

Nierziekten

Renale cysten en/of ciliopathieën (incl. Bardet-Biedl syndroom, nefronoftise en Joubert syndroom). NB: MUC1 VNTR Cytosine insertie doorgaans niet aantoonbaar.

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NEF17v23.2

Gennaam CreV4 (15X) Ssv7 (15X)

CEP55	100	99.9
TCTN3	100	100
PAX2	100	100
SUFU	100	100
ARL3	100	91.9
BBIP1	100	100
CDKN1C	86.4	82.6
TMEM138	100	100
TMEM216	100	100
GANAB	100	100
BBS1	100	100
LRP5	99.5	97.8
DHCR7	100	100
C2CD3	100	100
ALG8	100	96.4
DYNC2H1	100	98.1
ALG9	100	99.7
CEP164	100	100
TMEM218	100	100
HYLS1	100	100
TULP3	100	100
BBS10	100	100
CEP290	100	84.8
POC1B	100	99.1
CEP83	100	89.7
IFT81	100	99.3
TCTN1	100	97.3
TCTN2	100	100
ALG5	100	100
PIBF1	100	98.7
CLDN10	100	100
COL4A1	100	99.6
RPGRIP1	100	100
FAM179B	100	99.8
KIAA0586	95.4	99.8
IFT43	100	100
TTC8	100	96
DICER1	100	100
FAN1	100	100
MAPKBP1	100	100
BBS4	100	100
KIF7	99.8	97.3
IFT140	100	100

Gennaam CreV4 (15X) SSv7 (15X)

TSC2	100	100
PKD1	96.1	96.9
GLIS2	100	100
ANKS3	100	99.5
PMM2	100	100
UMOD	100	100
KIAA0556	100	100
ZNF423	100	100
SALL1	100	100
RPGRIP1L	96.4	96.9
BBS2	100	100
TMEM231	100	100
KIAA0753	100	100
TMEM107	100	100
B9D1	100	100
NEK8	100	100
HNF1B	100	100
SLC4A1	100	100
MKS1	100	100
PRKCSH	100	100
B9D2	100	100
CCDC114	100	100
MKKS	100	100
JAG1	100	100
KIF3B	100	100
IFT52	100	100
IFT27	100	100
CBY1	100	100
XPNPEP3	100	100
ATXN10	100	100
OFD1	100	99.1
COL4A5	100	99.7
GPC3	100	100
MAP7D3	99.8	94.9
ZIC3	100	100
CEP104	100	100
NPHP4	100	100
RERE	100	99.9
CCDC28B	100	100
CPT2	100	99.7
ALG6	100	98.9
NOTCH2	100	99.5
MUC1	96.2	96.2
PBX1	100	100
CDC73	100	100
KIF14	100	97.7
DDX59	100	100
REN	100	100
SLC41A1	100	100
CENPF	100	100

Gennaam CreV4 (15X) Ssv7 (15X)

EXOC8	100	100
SDCCAG8	100	100
WDR35	100	99
IFT172	100	100
DYNC2LI1	100	100
WDPCP	100	98.8
ATP6V1B1	100	100
ALMS1	100	99.9
NPHP1	100	96.6
TTC21B	100	100
BBS5	100	100
TMEM237	100	100
COL4A4	99.5	99.8
COL4A3	100	98.7
ARMC9	100	100
PDE6D	100	100
TRAF3IP1	100	99.9
AGXT	100	100
VHL	100	100
LZTFL1	100	93.4
ADAMTS9	99.8	99
ARL13B	100	100
ARL6	100	100
IFT57	99.8	99.8
IQCB1	100	100
SEC61A1	100	100
IFT122	100	100
NPHP3	100	98.4
DZIP1L	100	99.9
IFT80	100	99.2
DNAJB11	100	100
TCTEX1D2	100	100
EVC2	100	100
EVC	99	95.4
CC2D2A	100	99.8
WDR19	100	99.9
PKD2	100	100
BBS7	100	99.5
BBS12	100	100
INTU	100	100
SCLT1	100	95.7
NEK1	100	98
C5orf42	100	99
CEP120	100	100
TXNDC15	100	100
DCDC2	100	99.8
PKHD1	100	100
ICK	100	100
TBX18	100	99.9
SEC63	100	100

Gennaam CreV4 (15X) SSv7 (15X)

AHI1	100	100
RMND1	100	97.7
BBS9	100	99.5
CEP41	100	100
ATP6V0A4	100	100
NCAPG2	100	99.6
WDR60	100	99.6
PCM1	100	99.8
CSPP1	100	98.3
TMEM67	100	99.6
C8orf37	100	100
GLIS3	100	100
IFT74	100	95.7
ANKS6	98.1	96.7
SEC61B	99.9	100
INVS	100	100
TRIM32	100	100
CRB2	100	100
WDR34	100	99.7
TSC1	100	100
INPP5E	100	100
TMEM72	100	100
FAM149B1	100	100
DLG5	100	98.5