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). Zie https://www.dnadiagnostiek.nl/core-genen.
Per analyse is aangegeven waar een Core-NL pakket wordt aangeboden.

Rapportage nevenbevindingen: Met het aanvragen van WES-gebaseerde (pakket)analyses gaan we ervan uit dat aanvrager de kans op nevenbevindingen met de patiënt heeft besproken. Zie voor meer informatie: https://www.vkgn.org/nieuws/landelijk-beleid-voor-het-melden-van-nevenbevindingen-in-de-klinisch-genetische-diagnostiek/

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

Laboratorium handelingen en declaraties tbv WES gebaseerde analyses worden uitgevoerd bij Laboratorium Genoomanalyse, Amsterdam UMC, Locatie VUmc (RvA M130).

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NGS Arthrogrypose

Genen: 8

Methode: Pakket capture (AGPv5)

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:	42		
Methode:	Pakket capture		(SCDv8)
Kwaliteit:	Type A	**Incl CNV:**	Ja

GENE	TYPE
ABCC9	A
AKAP9	A
ANK2	A
CACNA1C	A
CALM1	A
CALM2	A
CALM3	A
CASQ2	A
CAV3	A
DPP6	A
GJA5	A
HCN4	A
HEY2	A
HOOK3	A
JPH2	A
KCNA5	A
KCND3	A
KCNE1	A
KCNE2	A
KCNH2	A
KCNJ2	A
KCNQ1	A
LAMP2	A
LMNA	A
MYL4	A
NKX2-5	A
NPPA	A
PKP2	A
PLN	A
PRKAG2	A
RANGRF	A
RRAD	A
RYR2	A
SCN5A	A
SLC4A3	A
SNTA1	A
TCAP	A
TECRL	A
TNNI3K	A
TNNT2	A
TRDN	A
TRPM4	A

NGS Plotse hartdood (SCD)

Genen: 54

Methode: Pakket capture (SCDv8)

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

GENE	TYPE
ABCC9	A
AKAP9	A
ANK2	A
CACNA1C	A
CALM1	A
CALM2	A
CALM3	A
CASQ2	A
CAV3	A
DPP6	A
DSC2	A
DSG2	A
DSP	A
FLNC	A
GJA5	A
HCN4	A
HEY2	A
HOOK3	A
JPH2	A
JUP	A
KCNA5	A
KCND3	A
KCNE1	A
KCNE2	A
KCNH2	A
KCNJ2	A
KCNQ1	A
LAMP2	A
LMNA	A
MYBPC3	A
MYH7	A
MYL2	A
MYL3	A
MYL4	A
NKX2-5	A
NPPA	A
PKP2	A
PLN	A
PRKAG2	A
RANGRF	A
RBM20	A
RRAD	A
RYR2	A
SCN5A	A

GENE	TYPE
SLC4A3	A
SNTA1	A
TCAP	A
TECRL	A
TNNI3	A
TNNI3K	A
TNNT2	A
TPM1	A
TRDN	A
TRPM4	A

NGS Brugada syndroom			
Genen:	1		
Methode:	Pakket capture		(SCDv8; BRSv1)
Kwaliteit:	Type A	**Incl CNV:**	Ja

GENE	TYPE
SCN5A	A

NGS Dragerschap Preconceptie

Genen: 50 (PCOv2; BMUTtypeAv1)

Methode: Pakket capture met filter voor bekende (waarsch.) pathogene (fouder)mutaties

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

GENE	TYPE
AGA	A
ALDH3A2	A
ARSA	A
ASPA	A
ASS1	A
BCKDHB	A
BLM	A
CBS	A
CFTR	A
CLN3	A
CLN5	A
COL17A1	A
CRTAP	A
DHCR7	A
ELP1	A; alleen exon 19, 20
FANCC	A
G6PC	A
GAA	A
GALC	A
HADHA	A
HBB	A
HEXA	A
HGSNAT	A; muv exon 1 (C)
HSD17B4	A
IDUA	A
IVD	A
LAMA3	A
LAMB3	A
LAMC2	A
MAN2B1	A
MCOLN1	A
MLC1	A
MUSK	A
NBN	A
NEB	A; alleen exonen 53-57
NPC1	A
PEX1	A
PEX12	A
PEX7	A
PMM2	A
POLG	A
PPT1	A
SACS	A
SGSH	A

GENE	TYPE
SMN1	A
SMPD1	A
TMEM216	A
TPP1	A
TSEN54	A; muv exon 1 (C)
TTPA	A

NGS Albinisme
Genen: 29
Methode: Pakket capture (ALBIv4; ALBItypeAv3)
Kwaliteit: Type A/C **Incl CNV:** Nee

GENE	TYPE
AP3B1	A
AP3D1	C
BLOC1S3	A
BLOC1S6	A
COL18A1	C
DTNBP1	A
FRMD7	A
GNAI3	C
GPR143	A
HPS1	A
HPS3	A
HPS4	A
HPS5	A
HPS6	A
LRMDA	A
LRMDA	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

NGS Cone-rod dystrophie

Genen: 260

Methode: Pakket capture (BHv6; CRDtypeAv5)

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
AHR	C
AIPL1	A
ALMS1	C
ARHGEF18	C
ARL2BP	C
ARL3	C
ARL6	C
ARSG	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
C8orf37	A
CA4	C
CABP4	C
CACNA1F	A
CACNA2D4	C
CAPN5	C
CC2D2A	C
CDH23	C

GENE	TYPE
FSCN2	C
FZD4	C
GDF6	C
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	A
GUCA1B	A
GUCY2D	A
HARS1	C
HCCS	C
HGSNAT	C
HK1	C
HMX1	C
IDH3A	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

GENE	TYPE
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	A
POC5	C
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	C
RP1L1	A
RP2	C
RP9	C
RPE65	C

NGS Cone-rod dystrophie

Genen: 260

Methode: Pakket capture (BHv6; CRDtypeAv5)

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

GENE	TYPE
CDH3	C
CDHR1	A
CEP164	C
CEP19	C
CEP250	C
CEP290	C
CEP41	C
CEP78	A
CERKL	A
CFAP410	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
CPLANE1	C
CRB1	C
CRX	A
CSPP1	C
CTNNA1	A
CWC27	C
CYP4V2	C
DHDDS	C
DHX38	C
DRAM2	C
DTHD1	C
EFEMP1	C
ELOVL4	A
EMC1	C
EXOSC2	C
EYS	C
FAM161A	C
FLVCR1	C
FOXE3	C

GENE	TYPE
MAPKAPK3	C
MERTK	C
MFN2	C
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
PAX6	C
PCARE	C
PCDH15	C
PCYT1A	C
PDE6A	C
PDE6B	C
PDE6C	A
PDE6G	C
PDE6H	A

GENE	TYPE
RPGR	C
RPGRIP1	A
RPGRIP1L	C
RS1	C
SAG	C
SCAPER	C
SDCCAG8	C
SEMA4A	A
SIX6	C
SLC24A1	C
SLC7A14	C
SNRNP200	C
SPATA7	C
SPP2	C
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	A
TMEM126A	C
TMEM237	C
TMEM67	C
TOPORS	C
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	C
TTLL5	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 (CSNBv2; CSNBtypeAv1)

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

Genen: 17

Methode: Pakket capture (DOAv1; DOAtypeAv1)

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
SLC25A46	A
SPG7	C
TIMM8A	A
TMEM126A	A
WFS1	A

NGS Glaucoom
Genen: 26
Methode: Pakket capture (GLAUv1; GLAUtypeAv1)
Kwaliteit: Type A/C **Incl CNV:** Nee

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

NGS Leber congenital amaurosis

Genen: 261

Methode: Pakket capture (BHv6; LCAtypeAv2)

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
AHR	C
AIPL1	A
ALMS1	C
ARHGEF18	C
ARL2BP	C
ARL3	C
ARL6	C
ARSG	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
C8orf37	C
CA4	C
CABP4	A
CACNA1F	C
CACNA2D4	C
CAPN5	C
CC2D2A	C
CDH23	C

GENE	TYPE
FSCN2	C
FZD4	C
GDF6	C
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	C
GUCA1B	C
GUCY2D	A
HARS1	C
HCCS	C
HGSNAT	C
HK1	C
HMX1	C
IDH3A	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

GENE	TYPE
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	C
POC5	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

NGS Leber congenital amaurosis

Genen: 261

Methode: Pakket capture (BHv6; LCAtypeAv2)

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

GENE	TYPE
CDH3	C
CDHR1	C
CEP164	C
CEP19	C
CEP250	C
CEP290	A
CEP41	C
CEP78	C
CERKL	C
CFAP410	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
CPLANE1	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

GENE	TYPE
MAPKAPK3	C
MERTK	C
MFN2	C
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
PAX6	C
PCARE	C
PCDH15	C
PCYT1A	C
PDE6A	C
PDE6B	C
PDE6C	C
PDE6G	C
PDE6H	C

GENE	TYPE
RPGR	C
RPGRIP1	A
RPGRIP1L	C
RS1	C
SAG	C
SCAPER	C
SDCCAG8	C
SEMA4A	C
SIX6	C
SLC24A1	C
SLC7A14	C
SNRNP200	C
SPATA7	A
SPP2	C
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	C
TMEM126A	C
TMEM237	C
TMEM67	C
TOPORS	C
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	C
TTLL5	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 Visus Overig
Genen: 260
Methode: Pakket capture; (BHv6)
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
AHR	C
AIPL1	C
ALMS1	C
ARHGEF18	C
ARL2BP	C
ARL3	C
ARL6	C
ARSG	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
C8orf37	C
CA4	C
CABP4	C
CACNA1F	C
CACNA2D4	C
CAPN5	C
CC2D2A	C
CDH23	C

GENE	TYPE
FSCN2	C
FZD4	C
GDF6	C
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	C
GUCA1B	C
GUCY2D	C
HARS1	C
HCCS	C
HGSNAT	C
HK1	C
HMX1	C
IDH3A	C
IDH3B	C
IFT140	C
IFT172	C
IFT27	C
IFT81	C
IMPDH1	C
IMPG1	C
IMPG2	C
INPP5E	C
INVS	C
IQCB1	C
ITM2B	C
JAG1	C
KCNJ13	C
KCNV2	C
KIAA1549	C
KIF11	C
KIZ	C
KLHL7	C
LCA5	C
LRAT	C
LRIT3	C
LRP5	C
LZTFL1	C
MAK	C

GENE	TYPE
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	C
POC5	C
POMGNT1	C
PRCD	C
PRDM13	C
PROM1	C
PRPF3	C
PRPF31	C
PRPF4	C
PRPF6	C
PRPF8	C
PRPH2	C
PRPS1	C
PXDN	C
RAB28	C
RBP3	C
RBP4	C
RCBTB1	C
RD3	C
RDH11	C
RDH12	C
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	C

NGS Visus Overig
Genen: 260
Methode: Pakket capture; (BHv6)
Kwaliteit: Type A/C **Incl CNV:** Nee

GENE	TYPE
CDH3	C
CDHR1	C
CEP164	C
CEP19	C
CEP250	C
CEP290	C
CEP41	C
CEP78	C
CERKL	C
CFAP410	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
CPLANE1	C
CRB1	C
CRX	C
CSPP1	C
CTNNA1	C
CWC27	C
CYP4V2	C
DHDDS	C
DHX38	C
DRAM2	C
DTHD1	C
EFEMP1	C
ELOVL4	C
EMC1	C
EXOSC2	C
EYS	C
FAM161A	C
FLVCR1	C
FOXE3	C

GENE	TYPE
MAPKAPK	C
MERTK	C
MFN2	C
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	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
PAX6	C
PCARE	C
PCDH15	C
PCYT1A	C
PDE6A	C
PDE6B	C
PDE6C	C
PDE6G	C
PDE6H	C

GENE	TYPE
RPGR	C
RPGRIP1	C
RPGRIP1L	C
RS1	C
SAG	C
SCAPER	C
SDCCAG8	C
SEMA4A	C
SIX6	C
SLC24A1	C
SLC7A14	C
SNRNP200	C
SPATA7	C
SPP2	C
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	C
TMEM126A	C
TMEM237	C
TMEM67	C
TOPORS	C
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	C
TTLL5	C
TUB	C
TULP1	C
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 Retinitis pigmentosa

Genen: 260

Methode: Pakket capture (BHv6; RPtypeAv4)

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
AHR	C
AIPL1	C
ALMS1	C
ARHGEF18	A
ARL2BP	A
ARL3	A
ARL6	A
ARSG	C
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
C8orf37	A
CA4	A
CABP4	C
CACNA1F	C
CACNA2D4	C
CAPN5	C
CC2D2A	C
CDH23	C

GENE	TYPE
FSCN2	A
FZD4	C
GDF6	C
GNAT1	C
GNAT2	C
GNB3	C
GNPTG	C
GPR179	C
GRK1	C
GRM6	C
GUCA1A	C
GUCA1B	A
GUCY2D	C
HARS1	C
HCCS	C
HGSNAT	C
HK1	C
HMX1	C
IDH3A	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

GENE	TYPE
PDZD7	C
PEX1	C
PEX2	C
PEX7	C
PGK1	C
PHYH	C
PITPNM3	C
PLA2G5	C
PNPLA6	C
POC1B	C
POC5	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

NGS Retinitis pigmentosa

Genen: 260

Methode: Pakket capture (BHv6; RPtypeAv4)

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

GENE	TYPE
CDH3	C
CDHR1	C
CEP164	C
CEP19	C
CEP250	C
CEP290	C
CEP41	C
CEP78	C
CERKL	A
CFAP410	C
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
CPLANE1	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

GENE	TYPE
MAPKAPK3	C
MERTK	A
MFN2	C
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
PAX6	C
PCARE	A
PCDH15	C
PCYT1A	C
PDE6A	A
PDE6B	A
PDE6C	C
PDE6G	A
PDE6H	C

GENE	TYPE
RPGR	C
RPGRIP1	C
RPGRIP1L	C
RS1	C
SAG	A
SCAPER	C
SDCCAG8	C
SEMA4A	A
SIX6	C
SLC24A1	C
SLC7A14	A
SNRNP200	A
SPATA7	A
SPP2	A
SRD5A3	C
TEAD1	C
TIMM8A	C
TIMP3	C
TMEM126A	C
TMEM237	C
TMEM67	C
TOPORS	A
TREX1	C
TRIM32	C
TRPM1	C
TSPAN12	C
TTC8	A
TTLL5	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 Hypothyreoidie

Genen: 58 (HTv3)

Methode: Pakket capture

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

THT	Thyreoidale (primaire) hypothyreoïdie
CHT	Centrale hypothyreoïdie
VGS	Verminderde gevoeligheid voor schildklierhormoon
AT	Aafwijkend (plasma) transporteiwit

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

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

NGS Chronisch lymfatische leukemie

Genen: 14

Methode: Pakket capture (CLLv1)

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

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

NGS Cardiomyopthie - Incl Core-NL

Genen: 56

Methode: Pakket capture (CMv18)

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

GENE	TYPE	Opmerking
ACTC1	A	Core-NL
ACTN2	A	Core-NL
ALPK3	A	
ANKRD1	A	Core-NL
BAG3	A	Core-NL
CALR3	A	Core-NL
CAV3	A	Core-NL
CDH2	A	
CRYAB	A	Core-NL
CSRP3	A	Core-NL
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
FHL2	A	
FKRP	A	
FLNC	A	
GLA	A	Core-NL
HCN4	A	
JPH2	A	Core-NL
JUP	A	Core-NL
LAMA4	A	Core-NL
LAMP2	A	Core-NL
LDB3	A	Core-NL
LMNA	A	Core-NL
MIB1	A	Core-NL
MYBPC3	A	Core-NL
MYH6	A	Core-NL
MYH7	A	Core-NL
MYL2	A	Core-NL
MYL3	A	Core-NL
MYLK3	A	
MYOZ2	A	Core-NL
MYPN	A	Core-NL
NEXN	A	Core-NL
PKP2	A	Core-NL
PLN	A	Core-NL
PPA2	A	
PRDM16	A	
PRKAG2	A	Core-NL
RBM20	A	Core-NL

GENE	TYPE	Opmerking
SCN5A	A	Core-NL
TAZ	A	Core-NL
TCAP	A	Core-NL
TMEM43	A	Core-NL
TNNC1	A	Core-NL
TNNI3	A	Core-NL
TNNI3K	A	
TNNT2	A	Core-NL
TPM1	A	Core-NL
TTN	A	
TTR	A	Core-NL
VCL	A	Core-NL

NGS Cowden

Genen: 5

Methode: Pakket capture (COWv3)

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

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

NGS Dyslipidemie
Genen: 29
Methode: Pakket capture (DLv4)
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
GPD1	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

NGS Lymphoedeem

Genen: 50

Methode: Pakket capture (LYMPHv7)

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

Kolom1	Kolom2
A2ML1	A
ABCC9	A
ADAMTS3	A
ALG8	A
BRAF	A
CBL	A
CCBE1	A
CDC42	A
CDK19	A
DCHS1	A
EMILIN1	A
EPHB4	A
FAT4	A
FLT4	A
FOXC2	A
GATA2	A
GJA1	A
GJC2	A
GLA	A
HGF	A
HRAS	A
INPPL1	A
KIF11	A
KIF7	A
KRAS	A
LYVE1	A
LZTR1	A
MAP2K1	A
MAP2K2	A
MET	A
MPI	A
NAGA	A
NRAS	A
PEPD	A
PIEZO1	A
PMM2	A
PTPN11	A
PTPN14	A
RAF1	A
RASA2	A
RELN	A
RIT1	A
RRAS	A
SHANK3	A

GENE	TYPE
SHOC2	A
SOS1	A
SOS2	A
SOX18	A
SPRED1	A
VEGFC	A

NGS LangeQT-syndroom

Genen: 11

Methode: Pakket capture (SCDv8;LQTv2)

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

GENE	TYPE
CACNA1C	A
CALM1	A
CALM2	A
CALM3	A
KCNE1	A
KCNE2	A
KCNH2	A
KCNJ2	A
KCNQ1	A
SCN5A	A
TRDN	A

NGS Metabool
Genen: 67
Methode: Pakket capture (MBSv4)
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
CYP27A1	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
MMAA	A
MMAB	A
MMACHC	A
MMADHC	A
MOCS1	A
MOCS2	A

GENE	TYPE
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: 122
Methode: Pakket capture (NEUROv7; CMTtypeAv5)
Kwaliteit: Type A/C **Incl CNV:** Nee

GENE	TYPE
AARS1	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
COX6A1	C
CTDP1	A
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
ELP1	C
FBLN5	C
FBXO38	C
FGD4	A
FIG4	C
FLVCR1	C
GALC	C
GAN	A
GARS1	C
GDAP1	A
GJB1	A
GJB3	A
GNB4	C
HARS1	C

GENE	TYPE
HINT1	A
HK1	C
HSPB1	A
HSPB3	C
HSPB8	A
IFRD1	C
IGHMBP2	A
INF2	C
KARS1	C
KIF1A	C
KIF1B	C
KIF5A	C
LAS1L	C
LITAF	A
LMNA	A
LRIG3	C
LRSAM1	A
MARS1	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
PEX16	C
PEX7	C
PHYH	C
PLA2G6	C
PLEKHG5	C
PMP2	C
PMP22	A
PRDM12	C
PRPS1	C
PRX	C

GENE	TYPE
RAB7A	A
REEP1	C
RETREG1	C
SBF1	C
SBF2	C
SCN10A	C
SCN11A	C
SCN9A	C
SEPTIN9	C
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
TFG	C
TRIM2	C
TRPV4	A
TUBB3	C
VCP	C
VRK1	C
WNK1	A
YARS1	C
TTR	C

NGS Obesitas
Genen: 16
Methode: Pakket capture (OBSv2)
Kwaliteit: Type A **Incl CNV:** Ja

GENE	TYPE
ALMS1	A
BDNF	A
CPE	A
GNAS	A
LEP	A
LEPR	A
MAGEL2	A
MC3R	A
MC4R	A
PCSK1	A
PHF6	A
POMC	A
SH2B1	A
SIM1	A
VPS13B	A
16p11.2	CNV analyse gehele regio

NGS Polyposis

Genen: 23 / 24

Methode: Pakket capture (PPv6)

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

GENE	TYPE	Kolom1
ACVRL1	A	
APC	A	Core-NL Polyposis
AXIN2	A	
BMPR1A	A	Core-NL Juvenile
CDH1	A	*optioneel*
ENG	A	
EPCAM	A	Core-NL Lynch
GREM1	A	
MLH1	A	Core-NL Lynch
MLH3	A	
MSH2	A	Core-NL Lynch
MSH3	A	
MSH6	A	Core-NL Lynch
MUTYH	A	Core-NL Polyposis
NTHL1	A	
PMS2	A	Core-NL Lynch
POLD1	A	
POLE	A	
PTEN	A	
RNF43	A	
SMAD4	A	Core-NL Juvenile
STK11	A	
TSC1	A	
TSC2	A	

NGS Segmental overgrowth

Genen: 13

Methode: Pakket capture (SOv3)

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

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

NGS Vaatmalformaties

Genen: 30

Methode: Pakket capture (VMv5)

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 (VMLDv1)

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

GENE	TYPE
DCHS1	A
FAT4	A

NGS Epilepsie

Genen: 393 AUMC_Epilepsie_v3
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
AARS1	C
ABAT	C
ABCC8	C
ACTB	C
ACTL6B	C
ACY1	C
ADAM22	C
ADSL	C
ALDH5A1	C
ALDH7A1	C
ALG1	C
ALG11	C
ALG13	C
ALG3	C
ALG6	C
AMACR	C
AMPD2	C
AMT	C
ANKRD11	C
AP3B2	C
ARHGEF9	C
ARID1B	C
ARV1	C
ARX	C
ASAH1	C
ASL	C
ASNS	C
ASXL3	C
ATAD1	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
CACNB4	C
CAD	C
CASK	C

Gene	Type
GLRB	C
GLUD1	C
GNAO1	C
GNB1	C
GOSR2	C
GPC3	C
GPHN	C
GRIA2	C
GRIA3	C
GRIK2	C
GRIN1	C
GRIN2A	C
GRIN2B	C
GRIN2D	C
GRN	C
HACE1	C
HADH	C
HCFC1	C
HCN1	C
HCN2	C
HDAC4	C
HLCS	C
HNRNPU	C
HPRT1	C
HSD17B10	C
HSD17B4	C
HUWE1	C
CILK1	C
IDH2	C
IER3IP1	C
IFIH1	C
INTS8	C
IQSEC2	C
IRF2BPL	C
ITPA	C
JAM3	C
KANSL1	C
KATNB1	C
KCNA1	C
KCNA2	C
KCNB1	C
KCNC1	C
KCND3	C
KCNH1	C

Gene	Type
PHGDH	C
PIGA	C
PIGH	C
PIGN	C
PIGO	C
PIGP	C
PIGT	C
PIGV	C
PLA2G6	C
PLCB1	C
PLP1	C
PLPBP	C
PMM2	C
PNKP	C
PNPO	C
POLG	C
PPP2R1A	C
PPP2R5D	C
PPP3CA	C
PPT1	C
PQBP1	C
PRF1	C
PRICKLE1	C
PRICKLE2	C
PRIMA1	C
PRPS1	C
PRRT2	C
PSAP	C
PSAT1	C
PSPH	C
PUM1	C
PURA	C
PYCR2	C
QARS1	C
RAB39B	C
RAI1	C
RANBP2	C
RARS2	C
RELN	C
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
ROGDI	C
RPS6KA3	C

NGS Epilepsie

Genen: 393 AUMC_Epilepsie_v3
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
CBS	C
CDKL5	C
CERS1	C
CERT1	C
CHD2	C
CHRNA2	C
CHRNA4	C
CHRNA7	C
CHRNB2	C
CIC	C
CLCN4	C
CLDN16	C
CLDN19	C
CLN3	C
CLN5	C
CLN6	C
CLN8	C
CNKSR2	C
CNNM2	C
CNTN2	C
CNTNAP2	C
COA8	C
COL4A1	C
COLGALT1	C
COQ2	C
COQ4	C
COQ8A	C
CPA6	C
CPS1	C
CPT1A	C
CPT2	C
CSNK2B	C
CSTB	C
CTNND2	C
CTSD	C
CTSF	C
CUL4B	C
CUX2	C
D2HGDH	C
DCX	C
DDX3X	C
DENND5A	C
DEPDC5	C
DHDDS	C

Gene	Type
KCNJ10	C
KCNJ11	C
KCNMA1	C
KCNQ2	C
KCNQ3	C
KCNQ5	C
KCNT1	C
KCTD7	C
KDM5C	C
KMT2A	C
KPNA7	C
KPTN	C
LAMB1	C
LGI1	C
LIAS	C
MBD5	C
MDH2	C
MECP2	C
MED12	C
MEF2C	C
MFF	C
MFSD8	C
MLC1	C
MOCS1	C
MOCS2	C
MPDU1	C
MPDZ	C
MTHFR	C
MTOR	C
MTRR	C
NACC1	C
NALCN	C
NANS	C
NAPB	C
NBEA	C
NDUFA1	C
NDUFA11	C
NDUFAF1	C
NDUFAF2	C
NDUFAF3	C
NDUFAF4	C
NDUFAF5	C
NDUFB3	C
NDUFB9	C

Gene	Type
RRM2B	C
SAMHD1	C
SATB2	C
SCARB2	C
SCN1A	C
SCN1B	C
SCN2A	C
SCN3A	C
SCN8A	C
SEPSECS	C
SERPINI1	C
SHANK3	C
SIK1	C
SLC12A5	C
SLC13A5	C
SLC16A1	C
SLC16A2	C
SLC19A3	C
SLC1A2	C
SLC1A3	C
SLC25A1	C
SLC25A15	C
SLC25A22	C
SLC2A1	C
SLC35A2	C
SLC6A1	C
SLC6A5	C
SLC6A8	C
SLC9A6	C
SMARCA2	C
SMC1A	C
SMPD4	C
SMS	C
SNAP25	C
SON	C
SPATA5	C
SPTAN1	C
SRPX2	C
ST3GAL3	C
ST3GAL5	C
STX1B	C
STXBP1	C
SUOX	C
SYN1	C

NGS Epilepsie

Genen: 393 AUMC_Epilepsie_v3
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
DLAT	C
DNAJC5	C
DNM1	C
DNM1L	C
DOCK7	C
DPAGT1	C
DPM1	C
DPM2	C
DPYD	C
DPYS	C
DYNC1H1	C
DYRK1A	C
EEF1A2	C
EFHC1	C
EGF	C
EHMT1	C
EPM2A	C
ETHE1	C
EXOSC3	C
FA2H	C
FARS2	C
FASN	C
FGD1	C
FGF12	C
FLNA	C
FOLR1	C
FOXG1	C
FOXRED1	C
FRMPD4	C
FRRS1L	C
FXYD2	C
GABBR2	C
GABRA1	C
GABRA2	C
GABRA3	C
GABRB2	C
GABRB3	C
GABRE	C
GABRG2	C
GAMT	C
GCK	C
GCSH	C
GLDC	C
GLRA1	C

Gene	Type
NDUFS1	C
NDUFS2	C
NDUFS3	C
NDUFS4	C
NDUFS6	C
NDUFV1	C
NDUFV2	C
NECAP1	C
NEDD4L	C
NEU1	C
NEXMIF	C
NGLY1	C
NHLRC1	C
NPRL2	C
NPRL3	C
NRXN1	C
NSDHL	C
NUBPL	C
OCLN	C
OFD1	C
OPHN1	C
PAFAH1B1	C
PAK3	C
PC	C
PCDH19	C
PDHA1	C
PDHB	C
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
PGAP1	C
PGAP3	C
PHF6	C

Gene	Type
SYNGAP1	C
SYNJ1	C
SYP	C
SZT2	C
TANGO2	C
TBC1D23	C
TBC1D24	C
TBCD	C
TBCE	C
TBCK	C
TCF4	C
TDP2	C
TOE1	C
TPP1	C
TREX1	C
TRIO	C
TRPM6	C
TSC1	C
TSC2	C
TSEN15	C
TSEN2	C
TSEN54	C
TUBA1A	C
TUBB2A	C
TUBB2B	C
TUBB4A	C
TUBG1	C
UBA5	C
UBE2A	C
UBE3A	C
UBTF	C
UGDH	C
UGP2	C
VPS53	C
WDR26	C
WDR45	C
WWOX	C
XK	C
YWHAG	C
ZDHHC9	C
ZEB2	C

NGS Bewegingsstoornissen			v3
Genen:	397		
Methode:	WES met filter		
Kwaliteit:	Type C	**Incl CNV:**	Nee

NB: Het bewegingspanel is een NGS gebaseerde techniek en detecteert daardoor GEEN repeatexpansies en grote deleties of duplicaties. Hierdoor is het met dit panel niet mogelijk om repeatexpansies in de spinocerebellaire ataxie genen te detecteren.

Gene	Type
AAAS	C
AARS2	C
ABCB7	C
ABCD1	C
ABHD12	C
ACTB	C
ADAR	C
ADCY5	C
ADGRG1	C
ADPRS	C
AFG3L2	C
AGTPBP1	C
AIMP1	C
ALDH18A1	C
ALDH3A2	C
ALS2	C
AMPD2	C
ANO10	C
ANO3	C
AP4B1	C
AP4E1	C
AP4M1	C
AP4S1	C
AP5Z1	C
APTX	C
ARG1	C
ARL6IP1	C
ARSA	C
ARX	C
ASPA	C
ATCAY	C
ATG5	C
ATL1	C
ATM	C
ATP13A2	C
ATP1A2	C
ATP1A3	C
ATP2B3	C
ATP6AP2	C
ATP7B	C

Gene	Type
FXN	C
GALC	C
GAN	C
GBA	C
GBA2	C
GBE1	C
GCDH	C
GCH1	C
GDAP2	C
GFAP	C
GJC2	C
GLB1	C
GNAL	C
GOSR2	C
GPR143	C
GRID2	C
GRIN1	C
GRIN2B	C
GRM1	C
HACE1	C
HEXB	C
HK1	C
HPCA	C
HPDL	C
HPRT1	C
HSD17B4	C
HSPD1	C
HTRA2	C
IBA57	C
IFRD1	C
ISCA2	C
ITPR1	C
JAM2	C
JAM3	C
KATNB1	C
KCNA1	C
KCNA2	C
KCNC1	C
KCNC3	C
KCND3	C

Gene	Type
QDPR	C
RAB18	C
RAB3GAP1	C
RAB3GAP2	C
RAD51	C
RARB	C
RARS1	C
RARS2	C
REEP1	C
RETREG1	C
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
RNF170	C
RNF216	C
RTN2	C
RUBCN	C
SACS	C
SAMD9L	C
SAMHD1	C
SCN11A	C
SCN1A	C
SCN2A	C
SCN8A	C
SCYL1	C
SEPSECS	C
SERAC1	C
SETX	C
SGCE	C
SIL1	C
SLC12A6	C
SLC16A2	C
SLC17A5	C
SLC19A3	C
SLC1A3	C
SLC20A2	C
SLC25A15	C
SLC2A1	C
SLC30A10	C
SLC33A1	C

NGS Bewegingsstoornissen			v3
Genen:	397		
Methode:	WES met filter		
Kwaliteit:	Type C	**Incl CNV:**	Nee

NB: Het bewegingspanel is een NGS gebaseerde techniek en detecteert daardoor GEEN repeatexpansies en grote deleties of duplicaties. Hierdoor is het met dit panel niet mogelijk om repeatexpansies in de spinocerebellaire ataxie genen te detecteren.

Gene	Type
ATP8A2	C
AUH	C
B4GALNT1	C
BCAP31	C
BCKDHA	C
BCKDHB	C
BSCL2	C
BTD	C
C12orf65	C
C19orf12	C
CA8	C
CACNA1A	C
CACNA1B	C
CACNA1E	C
CACNA1G	C
CACNB4	C
CAMTA1	C
CAPN1	C
CCDC88C	C
CCT5	C
CHMP1A	C
CHP1	C
CIZ1	C
CLCN2	C
CLCN4	C
CLPB	C
COASY	C
COL4A1	C
COL6A3	C
COQ2	C
COQ4	C
COQ8A	C
COQ9	C
COX20	C
CP	C
CPT1C	C
CSF1R	C
CSTB	C
CTBP1	C
CWF19L1	C

Gene	Type
KCNJ10	C
KCNJ6	C
KCNMA1	C
KCTD17	C
KCTD7	C
KIDINS220	C
KIF1A	C
KIF1C	C
KIF5A	C
KMT2B	C
L1CAM	C
LAMA1	C
LAMB1	C
LMNB1	C
MAG	C
MAPK8IP3	C
MARS2	C
MECP2	C
MECR	C
MFF	C
MICU1	C
MLC1	C
MMADHC	C
MME	C
MRE11	C
MTHFR	C
MTPAP	C
MTTP	C
MVK	C
MYBPC1	C
MYORG	C
NANS	C
NEFL	C
NEU1	C
NEXMIF	C
NFASC	C
NGLY1	C
NIPA1	C
NKX2-1	C
NKX6-2	C

Gene	Type
SLC39A14	C
SLC52A2	C
SLC52A3	C
SLC6A19	C
SLC6A3	C
SLC9A1	C
SMPD1	C
SNCA	C
SNORD118	C
SNX14	C
SOX10	C
SPART	C
SPAST	C
SPG11	C
SPG21	C
SPG7	C
SPR	C
SPTBN2	C
STUB1	C
SUCLA2	C
SUCLG1	C
SUMF1	C
SUOX	C
SURF1	C
SYNE1	C
SYT14	C
TAF1	C
TANGO2	C
TBC1D20	C
TBC1D23	C
TBCD	C
TDP1	C
TDP2	C
TECPR2	C
TENM4	C
TFG	C
TGM6	C
TH	C
THAP1	C
TIMM8A	C

NGS Bewegingsstoornissen			v3
Genen:	397		
Methode:	WES met filter		
Kwaliteit:	Type C	**Incl CNV:**	Nee

NB: Het bewegingspanel is een NGS gebaseerde techniek en detecteert daardoor GEEN repeatexpansies en grote deleties of duplicaties. Hierdoor is het met dit panel niet mogelijk om repeatexpansies in de spinocerebellaire ataxie genen te detecteren.

Gene	Type
CYB5R3	C
CYP27A1	C
CYP2U1	C
CYP7B1	C
DAB1	C
DARS1	C
DARS2	C
DBT	C
DCAF17	C
DCC	C
DCTN1	C
DDC	C
DDHD1	C
DDHD2	C
DHDDS	C
DLAT	C
DLD	C
DNAJC12	C
DNAJC3	C
DNAJC6	C
DNAL4	C
DNMT1	C
DPYS	C
DSTYK	C
ECHS1	C
EEF2	C
EIF2B1	C
EIF2B2	C
EIF2B3	C
EIF2B4	C
EIF2B5	C
EIF4G1	C
ELOVL4	C
ELOVL5	C
ENTPD1	C
ERLIN1	C
ERLIN2	C
ETHE1	C
EXOSC3	C
EXOSC5	C

Gene	Type
NOL3	C
NPC1	C
NPC2	C
NT5C2	C
NUP62	C
OCLN	C
OPA1	C
OPHN1	C
PACS2	C
PANK2	C
PARK7	C
PAX6	C
PCNA	C
PCYT2	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
PLD3	C
PLP1	C
PMM2	C
PMPCA	C
PNKD	C
PNKP	C
PNPLA6	C
POLG	C
POLR1C	C
POLR3A	C
POLR3B	C

Gene	Type
TMEM106E	C
TMEM240	C
TMEM67	C
TOE1	C
TOR1A	C
TPP1	C
TREM2	C
TREX1	C
TRPC3	C
TSEN2	C
TSEN54	C
TTBK2	C
TTC19	C
TTPA	C
TUBA1A	C
TUBB4A	C
TUBG1	C
TWNK	C
TYROBP	C
UBA5	C
UBAP1	C
UBQLN2	C
UCHL1	C
VAMP1	C
VARS2	C
VCP	C
VLDLR	C
VPS11	C
VPS13A	C
VPS13C	C
VPS13D	C
VPS16	C
VPS35	C
VPS37A	C
VPS53	C
VRK1	C
VWA3B	C
WASHC5	C
WDR26	C
WDR45	C

NGS Bewegingsstoornissen v3
Genen: 397
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

NB: Het bewegingspanel is een NGS gebaseerde techniek en detecteert daardoor GEEN repeatexpansies en grote deleties of duplicaties. Hierdoor is het met dit panel niet mogelijk om repeatexpansies in de spinocerebellaire ataxie genen te detecteren.

Gene	Type
FA2H	C
FAM126A	C
FAR1	C
FARS2	C
FAT2	C
FBXO7	C
FGF14	C
FLVCR1	C
FOLR1	C
FOXG1	C
FRMD7	C
FTL	C
FUS	C

Gene	Type
PPP2R2B	C
PREPL	C
PRF1	C
PRICKLE1	C
PRKCG	C
PRKN	C
PRKRA	C
PRRT2	C
PSAP	C
PTRH2	C
PTS	C
PUM1	C
PYCR2	C

Gene	Type
WDR73	C
WDR81	C
WFS1	C
WWOX	C
XK	C
XPR1	C
XRCC1	C
ZC4H2	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
TWNK	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

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
CRPPA	C
ITGA7	C
KBTBD13	C
KCNJ2	C
KLHL40	C
KLHL41	C
KLHL9	C
LAMA2	C
LAMA5	C
LAMP2	C
LARGE1	C
LDB3	C
LDHA	C

Gene	Type
LMNA	C
LMOD3	C
LPIN1	C
MEGF10	C
MICU1	C
MORC2	C
MSTN	C
MTM1	C
UNC13A	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
CAVIN1	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
SELENON	C
SGCA	C
SGCB	C
SGCD	C
SGCG	C
SLC18A2	C
SLC18A3	C
SLC25A1	C
SLC25A4	C
SLC52A2	C
SLC52A3	C
SLC5A7	C
SMCHD1	C
SPEG	C
STIM1	C
SYT2	C
TANGO2	C
TCAP	C
RXYLT1	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
RXYLT1	C
ZC4H2	C

Gene	Type

Gene	Type

NGS Amyotrofe Lateraal Sclerose (ALS)
Genen: 38
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
ALS2	C
ANG	C
ANXA11	C
ATXN2	C
C9orf72	C
CCNF	C
CHCHD10	C
CHMP2B	C
DAO	C
DCTN1	C
ErbB4	C
FIG4	C
FUS	C
GLE1	C
HNRNPA1	C
HNRNPA2B1	C
MAPT	C
MATR3	C
NEFH	C
NEK1	C
OPTN	C
PFN1	C
PRPH	C
PSEN1	C
SETX	C
SIGMAR1	C
SOD1	C
SPG11	C
SQSTM1	C
TAF15	C
TARDBP	C
TBK1	C
TREM2	C
TUBA4A	C
UBQLN2	C
UNC13A	C
VAPB	C
VCP	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
CRPPA	C
ITPR1	C
LARGE1	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
KNL1	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
CRIPT	C
CTNNB1	C
DHCR7	C
DIAPH1	C
DYRK1A	C
EFTUD2	C
EIF2S3	C
FOXG1	C
IER3IP1	C
KATNB1	C
KIFBP	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

Gene	Type
NIPBL	C
ORC1	C
ORC4	C
ORC6	C
PCNT	C
PHC1	C
PLK4	C
PNKP	C
PPP1R15B	C
PQBP1	C
PYCR2	C
QARS1	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: 31

Methode: WES met filter CdLv3

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

Gene	Type
ADNP	C
AFF4	C
ANKRD11	C
ARID1A	C
ARID1B	C
ASXL1	C
ASXL3	C
BRD4	C
CREBBP	C
EP300	C
ESCO2	C
HDAC8	C
KMT2A	C
MAU2	C
NIPBL	C
PACS1	C
PHF6	C
PMM2	C
RAD21	C
ROR2	C
SMARCA2	C
SMARCA4	C
SMARCB1	C
SMARCE1	C
SMC1A	C
SMC3	C
SOX11	C
TAF6	C
TBC1D24	C
UBE2A	C
WNT5A	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
A2ML1	C
AAAS	C
AARS1	C
AASS	C
ABAT	C
ABCC8	C
ABCC9	C
ABCD1	C
ABCD4	C
ABHD5	C
ACAD9	C
ACADS	C
ACAT1	C
ACBD6	C
ACD	C
ACO2	C
ACOX1	C
ACSF3	C
ACSL4	C
ACTB	C
ACTG1	C
ACTL6A	C
ACVR1	C
ACY1	C
ADAM22	C
ADAR	C
ADAT3	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

Gene	Type
FTCD	C
FTO	C
FTSJ1	C
FUCA1	C
FUT8	C
GABBR2	C
GABRA1	C
GABRA3	C
GABRB1	C
GABRB2	C
GABRB3	C
GABRD	C
GABRG2	C
GAD1	C
GALC	C
GALE	C
GALT	C
GAMT	C
GATAD2B	C
GATM	C
GCDH	C
GCH1	C
GCSH	C
GDI1	C
GFAP	C
GFM1	C
GFM2	C
GJA1	C
GJB1	C
GJC2	C
GK	C
GLB1	C
GLDC	C
GLI2	C
GLI3	C
GLIS3	C
GLUD1	C
GLYCTK	C
GM2A	C
GMPPA	C
GMPPB	C
GNAO1	C
GNAS	C
GNB1	C

Gene	Type
POLG	C
POLR1D	C
POLR3A	C
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
PPP2CA	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
PRUNE1	C
PSAP	C
PSAT1	C
PSMD12	C
PSPH	C
PTCH1	C
PTCHD1	C
PTDSS1	C
PTEN	C
PTF1A	C
PTPN11	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
AIMP2	C
AK1	C
AKT3	C
ALDH18A1	C
ALDH3A2	C
ALDH4A1	C
ALDH5A1	C
ALDH7A1	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
APTX	C
ARCN1	C
ARFGEF2	C

Gene	Type
GNB5	C
GNPAT	C
GNPTAB	C
GNPTG	C
GNS	C
GPAA1	C
GPC3	C
GPHN	C
GPSM2	C
GPT2	C
GRIA3	C
GRIA4	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
GTPBP2	C
GTPBP3	C
GUSB	C
HACE1	C
HADH	C
HADHA	C
HAX1	C
HCCS	C
HCFC1	C
HCN1	C
HDAC4	C
HDAC6	C
HDAC8	C
HECTD1	C
HECW2	C
HEPACAM	C
HERC1	C
HERC2	C
HESX1	C

Gene	Type
PTPN4	C
PTRH2	C
PTRHD1	C
PTS	C
PUF60	C
PUM1	C
PURA	C
PUS1	C
PUS3	C
PUS7	C
PYCR1	C
PYCR2	C
QARS1	C
QDPR	C
QRICH1	C
QRSL1	C
RAB11B	C
RAB18	C
RAB23	C
RAB27A	C
RAB39B	C
RAB3GAP1	C
RAB3GAP2	C
RAB40AL	C
RAC1	C
RAD21	C
RAF1	C
RAI1	C
RALA	C
RAP1A	C
RAP1B	C
RARB	C
RARS2	C
RASA2	C
RBBP8	C
RBFOX1	C
RBM10	C
RBM28	C
RBMX	C
RBPJ	C
RBSN	C
RCBTB1	C
RECQL4	C
RELN	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
ARG1	C
ARHGAP31	C
ARHGEF6	C
ARHGEF9	C
ARID1A	C
ARID1B	C
ARID2	C
ARIH1	C
ARL13B	C
ARL6	C
ARMC9	C
ARSA	C
ARSL	C
ARV1	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
ATAD1	C
ATAD3A	C
ATCAY	C
ATIC	C
ATL1	C
ATN1	C
ATP1A1	C
ATP1A2	C
ATP1A3	C
ATP2A2	C
ATP6AP2	C
ATP6V0A2	C
ATP6V1A	C
ATP6V1B2	C
ATP7A	C
ATP8A2	C
ATPAF2	C

Gene	Type
HEXA	C
HEXB	C
HGSNAT	C
HIBCH	C
HIKESHI	C
H1-4	C
H4C3	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
HSD17B4	C
HSPA9	C
HSPD1	C
HTRA2	C
HUWE1	C
HYLS1	C
IARS1	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
CHUK	C
IL1RAPL1	C
IMPA1	C
INPP4A	C

Gene	Type
RERE	C
REV3L	C
RFT1	C
RGS6	C
RHEB	C
RHOBTB2	C
RIPK4	C
RIT1	C
RLIM	C
RMND1	C
RMRP	C
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
RNASET2	C
RNF113A	C
RNF125	C
ROGDI	C
ROR2	C
RORA	C
RPGRIP1L	C
RPL10	C
RPS19	C
RPS6KA3	C
RRM2B	C
RSPRY1	C
RSRC1	C
RTEL1	C
RTN4IP1	C
RTTN	C
RUBCN	C
RUSC2	C
RXYLT1	C
SALL1	C
SAMD9	C
SAMHD1	C
SARS1	C
SASS6	C
SATB2	C
SBDS	C
SC5D	C
SCAPER	C
SCN1A	C
SCN1B	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
ATR	C
ATRX	C
AUH	C
AUTS2	C
AVPR2	C
B3GALNT2	C
B3GALT6	C
B3GLCT	C
B4GALNT1	C
B4GALT1	C
B4GALT7	C
B4GAT1	C
BBS1	C
BBS10	C
BBS12	C
BBS2	C
BBS4	C
BBS5	C
BBS7	C
BBS9	C
BCAP31	C
BCKDHA	C
BCKDHB	C
BCKDK	C
BCL11A	C
BCL11B	C
BCOR	C
BCORL1	C
BCS1L	C
BLM	C
BOLA3	C
BPTF	C
BRAF	C
BRAT1	C
BRD4	C
BRF1	C
BRPF1	C
BRSK2	C
BRWD3	C
BSCL2	C
BSND	C
BTD	C
BUB1B	C
C12orf4	C

Gene	Type
INPP5E	C
INPP5K	C
IQSEC2	C
IRF2BPL	C
IRF6	C
ISCA2	C
CRPPA	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
KDM5B	C
KDM5C	C
KDM6A	C
KDM6B	C
KIAA0586	C
KIAA1109	C
KIDINS220	C

Gene	Type
SCN2A	C
SCN3A	C
SCN8A	C
SCO1	C
SCO2	C
SCYL1	C
SDCCAG8	C
SDHA	C
SELENOI	C
SEMA3E	C
SEPSECS	C
SERAC1	C
SET	C
SETBP1	C
SETD1A	C
SETD1B	C
SETD2	C
SETD5	C
SF1	C
SGPL1	C
SGSH	C
SHANK2	C
SHANK3	C
SHH	C
SHOC2	C
SHROOM4	C
SIK1	C
SIL1	C
SIN3A	C
SIX3	C
SKI	C
SLC12A5	C
SLC12A6	C
SLC13A5	C
SLC16A2	C
SLC17A5	C
SLC19A3	C
SLC1A1	C
SLC1A2	C
SLC1A4	C
SLC25A1	C
SLC25A12	C
SLC25A15	C
SLC25A22	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
C12orf57	C
C12orf65	C
C2CD3	C
CA2	C
CA5A	C
CA8	C
CACNA1A	C
CACNA1C	C
CACNA1D	C
CACNA1E	C
CACNA1G	C
CACNA2D1	C
CACNG2	C
CAD	C
CAMK2A	C
CAMK2B	C
CAMTA1	C
CANT1	C
CAPN10	C
CARS2	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
CCNK	C
CDC42	C
CDC6	C
CDH11	C
CDH15	C
CDK10	C
CDK13	C
CDK16	C
CDK5	C
CDK5RAP2	C
CDK8	C

Gene	Type
KIF11	C
KIF14	C
KIF1A	C
KIFBP	C
KIF2A	C
KIF4A	C
KIF5C	C
KIF7	C
KIRREL3	C
KLF7	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
LAMB1	C
LAMC3	C
LAMP2	C
LARGE1	C
LARP7	C
LAS1L	C
LGI4	C
LIAS	C
LIG4	C
LINGO1	C
LINS1	C
LMAN2L	C
LMNB2	C
LONP1	C
LRP2	C
LRPPRC	C
LZTFL1	C
LZTR1	C

Gene	Type
SLC25A24	C
SLC2A1	C
SLC33A1	C
SLC35A1	C
SLC35A2	C
SLC35A3	C
SLC35C1	C
SLC39A12	C
SLC39A14	C
SLC39A8	C
SLC46A1	C
SLC4A4	C
SLC6A1	C
SLC6A17	C
SLC6A19	C
SLC6A3	C
SLC6A8	C
SLC6A9	C
SLC7A7	C
SLC9A1	C
SLC9A6	C
SLC9A7	C
SMAD4	C
SMAD6	C
SMARCA1	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
SNAP25	C
SNAP29	C
SNIP1	C
SNRPB	C
SNRPN	C
SNX14	C
SOBP	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
CDKL5	C
CDKN1C	C
CDON	C
CENPE	C
CENPF	C
CENPJ	C
CEP104	C
CEP120	C
CEP135	C
CEP152	C
CEP290	C
CEP41	C
CEP57	C
CEP83	C
CEP89	C
CERT1	C
CHAMP1	C
CHD1	C
CHD2	C
CHD3	C
CHD4	C
CHD7	C
CHD8	C
CHKB	C
CHMP1A	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
MAB21L1	C
MAB21L2	C
MACF1	C
MAF	C
MAG	C
MAGEL2	C
MAGT1	C
MAN1B1	C
MAN2B1	C
MANBA	C
MAOA	C
MAP1B	C
MAP2K1	C
MAP2K2	C
MAPK8IP3	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
MED13	C
MED13L	C
MED17	C
MED23	C
MED25	C
MEF2C	C
MEGF8	C
MEIS2	C
METTL23	C
MFF	C
MFSD2A	C
MFSD8	C
MGAT2	C
MGP	C
SON	C
SOS1	C
SOS2	C
SOX10	C
SOX11	C
SOX2	C
SOX3	C
SOX4	C
SOX5	C
SPART	C
SPAST	C
SPATA5	C
SPECC1L	C
SPG11	C
SPOCK1	C
SPR	C
SPRED1	C
SPTAN1	C
SPTBN2	C
SPTBN4	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
STRADA	C
STT3A	C
STT3B	C
STX1B	C
STX3	C
STXBP1	C
STXBP5L	C
STYXL1	C
SUCLA2	C
SUCLG1	C
SUMF1	C
SUOX	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
CNNM2	C
CNOT1	C
CNOT2	C
CNOT3	C
CNPY3	C
CNTNAP2	C
COA8	C
COASY	C
COG1	C
COG2	C
COG4	C
COG5	C
COG6	C
COG7	C
COG8	C
COL13A1	C
COL4A1	C
COL4A2	C
COLEC11	C
COQ2	C
COQ4	C
COQ8A	C
COQ9	C
COX10	C
COX15	C
COX6B1	C
CPE	C
CPLANE1	C
CPLX1	C
CPS1	C
CRADD	C
CRB2	C
CRBN	C
CREBBP	C
CRLF1	C
CSNK2A1	C
CSNK2B	C
CSPP1	C
CSTB	C
CTBP1	C
CTCF	C
CTDP1	C
CTNNA2	C
CTNNB1	C

Gene	Type
MICU1	C
MID1	C
MID2	C
MKKS	C
MKS1	C
MLC1	C
MLYCD	C
MMAA	C
MMAB	C
MMACHC	C
MMADHC	C
MMUT	C
MOCS1	C
MOCS2	C
MOGS	C
MPDU1	C
MPDZ	C
MPLKIP	C
MRPL3	C
MRPS22	C
MSL3	C
MSMO1	C
MTFMT	C
MTHFR	C
MTM1	C
MTOR	C
MTR	C
MTRR	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

Gene	Type
SURF1	C
SUZ12	C
SVBP	C
SYN1	C
SYNCRIP	C
SYNE1	C
SYNGAP1	C
SYNJ1	C
SYP	C
SYT1	C
SYT14	C
SZT2	C
TAF1	C
TAF13	C
TAF2	C
TAF6	C
TANC2	C
TANGO2	C
TAT	C
TBC1D20	C
TBC1D23	C
TBC1D24	C
TBC1D7	C
TBCD	C
TBCE	C
TBCK	C
TBL1XR1	C
TBP	C
TBR1	C
TBX1	C
TCF20	C
TCF4	C
TCF7L2	C
TCN2	C
TCTN2	C
TCTN3	C
TDP2	C
TECPR2	C
TECR	C
TELO2	C
TFAP2A	C
TG	C
TGDS	C
TGFBR1	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
CTNND1	C
CTNND2	C
CTSA	C
CTSD	C
CTTNBP2	C
CUBN	C
CUL4B	C
CUX1	C
CUX2	C
CWC27	C
CWF19L1	C
CXorf56	C
CYB5R3	C
CYP27A1	C
CYP2U1	C
D2HGDH	C
DAG1	C
DARS1	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
DHDDS	C
DHFR	C
DHTKD1	C
DHX30	C
DIAPH1	C
DIP2B	C
DIS3L2	C
DKC1	C

Gene	Type
NBEA	C
NBN	C
NDE1	C
NDP	C
NDST1	C
NDUFA1	C
NDUFA11	C
NDUFA12	C
NDUFA13	C
NDUFA2	C
NDUFAF3	C
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
NECTIN1	C
NEDD4L	C
NEU1	C
NEXMIF	C
NF1	C
NFATC1	C
NFIA	C
NFIB	C
NFIX	C
NFU1	C
NGLY1	C
NHS	C
NIPBL	C
NKX2-1	C
NLGN3	C
NLGN4X	C
NLRP3	C
NONO	C
NOS1	C
NOTCH1	C

Gene	Type
TGFBR2	C
TGIF1	C
TH	C
THOC2	C
THOC6	C
THRB	C
TIMM50	C
TIMM8A	C
TINF2	C
TKT	C
TLK2	C
TMCO1	C
TMEM135	C
TMEM165	C
TMEM216	C
TMEM231	C
TMEM237	C
TMEM240	C
TMEM67	C
TMEM70	C
TMLHE	C
TMTC3	C
TNIK	C
TOE1	C
TP53RK	C
TPI1	C
TPO	C
TPP1	C
TPRKB	C
TRAF7	C
TRAIP	C
TRAPPC11	C
TRAPPC6B	C
TRAPPC9	C
TREX1	C
TRIM32	C
TRIO	C
TRIP12	C
TRIP4	C
TRIT1	C
TRMT1	C
TRMT10A	C
TRMT10C	C
TRNT1	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
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
DPF2	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
EED	C
EEF1A2	C
EFNB2	C
EFTUD2	C
EHMT1	C
EIF2AK3	C
EIF2S3	C
EIF3F	C
EIF4A3	C
EIF4G1	C

Gene	Type
NPC1	C
NPC2	C
NPHP1	C
NR2F1	C
NR4A2	C
NRAS	C
NRXN1	C
NSD1	C
NSD2	C
NSDHL	C
NSUN2	C
NT5C2	C
NTRK1	C
NTRK2	C
NUBPL	C
NUP37	C
NUP62	C
NUS1	C
NXF5	C
OAT	C
OCLN	C
OCRL	C
ODC1	C
OFD1	C
OGT	C
OPHN1	C
ORC1	C
ORC6	C
OSGEP	C
OTC	C
OTUD6B	C
OTX2	C
P4HTM	C
PACS1	C
PACS2	C
PAFAH1B1	C
PAH	C
PAK3	C
PANK2	C
PANX1	C
PARN	C
PARP1	C
PAX1	C
PAX6	C

Gene	Type
TRRAP	C
TSC1	C
TSC2	C
TSEN15	C
TSEN2	C
TSEN54	C
TSFM	C
TSHB	C
TSHR	C
TSPAN7	C
TTC19	C
TTC37	C
TTC8	C
TTI2	C
TUBA1A	C
TUBA8	C
TUBB	C
TUBB2A	C
TUBB2B	C
TUBB3	C
TUBB4A	C
TUBG1	C
TUBGCP4	C
TUBGCP6	C
TUSC3	C
TWIST1	C
TWIST2	C
TWNK	C
UBA5	C
UBE2A	C
UBE3A	C
UBE3B	C
UBR1	C
UBR7	C
UBTF	C
UFC1	C
UFM1	C
UNC13A	C
UNC80	C
UPB1	C
UPF3B	C
UQCRQ	C
UROC1	C
USP27X	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
ELAC2	C
ELOVL4	C
ELOVL5	C
ELP2	C
EMC1	C
EML1	C
EMX2	C
ENTPD1	C
EP300	C
EPB41L1	C
EPG5	C
ERCC1	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
EXOSC9	C
EXT2	C
EXTL3	C
EZH2	C
EZR	C
FA2H	C
FAM126A	C
FAM20C	C
FAR1	C
FARSB	C
FASN	C
FAT4	C
FBN1	C
FBXL3	C
FBXL4	C
FBXO11	C
FBXO31	C
FGD1	C

Gene	Type
PAX8	C
PBX1	C
PC	C
PCCA	C
PCCB	C
PCDH19	C
PCGF2	C
PCLO	C
PCNT	C
PCYT2	C
PDE4D	C
PDHA1	C
PDHX	C
PDP1	C
PDSS1	C
PDSS2	C
PEPD	C
PET100	C
PEX1	C
PEX10	C
PEX11B	C
PEX12	C
PEX13	C
PEX16	C
PEX19	C
PEX2	C
PEX26	C
PEX3	C
PEX5	C
PEX6	C
PEX7	C
PGAP1	C
PGAP2	C
PGAP3	C
PGK1	C
PGM3	C
PHF21A	C
PHF6	C
PHF8	C
PHGDH	C
PHIP	C
PI4KA	C
PIGA	C
PIGC	C

Gene	Type
USP7	C
USP9X	C
VAMP1	C
VAMP2	C
VLDLR	C
VPS11	C
VPS13B	C
VPS37A	C
VPS53	C
VRK1	C
VWA3B	C
WAC	C
WARS2	C
WASF1	C
WASHC4	C
WDPCP	C
WDR13	C
WDR19	C
WDR26	C
WDR4	C
WDR45	C
WDR45B	C
WDR62	C
WDR73	C
WDR81	C
WFS1	C
WWOX	C
XPA	C
XPNPEP3	C
XRCC4	C
XYLT1	C
XYLT2	C
YAP1	C
YME1L1	C
YWHAE	C
YWHAG	C
YY1	C
ZBTB11	C
ZBTB16	C
ZBTB18	C
ZBTB20	C
ZBTB24	C
ZBTB40	C
ZC3H14	C

NGS Intellectual disability

Genen: 1393

Methode: WES met filter IDv12

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

Gene	Type
FGF12	C
FGF14	C
FGFR1	C
FGFR2	C
FGFR3	C
FH	C
FIBP	C
FIGN	C
FKRP	C
FKTN	C
FLNA	C
FLVCR1	C
FLVCR2	C
FMN2	C
FMR1	C
FOLR1	C
FOXG1	C
FOXP1	C
FOXP2	C
FOXRED1	C
FRAS1	C
FREM2	C
FRMD4A	C
FRMPD4	C
FRRS1L	C

Gene	Type
PIGG	C
PIGL	C
PIGN	C
PIGO	C
PIGT	C
PIGV	C
PIGW	C
PIGY	C
PIK3CA	C
PIK3R2	C
PLA2G6	C
PLAA	C
PLCB1	C
PLK4	C
PLP1	C
PLPBP	C
PLXND1	C
PMM2	C
PMPCA	C
PMPCB	C
PNKP	C
PNP	C
PNPLA6	C
POC1A	C
POGZ	C

Gene	Type
ZC4H2	C
ZCCHC12	C
ZCCHC8	C
ZDHHC15	C
ZDHHC9	C
ZEB2	C
ZFYVE26	C
ZIC1	C
ZIC2	C
ZMIZ1	C
ZMYM3	C
ZMYND11	C
ZNF148	C
ZNF292	C
ZNF407	C
ZNF41	C
ZNF462	C
ZNF526	C
ZNF592	C
ZNF674	C
ZNF711	C
ZNF81	C
ZSWIM6	C

NGS Neonatale Diabetes-Mellitus
Genen: 21
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
ABCC8	C
EIF2AK3	C
FOXP3	C
GATA4	C
GATA6	C
GCK	C
GLIS3	C
HNF1B	C
IER3IP1	C
INS	C
KCNJ11	C
MNX1	C
NEUROD1	C
NEUROG3	C
NKX2-2	C
PDX1	C
PTF1A	C
RFX6	C
SLC19A2	C
SLC2A2	C
ZFP57	C

NGS Hyperinsulinisme
Genen: 15
Methode: WES met filter
Kwaliteit: Type C **Incl CNV:** Nee

Gene	Type
ABCC8	C
CACNA1D	C
FOXA2	C
GCK	C
GLUD1	C
HADH	C
HK1	C
HNF1A	C
HNF4A	C
INS	C
INSR	C
KCNJ11	C
PGM1	C
SLC16A1	C
UCP2	C

NGS Primaire immunodeficientie (PID)

Genen: 431

Methode: WES met filter v9

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

Gene	Type
ACD	C
ACP5	C
ACTB	C
ADA	C
ADA2	C
ADAM17	C
ADAR	C
AGA	C
AICDA	C
AIRE	C
AK2	C
ALG13	C
ALPI	C
ANGPT1	C
AP1S3	C
AP3B1	C
AP3D1	C
APOL1	C
ARHGEF1	C
ARPC1B	C
ATM	C
ATP6AP1	C
B2M	C
BACH2	C
BCL10	C
BCL11B	C
BLK	C
BLM	C
BLNK	C
BLOC1S6	C
BTK	C
C1QA	C
C1QB	C
C1QC	C
C1R	C
C1S	C
C2	C
C3	C
C5	C
C6	C
C7	C
C8A	C
C8B	C
C8G	C

Gene	Type
G6PC3	C
G6PD	C
GATA2	C
GFI1	C
GINS1	C
GJC2	C
GRHL2	C
GTF2H5	C
HAVCR2	C
HAX1	C
HELLS	C
HMOX1	C
HYOU1	C
ICOS	C
ICOSLG	C
IFIH1	C
IFNAR1	C
IFNAR2	C
IFNG	C
IFNGR1	C
IFNGR2	C
IGHM	C
IGLL1	C
IKBKB	C
IKBKG	C
IKZF1	C
IL10	C
IL10RA	C
IL10RB	C
IL12B	C
IL12RB1	C
IL17F	C
IL17RA	C
IL17RC	C
IL18BP	C
IL1RN	C
IL2	C
IL21	C
IL21R	C
IL2RA	C
IL2RB	C
IL2RG	C
IL36RN	C
IL6R	C

Gene	Type
PRF1	C
PRKCD	C
PRKDC	C
PRPS1	C
PSENEN	C
PSMA3	C
PSMB4	C
PSMB8	C
PSMB9	C
PSMG2	C
PSTPIP1	C
PTPN22	C
PTPRC	C
RAB27A	C
RAC2	C
RAG1	C
RAG2	C
RANBP2	C
RASGRP1	C
RASGRP2	C
RBCK1	C
RC3H1	C
RECQL4	C
RELB	C
RFX5	C
RFXANK	C
RFXAP	C
RHOH	C
RIPK1	C
RMRP	C
RNASEH2A	C
RNASEH2B	C
RNASEH2C	C
RNF168	C
RNF31	C
RNU4ATAC	C
RORC	C
RPSA	C
RSPH9	C
RTEL1	C
SAMD9	C
SAMD9L	C
SAMHD1	C
SBDS	C

NGS Primaire immunodeficientie (PID)

Genen: 431

Methode: WES met filter v9

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

Gene	Type
C9	C
CA2	C
CARD11	C
CARD14	C
CARD9	C
CARMIL2	C
CASP10	C
CASP8	C
CAVIN1	C
CCBE1	C
CD19	C
CD247	C
CD27	C
CD3D	C
CD3E	C
CD3G	C
CD40	C
CD40LG	C
CD46	C
CD55	C
CD59	C
CD70	C
CD79A	C
CD79B	C
CD81	C
CD8A	C
CDCA7	C
CDKN2B	C
CEBPE	C
CFB	C
CFD	C
CFH	C
CFHR1	C
CFHR3	C
CFHR5	C
CFI	C
CFP	C
CFTR	C
CHD7	C
CIB1	C
CIITA	C
CLCN7	C
CLEC4D	C
CLEC7A	C

Gene	Type
IL6ST	C
IL7R	C
INO80	C
INSR	C
IRAK1	C
IRAK4	C
IRF2BP2	C
IRF3	C
IRF4	C
IRF7	C
IRF8	C
IRF9	C
ISG15	C
ITCH	C
ITGB2	C
ITK	C
IVNS1ABP	C
JAGN1	C
JAK1	C
JAK2	C
JAK3	C
KDM6A	C
KMT2D	C
KRAS	C
LACC1	C
LAMTOR2	C
LAT	C
LCK	C
LIG1	C
LIG4	C
LPIN2	C
LRBA	C
LRRC8A	C
LTBP3	C
LYST	C
MAGT1	C
MAL	C
MALT1	C
MAN2B1	C
MANBA	C
MAP3K14	C
MASP2	C
MBL2	C
MC2R	C

Gene	Type
SEC61A1	C
SEMA3E	C
SERAC1	C
SERPING1	C
SH2B3	C
SH2D1A	C
SH3BP2	C
SH3KBP1	C
SKIV2L	C
SLC29A3	C
SLC35A1	C
SLC35C1	C
SLC37A4	C
SLC39A4	C
SLC39A7	C
SLC46A1	C
SLC7A7	C
SMARCAL1	C
SMARCD2	C
SNX10	C
SOCS1	C
SOCS4	C
SP110	C
SPINK5	C
SPPL2A	C
SRP54	C
SRP72	C
STAT1	C
STAT2	C
STAT3	C
STAT4	C
STAT5B	C
STAT6	C
STIM1	C
STING1	C
STK4	C
STN1	C
STX11	C
STXBP2	C
TAP1	C
TAP2	C
TAPBP	C
TAZ	C
TBX1	C

NGS Primaire immunodeficientie (PID)

Genen: 431

Methode: WES met filter v9

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

Gene	Type
CLPB	C
COPA	C
CORO1A	C
CR2	C
CREBBP	C
CSF2RA	C
CSF2RB	C
CSF3R	C
CTC1	C
CTLA4	C
CTPS1	C
CTSC	C
CXCR4	C
CYBA	C
CYBB	C
CYBC1	C
DBR1	C
DCLRE1B	C
DCLRE1C	C
DDX58	C
DEF6	C
DGAT1	C
DHFR	C
DKC1	C
DNAJC21	C
DNASE1	C
DNASE1L3	C
DNASE2	C
DNMT3B	C
DOCK2	C
DOCK8	C
ELANE	C
ELF4	C
EPG5	C
ERCC2	C
ERCC3	C
ERCC6L2	C
EXTL3	C
F12	C
FAAP24	C
FADD	C
FAS	C
FASLG	C
FAT4	C

Gene	Type
MCM4	C
MEFV	C
MOGS	C
MRE11	C
MRTFA	C
MS4A1	C
MSN	C
MTHFD1	C
MVK	C
MYD88	C
MYSM1	C
NBAS	C
NBN	C
NCF1	C
NCF2	C
NCF4	C
NCKAP1L	C
NCSTN	C
NFAT5	C
NFE2L2	C
NFKB1	C
NFKB2	C
NFKBIA	C
NHEJ1	C
NHP2	C
NLRC4	C
NLRP1	C
NLRP12	C
NLRP3	C
NOD2	C
NOP10	C
NRAS	C
NSMCE3	C
OAS1	C
ORAI1	C
OSTM1	C
OTULIN	C
PARN	C
PAX5	C
PBX1	C
PCCA	C
PCCB	C
PEPD	C
PGM3	C

Gene	Type
TCF3	C
TCIRG1	C
TCN2	C
TERC	C
TERT	C
TET2	C
TFRC	C
TGFB1	C
THBD	C
TICAM1	C
TINF2	C
TIRAP	C
TLR3	C
TLR4	C
TLR7	C
TLR8	C
TMC6	C
TMC8	C
TNFAIP3	C
TNFRSF11A	C
TNFRSF13B	C
TNFRSF13C	C
TNFRSF1A	C
TNFRSF4	C
TNFRSF9	C
TNFSF11	C
TNFSF12	C
TOP2B	C
TPP2	C
TRAC	C
TRAF3	C
TRAF3IP2	C
TREX1	C
TRIM22	C
TRNT1	C
TTC37	C
TTC7A	C
TYK2	C
UBA1	C
UNC13D	C
UNC93B1	C
UNG	C
USB1	C
USP18	C

NGS Primaire immunodeficientie (PID)

Genen: 431

Methode: WES met filter v9

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

Gene	Type
FCGR1A	C
FCGR2A	C
FCGR2B	C
FCGR3A	C
FCGR3B	C
FCHO1	C
FCN3	C
FERMT3	C
FOXN1	C
FOXP3	C
FPR1	C
G6PC	C

Gene	Type
PIGA	C
PIK3CD	C
PIK3R1	C
PLCG2	C
PLEKHM1	C
PLG	C
PMM2	C
PNP	C
POLA1	C
POLE2	C
POMP	C
POT1	C

Gene	Type
VAV1	C
VPS13B	C
VPS45	C
WAS	C
WDR1	C
WIPF1	C
WRAP53	C
XIAP	C
ZAP70	C
ZBTB24	C
ZNF341	C