



## VYŠETŘOVANÉ NGS PANELE - SEZNAM GENŮ

**KLINICKÝ EXOM:** 4800 genů

seznam viz: <https://www.vfn.cz/pacienti/kliniky-ustavy/ustav-biologie-a-lekarske-genetiky/laborator/>

### NEURO panel (177 genů):

A2M, ABCA7, ADCY5, ADH1C, ANO3, APOE, APP, ARSB, ATP13A2, ATP1A3, ATXN2, ATXN3, ATXN8OS, BCAP31, BCKDK, BDNF, BST1, C9orf72, CACNA1B, CACNA1G, CACNA1S, CCFN, CLCN1, COMT, COQ2, CSF1R, CYP27A1, CYP2D6, DCAF17, DCTN1, DLG2, DNAJC12, DNAJC13, DNAJC6, DRD3, DUSP10, EIF2AK3, EIF4EBP1, EIF4G1, ELAVL4, FA2H, FAM71A, FAM83A, FBXO7, FIG4, FITM2, FUS, GAK, GBA, GCH1, GIGYF2, GLUD2, GNAL, GPATCH2L, GPNMB, GRIA1, GRIA2, GRIA3, GRIA4, GRIK1, GRIK2, GRIK3, GRIK4, GRIK5, GRIN1, GRIN2A, GRIN2B, GRIN2C, GRIN2D, GRIN3A, GRIN3B, GRN, HFE, HSPA9, HTRA2, CHCHD10, CHCHD2, CHMP2B, INPP5F, KCNJ2, KCNMA1, KMT2B, LIN28A, LRP10, LRRK2, MAOB, MAPT, MC1R, MECR, MIPEP, MIR4697, MOBP, MPO, NOS3, NPC1, NPC2, NR4A2, NUCKS1, OPTN, PARK7, PARL, PDE10A, PDGFB, PDGFRB, PINK1, PLA2G6, PLAU, PNKD, PODXL, POLG, PRKAG2, PRKN, PRKRA, PRNP, PRRT2, PSEN1, PSEN2, PTPRH, RAB29, RAB39B, RIT2, RNF11, RNF216, RUNX2, SCARB2, SCN4A, SGCE, SHC2, SIGMAR1, SIPA1L2, SLC1A4, SLC20A2, SLC2A1, SLC39A14, SLC6A3, SLCO1A2, SMPD1, SNCA, SNCAIP, SNCB, SOD1, SOD2, SORL1, SPG11, SPR, SQSTM1, STK39, STX6, SYNJ1, SYT11, TARDBP, TBK1, TBP, TH, THAP1, TIA1, TMEM175, TMEM229B, TMEM230, TMPRSS9, TNF, TOR1A, TREM2, TRPM7, TUBA4A, UBE3A, UBQLN2, UBTF, UHRF1BP1L, UCHL1, USP24, VAC14, VCP, VPS13A, VPS13C, VPS35, XPR1

### OIDIP panel v1 (263 genů):

ABCA1, ABCG5, ABCG8, ACAN, ACE, ACTN4, ADAMTS13, AGT, AGTR1, AGXT, AIP, AKR1C2, AKR1C4, ALB, ALG8, ALG9, AMH, AMHR2, ANGPTL3, ANLN, ANOS1, APOA1, APOA5, APOB, APOC2, APOE, APOL1, AQP2, AR, ARHGDI, ATP1A2, AVPR2, BMP2, BMPR1B, BRAF, BSND, BTK, C3, CACNA1A, CACNA1D, CACNA1H, CALCA, CALCB, CALCL, CASR, CBL, CBX2, CCDC8, CD2AP, CD46, CDKL5, CDKN1B, CETP, CFB, CFH, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCNKA, CLCNKB, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COMP, COQ8B, CRB2, CRCP, CREBBP, CUL7, CYP11B1, CYP11B2, DGKE, DHH, DIO1, DIO2, DIO3, DNAJB11, DUOX1, DUOX2, DUOXA1, DUOXA2, DVL1, DZIP1L, EP300, FBN1, FBN2, FGF8, FGFR1, FGFR2, FGFR3, FN1, FOXA2, FOXE1, FOXG1, GANAB, GDF5, GH1, GHR, GHRHR, GLI2, GLI3, GNAS, GPD1, GPIHBP1, GRHR, HESX1, HNF1B, HOGA1, HRAS, HSD17B3, CHD7, IGF1, IGF1R, IGF2, IGFALS, IGSF1, IHH, INF2, INSR, IRS4, IYD, KCNJ1, KCNJ5, KRAS, LAMB2, LCAT, LDLR, LDLRAP1, LEP, LEPR, LEPROT, LHCGR, LHX3, LHX4, LIPA, LIPC, LMF1, LMX1B, LPL, LRP5, LZTR1, MAGED2, MAGI2, MAP2K1, MAP2K2, MAP3K1, MATN3, MC3R, MC4R, MCM9, MECP2, MEN1, MYH9, MYO1E, NF1, NF2, NKX2-1, NKX2-5, NPHS1, NPHS2, NPPC, NPR2, NR0B1, NR5A1, NRAS, OBSL1, OCRL, OFD1, OTX2, PAH, PAX2, PAX8, PCSK1, PCSK9, PITX2, PKD2, PKHD1, PLCE1, POMC, POU1F1, PRKAR1A, PRKCSH, PROK2, PROKR2, PROP1, PRRT2, PTH1R, PTHLH, PTPN11, PTPRO, RAF1, RAMP1, RASA2, REN, RIT1, ROR2, RRAS, RUNX2, SAR1B, SCN1A, SEC61A1, SEC61B, SEC63, SECISBP2, SERPINA7, SHH, SHOC2, SHOX, SLC12A1, SLC12A3, SLC16A2, SLC1A3, SLC26A2, SLC26A4, SLC4A4, SLC5A5, SMARCB1, SOS1, SOS2, SOX10, SOX2, SOX3, SOX9, SRD5A2, SRY, STAP1, STAT5B, TBL1X, TG, TGFBR1, TGFBR2, THBD, THRA, THRB, TMEM67, TPO, TRH, TRHR, TRPC6, TRPS1, TSC1, TSC2, TSHB, TSHR, UBE3A, UMOD, VHL, WNT5A, WT1, ZFPM2

**OIDIP v1 – cílená analýza 62 SNPs:** rs2479409, rs629301, rs1367117, rs4299376, rs1564348, rs1800562, rs3757354, rs11220462, rs8017377, rs6511720, rs429358, rs7412, rs2187668, rs7454108, rs4639334, rs2395182, rs7775228, rs4713586, rs2066844, rs2066845, rs2066847, rs1801133, rs1801131, rs1801394, rs9923231, rs1799853, rs1057910, rs2108622, rs56040400, rs373359894, rs6025, rs1800595, rs1799963, rs5918, rs4988235, rs3753394, rs800292, rs1061170, rs3753396, rs1410996, rs1065489, rs2796267, rs2796268, rs1962149, rs859705, rs7144, rs863223306, rs587777176, rs72648337, rs1131691067, rs752405769, rs730880269, rs60284988, rs1313468278, rs727503425, rs1600152215, rs1239450803, rs794726837, rs17782313, rs751141, rs75039782, rs1805177



## VYŠETŘOVANÉ NGS PANELY - SEZNAM GENŮ

### OIDIP® panel v2 (272 genů): geny navíc jsou zvýrazněny

ABCA1, ABCG5, ABCG8, ACAN, ACE, ACTN4, ADAMTS13, AGT, AGTR1, AGXT, AIP, AKR1C2, AKR1C4, ALB, ALG8, ALG9, AMH, AMHR2, ANGPTL3, ANLN, ANOS1, APOA1, APOA5, APOB, APOC2, APOE, APOL1, AQP2, AR, ARHGDI, ATP1A2, AVPR2, BMP2, BMPR1B, BRAF, BSND, BTK, C3, CACNA1A, CACNA1D, CACNA1H, CALCA, CALCB, CALCL, CASR, CBL, CBX2, CCDC8, CD2AP, CD46, CDKL5, CDKN1B, CETP, CFB, CFH, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCNKA, CLCNKB, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COMP, COQ8B, CRB2, CRCP, CREBBP, CUL7, CYP11B1, CYP11B2, DGKE, **DHCR7**, DHH, DIO1, DIO2, DIO3, DNAJB11, DUOX1, DUOX2, DUOXA1, DUOXA2, DVL1, DZIP1L, EP300, FBN1, FBN2, FGF8, FGFR1, FGFR2, FGFR3, FN1, FOXA2, FOXE1, FOXG1, GANAB, GDF5, GH1, GHR, GHRHR, **GJB2**, GLI2, GLI3, GNAS, GPD1, GPIHBP1, **GREB1**, **GREB1L**, GRHPR, HESX1, HNF1B, HOGA1, HRAS, HSD17B3, CHD7, IGF1, IGF1R, IGF2, IGFALS, IGSF1, IHH, INF2, INSR, IRS4, IYD, KCNJ1, KCNJ5, KRAS, LAMB2, LCAT, LDLR, LDLRAP1, LEP, LEPR, LEPROT, LHCGR, LHX3, LHX4, LIPA, LIPC, LMF1, LMX1B, LPL, LRP5, LZTR1, MAGED2, MAGI2, MAP2K1, MAP2K2, MAP3K1, MATN3, MC3R, MC4R, MCM9, MECP2, MEN1, **MRAS**, MYH9, MYO1E, NF1, NF2, NKX2-1, NKX2-5, NPHS1, NPHS2, NPPC, NPR2, NROB1, NR5A1, NRAS, OBSL1, OCRL, OFD1, OTX2, PAH, PAX2, PAX8, PCSK1, PCSK9, PITX2, PKD2, PKHD1, PLCE1, POMC, POU1F1, **PPP1CB**, PRKAR1A, PRKCSH, PROK2, PROKR2, PROP1, PRRT2, PTH1R, PTHLH, PTPN11, PTPRO, RAF1, RAMP1, RASA2, REN, RIT1, ROR2, RRAS, **RRAS2**, RUNX2, SAR1B, SCN1A, SEC61A1, SEC61B, SEC63, SECISBP2, SERPINA7, SHH, SHOC2, SHOX, SLC12A1, SLC12A3, SLC16A2, SLC1A3, SLC26A2, SLC26A4, SLC4A4, SLC5A5, SMARCB1, SOS1, SOS2, SOX10, SOX2, SOX3, SOX9, **SPRED1**, **SPRED2**, SRD5A2, SRY, STAP1, STAT5B, TBL1X, TG, TGFB1, TGFB2, THBD, THRA, THRB, TMEM67, TPO, TRH, TRHR, TRPC6, TRPS1, TSC1, TSC2, TSHB, TSHR, UBE3A, UMOD, VHL, WNT5A, WT1, ZFPM2

**OIDIP v2 – cílená analýza** (navíc ve verzi v2): ANXA5 (NM\_001154) - promotor; FSHR (NM\_000145) - exon 6, SMN1 (NM\_000344) - exon 7 a 8

### OIDIP® panel v3 (275 genů): geny navíc (oproti v2) jsou zvýrazněny

ABCA1, ABCG5, ABCG8, ACAN, ACE, ACTN4, ADAMTS13, AGT, AGTR1, AGXT, AIP, AKR1C2, AKR1C4, ALB, **ALG5**, ALG8, ALG9, AMH, AMHR2, ANGPTL3, ANLN, ANOS1, APOA1, APOA5, APOB, APOC2, APOE, APOL1, AQP2, AR, ARHGDI, ATP1A2, AVPR2, BMP2, BMPR1B, BRAF, BSND, BTK, C3, CACNA1A, CACNA1D, CACNA1H, CALCA, CALCB, CALCL, CASR, CBL, CBX2, CCDC8, CD2AP, CD46, CDKL5, CDKN1B, CETP, CFB, CFH, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCNKA, CLCNKB, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COMP, COQ8B, CRB2, CRCP, CREBBP, CUL7, CYP11B1, CYP11B2, DGKE, DHCR7, DHH, DIO1, DIO2, DIO3, DNAJB11, DUOX1, DUOX2, DUOXA1, DUOXA2, DVL1, DZIP1L, EP300, FBN1, FBN2, FGF8, FGFR1, FGFR2, FGFR3, FN1, FOXA2, FOXE1, FOXG1, GANAB, GDF5, GH1, GHR, GHRHR, GJB2, GLI2, GLI3, GNAS, GPD1, GPIHBP1, GREB1, GREB1L, GRHPR, HESX1, HNF1B, HOGA1, HRAS, HSD17B3, CHD7, **IFT140**, IGF1, IGF1R, IGF2, IGFALS, IGSF1, IHH, INF2, INSR, IRS4, IYD, KCNJ1, KCNJ5, KRAS, LAMB2, LCAT, LDLR, LDLRAP1, LEP, LEPR, LEPROT, LHCGR, LHX3, LHX4, LIPA, LIPC, LMF1, LMX1B, LPL, LRP5, LZTR1, MAGED2, MAGI2, MAP2K1, MAP2K2, MAP3K1, MATN3, MC3R, MC4R, MCM9, MECP2, MEN1, MRAS, MYH9, MYO1E, NF1, NF2, NKX2-1, NKX2-5, NPHS1, NPHS2, NPPC, NPR2, NROB1, NR5A1, NRAS, OBSL1, OCRL, OFD1, OTX2, PAH, PAX2, PAX8, PCSK1, PCSK9, PITX2, PKD2, PKHD1, PLCE1, **PMM2**, POMC, POU1F1, PPP1CB, PRKAR1A, PRKCSH, PROK2, PROKR2, PROP1, PRRT2, PTH1R, PTHLH, PTPN11, PTPRO, RAF1, RAMP1, RASA2, REN, RIT1, ROR2, RRAS, RRAS2, RUNX2, SAR1B, SCN1A, SEC61A1, SEC61B, SEC63, SECISBP2, SERPINA7, SHH, SHOC2, SHOX, SLC12A1, SLC12A3, SLC16A2, SLC1A3, SLC26A2, SLC26A4, SLC4A4, SLC5A5, SMARCB1, SOS1, SOS2, SOX10, SOX2, SOX3, SOX9, SPRED1, SPRED2, SRD5A2, SRY, STAP1, STAT5B, TBL1X, TG, TGFB1, TGFB2, THBD, THRA, THRB, TMEM67, TPO, TRH, TRHR, TRPC6, TRPS1, TSC1, TSC2, TSHB, TSHR, UBE3A, UMOD, VHL, WNT5A, WT1, ZFPM2

**OIDIP v3 – cílená analýza** - shodné s v2



## VŠEOBECNÁ FAKULTNÍ NEMOCNICE V PRAZE

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Ústav biologie a lékařské genetiky | Albertov 2048/4, 128 00 Praha 2

## VYŠETŘOVANÉ NGS PANELY - SEZNAM GENŮ

**SEZNAM AKREDITOVANÝCH METOD S UPLATNĚNÝM FLEXIBILNÍM ROZSAHEM AKREDITACE JE UVEDEN V DOKUMENTU „SEZNAM ČINNOSTÍ V RÁMCI FLEXIBILNÍHO ROZSAHU AKREDITACE“.**

Vše je dostupné na: <https://www.vfn.cz/pacienti/kliniky-ustavy/ustav-biologie-a-lekarske-genetiky/laborator/>

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