

Index NGS Pakketten Amsterdam UMC, Locatie AMC

NGS panel genen kunnen met verschillende kwaliteit worden geanalyseerd. Om meer inzicht in deze kwaliteit te krijgen worden de testen als volgt ingedeeld (*Matthijs G et al., Eur J Hum Genet 2015; doi: 10.1038*).

- **Kwaliteit Type A:** alle genen wordt volledig dekkend geanalyseerd. Gebieden met een lage coverage (<30 reads) in de NGS test worden alsnog met behulp van Sanger sequencen geanalyseerd.
- **Kwaliteit Type C:** alleen NGS analyse. Er wordt geen aanvullende analyse gedaan van eventuele gebieden met een lage coverage.

Core-NL: hiermee wordt bedoeld dat de genen uit deze lijst essentieel geacht worden voor het stellen van een betrouwbare diagnose (*Weiss MM et al., Human Mut 2013; 34: 1313-1321*). Deze lijst is in landelijk overleg met de Nederlandse laboratoria en kliniek tot stand gekomen. Een core gen wordt volledig dekkend geanalyseerd (kwaliteit A).

In aangegeven NGS pakketten wordt copy number variation (CNV) analyse gedaan om exon deleties/duplicaties te detecteren. Deze CNV test werkt UITSLUITEND betrouwbaar op ingestuurd EDTA bloed en NIET op ingestuurd DNA

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NGS Microcefalie
NGS Pontocerebellaire Hypoplasie (PCH)
NGS Spieraandoeningen

NGS Arthrogrypose

Genen: 8

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
ECEL1	A
MYBPC1	A
MYH3	A
MYH8	A
PIEZO2	A
TNNI2	A
TNNT3	A
TPM2	A

NGS Aritmie

Genen: 55
Methode: Pakket capture
Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
TNNI3K	A
FPGT-TNNI3K	A
MYL4	A
GNB2	A
TECRL	A
ABCC9	A
AKAP9	A
ANK2	A
ASPH	A
CACNA1C	A
CACNA1D	A
CACNA2D1	A
CACNB2	A
CALM1	A
CALM2	A
CALM3	A
CASQ2	A
CAV3	A
DPP6	A
GJA5	A
GPD1L	A
HCN4	A
JPH2	A
KCNA5	A
KCND3	A
KCNE1	A
KCNE2	A
KCNE3	A
KCNE5	A
KCNH2	A
KCNJ2	A
KCNJ5	A
KCNJ8	A
KCNQ1	A
LAMP2	A
LMNA	A
NKX2-5	A
NPPA	A
PKP2	A
PLN	A
PPA2	A
PRKAG2	A
RANGRF	A
RYR2	A

NGS Aritmie

Genen: 55
Methode: Pakket capture
Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
SCN10A	A
SCN1B	A
SCN2B	A
SCN3B	A
SCN4B	A
SCN5A	A
SLMAP	A
SNTA1	A
TNNT2	A
TRDN	A
TRPM4	A

NGS Dragerschap Preconceptie**Genen:** 50

Pakket capture met filter voor bekende (waarsch.) pathogene

Methode: (fouder)mutaties**Kwaliteit:** Type C **Incl CNV:** Ja

GENE	TYPE
AGA	C
ALDH3A2	C
ARSA	C
ASPA	C
ASS1	C
BCKDHB	C
BLM	C
CBS	C
CFTR	C
CLN3	C
CLN5	C
COL17A1	C
CRTAP	C
DHCR7	C
ELP1	C
FANCC	C
G6PC	C
GAA	C
GALC	C
HADHA	C
HBB	C
HEXA	C
HGSNAT	C
HSD17B4	C
IDUA	C
IVD	C
LAMA3	C
LAMB3	C
LAMC2	C
MAN2B1	C
MCOLN1	C
MLC1	C
MUSK	C
NBN	C
NEB	C
NPC1	C
PEX1	C
PEX12	C
PEX7	C
PMM2	C
POLG	C

NGS Dragerschap Preconceptie**Genen:** 50

Pakket capture met filter voor bekende (waarsch.) pathogene

Methode: (fouder)mutaties**Kwaliteit:** Type C **Incl CNV:** Ja

GENE	TYPE
PPT1	C
SACS	C
SGSH	C
SMN1	C
SMPD1	C
TMEM216	C
TPP1	C
TSEN54	C
TTPA	C

NGS Albinisme

Genen: 29

Methode: Pakket capture

Kwaliteit: Type A/C **Incl CNV:** Nee

GENE	TYPE
AP3B1	A
AP3D1	C
BLOC1S3	A
BLOC1S6	A
LRMDA	A
COL18A1	C
DTNBP1	A
FRMD7	A
GNAI3	C
GPR143	A
HPS1	A
HPS3	A
HPS4	A
HPS5	A
HPS6	A
LYST	A
MC1R	A
MITF	A
MLPH	A
MYO5A	A
OCA2	A
PAX6	C
RAB27A	A
SLC24A5	A
SLC38A8	A
SLC45A2	A
TYR	A
TYRP1	A
LRMDA	A

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
ABCA4	A
ABCB6	C
ABCC6	C
ABHD12	C
ACO2	C
ADAM9	A
ADAMTS18	C
ADGRA3	C
ADGRV1	C
ADIPOR1	C
AHI1	C
AIPL1	A
ALMS1	C
ARHGEF18	C
ARL2BP	C
ARL3	C
ARL6	C
ASRGL1	C
ATF6	A
ATXN7	C
BBIP1	C
BBS1	C
BBS10	C
BBS12	C
BBS2	C
BBS4	C
BBS5	C
BBS7	C
BBS9	C
BCOR	C
BEST1	A
BMP4	C
C12orf65	C
C1QTNF5	A
C21orf2	C
C2orf71	C
C5orf42	C
C8orf37	A
CA4	C
CABP4	C
CACNA1F	A
CACNA2D4	A
CAPN5	C
CC2D2A	C

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
CDH23	C
CDH3	C
CDHR1	A
CEP164	C
CEP250	C
CEP290	C
CEP41	C
CEP78	A
CERKL	A
CFH	A
CHM	C
CIB2	C
CLN3	C
CLRN1	C
CNGA1	C
CNGA3	A
CNGB1	C
CNGB3	A
CNNM4	A
COL11A1	C
COL2A1	C
COL9A1	C
COL9A2	C
CRB1	C
CRX	A
CSPP1	C
CTNNA1	A
CWC27	C
CYP4V2	C
DHDDS	C
DHX38	C
DRAM2	C
DTHD1	C
EFEMP1	A
ELOVL4	A
EMC1	C
EXOSC2	C
EYS	C
FAM161A	C
FLVCR1	C
FOXE3	C
FSCN2	A
FZD4	C
GDF6	C

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	A
GUCA1B	A
GUCY2D	A
HARS	C
HCCS	C
HGSNAT	C
HK1	C
HK1	C
HMX1	C
IDH3B	C
IFT140	C
IFT172	C
IFT27	C
IFT81	C
IMPDH1	C
IMPG1	A
IMPG2	C
INPP5E	C
INVS	C
IQCB1	C
ITM2B	C
JAG1	C
KCNJ13	C
KCNV2	A
KIAA1549	C
KIF11	C
KIZ	C
KLHL7	C
LCA5	C
LRAT	C
LRIT3	C
LRP5	C
LZTFL1	C
MAK	C
MAPKAPK3	C
MERTK	C
MFN2	C

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
MFRP	C
MFSD8	A
MIR204	C
MKKS	C
MKS1	C
MMACHC	A
MTTP	C
MVK	C
MYO7A	C
NDP	C
NEK2	C
NEUROD1	C
NMNAT1	C
NPHP1	C
NPHP3	C
NPHP4	C
NR2E3	C
NR2F1	C
NRL	C
NYX	C
OAT	C
OFD1	C
OPA1	C
OPA3	C
OPN1LW	C
OPN1MW	C
OPN1SW	C
OR2W3	C
OTX2	C
PANK2	C
PAX2	C
PCDH15	C
PCYT1A	C
PDE6A	C
PDE6B	C
PDE6C	A
PDE6G	C
PDE6H	A
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
PITPNM3	A
PLA2G5	C
PNPLA6	C
POC1B	A
POMGNT1	C
PRCD	C
PRDM13	C
PROM1	A
PRPF3	C
PRPF31	C
PRPF4	C
PRPF6	C
PRPF8	C
PRPH2	A
PRPS1	C
PXDN	C
RAB28	A
RBP3	C
RBP4	C
RCBTB1	C
RD3	C
RDH11	C
RDH12	C
RDH5	A
REEP6	C
RGR	C
RGS9	C
RGS9BP	C
RHO	C
RIMS1	A
RLBP1	C
ROM1	C
RP1	A
RP1L1	A
RP2	C
RP9	C
RPE65	C
RPGR	A
RPGRIP1	A
RPGRIP1L	C
RS1	C
SAG	C
SDCCAG8	C
SEMA4A	A

NGS Cone-rod dystrophie**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
SIX6	C
SLC24A1	C
SLC7A14	C
SNRNP200	C
SPATA7	C
SPP2	C
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	A
TMEM126	C
TMEM237	C
TMEM67	C
TOPORS	C
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	C
TLL5	A
TUB	C
TULP1	C
UNC119	A
USH1C	C
USH1G	C
USH2A	C
VCAN	C
WDPCP	C
WDR19	C
WFS1	C
WHRN	C
ZNF408	C
ZNF423	C
ZNF513	C

NGS Congenitale stationaire nachtblindheid

Genen: 20

Methode: Pakket capture

Kwaliteit: Type A/C **Incl CNV:** Nee

GENE	TYPE
CABP4	A
CACNA1F	A
CACNA2D4	A
CHM	C
CYP4V2	A
GNAT1	A
GNB3	A
GPR179	A
GRK1	A
GRM6	A
LRIT3	A
NYX	A
PDE6B	A
RDH5	A
RHO	A
RLBP1	A
RPE65	A
SAG	A
SLC24A1	A
TRPM1	A

NGS Optic atrophy prevention**Genen:** 18**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
ACO2	A
AUH	A
C12orf65	A
CISD2	A
MFN2	A
MTPAP	A
NBAS	A
NDUFS1	A
NR2F1	A
OPA1	A
OPA3	A
RTN4IP1	A
SLC24A1	A
SLC25A46	A
TIMM8A	A
TMEM126	A
WFS1	A
SPG7	C

NGS Glaucoom**Genen:** 26**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
CYP1B1	A
FOXC1	A
MYOC	A
OPTN	A
PITX2	A
TBK1	A
ASB10	C
B3GLCT	C
BEST1	C
COL1A1	C
EFEMP1	C
GJA1	C
GLIS3	C
LMX1B	C
LOXL1	C
LTBP2	C
NTF4	C
OPA1	C
PAX6	C
POMGNT1	C
PRPF8	C
PXDN	C
SBF2	C
SH3PXD2B	C
SLC4A4	C
TEK	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
ABCA4	C
ABCB6	C
ABCC6	C
ABHD12	C
ACO2	C
ADAM9	C
ADAMTS18	C
ADGRA3	C
ADGRV1	C
ADIPOR1	C
AHI1	C
AIPL1	C
ALMS1	C
ARHGEF18	C
ARL2BP	C
ARL3	C
ARL6	C
ASRGL1	C
ATF6	C
ATXN7	C
BBIP1	C
BBS1	C
BBS10	C
BBS12	C
BBS2	C
BBS4	C
BBS5	C
BBS7	C
BBS9	C
BCOR	C
BEST1	C
BMP4	C
C12orf65	C
C1QTNF5	C
C21orf2	C
C2orf71	C
C5orf42	C
C8orf37	C
CA4	C
CABP4	A
CACNA1F	C
CACNA2D4	C
CAPN5	C
CC2D2A	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
CDH23	C
CDH3	C
CDHR1	C
CEP164	C
CEP250	C
CEP290	A
CEP41	C
CEP78	C
CERKL	C
CFH	C
CHM	C
CIB2	C
CLN3	C
CLRN1	C
CNGA1	C
CNGA3	C
CNGB1	C
CNGB3	C
CNNM4	C
COL11A1	C
COL2A1	C
COL9A1	C
COL9A2	C
CRB1	A
CRX	A
CSPP1	C
CTNNA1	C
CWC27	C
CYP4V2	C
DHDDS	C
DHX38	C
DRAM2	C
DTHD1	A
EFEMP1	C
ELOVL4	C
EMC1	C
EXOSC2	C
EYS	C
FAM161A	C
FLVCR1	C
FOXE3	C
FSCN2	C
FZD4	C
GDF6	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	C
GUCA1B	C
GUCY2D	A
HARS	C
HCCS	C
HGSNAT	C
HK1	C
HK1	C
HMX1	C
IDH3B	C
IFT140	C
IFT172	C
IFT27	C
IFT81	C
IMPDH1	A
IMPG1	C
IMPG2	C
INPP5E	C
INVS	C
IQCB1	A
ITM2B	C
JAG1	C
KCNJ13	A
KCNV2	C
KIAA1549	C
KIF11	C
KIZ	C
KLHL7	C
LCA5	A
LRAT	A
LRIT3	C
LRP5	C
LZTFL1	C
MAK	C
MAPKAPK3	C
MERTK	C
MFN2	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
MFRP	C
MFSD8	C
MIR204	C
MKKS	C
MKS1	C
MMACHC	C
MTTP	C
MVK	C
MYO7A	C
NDP	C
NEK2	C
NEUROD1	C
NMNAT1	A
NPHP1	C
NPHP3	C
NPHP4	C
NR2E3	C
NR2F1	C
NRL	C
NYX	C
OAT	C
OFD1	C
OPA1	C
OPA3	C
OPN1LW	C
OPN1MW	C
OPN1SW	C
OR2W3	C
OTX2	A
PANK2	C
PAX2	C
PCDH15	C
PCYT1A	C
PDE6A	C
PDE6B	C
PDE6C	C
PDE6G	C
PDE6H	C
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	C
POMGNT1	C
PRCD	C
PRDM13	C
PROM1	C
PRPF3	C
PRPF31	C
PRPF4	C
PRPF6	C
PRPF8	C
PRPH2	A
PRPS1	C
PXDN	C
RAB28	C
RBP3	C
RBP4	C
RCBTB1	C
RD3	A
RDH11	C
RDH12	A
RDH5	C
REEP6	C
RGR	C
RGS9	C
RGS9BP	C
RHO	C
RIMS1	C
RLBP1	C
ROM1	C
RP1	C
RP1L1	C
RP2	C
RP9	C
RPE65	A
RPGR	A
RPGRIP1	A
RPGRIP1L	C
RS1	C
SAG	C
SDCCAG8	C
SEMA4A	C

NGS Leber congenital amaurosis**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
SIX6	C
SLC24A1	C
SLC7A14	C
SNRNP200	C
SPATA7	A
SPP2	C
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	C
TMEM126	C
TMEM237	C
TMEM67	C
TOPORS	C
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	C
TLL5	C
TUB	C
TULP1	A
UNC119	C
USH1C	C
USH1G	C
USH2A	C
VCAN	C
WDPCP	C
WDR19	C
WFS1	C
WHRN	C
ZNF408	C
ZNF423	C
ZNF513	C

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
ABCA4	A	C	A	C
ABCB6	C	C	C	C
ABCC6	C	C	C	C
ABHD12	C	C	C	C
ACO2	C	C	C	C
ADAM9	A	C	C	C
ADAMTS18	C	C	C	C
ADGRA3	C	C	C	C
ADGRV1	C	C	C	C
ADIPOR1	C	C	C	C
AHI1	C	C	C	C
AIPL1	A	A	C	C
ALMS1	C	C	C	C
ARHGEF18	C	C	A	C
ARL2BP	C	C	A	C
ARL3	C	C	A	C
ARL6	C	C	A	C
ASRGL1	C	C	C	C
ATF6	A	C	C	C
ATXN7	C	C	C	C
BBIP1	C	C	C	C
BBS1	C	C	A	C
BBS10	C	C	C	C
BBS12	C	C	C	C
BBS2	C	C	A	C
BBS4	C	C	C	C
BBS5	C	C	C	C
BBS7	C	C	C	C
BBS9	C	C	C	C
BCOR	C	C	C	C
BEST1	A	C	A	C
BMP4	C	C	C	C
C12orf65	C	C	C	C
C1QTNF5	A	C	C	C
C21orf2	A	C	C	C
C2orf71	C	C	A	C
C5orf42	C	C	C	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
C8orf37	A	C	A	C
CA4	C	C	A	C
CABP4	C	A	C	C
CACNA1F	A	C	C	C
CACNA2D4	A	C	C	C
CAPN5	C	C	C	C
CC2D2A	C	C	C	C
CDH23	C	C	C	C
CDH3	C	C	C	C
CDHR1	A	C	C	C
CEP164	C	C	C	C
CEP250	C	C	C	C
CEP290	C	A	C	C
CEP41	C	C	C	C
CEP78	A	C	C	C
CERKL	A	C	A	C
CFH	A	C	C	C
CHM	C	C	C	C
CIB2	C	C	C	C
CLN3	C	C	C	C
CLRN1	C	C	A	C
CNGA1	C	C	A	C
CNGA3	A	C	C	C
CNGB1	C	C	A	C
CNGB3	A	C	C	C
CNNM4	A	C	C	C
COL11A1	C	C	C	C
COL2A1	C	C	C	C
COL9A1	C	C	C	C
COL9A2	C	C	C	C
CRB1	C	A	A	C
CRX	A	A	A	C
CSPP1	C	C	C	C
CTNNA1	A	C	C	C
CWC27	C	C	C	C
CYP4V2	C	C	A	C
DHDDS	C	C	A	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
DHX38	C	C	A	C
DRAM2	C	C	C	C
DTHD1	C	A	C	C
EFEMP1	A	C	C	C
ELOVL4	A	C	C	C
EMC1	C	C	A	C
EXOSC2	C	C	C	C
EYS	C	C	A	C
FAM161A	C	C	A	C
FLVCR1	C	C	C	C
FOXE3	C	C	C	C
FSCN2	A	C	A	C
FZD4	C	C	C	C
GDF6	C	C	C	C
GNAT1	C	C	C	C
GNAT2	C	C	C	C
GNB3	C	C	C	C
GNPTG	C	C	C	C
GPR179	C	C	C	C
GRK1	C	C	C	C
GRM6	C	C	C	C
GUCA1A	A	C	C	C
GUCA1B	A	C	A	C
GUCY2D	A	A	C	C
HARS	C	C	C	C
HCCS	C	C	C	C
HGSNAT	C	C	C	C
HK1	C	C	C	C
HK1	C	C	C	C
HMX1	C	C	C	C
IDH3B	C	C	A	C
IFT140	C	C	C	C
IFT172	C	C	A	C
IFT27	C	C	C	C
IFT81	C	C	C	C
IMPDH1	C	A	A	C
IMPG1	A	C	C	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
IMPG2	C	C	A	C
INPP5E	C	C	C	C
INVS	C	C	C	C
IQCB1	C	A	C	C
ITM2B	C	C	C	C
JAG1	C	C	C	C
KCNJ13	C	A	C	C
KCNV2	A	C	C	C
KIAA1549	C	C	C	C
KIF11	C	C	C	C
KIZ	C	C	A	C
KLHL7	C	C	A	C
LCA5	C	A	C	C
LRAT	C	A	A	C
LRIT3	C	C	C	C
LRP5	C	C	C	C
LZTFL1	C	C	C	C
MAK	C	C	A	C
MAPKAPK3	C	C	C	C
MERTK	C	C	A	C
MFN2	C	C	C	C
MFRP	C	C	C	C
MFSD8	A	C	C	C
MIR204	C	C	C	C
MKKS	C	C	C	C
MKS1	C	C	C	C
MMACHC	A	C	C	C
MTTP	C	C	C	C
MVK	C	C	A	C
MYO7A	C	C	C	C
NDP	C	C	C	C
NEK2	C	C	C	C
NEUROD1	C	C	C	C
NMNAT1	C	A	C	C
NPHP1	C	C	C	C
NPHP3	C	C	C	C
NPHP4	C	C	C	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
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NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
NR2E3	C	C	A	C
NR2F1	C	C	C	C
NRL	C	C	A	C
NYX	C	C	C	C
OAT	C	C	C	C
OFD1	C	C	A	C
OPA1	C	C	C	C
OPA3	C	C	C	C
OPN1LW	C	C	C	C
OPN1MW	C	C	C	C
OPN1SW	C	C	C	C
OR2W3	C	C	C	C
OTX2	C	A	C	C
PANK2	C	C	C	C
PAX2	C	C	C	C
PCDH15	C	C	C	C
PCYT1A	C	C	C	C
PDE6A	C	C	A	C
PDE6B	C	C	A	C
PDE6C	A	C	C	C
PDE6G	C	C	A	C
PDE6H	A	C	C	C
PDZD7	C	C	C	C
PEX1	C	C	C	C
PEX2	C	C	C	C
PEX7	C	C	C	C
PGK1	C	C	C	C
PHYH	C	C	C	C
PITPNM3	A	C	C	C
PLA2G5	C	C	C	C
PNPLA6	C	C	C	C
POC1B	A	C	C	C
POMGNT1	C	C	A	C
PRCD	C	C	A	C
PRDM13	C	C	C	C
PROM1	A	C	A	C
PRPF3	C	C	A	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
PRPF31	C	C	A	C
PRPF4	C	C	A	C
PRPF6	C	C	A	C
PRPF8	C	C	A	C
PRPH2	A	A	A	C
PRPS1	C	C	C	C
PXDN	C	C	C	C
RAB28	A	C	C	C
RBP3	C	C	A	C
RBP4	C	C	C	C
RCBTB1	C	C	C	C
RD3	C	A	C	C
RDH11	C	C	C	C
RDH12	C	A	A	C
RDH5	A	C	C	C
REEP6	C	C	A	C
RGR	C	C	A	C
RGS9	C	C	C	C
RGS9BP	C	C	C	C
RHO	C	C	A	C
RIMS1	A	C	C	C
RLBP1	C	C	A	C
ROM1	C	C	A	C
RP1	C	C	A	C
RP1L1	A	C	A	C
RP2	C	C	A	C
RP9	C	C	A	C
RPE65	C	A	A	C
RPGR	C	C	C	C
RPGRIP1	A	A	C	C
RPGRIP1L	C	C	C	C
RS1	C	C	C	C
SAG	C	C	A	C
SDCCAG8	C	C	C	C
SEMA4A	A	C	A	C
SIX6	C	C	C	C
SLC24A1	C	C	C	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Blindheid panels (BHv5)**Genen:** 254**Methode:** Pakket capture; Filter per type aandoeng**Kwaliteit:** Type A/C **Incl CNV:** Nee

Gen	Kwaliteit CRD	Kwaliteit LCA	Kwaliteit RP	Kwaliteit OVR
SLC7A14	C	C	A	C
SNRNP200	C	C	A	C
SPATA7	C	A	A	C
SPP2	C	C	A	C
SRD5A3	C	C	C	C
TEAD1	C	C	C	C
TIMM8A	C	C	C	C
TIMP3	A	C	C	C
TMEM126	C	C	C	C
TMEM237	C	C	C	C
TMEM67	C	C	C	C
TOPORS	C	C	A	C
TREX1	C	C	C	C
TRIM32	C	C	C	C
TRPM1	C	C	C	C
TSPAN12	C	C	C	C
TTC8	C	C	A	C
TLL5	A	C	C	C
TUB	C	C	C	C
TULP1	C	A	A	C
UNC119	A	C	C	C
USH1C	C	C	C	C
USH1G	C	C	C	C
USH2A	C	C	A	C
VCAN	C	C	C	C
WDPCP	C	C	C	C
WDR19	C	C	C	C
WFS1	C	C	C	C
WHRN	C	C	C	C
ZNF408	C	C	C	C
ZNF423	C	C	C	C
ZNF513	C	C	A	C

CRD = Cone/rod dystrofie
en Maculadegeneratie
LCA = Leber Congenitale Amaurosis
RP = Retinitis Pigmentosa
OVR = Overig blindheid

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
ABCA4	A
ABCB6	C
ABCC6	C
ABHD12	C
ACO2	C
ADAM9	C
ADAMTS18	C
ADGRA3	C
ADGRV1	C
ADIPOR1	C
AHI1	C
AIPL1	C
ALMS1	C
ARHGEF18	A
ARL2BP	A
ARL3	A
ARL6	A
ASRGL1	C
ATF6	C
ATXN7	C
BBIP1	C
BBS1	A
BBS10	C
BBS12	C
BBS2	A
BBS4	C
BBS5	C
BBS7	C
BBS9	C
BCOR	C
BEST1	A
BMP4	C
C12orf65	C
C1QTNF5	C
C21orf2	C
C2orf71	C
C5orf42	C
C8orf37	A
CA4	A
CABP4	C
CACNA1F	C
CACNA2D4	C
CAPN5	C
CC2D2A	C

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
CDH23	C
CDH3	C
CDHR1	C
CEP164	C
CEP250	C
CEP290	C
CEP41	C
CEP78	C
CERKL	A
CFH	C
CHM	C
CIB2	C
CLN3	C
CLRN1	A
CNGA1	A
CNGA3	C
CNGB1	A
CNGB3	C
CNNM4	C
COL11A1	C
COL2A1	C
COL9A1	C
COL9A2	C
CRB1	A
CRX	A
CSPP1	C
CTNNA1	C
CWC27	C
CYP4V2	A
DHDDS	A
DHX38	A
DRAM2	C
DTHD1	C
EFEMP1	C
ELOVL4	C
EMC1	A
EXOSC2	C
EYS	A
FAM161A	A
FLVCR1	C
FOXE3	C
FSCN2	A
FZD4	C
GDF6	C

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	C
GUCA1B	A
GUCY2D	C
HARS	C
HCCS	C
HGSNAT	C
HK1	C
HK1	C
HMX1	C
IDH3B	A
IFT140	C
IFT172	A
IFT27	C
IFT81	C
IMPDH1	A
IMPG1	C
IMPG2	A
INPP5E	C
INVS	C
IQCB1	C
ITM2B	C
JAG1	C
KCNJ13	C
KCNV2	C
KIAA1549	C
KIF11	C
KIZ	A
KLHL7	A
LCA5	C
LRAT	A
LRIT3	C
LRP5	C
LZTFL1	C
MAK	A
MAPKAPK3	C
MERTK	A
MFN2	C

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
MFRP	C
MFSD8	C
MIR204	C
MKKS	C
MKS1	C
MMACHC	C
MTTP	C
MVK	A
MYO7A	C
NDP	C
NEK2	C
NEUROD1	C
NMNAT1	C
NPHP1	C
NPHP3	C
NPHP4	C
NR2E3	A
NR2F1	C
NRL	A
NYX	C
OAT	C
OFD1	A
OPA1	C
OPA3	C
OPN1LW	C
OPN1MW	C
OPN1SW	C
OR2W3	C
OTX2	C
PANK2	C
PAX2	C
PCDH15	C
PCYT1A	C
PDE6A	A
PDE6B	A
PDE6C	C
PDE6G	A
PDE6H	C
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	C
POMGNT1	A
PRCD	A
PRDM13	C
PROM1	A
PRPF3	A
PRPF31	A
PRPF4	A
PRPF6	A
PRPF8	A
PRPH2	A
PRPS1	C
PXDN	C
RAB28	C
RBP3	A
RBP4	C
RCBTB1	C
RD3	C
RDH11	C
RDH12	A
RDH5	C
REEP6	A
RGR	A
RGS9	C
RGS9BP	C
RHO	A
RIMS1	C
RLBP1	A
ROM1	A
RP1	A
RP1L1	A
RP2	A
RP9	A
RPE65	A
RPGR	C
RPGRIP1	C
RPGRIP1L	C
RS1	C
SAG	A
SDCCAG8	C
SEMA4A	A

NGS Retinitis pigmentosa**Genen:** 254**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
SIX6	C
SLC24A1	C
SLC7A14	A
SNRNP200	A
SPATA7	A
SPP2	A
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	C
TMEM126	C
TMEM237	C
TMEM67	C
TOPORS	A
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	A
TLL5	C
TUB	C
TULP1	A
UNC119	C
USH1C	C
USH1G	C
USH2A	A
VCAN	C
WDPCP	C
WDR19	C
WFS1	C
WHRN	C
ZNF408	C
ZNF423	C
ZNF513	A

NGS Cardiovasculaire screening

Genen: 41

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
KCNQ1	A
KCNH2	A
SCN5A	A
LMNA	A
PKP2	A
RYR2	A
MYBPC3	A
MYH7	A
TNNT2	A
TNNI3	A
TTN	A
DPP6	A
HCN4	A
KCNJ2	A
LAMP2	A
PLN	A
PRKAG2	A
BAG3	A
DES	A
DSP	A
GLA	A
MYL2	A
RBM20	A
TAZ	A
TPM1	A
TTR	A
LDLR	A
APOB	A
LPL	A
APOE	A
SLCO1B1	A
FBN1	A
TGFBR1	A
TGFBR2	A
TGFB2	A
TGFB3	A
ACTA2	A
COL3A1	A
SMAD3	A
PRKG1	A
MYH11	A

NGS Hypothyreoidie**Genen:** 58**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Ja

Gen	Kwaliteit	THT	CHT
ALB	A		
ANOS1	A		
BMP4	A		
CDCA8	A		
CHD7	A		
CRYM	A		
DIO1	A		
DIO2	A		
DIO3	A		
DUOX1	A		
DUOX2	A		
DUOXA1	A	THT	
DUOXA2	A	THT	
FGF8	A		
FGFR1	A		
FOXE1	A	THT	
GLI2	A		
GLIS3	A		
GNAS	A		
HESX1	A		CHT
HOXA3	A	THT	
IGSF1	A		CHT
IRS4	A		CHT
IYD	A	THT	
JAG1	A		
LHX3	A		CHT
LHX4	A		CHT
NKX2-1	A	THT	
NTN1	A		
OTX2	A		CHT
PAX6	A		
PAX8	A	THT	
POU1F1	A		
PROK2	A		
PROKR2	A		
PROP1	A		CHT
SECISBP2	A		
SERPINA7	A		
SHH	A		
SLC16A10	A		
SLC16A2	A		
SLC26A4	A	THT	
SLC5A5	A	THT	
SLC7A7	A		

NGS Hypothyreoidie

Genen: 58

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

Gen	Kwaliteit	THT	CHT
SLCO1C1	A		
SOX2	A		CHT
SOX3	A		CHT
TBL1X	A		CHT
TG	A	THT	
THRA	A		
THRB	A		
TPO	A	THT	
TPST2	A	THT	
TRH	A	CHT	CHT
TRHR	A	CHT	CHT
TSHB	A	CHT	CHT
TSHR	A	THT	
TTR	A		

NGS Chronisch lymfatische leukemie

Genen: 14

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Nee

GENE	TYPE
EGR2	A
BIRC3	A
ATM	A
PLCG2	A
RPS15	A
TP53	A
XPO1	A
SF3B1	A
MYD88	A
FBXW7	A
NFKBIE	A
POT1	A
NOTCH1	A
BTK	A

NGS Cardiomyopathie**Genen:** 57**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Ja

GENE	TYPE	Opmerking
ACTC1	A	Core-NL
ACTN2	A	Core-NL
ANKRD1	A	
BAG3	A	Core-NL
CALR3	A	Core-NL
CAV3	A	Core-NL
CRYAB	A	Core-NL
CSRP3	A	
CTNNA3	A	Core-NL
DES	A	Core-NL
DSC2	A	Core-NL
DSG2	A	Core-NL
DSP	A	Core-NL
EMD	A	Core-NL
FHL1	A	Core-NL
GLA	A	Core-NL
JPH2	A	Core-NL
JUP	A	
LAMA4	A	
LAMP2	A	
LDB3	A	
LMNA	A	Core-NL
MIB1	A	
MYBPC3	A	Core-NL
MYH6	A	Core-NL
MYH7	A	Core-NL
MYL2	A	Core-NL
MYL3	A	Core-NL
MYOZ2	A	Core-NL
MYPN	A	Core-NL
NEXN	A	Core-NL
PKP2	A	Core-NL
PLN	A	Core-NL
PRKAG2	A	Core-NL
RBM20	A	Core-NL
SCN5A	A	
TAZ	A	Core-NL
TCAP	A	Core-NL
TMEM43	A	Core-NL
TNNC1	A	Core-NL
TNNI3	A	Core-NL
TNNT2	A	
TPM1	A	
TTR	A	Core-NL

NGS Cardiomyopathie**Genen:** 57**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Ja

GENE	TYPE	Opmerking
VCL	A	Core-NL
PRDM16	A	Core-NL
HCN4	A	Core-NL
ALPK3	A	Core-NL
FHL2	A	Core-NL
CDH2	A	Core-NL
FLNC	A	Core-NL
PPA2	A	
TTN	A	Core-NL
FKRP	A	Core-NL
TNNI3K	A	
FPGT-TNNI	A	Core-NL
MYLK3	A	Core-NL

NGS Cowden**Genen:** 5**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Nee

GENE	TYPE
AKT1	A
PIK3CA	A
PTEN	A
SDHB	A
SDHD	A

NGS Dyslipidemie

Genen: 29

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
ABCA1	A
ABCG5	A
ABCG8	A
ANGPTL3	A
APOA1	A
APOA5	A
APOB	A
APOC2	A
APOC3	A
APOE	A
CETP	A
CYP27A1	A
CYP7A1	A
GPIHBP1	A
LCAT	A
LDLR	A
LDLRAP1	A
LIPA	A
LIPC	A
LIPG	A
LMF1	A
LPL	A
MTTP	A
MYLIP	A
PCSK9	A
SAR1B	A
SCARB1	A
SLCO1B1	A
STAP1	A

NGS Lymphoedeem

Genen: 46

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
RIT1	A
LZTR1	A
RASA2	A
INPPL1	A
PIEZO1	A
A2ML1	A
RRAS	A
CDC42	A
SHANK3	A
KIF7	A
SOS2	A
ABCC9	A
ALG8	A
BRAF	A
CBL	A
CCBE1	A
CDK19	A
DCHS1	A
FAT4	A
FLT4	A
FOXC2	A
GATA2	A
GJA1	A
GJC2	A
GLA	A
HGF	A
HRAS	A
KIF11	A
KRAS	A
MAP2K1	A
MAP2K2	A
MET	A
MPI	A
NAGA	A
NRAS	A
PEPD	A
PMM2	A
PTPN11	A
PTPN14	A
RAF1	A
RELN	A
SHOC2	A
SOS1	A
SOX18	A

NGS Lymphoedeem

Genen: 46

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
VEGFC	A
EPHB4	A

NGS LangeQT-syndroom

Genen: 5

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
KCNE1	A
KCNE2	A
KCNH2	A
KCNQ1	A
SCN5A	A

NGS Metabool**Genen:** 73**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Nee

GENE	TYPE
ABCD1	A
ADSL	A
ALAD	A
ALDH7A1	A
APTX	A
ARG1	A
ARSA	A
ASL	A
ASS1	A
ATP7B	A
BCKDHA	A
BCKDHB	A
CBS	A
COQ2	A
COQ8A	A
COQ9	A
CPOX	A
CPS1	A
CYP1A2	A
CYP27A1	A
CYP2C19	A
CYP2C9	A
CYP2D6	A
CYP3A4	A
CYP3A5	A
DBT	A
DDC	A
DLD	A
GAMT	A
GATM	A
GCDH	A
GCH1	A
GNS	A
HEXA	A
HGSNAT	A
HLCS	A
HMBS	A
HPRT1	A
IDS	A
IDUA	A
IVD	A
LMBRD1	A
MAN2B1	A
MLYCD	A

NGS Metabool**Genen:** 73**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Nee

GENE	TYPE
MMAA	A
MMAB	A
MMACHC	A
MMADHC	A
MOCS1	A
MOCS2	A
MTHFR	A
MTR	A
MTRR	A
MUT	A
NAGLU	A
NAGS	A
NPC1	A
NPC2	A
OTC	A
PCBD1	A
PCCA	A
PCCB	A
PDSS1	A
PDSS2	A
PNP	A
PNPO	A
PPOX	A
PTS	A
QDPR	A
SGSH	A
SLC2A1	A
SLC6A19	A
SPR	A

NGS Neuropathie / Charcot Marie Tooth**Genen:** 123**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
AARS	C
ABHD12	C
AIFM1	C
ARHGEF10	C
ARSA	C
ATL1	C
ATL3	C
ATM	C
ATP1A1	C
ATP7A	C
BICD2	C
BSCL2	C
CCT5	C
COX10	C
COX6A1	C
CTDP1	C
DCAF8	C
DCTN1	C
DCTN2	C
DGAT2	C
DHTKD1	C
DNAH10	C
DNAJB2	C
DNAJB5	C
DNAJC3	C
DNM2	C
DNMT1	C
DRP2	C
DST	C
DYNC1H1	C
EGR2	A
RETREG1	C
FBLN5	C
FBXO38	C
FGD4	A
FIG4	C
FLVCR1	C
GALC	C
GAN	A
GARS	A
GDAP1	A
GJB1	A
GJB3	A
GNB4	C

NGS Neuropathie / Charcot Marie Tooth**Genen:** 123**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
HARS	C
HINT1	A
HK1	C
HSPB1	A
HSPB3	C
HSPB8	A
IFRD1	C
IGHMBP2	A
INF2	C
KARS	C
KIF1A	C
KIF1B	C
KIF5A	C
LAS1L	C
LITAF	A
LMNA	A
LRIG3	C
LRSAM1	A
MARS	C
MED25	C
MFN2	A
MME	C
MORC2	C
MPZ	A
MTMR2	A
MYH14	C
MYO1A	C
NAGLU	C
NDRG1	C
NEFH	C
NEFL	A
NGF	A
NTRK1	C
PDK3	A
PEX1	C
PEX7	C
PHYH	C
PLA2G6	C
PLEKHG5	C
PMP2	A
PMP22	A
PRDM12	C
PRPS1	C
PRX	C

NGS Neuropathie / Charcot Marie Tooth**Genen:** 123**Methode:** Pakket capture**Kwaliteit:** Type A/C **Incl CNV:** Nee

GENE	TYPE
RAB7A	A
REEP1	C
ELP1	C
SBF1	C
SBF2	C
SCN10A	C
SCN11A	C
SCN9A	C
SEPT9	A
SETX	C
SH3TC2	A
SIGMAR1	C
SLC12A6	C
SLC25A46	C
SLC52A2	C
SLC52A3	C
SLC5A7	C
SOX10	C
SPG11	C
SPTLC1	A
SPTLC2	C
SPTLC3	C
SURF1	C
TBCE	C
TDP1	C
TEKT3	C
TFG	C
TRIM2	C
TRPV4	A
TUBB3	C
VCP	C
VRK1	C
WNK1	A
YARS	C
PEX16	C

NGS Obesitas

Genen: 16

Methode: Pakket capture

Kwaliteit: Type A/C **Incl CNV:** Ja

GENE	TYPE
GNAS	C
MC3R	C
MC4R	A
SIM1	C
LEP	C
LEPR	C
PCSK1	C
POMC	C
ALMS1	C
VPS13B	C
PHF6	C
MAGEL2	C
BDNF	C
NPY4R	C
CPE	C
SH2B1	C

NGS Polyposis**Genen:** 22 / 23**Methode:** Pakket capture**Kwaliteit:** Type A **Incl CNV:** Ja

GENE	TYPE	Kolom1
ACVRL1	A	
APC	A	
AXIN2	A	
BMPR1A	A	Core-NL Juvenile
CDH1	A	optioneel
ENG	A	
EPCAM	A	Core-NL Lynch
MLH1	A	Core-NL Lynch
MSH2	A	Core-NL Lynch
MSH3	A	
MSH6	A	Core-NL Lynch
MUTYH	A	
NTHL1	A	
PMS2	A	Core-NL Lynch
POLD1	A	
POLE	A	
preGREM	A	
PTEN	A	
RNF43	A	
SMAD4	A	Core-NL Juvenile
STK11	A	
TSC1	A	
TSC2	A	

NGS Segmental overgrowth

Genen: 13

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Nee

GENE	TYPE
AKT1	A
AKT3	A
MTOR	A
PIK3CA	A
PIK3R2	A
PTEN	A
TSC1	A
TSC2	A
GNA11	A
GNAQ	A
HRAS	A
KRAS	A
NRAS	A

NGS Vaatmalformaties

Genen: 30

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
ACVRL1	A
AKT1	A
ANTXR1	A
BMPR2	A
CAV1	A
CCM2	A
DOCK6	A
EIF2AK4	A
ELMO2	A
ENG	A
EPHB4	A
FOXF1	A
GDF2	A
GLMN	A
GNAQ	A
KCNK3	A
KDR	A
KRAS	A
KRIT1	A
MAP2K1	A
MAP3K3	A
PDCD10	A
PIK3CA	A
PTEN	A
RASA1	A
SMAD4	A
SMAD9	A
SOX18	A
STAMBP	A
TEK	A

NGS Van Maldergem syndroom

Genen: 2

Methode: Pakket capture

Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
FAT4	A
DCHS1	A

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
AARS	C
ABAT	C
ABCC8	C
ACTB	C
ACY1	C
ADCK3	C
ADSL	C
ALDH7A1	C
ALG1	C
ALG11	C
ALG13	C
ALG3	C
ALG6	C
AMACR	C
AMT	C
APOPT1	C
ARHGEF9	C
ARID1B	C
ARX	C
ASAH1	C
ASL	C
ATP1A2	C
ATP1A3	C
ATP6AP2	C
ATP7A	C
ATRX	C
AUTS2	C
BOLA3	C
BRAT1	C
BTD	C
CACNA1A	C
CACNA1E	C
CACNA2D2	C
CASK	C
CDKL5	C
CHD2	C
CHRNA2	C
CHRNA4	C
CHRNA2	C
CLDN16	C
CLDN19	C
CLN3	C
CLN5	C
CLN6	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
CLN8	C
CNNM2	C
CNTN2	C
CNTNAP2	C
COL4A1	C
COL4A3BP	C
COQ2	C
COQ4	C
CPA6	C
CPS1	C
CPT1A	C
CPT2	C
CSTB	C
CTSD	C
CTSF	C
CUL4B	C
D2HGDH	C
DCX	C
DEPDC5	C
DLAT	C
DNAJC5	C
DNM1	C
DOCK7	C
DPAGT1	C
DPM1	C
DPM2	C
DPYD	C
DYNC1H1	C
DYRK1A	C
EEF1A2	C
EGF	C
EHMT1	C
EPM2A	C
FA2H	C
FARS2	C
FGD1	C
FLNA	C
FOLR1	C
FOXG1	C
FOXRED1	C
FXYD2	C
GABRA1	C
GABRB3	C
GABRG2	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
GAMT	C
GCK	C
GCSH	C
GLDC	C
GLRA1	C
GLRB	C
GLUD1	C
GNAO1	C
GOSR2	C
GPC3	C
GPHN	C
GRIA3	C
GRIN1	C
GRIN2A	C
GRIN2B	C
GRN	C
HADH	C
HCN1	C
HDAC4	C
HLCS	C
HNRNPU	C
HSD17B10	C
HSD17B4	C
IDH2	C
IER3IP1	C
IFIH1	C
IQSEC2	C
JAM3	C
KANSL1	C
KCNA1	C
KCNA2	C
KCNB1	C
KCNC1	C
KCNH1	C
KCNJ10	C
KCNJ11	C
KCNMA1	C
KCNQ2	C
KCNQ3	C
KCNT1	C
KCTD7	C
KDM5C	C
KIAA2022	C
KPTN	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
LG11	C
LIAS	C
MBD5	C
MECP2	C
MED12	C
MEF2C	C
MFSD8	C
MLC1	C
MOCS1	C
MOCS2	C
MPDU1	C
MTHFR	C
MTOR	C
NDUFA1	C
NDUFA11	C
NDUFAF1	C
NDUFAF2	C
NDUFAF3	C
NDUFAF4	C
NDUFAF5	C
NDUFB3	C
NDUFB9	C
NDUFS1	C
NDUFS2	C
NDUFS3	C
NDUFS4	C
NDUFS6	C
NDUFV1	C
NDUFV2	C
NECAP1	C
NEDD4L	C
NGLY1	C
NHLRC1	C
NPRL2	C
NPRL3	C
NRXN1	C
NUBPL	C
OFD1	C
OPHN1	C
PAK3	C
PC	C
PCDH19	C
PDHA1	C
PDHB	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
PDP1	C
PDX1	C
PET100	C
PEX1	C
PEX10	C
PEX12	C
PEX13	C
PEX14	C
PEX16	C
PEX19	C
PEX26	C
PEX3	C
PEX5	C
PEX6	C
PGAP3	C
PHF6	C
PHGDH	C
PIGA	C
PIGN	C
PIGO	C
PIGT	C
PLA2G6	C
PLCB1	C
PLP1	C
PMM2	C
PNKP	C
PNPO	C
POLG	C
PPP2R1A	C
PPT1	C
PQBP1	C
PRICKLE1	C
PRICKLE2	C
PRRT2	C
PURA	C
PYCR2	C
QARS	C
RAB39B	C
RARS2	C
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
ROGDI	C
RPS6KA3	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
RRM2B	C
SAMHD1	C
SCARB2	C
SCN1A	C
SCN1B	C
SCN2A	C
SCN8A	C
SIK1	C
SLC13A5	C
SLC16A1	C
SLC19A3	C
SLC25A1	C
SLC25A15	C
SLC25A22	C
SLC2A1	C
SLC35A2	C
SLC6A1	C
SLC6A8	C
SLC9A6	C
SMARCA2	C
SMC1A	C
SMS	C
SPTAN1	C
ST3GAL3	C
ST3GAL5	C
STX1B	C
STXBP1	C
SUOX	C
SYN1	C
SYNGAP1	C
SYP	C
SZT2	C
TANGO2	C
TBC1D24	C
TBCE	C
TCF4	C
TDP2	C
TPP1	C
TREX1	C
TRPM6	C
TSC1	C
TSC2	C
TSEN54	C
TUBA1A	C

NGS Epilepsie

Genen: 270

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
TUBB2A	C
TUBG1	C
UBE3A	C
WWOX	C
XK	C
ZEB2	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
AARS2	C
ABCB7	C
ABCD1	C
ABHD12	C
ACTB	C
ADAR	C
ADCK3	C
ADCY5	C
ADGRG1	C
AFG3L2	C
ALDH18A1	C
ALDH3A2	C
ALS2	C
ANO10	C
ANO3	C
AP4B1	C
AP4E1	C
AP4M1	C
AP4S1	C
AP5Z1	C
APTX	C
ARG1	C
ARSA	C
ARX	C
ASPA	C
ATCAY	C
ATL1	C
ATM	C
ATP13A2	C
ATP1A2	C
ATP1A3	C
ATP2B3	C
ATP7B	C
B4GALNT1	C
BCAP31	C
BCKDHA	C
BCKDHB	C
BSCL2	C
C10orf2	C
C12orf65	C
C19orf12	C
CA8	C
CACNA1A	C
CACNA1G	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
CACNB4	C
CAMTA1	C
CAPN1	C
CCT5	C
CIZ1	C
COASY	C
COL4A1	C
COQ2	C
COQ9	C
COX20	C
CP	C
CSF1R	C
CSTB	C
CYP27A1	C
CYP2U1	C
CYP7B1	C
DBT	C
DCAF17	C
DCC	C
DCTN1	C
DDC	C
DDHD1	C
DDHD2	C
DLAT	C
DLD	C
DNAL4	C
DNMT1	C
ECHS1	C
EIF2B1	C
EIF2B2	C
EIF2B3	C
EIF2B4	C
EIF2B5	C
EIF4G1	C
ELOVL5	C
ERLIN2	C
FA2H	C
FAR1	C
FBXO7	C
FGF14	C
FLVCR1	C
FOLR1	C
FRMD7	C
FTL	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
GALC	C
GAN	C
GBA	C
GBA2	C
GCDH	C
GCH1	C
GFAP	C
GJC2	C
GLB1	C
GNAL	C
GOSR2	C
GPR143	C
GRID2	C
GRM1	C
HEXB	C
HPRT1	C
HSPD1	C
ITPR1	C
KCNA1	C
KCNC1	C
KCNC3	C
KCND3	C
KCNJ10	C
KCNJ6	C
KCNMA1	C
KCTD7	C
KIAA0196	C
KIAA0226	C
KIAA2022	C
KIF1A	C
KIF1C	C
KIF5A	C
KMT2B	C
L1CAM	C
LAMA1	C
MARS2	C
MECP2	C
MECR	C
MICU1	C
MLC1	C
MMADHC	C
MRE11A	C
MTHFR	C
MTPAP	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
MTTP	C
NIPA1	C
NKX2-1	C
NOL3	C
NPC1	C
NPC2	C
NUP62	C
OPA1	C
PANK2	C
PAX6	C
PDE10A	C
PDE8B	C
PDGFB	C
PDGFRB	C
PDHA1	C
PDHX	C
PDSS1	C
PDSS2	C
PDYN	C
PEX10	C
PEX2	C
PEX7	C
PHYH	C
PIK3R5	C
PLA2G6	C
PLP1	C
PMM2	C
PMPCA	C
PNKD	C
PNKP	C
PNPLA6	C
POLG	C
POLR3A	C
POLR3B	C
PREPL	C
PRKCG	C
PRKRA	C
PRRT2	C
PYCR2	C
RAB18	C
RAB3GAP1	C
RAB3GAP2	C
RAD51	C
REEP1	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
RNF170	C
RNF216	C
RTN2	C
SACS	C
SAMHD1	C
SCN11A	C
SCN8A	C
SERAC1	C
SETX	C
SGCE	C
SIL1	C
SLC12A6	C
SLC16A2	C
SLC19A3	C
SLC1A3	C
SLC20A2	C
SLC25A15	C
SLC2A1	C
SLC30A10	C
SLC33A1	C
SLC39A14	C
SLC52A2	C
SLC52A3	C
SLC6A3	C
SLC9A1	C
SMPD1	C
SNCA	C
SNX14	C
SOX10	C
SPAST	C
SPG11	C
SPG20	C
SPG21	C
SPG7	C
SPR	C
SPTBN2	C
STUB1	C
SUOX	C
SYNE1	C
TAF1	C
TANGO2	C

NGS Bewegingsstoornissen

Genen: 256

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
TBC1D20	C
TDP1	C
TECPR2	C
TENM4	C
TGM6	C
TH	C
THAP1	C
TIMM8A	C
TMEM240	C
TMEM67	C
TOR1A	C
TPP1	C
TREM2	C
TREX1	C
TSEN54	C
TTBK2	C
TTC19	C
TTPA	C
TUBA1A	C
TUBB4A	C
TUBG1	C
TYROBP	C
VAMP1	C
VCP	C
VLDLR	C
VPS13A	C
VPS37A	C
VRK1	C
WDR45	C
WDR81	C
WWOX	C
XK	C
XPR1	C
ZFYVE26	C
ZFYVE27	C
ZNF592	C

NGS Spieraandoeningen

Genen: 172

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
ACADVL	C
ACTA1	C
ACVR1	C
AGL	C
AGRN	C
ALG14	C
ALG2	C
ANO5	C
ATP2A1	C
ATP7A	C
B3GALNT2	C
B4GAT1	C
BAG3	C
BICD2	C
BIN1	C
C10ORF2	C
CACNA1S	C
CAPN3	C
CASQ1	C
CAV3	C
CAVIN1	C
CCDC78	C
CFL2	C
CHAT	C
CHCHD10	C
CHKB	C
CHRNA1	C
CHRNB1	C
CHRND	C
CHRNE	C
CLCN1	C
CNTN1	C
COL12A1	C
COL13A1	C
COL6A1	C
COL6A2	C
COL6A3	C
COLQ	C
CPT2	C
CRYAB	C
DAG1	C
DES	C
DMD	C
DNA2	C

NGS Spieraandoeningen

Genen: 172

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
DNAJB6	C
DNM2	C
DOK7	C
DPAGT1	C
DPM1	C
DPM2	C
DPM3	C
DYNC1H1	C
DYSF	C
ECEL1	C
EMD	C
ENO3	C
ERBB3	C
EXOSC8	C
FAM111B	C
FHL1	C
FKBP14	C
FKRP	C
FKTN	C
FLNC	C
GAA	C
GBE1	C
GFPT1	C
GMPPB	C
GNE	C
GYG1	C
GYS1	C
HSPG2	C
IGHMBP2	C
INPP5K	C
ISCU	C
ISPD	C
ITGA7	C
KBTBD13	C
KCNJ2	C
KLHL40	C
KLHL41	C
KLHL9	C
LAMA2	C
LAMA5	C
LAMP2	C
LARGE	C
LDB3	C
LDHA	C

NGS Spieraandoeningen

Genen: 172

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
LMNA	C
LMOD3	C
LPIN1	C
MEGF10	C
MICU1	C
MORC2	C
MSTN	C
MTM1	C
Munc13-1	C
MUSK	C
MYF6	C
MYH2	C
MYH3	C
MYH7	C
MYO9A	C
MYOT	C
NAPA	C
NEB	C
OPA1	C
ORAI1	C
PABPN1	C
PFKM	C
PGAM2	C
PGK1	C
PGM1	C
PHKA1	C
PIP5K1C	C
PLEC	C
PNPLA2	C
POMGNT1	C
POMGNT2	C
POMK	C
POMT1	C
POMT2	C
PREPL	C
PRPS1	C
PTRF	C
PYGM	C
PYROXD1	C
RAPSN	C
RBCK1	C
RPH3A	C
RRM2B	C
RYR1	C

NGS Spieraandoeningen

Genen: 172

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
SCN4A	C
SEPN1	C
SGCA	C
SGCB	C
SGCD	C
SGCG	C
SLC5A7	C
SLC18A2	C
SLC18A3	C
SLC25A1	C
SLC52A2	C
SLC52A3	C
SLC25A4	C
SMCHD1	C
SPEG	C
STIM1	C
SYT2	C
TANGO2	C
TCAP	C
TMEM5	C
TNNI2	C
TNNT1	C
TNPO3	C
TPM2	C
TPM3	C
TRAPPC11	C
TRIM32	C
TRIP4	C
TRPV4	C
TTC19	C
TTN	C
UBA1	C
VAMP1	C
VCP	C
VIPAS39	C
VMA21	C
VRK1	C
XK	C
YARS	C
ZC4H2	C

NGS Amyotrofe Lateraal Sclerose (ALS)

Genen: 38

Methode: WES met filter

Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
OPTN	C
SETX	C
SOD1	C
TARDBP	C
UBQLN2	C
VCP	C
FUS	C
SQSTM1	C
VAPB	C
ALS2	C
ANG	C
CHMP2B	C
PFN1	C
SIGMAR1	C
TBK1	C
SPG11	C
CHCHD10	C
FIG4	C
DCTN1	C
HNRNPA1	C
MATR3	C
ErbB4	C
TUBA4A	C
C9orf72	C
PRPH	C
NEFH	C
HNRNPA2B1	C
ATXN2	C
MAPT	C
TAF15	C
NEK1	C
PSEN1	C
TREM2	C
DAO	C
UNC13A	C
ANXA11	C
CCNF	C
GLE1	C

NGS Pontocerebellaire Hypoplasie (PCH)**Genen:** 37**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
AMPD2	C
ATXN2	C
ATXN7	C
CASK	C
CHMP1A	C
CLP1	C
DKC1	C
EXOSC3	C
EXOSC8	C
EXOSC9	C
FKRP	C
FKTN	C
ISPD	C
ITPR1	C
LARGE	C
PCLO	C
PMM2	C
POMGNT1	C
POMT1	C
POMT2	C
RARS2	C
RELN	C
SEPSECS	C
TBC1D23	C
TOE1	C
TSEN15	C
TSEN2	C
TSEN34	C
TSEN54	C
TUBA1A	C
TUBA8	C
TUBB	C
TUBB2B	C
TUBB3	C
VLDLR	C
VPS53	C
VRK1	C

NGS Microcefalie**Genen:** 86**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
AGMO	C
ANKLE2	C
ARFGEF2	C
ASPM	C
ASXL3	C
ATR	C
ATRX	C
CASC5	C
CASK	C
CDC6	C
CDK5RAP2	C
CDK6	C
CDKL5	C
CDT1	C
CENPE	C
CENPJ	C
CEP135	C
CEP152	C
CEP63	C
CIT	C
COPB2	C
CRIP1	C
CTNNA1	C
DHCR7	C
DIAPH1	C
DYRK1A	C
EFTUD2	C
EIF2S3	C
FOXP1	C
IER3IP1	C
KATNB1	C
KIAA1279	C
KIF11	C
KIF14	C
LIG4	C
MCPH1	C
MECP2	C
MED17	C
MFSD2A	C
MYCN	C
NBN	C
NDE1	C
NHEJ1	C
NIN	C

NGS Microcefalie**Genen:** 86**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
NIPBL	C
ORC1	C
ORC4	C
ORC6	C
PCNT	C
PHC1	C
PLK4	C
PNKP	C
PPP1R15B	C
PQBP1	C
PYCR2	C
QARS	C
RAB18	C
RAB3GAP1	C
RAB3GAP2	C
RAD50	C
RBBP8	C
SASS6	C
SLC1A4	C
SLC25A19	C
SLC2A1	C
SLC9A6	C
SOX11	C
SPATA5	C
STAMBP	C
STIL	C
TBC1D20	C
TCF4	C
TRAPPC9	C
TRMT10A	C
TSEN2	C
TSEN34	C
TSEN54	C
TUBB3	C
TUBGCP4	C
TUBGCP6	C
UBE3A	C
WDFY3	C
WDR62	C
WWOX	C
ZEB2	C
ZNF335	C

NGS Cornelia de Lange**Genen:** 23**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
ADNP	C
AFF4	C
ANKRD11	C
ARID1A	C
ARID1B	C
CREBBP	C
EP300	C
ESCO2	C
HDAC8	C
NIPBL	C
PHF6	C
RAD21	C
ROR2	C
SMARCA2	C
SMARCA4	C
SMARCB1	C
SMARCE1	C
SMC1A	C
SMC3	C
SOX11	C
TAF6	C
TBC1D24	C
WNT5A	C
DHCR7	C
DIAPH1	C
DYRK1A	C
EFTUD2	C
EIF2S3	C
FOXG1	C
IER3IP1	C
KATNB1	C
KIAA1279	C
KIF11	C
KIF14	C
LIG4	C
MCPH1	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
A2ML1	C
AAAS	C
AARS	C
AASS	C
ABAT	C
ABCC9	C
ABCD1	C
ABCD4	C
ABHD5	C
ACAD9	C
ACBD6	C
ACD	C
ACO2	C
ACOX1	C
ACSF3	C
ACSL4	C
ACTB	C
ACTG1	C
ACVR1	C
ACY1	C
ADAM22	C
ADAR	C
ADAT3	C
ADCK3	C
ADGRG1	C
ADK	C
ADNP	C
ADRA2B	C
ADSL	C
AFF2	C
AFF4	C
AFG3L2	C
AGA	C
AGO2	C
AGPAT2	C
AGTR2	C
AHCY	C
AHDC1	C
AHI1	C
AIFM1	C
AIMP1	C
AK1	C
AKT3	C
ALDH18A1	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
ALDH3A2	C
ALDH4A1	C
ALDH5A1	C
ALG1	C
ALG11	C
ALG12	C
ALG13	C
ALG2	C
ALG3	C
ALG6	C
ALG8	C
ALG9	C
ALMS1	C
ALX1	C
ALX4	C
AMER1	C
AMMECR1	C
AMPD2	C
AMT	C
ANK3	C
ANKH	C
ANKLE2	C
ANKRD11	C
ANO10	C
ANTXR1	C
AP1S1	C
AP1S2	C
AP3B1	C
AP3B2	C
AP3D1	C
AP4B1	C
AP4E1	C
AP4M1	C
AP4S1	C
AP5Z1	C
APC2	C
APOPT1	C
APTX	C
ARFGEF2	C
ARG1	C
ARHGAP31	C
ARHGEF6	C
ARHGEF9	C
ARID1A	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
ARID1B	C
ARID2	C
ARIH1	C
ARL13B	C
ARL6	C
ARMC9	C
ARSA	C
ARSE	C
ARX	C
ASAH1	C
ASCC1	C
ASCL1	C
ASH1L	C
ASL	C
ASNS	C
ASPA	C
ASPM	C
ASS1	C
ASXL1	C
ASXL2	C
ASXL3	C
ATCAY	C
ATIC	C
ATN1	C
ATP1A2	C
ATP1A3	C
ATP2A2	C
ATP6AP2	C
ATP6V0A2	C
ATP6V1B2	C
ATP7A	C
ATP8A2	C
ATPAF2	C
ATR	C
ATRX	C
AUH	C
AUTS2	C
AVPR2	C
B3GALNT2	C
B3GALT6	C
B3GALT	C
B4GALNT1	C
B4GALT1	C
B4GALT7	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
B4GAT1	C
BBS1	C
BBS10	C
BBS12	C
BBS2	C
BBS4	C
BBS5	C
BBS7	C
BBS9	C
BCAP31	C
BCKDHA	C
BCKDHB	C
BCL11A	C
BCOR	C
BCORL1	C
BCS1L	C
BLM	C
BRAF	C
BRAT1	C
BRD4	C
BRF1	C
BRPF1	C
BRWD3	C
BSCL2	C
BSND	C
BTD	C
BUB1B	C
C10orf2	C
C11ORF73	C
C12orf4	C
C12orf57	C
C12orf65	C
C2CD3	C
C5orf42	C
CA2	C
CA5A	C
CA8	C
CACNA1A	C
CACNA1C	C
CACNA1G	C
CACNA2D1	C
CACNG2	C
CAD	C
CAMK2A	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
CAMK2B	C
CAMTA1	C
CAPN10	C
CARS2	C
CASC5	C
CASK	C
CBL	C
CBS	C
CC2D1A	C
CC2D2A	C
CCBE1	C
CCDC115	C
CCDC174	C
CCDC22	C
CCDC78	C
CCDC88A	C
CCDC88C	C
CCND2	C
CDH15	C
CDK10	C
CDK13	C
CDK16	C
CDK5	C
CDK5RAP2	C
CDKL5	C
CDKN1C	C
CDON	C
CENPE	C
CENPF	C
CENPJ	C
CEP104	C
CEP120	C
CEP135	C
CEP152	C
CEP290	C
CEP41	C
CEP83	C
CEP89	C
CHAMP1	C
CHD1	C
CHD2	C
CHD3	C
CHD4	C
CHD7	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
CHD8	C
CHKB	C
CHP1	C
CHRNA4	C
CIC	C
CIP2A	C
CIT	C
CKAP2L	C
CLCN4	C
CLCNKB	C
CLIC2	C
CLIP1	C
CLN3	C
CLN5	C
CLN6	C
CLN8	C
CLP1	C
CLPB	C
CLTC	C
CLTCL1	C
CNKSR2	C
CNNM2	C
CNTNAP2	C
COASY	C
COG1	C
COG2	C
COG4	C
COG5	C
COG6	C
COG7	C
COG8	C
COL13A1	C
COL4A1	C
COL4A2	C
COL4A3BP	C
COLEC11	C
COQ2	C
COQ4	C
COQ9	C
COX10	C
COX15	C
COX6B1	C
CPE	C
CPS1	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
CRADD	C
CRB2	C
CRBN	C
CREBBP	C
CRLF1	C
CSNK2A1	C
CSPP1	C
CSTB	C
CTBP1	C
CTCF	C
CTDP1	C
CTNNB1	C
CTNND1	C
CTNND2	C
CTSA	C
CTSD	C
CTTNBP2	C
CUBN	C
CUL4B	C
CUX2	C
CWF19L1	C
CYB5R3	C
CYP27A1	C
CYP2U1	C
D2HGDH	C
DAG1	C
DARS2	C
DBT	C
DCAF17	C
DCC	C
DCHS1	C
DCPS	C
DCX	C
DDC	C
DDHD2	C
DDOST	C
DDX11	C
DDX3X	C
DDX59	C
DEAF1	C
DENND5A	C
DEPDC5	C
DHCR24	C
DHCR7	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
DHDDS	C
DHFR	C
DHTKD1	C
DHX30	C
DIAPH1	C
DIP2B	C
DKC1	C
DLD	C
DLG3	C
DLG4	C
DMD	C
DMPK	C
DMRTA2	C
DNAJC12	C
DNAJC19	C
DNM1	C
DNM1L	C
DNMT3A	C
DNMT3B	C
DOCK6	C
DOCK7	C
DOCK8	C
DOLK	C
DONSON	C
DPAGT1	C
DPH1	C
DPM1	C
DPP6	C
DPYD	C
DPYS	C
DST	C
DVL1	C
DYM	C
DYNC1H1	C
DYRK1A	C
EBF3	C
EBP	C
ECHS1	C
EDC3	C
EDNRA	C
EEF1A2	C
EFTUD2	C
EHMT1	C
EIF2AK3	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
EIF2S3	C
EIF4A3	C
EIF4G1	C
ELAC2	C
ELOVL4	C
ELOVL5	C
ELP2	C
EMC1	C
EMX2	C
ENTPD1	C
EP300	C
EPB41L1	C
EPG5	C
ERCC2	C
ERCC3	C
ERCC5	C
ERCC6	C
ERCC8	C
ERLIN2	C
ESCO2	C
ETFB	C
ETHE1	C
EXOC6B	C
EXOC8	C
EXOSC2	C
EXOSC3	C
EXOSC8	C
EXT2	C
EZH2	C
EZR	C
FA2H	C
FAM126A	C
FAR1	C
FASN	C
FAT4	C
FBN1	C
FBXL4	C
FBXO11	C
FBXO31	C
FGD1	C
FGF12	C
FGF14	C
FGFR1	C
FGFR2	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
FGFR3	C
FH	C
FIBP	C
FIGN	C
FKRP	C
FKTN	C
FLNA	C
FLVCR1	C
FMN2	C
FMR1	C
FOXG1	C
FOXP1	C
FOXP2	C
FRAS1	C
FREM2	C
FRMD4A	C
FRMPD4	C
FRRS1L	C
FTCD	C
FTO	C
FTSJ1	C
FUCA1	C
GABBR2	C
GABRA1	C
GABRB1	C
GABRB3	C
GABRD	C
GAD1	C
GALE	C
GALT	C
GAMT	C
GATAD2B	C
GATM	C
GCDH	C
GCH1	C
GCSH	C
GDI1	C
GFAP	C
GFM2	C
GJA1	C
GJB1	C
GJC2	C
GK	C
GLB1	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
GLDC	C
GLI2	C
GLI3	C
GLYCTK	C
GM2A	C
GMPPA	C
GMPPB	C
GNAO1	C
GNAS	C
GNB1	C
GNB5	C
GNPAT	C
GNPTAB	C
GNS	C
GPC3	C
GPHN	C
GPT2	C
GRIA3	C
GRID2	C
GRIK2	C
GRIN1	C
GRIN2A	C
GRIN2B	C
GRIN2D	C
GRIN3B	C
GRIP1	C
GRM1	C
GRN	C
GSE1	C
GSS	C
GTF2H5	C
GTPBP3	C
GUSB	C
HACE1	C
HAX1	C
HCCS	C
HCFC1	C
HCN1	C
HDAC4	C
HDAC6	C
HDAC8	C
HECTD1	C
HECW2	C
HEPACAM	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
HERC1	C
HERC2	C
HESX1	C
HEXA	C
HEXB	C
HGSNAT	C
HIST1H1E	C
HIST1H4C	C
HIVEP2	C
HLCS	C
HMGCL	C
HNMT	C
HNRNPH1	C
HNRNPH2	C
HNRNPK	C
HNRNPU	C
HOXA1	C
HPD	C
HPRT1	C
HRAS	C
HSD17B10	C
HSPA9	C
HSPD1	C
HUWE1	C
HYLS1	C
IARS	C
IARS2	C
IBA57	C
IDS	C
IDUA	C
IER3IP1	C
IFIH1	C
IFT122	C
IFT172	C
IFT81	C
IGBP1	C
IGF1	C
IGF1R	C
IKBKG	C
IKKA	C
IL1RAPL1	C
IMPA1	C
INPP4A	C
INPP5E	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
INPP5K	C
IQSEC2	C
IRF6	C
ISCA2	C
ISPD	C
ITGA7	C
ITPA	C
ITPR1	C
IVD	C
JAG1	C
JAM3	C
JMJD1C	C
KALRN	C
KANK1	C
KANSL1	C
KAT6A	C
KAT6B	C
KATNB1	C
KCNA2	C
KCNA4	C
KCNB1	C
KCNC1	C
KCNC3	C
KCNH1	C
KCNJ10	C
KCNJ11	C
KCNJ6	C
KCNK9	C
KCNMA1	C
KCNQ2	C
KCNQ3	C
KCNQ5	C
KCNT1	C
KCTD7	C
KDM1A	C
KDM5C	C
KDM6A	C
KDM6B	C
KIAA0226	C
KIAA0586	C
KIAA1033	C
KIAA1109	C
KIAA1279	C
KIAA2022	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
KIDINS220	C
KIF11	C
KIF14	C
KIF1A	C
KIF2A	C
KIF4A	C
KIF5C	C
KIF7	C
KIRREL3	C
KLF8	C
KLHL15	C
KMT2A	C
KMT2B	C
KMT2C	C
KMT2D	C
KMT2E	C
KMT5B	C
KNL1	C
KPTN	C
KRAS	C
KRBOX4	C
L1CAM	C
L2HGDH	C
LAMA1	C
LAMA2	C
LAMC3	C
LAMP2	C
LARGE	C
LARP7	C
LAS1L	C
LIAS	C
LIG4	C
LINGO1	C
LINS	C
LMAN2L	C
LMNB2	C
LONP1	C
LRP2	C
LRPPRC	C
LZTFL1	C
LZTR1	C
MAB21L2	C
MAF	C
MAG	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
MAGEL2	C
MAGT1	C
MAN1B1	C
MAN2B1	C
MANBA	C
MAOA	C
MAP2K1	C
MAP2K2	C
MAPRE2	C
MARS2	C
MASP1	C
MAT1A	C
MBD5	C
MBOAT7	C
MBTPS2	C
MCCC1	C
MCCC2	C
MCOLN1	C
MCPH1	C
MDH2	C
MECP2	C
MECR	C
MED12	C
MED13L	C
MED17	C
MED23	C
MED25	C
MEF2C	C
MEIS2	C
METTL23	C
MFF	C
MFSD2A	C
MFSD8	C
MGAT2	C
MICU1	C
MID1	C
MID2	C
MKKS	C
MKS1	C
MLC1	C
MLYCD	C
MMAA	C
MMAB	C
MMACHC	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
MMADHC	C
MOCS1	C
MOCS2	C
MOGS	C
MPDU1	C
MPDZ	C
MPLKIP	C
MRPL3	C
MRPS22	C
MSL3	C
MTFMT	C
MTHFR	C
MTM1	C
MTOR	C
MTR	C
MTRR	C
MUT	C
MVK	C
MYCN	C
MYH10	C
MYH9	C
MYO5A	C
MYT1L	C
NAA10	C
NAA15	C
NACC1	C
NADK2	C
NAGA	C
NAGLU	C
NALCN	C
NANS	C
NAPB	C
NARS2	C
NBEA	C
NBN	C
NDE1	C
NDP	C
NDST1	C
NDUFA1	C
NDUFA11	C
NDUFA12	C
NDUFA13	C
NDUFA2	C
NDUFAF3	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
NDUFAF5	C
NDUFB11	C
NDUFS1	C
NDUFS2	C
NDUFS3	C
NDUFS4	C
NDUFS6	C
NDUFS7	C
NDUFS8	C
NDUFV1	C
NDUFV2	C
NEB	C
NECAP1	C
NEDD4L	C
NEU1	C
NF1	C
NFATC1	C
NFIA	C
NFIB	C
NFIX	C
NGLY1	C
NHS	C
NIPBL	C
NKX2-1	C
NLGN3	C
NLGN4X	C
NLRP3	C
NONO	C
NOS1	C
NOTCH1	C
NPC1	C
NPC2	C
NPHP1	C
NR2F1	C
NRAS	C
NRXN1	C
NSD1	C
NSD2	C
NSDHL	C
NSUN2	C
NTRK1	C
NUP62	C
NUS1	C
NXF5	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
OAT	C
OCLN	C
OCRL	C
ODC1	C
OFD1	C
OGT	C
OPHN1	C
ORC1	C
OSGEP	C
OTC	C
OTUD6B	C
P4HTM	C
PACS1	C
PAFAH1B1	C
PAH	C
PAK3	C
PANK2	C
PANX1	C
PARP1	C
PAX1	C
PAX6	C
PAX8	C
PBX1	C
PC	C
PCCA	C
PCCB	C
PCDH19	C
PCGF2	C
PCLO	C
PCNT	C
PCYT2	C
PDE4D	C
PDHA1	C
PDP1	C
PDSS1	C
PDSS2	C
PEPD	C
PET100	C
PEX1	C
PEX10	C
PEX11B	C
PEX12	C
PEX13	C
PEX16	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
PEX19	C
PEX2	C
PEX26	C
PEX3	C
PEX5	C
PEX6	C
PEX7	C
PGAP1	C
PGAP2	C
PGAP3	C
PGK1	C
PGM3	C
PHF6	C
PHF8	C
PHGDH	C
PHIP	C
PI4KA	C
PIGA	C
PIGC	C
PIGG	C
PIGL	C
PIGN	C
PIGO	C
PIGT	C
PIGV	C
PIGW	C
PIGY	C
PIK3CA	C
PIK3R2	C
PLA2G6	C
PLCB1	C
PLK4	C
PLP1	C
PLXND1	C
PMM2	C
PMPCA	C
PNKP	C
PNP	C
PNPLA6	C
POC1A	C
POGZ	C
POLG	C
POLR1D	C
POLR3A	C

NGS Intellectual disability**Genen:** 1228**Methode:** WES met filter**Kwaliteit:** Type C **Incl CNV:** Nee

Gene	Type
POLR3B	C
POMGNT1	C
POMGNT2	C
POMK	C
POMT1	C
POMT2	C
PORCN	C
POU1F1	C
POU3F2	C
POU3F3	C
PPM1D	C
PPOX	C
PPP1CB	C
PPP1R15B	C
PPP2R1A	C
PPP2R5B	C
PPP2R5C	C
PPP2R5D	C
PPP3CA	C
PPT1	C
PQBP1	C
PRF1	C
PRKAR1A	C
PRMT7	C
PRODH	C
PRPS1	C
PRR12	C
PRRT2	C
PRSS12	C
PSAP	C
PSAT1	C
PSPH	C
PTCH1	C
PTCHD1	C
PTDSS1	C
PTEN	C
PTPN11	C
PTPN4	C
PTRH2	C
PTRHD1	C
PTS	C
PUF60	C
PUM1	C
PURA	C

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Gene	Type
PUS1	C
PUS3	C
PUS7	C
PVRL1	C
PYCR1	C
PYCR2	C
QARS	C
QDPR	C
QRSL1	C
RAB11B	C
RAB18	C
RAB27A	C
RAB39B	C
RAB3GAP1	C
RAB3GAP2	C
RAB40AL	C
RAC1	C
RAD21	C
RAF1	C
RAI1	C
RAP1A	C
RAP1B	C
RARB	C
RARS2	C
RASA2	C
RBFOX1	C
RBM10	C
RBM28	C
RBMX	C
RBPJ	C
RBSN	C
RCBTB1	C
RELN	C
RERE	C
REV3L	C
RFT1	C
RGS6	C
RHEB	C
RIPK4	C
RIT1	C
RLIM	C
RMND1	C
RMRP	C
RNASEH2A	C

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Gene	Type
RNASEH2B	C
RNASEH2C	C
RNASET2	C
RNF113A	C
RNF125	C
ROGDI	C
RPGRIP1L	C
RPL10	C
RPS6KA3	C
RSPRY1	C
RTEL1	C
RTTN	C
RUSC2	C
SALL1	C
SASS6	C
SATB2	C
SBDS	C
SC5D	C
SCAPER	C
SCN1A	C
SCN1B	C
SCN2A	C
SCN8A	C
SCO1	C
SCO2	C
SCYL1	C
SDHA	C
SEMA3E	C
SEPSECS	C
SERAC1	C
SET	C
SETBP1	C
SETD1A	C
SETD1B	C
SETD2	C
SETD5	C
SF1	C
SGSH	C
SHANK2	C
SHANK3	C
SHH	C
SHOC2	C
SHROOM4	C
SIK1	C

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Gene	Type
SIL1	C
SIN3A	C
SIX3	C
SKI	C
SLC12A6	C
SLC13A5	C
SLC16A2	C
SLC17A5	C
SLC19A3	C
SLC1A1	C
SLC1A2	C
SLC1A4	C
SLC25A12	C
SLC25A15	C
SLC25A22	C
SLC2A1	C
SLC33A1	C
SLC35A1	C
SLC35A2	C
SLC35C1	C
SLC39A12	C
SLC39A8	C
SLC4A4	C
SLC6A1	C
SLC6A17	C
SLC6A3	C
SLC6A8	C
SLC7A7	C
SLC9A1	C
SLC9A6	C
SMAD4	C
SMAD6	C
SMARCA2	C
SMARCA4	C
SMARCB1	C
SMARCC2	C
SMARCE1	C
SMC1A	C
SMC3	C
SMG9	C
SMO	C
SMOC1	C
SMPD1	C
SMS	C

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Gene	Type
SNAP25	C
SNAP29	C
SNIP1	C
SNRPB	C
SNRPN	C
SNX14	C
SOBP	C
SON	C
SOS1	C
SOS2	C
SOX10	C
SOX11	C
SOX2	C
SOX3	C
SOX5	C
SPAST	C
SPATA5	C
SPECC1L	C
SPG11	C
SPG20	C
SPOCK1	C
SPRED1	C
SPTAN1	C
SPTBN2	C
SRCAP	C
SRD5A3	C
SRPX2	C
SSR4	C
ST3GAL3	C
ST3GAL5	C
STAG1	C
STAG2	C
STAMBP	C
STIL	C
STIM1	C
STRA6	C
STT3A	C
STT3B	C
STX1B	C
STX3	C
STXBP1	C
STXBP5L	C
STYXL1	C
SUCLA2	C

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Gene	Type
SUCLG1	C
SUMF1	C
SUOX	C
SURF1	C
SYN1	C
SYNCRIP	C
SYNE1	C
SYNGAP1	C
SYP	C
SYT14	C
SZT2	C
TAF1	C
TAF13	C
TAF2	C
TAF6	C
TANC2	C
TANGO2	C
TAT	C
TBC1D20	C
TBC1D24	C
TBC1D7	C
TBCD	C
TBCE	C
TBCK	C
TBL1XR1	C
TBP	C
TBR1	C
TCF20	C
TCF4	C
TCF7L2	C
TCN2	C
TCTN3	C
TDP2	C
TECPR2	C
TECR	C
TELO2	C
TFAP2A	C
TG	C
TGDS	C
TGFBR1	C
TGFBR2	C
TGIF1	C
TH	C
THOC2	C

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Gene	Type
THOC6	C
THRB	C
TIMM8A	C
TINF2	C
TKT	C
TLK2	C
TMCO1	C
TMEM135	C
TMEM165	C
TMEM231	C
TMEM237	C
TMEM240	C
TMEM67	C
TMEM70	C
TMLHE	C
TNIK	C
TOE1	C
TP53RK	C
TPI1	C
TPO	C
TPP1	C
TPRKB	C
TRAIP	C
TRAPPC11	C
TRAPPC9	C
TREX1	C
TRIM32	C
TRIO	C
TRIP12	C
TRIP4	C
TRMT1	C
TRMT10A	C
TRMT10C	C
TRNT1	C
TSC1	C
TSC2	C
TSEN15	C
TSEN54	C
TSHB	C
TSHR	C
TSPAN7	C
TTC19	C
TTC37	C
TTC8	C

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Gene	Type
TTI2	C
TUBA1A	C
TUBA8	C
TUBB	C
TUBB2B	C
TUBB3	C
TUBB4A	C
TUBG1	C
TUBGCP4	C
TUBGCP6	C
TUSC3	C
TWIST1	C
TWIST2	C
UBA5	C
UBE2A	C
UBE3A	C
UBE3B	C
UBR1	C
UBR7	C
UBTF	C
UNC80	C
UPB1	C
UPF3B	C
UQCRQ	C
UROC1	C
USP27X	C
USP7	C
USP9X	C
VAMP2	C
VLDLR	C
VPS11	C
VPS13B	C
VPS37A	C
VPS53	C
VRK1	C
VWA3B	C
WAC	C
WARS2	C
WDR13	C
WDR19	C
WDR26	C
WDR4	C
WDR45	C
WDR62	C

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Gene	Type
WDR73	C
WDR81	C
WFS1	C
WWOX	C
XPA	C
XPNPEP3	C
XRCC4	C
XYL1	C
XYL2	C
YAP1	C
YME1L1	C
YWHAE	C
YY1	C
ZBTB16	C
ZBTB18	C
ZBTB20	C
ZBTB24	C
ZBTB40	C
ZC3H14	C
ZC4H2	C
ZCCHC12	C
ZCCHC8	C
ZDHHC15	C
ZDHHC9	C
ZEB2	C
ZFYVE26	C
ZIC1	C
ZIC2	C
ZMYM3	C
ZMYND11	C
ZNF148	C
ZNF292	C
ZNF407	C
ZNF41	C
ZNF526	C
ZNF592	C
ZNF674	C
ZNF711	C
ZNF81	C
ZSWIM6	C