
| ImmunoGenePlex - NGS panel | IL-1b, IL-1b, TNFa, IL-4, IL-6, IL-10, IL-13 |

| **Address of the laboratory:** | **Information on sample acceptance:** |
|---|---|
| KITGEN<br>Laboratory for molecular genetics<br>V Hurkach 3<br>158 00 Prague<br>CZECH REPUBLIC<br>www.kitgen.eu<br>mobile: +420 732 517 266<br>e-mail: info@kitgen.eu | Date and time of sample acceptance by the laboratory:<br><br><br><br>Sample accepted by / person responsible: |