CLINICAL EVALUATION PROTOCOL

Personal data

Name:_____ Sex:_____

Birth date and place:_____

History of present illness

Symptoms:_____

_____

_____

Age of: onset:_____ fundoscopic diagnosis:_____ NGS evaluation:_____

Obstetrical and perinatal background

_____

_____

_____

Medical background

Neurodevelopment: • normal   • delayed

Other neurological comorbidities:_____

Infectious disease:_____

Dermatological comorbidities:_____

Other ocular comorbidities:_____

Otorhinolaryngological comorbidities:_____

Cardiovascular comorbidities:_____

Gastrointestinal comorbidities:_____

Genitourinary comorbidities:_____

Orthopedical comorbidities:_____

Other:_____

Familial history

Parental consanguinity: • no   • yes, specify:_____

Recurrence of the condition:_____

Other relevant conditions:_____

Ethnic background:_____

Physical examination

Anthropometry:_____

Dysmorphological features (face, torso, and limbs): _____

_____

Complementary exams

Fundoscopy:_____

_____

Other ophthalmological exams:_____

_____

Others:_____

_____

_____

Supplementary material: List of genes of the Gene Panel Sequencing for Retinitis pigmentosa, in alphabetic order.

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADAMTSL4, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C10orf11, C12orf65, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CHM, CIB2, CISD2, CLCC1, CLN3, CLN5, CLN6, CLN8, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CSPP1, CTNNA1, CTSD, CWC27, CYP4V2, DHDDS, DHX32, DHX38, DNAJC17, DRAM2, DSCAML1, DTHD1, EFEMP1, ELOVL4, EMC1, ERCC6, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GNS, GPR143, GPR179, GPR45, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMCN1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT80, IFT81, IFT88, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA0586, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LYST, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSD8, MIR204, MKKS, MKS1, MPDZ, MTPAP, MTTP, MYO7A, NAGLU, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OR2W3, OTX2, P3H2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC24A5, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMED7, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WDR34, WFS1, WHRN, ZNF408, ZNF423, ZNF513.