Metodika – panel genů:

Cílená masivní paralelní sekvenace (KAPA HyperCap, Roche; MGI) exonů a přilehlých intronových oblastí genů spojených s následujícími skupinami onemocnění

1. Epidermolysis bullosa: *CD151, COL7A1, COL17A1, DSG3, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT5, KRT14, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, SLC39A4*
2. Peeling skin syndrom*: CAST, CDSN, CSTA, CTSB, CHST8, FLG2, SERPINB8, TGM5*
3. Dowling-Degosova nemoc: *KRT5, POFUT1, POGLUT1, PSENEN*
4. Ichtyóza: *ABCA12, ABHD5, ALDH3A2, ALOXE3, ALOX12B, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CYP4F22, ELOVL1, ELOVL4, FLG, GJB2, KRT1, KRT2, KRT10, LIPN, MBTPS2, NIPAL4, PHYH, PIGL, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, STS, ST14, SULT2B1, TGM1, VPS33B*
5. Palmoplantární keratodermie: *AAGAB, AQP5, CTSC, DSC2, DSG1, DSP, GJA1, GJB2, JUP, KANK2, KRT1, KRT9, KRT6C, KRT16, LOR, MVK, PKP1, RHBDF2, RSPO1, SERPINB7, SMARCAD1, SLURP1, SNAP29, TRPV3, VPS33B*
6. Anomálie nehtů
   a. Izolované: *COL7A1, FZD6, HPGD, PLCD1, RSPO4*
   b. Syndromické: *ATP6V1B2, FOXN1, KRT6A, KRT6B, KRT16, KRT17, LMX1B*
7. Steatocystoma multiplex: *KRT17*
8. Ehlers-Danlosův syndrom, Marfanův syndrom a Loeys-Dietzův syndrom: *ABL1, ACTA2, ADAMTS2, AEBP1, ATP6V1A, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CHD4, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, FBN1, FBN2, FKBP14, FLNA, FLNB, FOXE3, GATA4, GORAB, CHST14, CHST3, IPO8, KIF22, LOX, MAT2A, MFAP5, MYH11, MYLK, NKX2-5, NOTCH1, NPR3, P4HA1, PIEZO2, PLOD1, PLOD3, PRDM5, PRKG1, RIN2, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, TAB2, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469*
9. Ektodermální dysplázie: *CDH3, EDA, EDAR, EDARADD, GJB6, GRHL2, HOXC13, IFT122, IKBKG, KDF1, KREMEN1, KRT74, KRT85, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PRKD1, TP63, TSPEAR, TWIST2, WNT10A*
10. Erythrokeratoderma variabilis: *GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4*
11. Pseudoxanthoma elasticum: *ABCC6, ENPP1, GGCX*
12. Mastocytóza: *KIT*
13. Albinismus
    a. Okulokutánní albinismus: *DCT, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1*
    b. Okulární albinismus: *GPR143*
    c. Heřmanského-Pudlákův syndrom: *AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6*
    d. Chediak-Higashiho syndrom: *LYST*
    e. Griscelliho syndrom: *MLPH, MYO5A, RAB27A*
    f. Piebaldismus*: KIT, SNAI2*
14. Waardenburgův syndrom: *EDN3, EDNRB, MITF, PAX3, SOX10, SNAI2*
15. Xeroderma pigmentosum: *DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC*
16. Trichothiodystrofie: *AARS1, ERCC2, ERCC3, GTF2E2, GTF2H5, MARS1, MPLKIP, RNF113A, TARS1*
17. UV-senzitivní syndrom: *ERCC6, ERCC8, UVSSA*
18. Buschke-Ollendorffův syndrom: *LEMD3*
19. Darierova nemoc: *ATP2A2*
20. Benigní familiární pemfigus [Haileyova-Haileyova nemoc]: *ATP2C1*
21. Fokální dermální hypoplázie: *PORCN*
22. Hypotrichóza: *APCDD1, C3orf52, CDSN, CLDN1, DSC3, DSG4, EPS8L3, FOXN1, HR, HRURF, KRT25, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE*
23. Monilethrix: *DSG4, KRT81, KRT83, KRT86*
24. Syndrom neučesatelných vlasů: *PADI3, TCHH, TGM3*
25. Ageneze zubů: *AXIN2, CLDN1, EDA, GREM2, KDF1, KREMEN1, LRP6, MSX1, PAX9, TSPEAR, WNT10A, WNT10B*
26. Amelogenesis imperfecta: *ACP4, AMBN, AMELX, AMTN, CNNM4, DLX3, DSPP, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, LTBP3, MMP20, ODAPH, RELT, SLC24A4, SMOC2, STIM1, WDR72*
27. Onemocnění skeletu
    a. Osteogenesis imperfecta: *ALPL, ANO5, AP2S1, B4GALT7, BMP1, CBS, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, CYP24A1, CYP27B1, DMP1, ENPP1, FGF23, FKBP10, GNA11,*

IFITM5, KDELR2, LRP5, MESD, NOTCH2, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SLC34A1, SLC34A3, SP7, SPARC, TENT5A, TMEM38B, VDR, WNT1

b. Osteopetróza: *AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, FAM20C, GJA1, LRP5, OSTM1, PLEKHM1, SLC29A3, SNX10, SOST, TCIRG1, TGFB1, TNFRSF11A, TNFSF11*

c. Sticklerův syndrom: *COL2A1, COL9A1, COL11A1, COL11A2*

d. Izolovaný nízký vzrůst: *ACAN, BTK, FGFR3, GH1, GHR, GHRHR, GHSR, IGF1, IGF2, IGFALS, IHH, NPPC, NPR2, PTPN11, SHOX, STAT5B*

e. Skeletální dysplázie: *ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF4, AGA, AGPS, AIFM1, AKT1, ALX1, ALX3, ALX4, AMMECR1, ANKRD11, ANTXR2, ARCN1, ARHGAP31, ARSB, ARSL, ASCC1, ASXL1, ASXL2, ATR, B3GALT6, B3GAT3, BGN, BHLHA9, BMP2, BMPER, BMPR1B, BPNT2, BRAF, BTK, C2CD3, CANT1, CASR, CBL, CBS, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP120, CEP152, CEP290, CEP63, CFAP410, CILK1, CKAP2L, COL10A1, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CPLANE1, CREBBP, CRIPT, CSGALNACT1, CSPP1, CTSA, CUL7, CYP26B1, CYP2R1, DDR2, DDX59, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX5, DNA2, DNMT3A, DOCK6, DONSON, DVL1, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EFNB1, EFTUD2, EIF2AK3, EOGT, EP300, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAM111A, FBLN1, FBN1, FBXW4, FGD1, FGF10, FGF16, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNB, FMN1, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLI2, GLI3, GMNN, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GPC6, GPX4, GREM1, GSC, GUSB, GZF1, HDAC4, HDAC8, HES7, HGSNAT, HOXA13, HOXD13, HPGD, HRAS, HSPG2, CHD4, CHST14, CHSY1, IARS2, IDH1, IDH2, IDS, IDUA, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGFALS, IHH, IL11RA, IL1RN, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KIF7, KMT2A, KMT2D, KRAS, LARP7, LBR, LEMD3, LFNG, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRRK1, LTBP2, LTBP3, LZTR1, MAFB, MAGEL2, MAN2B1, MAP2K1, MAP2K2, MAP3K7, MATN3, MBTPS1, MCM5, MED12, MEGF8, MEOX1, MESP2, MGP, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEPRO, NEU1, NF1, NFIX, NIN, NIPBL, NKAP, NKX3-2, NOG, NOTCH2, NPPC, NPR2, NPR3, NRAS, NSD1, NSDHL, NSMCE2, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, PAM16, PAPSS2, PCNT, PCYT1A, PDE3A, PDE4D, PEX14, PEX5, PEX7, PGM3, PHGDH, PIGV, PIK3C2A, PIK3CA, PIK3R1, PISD, PITX1, PKDCC, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPP3CA, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, RAB23, RAB33B, RAD21, RAF1, RASA2, RBBP8, RBPJ, RECQL4, RIPPLY2, RIT1, ROR2, RPGRIP1L, RPL13, RRAS, RSPRY1, RTTN, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SLC10A7, SLC17A5, SLC26A2, SLC35D1, SLCO2A1, SMAD4, SMAD6, SMARCAL1, SMARCB1, SMC1A, SMC3, SMOC1, SMS, SNRPB, SOS1, SOX9, SRCAP, STAMBP, STAT5B, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCOF1, TCTN3, TMCO1, TMEM107, TMEM216, TMEM67, TNFRSF11B, TONSL, TP63, TRAIP, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TTC21B, TWIST1, TYROBP, UBA2, UFSP2, UNC45A, WDR19, WDR35, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZIC1, ZMPSTE24*

28. RASopatie a neurofibromatóza: *A2ML1, ACTB, ACTG1, BRAF, CBL, CFC1, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NF1, NF2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, RREB1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, SPRED2, SPRY1, SPRY2, SYNGAP1*