

### Panel dysmorfia



Badanie to jest zaprojektowane w celu diagnozy pacjentów cierpiących na zespół dysmorficzny. Panel obejmuje 776 genów powiązanych z zaburzeniami takimi jak RASopatie, nerwiakowłókniaćkowatość typu 1 czy zespołem Noonan. Wykonywany jest metodą sekwencjonowania nowej generacji (NGS) wraz z analizą CNV.

#### Zespoły i zaburzenia objęte zakresem m. in.:

- ✓ Zespół Bardeta-Biedla
- ✓ Zespół sercowo-twarzowo-skórny
- ✓ Malformacje jamiste mózgu
- ✓ Rozszczep wargi i podniebienia
- ✓ Zespół Cornellii de Lange
- ✓ Dysplazja szkieletowa ciliopatyczna
- ✓ Kraniosynostoza i zaburzenia twarzoczaszki
- ✓ Zespół heterotaksji
- ✓ Choroba Hirschsprunga
- ✓ Lissencefalia i wady rozwojowe mózgu
- ✓ Dysplazja przynasadowa i mnoga nasadowa
- ✓ Neurofibromatoza
- ✓ Zespoły Noonana-RASopatii
- ✓ Zespół Seckela
- ✓ Rozległa dysplazja szkieletowa
- ✓ Zespół Sticklera
- ✓ Stwardnienie guzowate
- ✓ Zespół Waardenburga

CENA  
**3890 ZŁ**

Czas oczekiwania na wynik:  
**25 dni roboczych**

Analizowane geny: A2ML1, ABCA12, ABCB6, ABCC6, ABL1, ACP5, ACTA1, ACTA2, ACTB, ACTG1, ACVR2B, ADAMTS18, ADAMTS2, ADAMTSL2, ADGRG1, ADGRG6, AEBP1, AFF4, AGPS, AGRN, AHDC1, AHII, AKRIC4, AKT3, ALDH18A1, ALDH1A3, ALG2, ALPL, ALX1, ALX4, AMELX, AMER1, AMH, AMHR2, AMPD2, ANKH, ANKLE2, ANKRD11, ANKS6, ANO5, ANOS1, AP4M1, ARFGF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARSL, ARX, ASPM, ASXL1, ASXL3, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1E1, ATP7A, ATR, ATRX, B3GALNT2, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCL11A, BCOR, BGN, BIN1, BMP1, BMP2, BMP4, BMPRI1B, BNC2, BRAF, C1R, C1S, CANT1, CASK, CASR, CBL, CBS, CC2D2A, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2, CCN6, CCNO, CDC45, CDH1, CDK13, CDK5RAP2, CDKN1C, CDON, CENPF, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP418, CFAP53, CFL2, CHAT, CHD4, CHD7, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CILK1, CIT, CLCN5, CLP1, CNOT1, CNTNAP1, COASY, COG5, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COX7B, CPAP, CPLANE1, CRB2, CREB3L1, CREBBP, CRELD1, CRIPT, CRPPA, CRTAP, CSGALNACT1, CSPP1, CTNNA2, CTNND1, CUL4B, CUL7, CYB5A, CYP19A1, CYP1B1, CYP26B1, DAG1, DCC, DCHS1, DCX, DDR2, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DMP1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB13, DNAL1, DNM2, DOCK6, DOK7, DPAGT1, DPF2, DRC1, DSE, DVL3, DYM, DYNCH1, DYNCH2, DYNCH21, DYNCH212, DYNCH211, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFN1, EFTUD2, EGR2, EIF2AK3, EIF2S3, ELN, ENPP1, EOGT, EPG5, ERCC1, ERCC2, ERCC5, ERCC6, ERF, ESCO2, EVC, EVC2, EXOSC3, EXOSC9, EXT1, EXT2, EYA1, FAM20C, FAS, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL4, FBXW11, FGD1, FGF10, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLCN, FLNA, FLNB, FOXC1, FOXE1, FOXL2, FRAS1, FREM1, FREM2, FZD6, GAS8, GATA4, GBA1, GBE1, GDF1, GDF3, GDF5, GDF6, GDNF, GFPT1, GGCX, GJA1, GLE1, GLI2, GLI3, GMPPB, GNAS, GNPAT, GORAB, GPC3, GPC6, GRHL3, GRIPI1, HBA1, HCCS, HDAC8, HES7, HESX1, HEXA, HMX1, HOXA13, HOXD13, HRAS, HSPG2, HUWE1, HYDIN, HYL1, IER3IP1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IGF1R, IHH, IL11RA, INPP5E, INPPL1, INVS, IPO8, IRF6, ITGB4, JAG1, KAT6B, KATNB1, KBTBD13, KDM5C, KDM6A, KIAA0586, KIF11, KIF14, KIF22, KIF2A, KIF5C, KIF7, KIFBP, KIT, KLHL40, KLHL41, KMT2A, KMT2D, KNL1, KRAS, KRIT1, LICAM, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LARGE1, LBR, LFNG, LIFR, LMNA, LMOD3, LMX1B, LOX, LRP2, LRP4, LRP5, LTBP3, LZTFL1, LZTR1, MAB21L2, MACF1, MAFB, MAGEL2, MAP2K1, MAP2K2, MASP1, MATN3, MBTPS2, MCIDAS, MCPH1, MED12, MED13L, MED17, MEGF8, MEIS2, MEOX1, MESP2, MFAP5, MFRP, MFSD2A, MGP, MID1, MITF, MKKS, MKS1, MMP13, MMP15, MMP21, MMP9, MN1, MRAS, MSMO1, MSX1, MSX2, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTM1, MUSK, MYBPC1, MYCN, MYH11, MYH2, MYH3, MYH8, MYLK, MYO18B, MYO7A, MYO9A, NAA10, NALCN, NBAS, NCAPD3, NDE1, NDP, NEB, NECTIN1, NEK1, NEK8, NEK9, NEPRO, NF1, NF2, NHEJ1, NIPBL, NKX2-5, NKX3-2, NME8, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPR2, NR2F2, NRAS, NRG1, NSD1, NSDHL, NSUN2, NT5E, OBSL1, OCRL, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, OPHN1, ORC1, OTX2, P3H1, P4HB, PAFAH1B1, PAPSS2, PAX2, PAX3, PAX6, PAX7, PAX9, PCNT, PDCD10, PDE4D, PDE6D, PEX7, PGM1, PHEX, PHF6, PHF8, PHYH, PIBF1, PIEZO2, PIGA, PIGV, PIP5K1C, PITX2, PKD1L1, PKD2, PKHD1, PLEKHA7, PLK4, PLOD1, PLOD2, PLP1, PMM2, PNKP, PNPLA6, POLA1, POLR1C, POLR1D, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PORCN, PPARC, PPIB, PPP1CB, PQBP1, PRDM5, PREPL, PRKARIA, PRKG1, PRPH2, PRSS56, PTCH1, PTH1R, PTHLH, PTPN11, PXDN, PYCR1, PYCR2, QARS1, RAB18, RAB23, RAB3GAP1, RAB3GAP2, RAD21, RAF1, RAPSN, RARB, RARS2, RASA1, RASA2, RAX, RBBP8, RBP4, RBPJ, RELN, RET, RHO, RIN2, RIPK4, RIPPLY2, RIT1, ROBO3, ROR2, RRGRIPI1, RPL10, RPS6KA3, RSPH1, RSPH3, RSPH4A, RSPH9, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SASS6, SATB2, SBDS, SCN4A, SDCCAG8, SEC24D, SELENON, SEMA3E, SEPSECS, SERPINF1, SERPINH1, SF3B4, SH3PXD2B, SHH, SHOC2, SHROOM4, SIX3, SIX6, SKI, SLC18A3, SLC25A19, SLC26A2, SLC2A10, SLC34A3, SLC35D1, SLC38A8, SLC39A13, SLC5A7, SLC9A6, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMCHD1, SMOC1, SMS, SNAI2, SNAP29, SNTG1, SOS1, SOS2, SOX10, SOX11, SOX2, SOX9, SP7, SPAG1, SPARC, SPECC1L, SPRED1, SPRY4, STAMBP, STIL, STRA6, SUFU, SYT2, TAF6, TBC1D20, TBC1D23, TBC1D24, TBX1, TBX15, TBX2, TBX22, TBX3, TBX5, TBX6, TCF12, TCF4, TCOF1, TCTN1, TCTN2, TCTN3, TENM3, TENT5A, TFAP2A, TGDS, TGFB1, TGFB2, TGFB3, TGFBRI1, TGFBRI2, TGIF1, TK2, TMCO1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMTC3, TNFRSF11B, TNNT1, TNNT2, TNNT3, TNXB, TOE1, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRIM32, TRIP11, TRMT10A, TRPS1, TRPV4, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TTC21B, TTC8, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TXNL4A, TYR, UBA1, VAMP1, VCAN, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, WDR11, WDR19, WDR35, WDR62, WNT1, WNT5A, WNT7A, XYLTI1, YWHAE, ZBTB24, ZC4H2, ZEB2, ZFPM2, ZIC1, ZIC2, ZIC3, ZMYND10, ZNF335, ZNF423, ZNF469, ZNF699, ZSWIM6