

V1, Stand: April 2023

## Genreferenzliste NGS-Genpanel TruSight Cardio

Gen	OMIM	LRG Transkript ID	LRG Protein ID	HGNC-ID
ABCC9	<a href="#">601439</a>	NM_020297.2	LRG_377p1	60
ABCG5	<a href="#">605459</a>	NM_022436.3	LRG_1181p1	13886
ABCG8	<a href="#">605460</a>	NM_022437.3	LRG_1182p1	13887
ACTA1	<a href="#">102610</a>	NM_001100.3	LRG_429p1	129
ACTA2	<a href="#">102620</a>	NM_001613.2	LRG_781p1	130
ACTC1	<a href="#">102540</a>	NM_005159.4	LRG_388p1	143
ACTN2	<a href="#">102573</a>	NM_001103.3	LRG_436p1	164
AKAP9	<a href="#">604001</a>	NM_005751.4	LRG_331p1	379
ALMS1	<a href="#">606844</a>	NM_001378454.1	LRG_741p1	428
ANK2	<a href="#">106410</a>	NM_001148.4	LRG_327p1	493
ANKRD1	<a href="#">609599</a>	NM_014391.2	LRG_379p1	15819
APOA4	<a href="#">107690</a>	NM_000482.4	-	602
APOA5	<a href="#">606368</a>	NM_052968.5	-	17288
APOB	<a href="#">107730</a>	NM_000384.2	-	603
APOC2	<a href="#">608083</a>	NM_000483.5	-	609
APOE	<a href="#">107741</a>	NM_000041.3	-	613
BAG3	<a href="#">603883</a>	NM_004281.4	LRG_742p1	939
BRAF	<a href="#">619589</a>	NM_004333.6	LRG_299p1	1097
CACNA1C	<a href="#">114205</a>	NM_000719.6	LRG_334p1	1390
CACNA2D1	<a href="#">114204</a>	NM_000722.3	LRG_437p1	1399
CACNB2	<a href="#">600003</a>	NM_201590.2	LRG_381p2	1402
CALM1	<a href="#">114180</a>	NM_006888.4	-	1442
CALR3	<a href="#">611414</a>	NM_145046.4	LRG_422p1	20407
CASQ2	<a href="#">114251</a>	NM_001232.3	LRG_404p1	1513
CAV3	<a href="#">601253</a>	NM_001234.4	LRG_329p1	1529
CBL	<a href="#">165360</a>	NM_005188.4	LRG_608p1	1541
CBS	<a href="#">613381</a>	NM_000071.2	LRG_777p1	1550
CETP	<a href="#">118470</a>	NM_000078.3	-	1869

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COL3A1	<a href="#">120180</a>	NM_000090.4	LRG_3p1	2201
COL5A1	<a href="#">120215</a>	NM_000093.4	LRG_737p1	2209
COL5A2	<a href="#">120190</a>	NM_000393.4	LRG_738p1	2210
COX15	<a href="#">603646</a>	NM_004376.7	LRG_406p2	2263
CRELD1	<a href="#">607170</a>	NM_001031717.4	-	14630
CRYAB	<a href="#">123590</a>	NM_001885.3	LRG_407p1	2389
CSRP3	<a href="#">600824</a>	NM_003476.4	LRG_440p1	2472
DES	<a href="#">125660</a>	NM_001927.3	LRG_380p1	2770
DMD	<a href="#">300377</a>	NM_004006.2	LRG_199p1	2928
DNAJC19	<a href="#">608977</a>	NM_145261.4	LRG_743p1	30528
DOLK	<a href="#">610746</a>	NM_014908.4	LRG_744p1	23406
DPP6	<a href="#">126141</a>	NM_130797.4	-	3010
DSC2	<a href="#">125645</a>	NM_024422.3	LRG_400p2	3036
DSG2	<a href="#">125671</a>	NM_001943.3	LRG_397p1	3049
DSP	<a href="#">125647</a>	NM_004415.2	LRG_423p1	3052
DTNA	<a href="#">601239</a>	NM_001390.4	LRG_756p1	3057
EFEMP2	<a href="#">604633</a>	NM_016938.5	-	3219
ELN	<a href="#">130160</a>	NM_001278939.2	-	3327
EMD	<a href="#">300384</a>	NM_000117.2	LRG_745p1	3331
EYA4	<a href="#">603550</a>	NM_004100.5	LRG_418p1	3522
FBN1	<a href="#">134797</a>	NM_000138.4	LRG_778p1	3603
FBN2	<a href="#">612570</a>	NM_001999.4	-	3604
FHL1	<a href="#">300163</a>	NM_001159702.3	LRG_739p2	3702
FHL2	<a href="#">602633</a>	NM_001450.3	LRG_740p1	3703
FKRP	<a href="#">606596</a>	NM_02301.5	LRG_761p1	17997
FKTN	<a href="#">607440</a>	NM_001079802.2	LRG_434p1	3622
FXN	<a href="#">606829</a>	NM_000144.5	LRG_339p1	3951
GAA	<a href="#">606800</a>	NM_000152.3	LRG_673p1	4065
GATAD1	<a href="#">614518</a>	NM_021167.5	LRG_746p1	29941
GCKR	<a href="#">600842</a>	NM_001486.4	-	4196
GJA5	<a href="#">121013</a>	NM_005266.6	-	4279
GLA	<a href="#">300644</a>	NM_000169.2	LRG_672p1	4296
GPD1L	<a href="#">611778</a>	NM_015141.3	LRG_419p1	28956
GPIHBP1	<a href="#">612757</a>	NM_178172.6	-	24945
HADHA	<a href="#">600890</a>	NM_000182.5	LRG_747p1	4802
HCN4	<a href="#">605206</a>	NM_005477.2	-	16882
HFE	<a href="#">613609</a>	NM_000410.3	LRG_748p1	4886
HRAS	<a href="#">190020</a>	NM_005343.4	LRG_506p1	5173
HSPB8	<a href="#">608014</a>	NM_014365.3	LRG_249p1	30171
ILK	<a href="#">602366</a>	NM_004517.4	LRG_444p1	6040

Gen	OMIM	LRG Transkript ID	LRG Protein ID	HGNC-ID
JAG1	<a href="#">601920</a>	NM_000214.2	LRG_1191p1	6188
JPH2	<a href="#">605267</a>	NM_020433.4	LRG_394p1	14202
JUP	<a href="#">173325</a>	NM_002230.2	LRG_401p2	6207
KCNA5	<a href="#">176267</a>	NM_002234.3	-	6224
KCND3	<a href="#">605411</a>	NM_004980.4	LRG_445p1	6239
KCNE1	<a href="#">176261</a>	NM_000219.5	LRG_290p1	6240
KCNE2	<a href="#">603796</a>	NM_172201.1	LRG_291p1	6242
KCNE3	<a href="#">604433</a>	NM_005472.4	LRG_439p1	6243
KCNE5	<a href="#">300328</a>	NM_012282.2	LRG_1083p1	6241
KCNH2	<a href="#">152427</a>	NM_000238.3	LRG_288p1	6251
KCNJ2	<a href="#">600681</a>	NM_000891.2	LRG_328p1	6263
KCNJ3	<a href="#">601534</a>	NM_002239.3	-	6264
KCNJ5	<a href="#">600734</a>	NM_000890.3	LRG_333p1	6266
KCNJ8	<a href="#">600935</a>	NM_004982.3	-	6269
KCNQ1	<a href="#">607542</a>	NM_000218.2	LRG_287p1	6294
KLF10	<a href="#">601878</a>	NM_005655.4	LRG_758p1	11810
KRAS	<a href="#">190070</a>	NM_033360.4	LRG_344p2	6407
LAMA2	<a href="#">156225</a>	NM_000426.3	LRG_409p1	6482
LAMA4	<a href="#">600133</a>	NM_001105206.3	LRG_433p2	6484
LAMP2	<a href="#">309060</a>	NM_002294.2	LRG_749p1	6501
LDB3	<a href="#">605906</a>	NM_007078.2	LRG_385p1	15710
LDLR	<a href="#">606945</a>	NM_000527.4	LRG_274p1	6547
LDLRAP1	<a href="#">605747</a>	NM_015627.3	LRG_276p1	18640
LMF1	<a href="#">611761</a>	NM_022773.4	-	14154
LMNA	<a href="#">150330</a>	NM_170707.3	LRG_254p2	6636
LPL	<a href="#">609708</a>	NM_000237.3	LRG_1298p1	6677
LTBP2	<a href="#">602091</a>	NM_000428.3	-	6715
MAP2K1	<a href="#">176872</a>	NM_002755.4	LRG_725p1	6840
MAP2K2	<a href="#">601263</a>	NM_030662.4	LRG_750p1	6842
MIB1	<a href="#">608677</a>	NM_020774.3	LRG_759p1	21086
MURC	<a href="#">617714</a>	NM_001018116.2	LRG_760p1	33742
MYBPC3	<a href="#">600958</a>	NM_000256.3	LRG_386p1	7551
MYH11	<a href="#">160745</a>	NM_001040114.1	LRG_1401p2	7569
MYH6	<a href="#">160710</a>	NM_002471.3	LRG_389p1	7576
MYH7	<a href="#">160760</a>	NM_000257.3	LRG_384p1	7577
MYL2	<a href="#">160781</a>	NM_000432.3	LRG_393p1	7583
MYL3	<a href="#">160790</a>	NM_000258.2	LRG_395p1	7584
MYLK	<a href="#">600922</a>	NM_053025.4	-	7590
MYLK2	<a href="#">606566</a>	NM_033118.3	LRG_392p1	16243
MYO6	<a href="#">600970</a>	NM_004999.4	LRG_438p1	7605

Gen	OMIM	LRG Transkript ID	LRG Protein ID	HGNC-ID
MYOZ2	<a href="#">605602</a>	NM_016599.4	LRG_396p1	1330
MYPN	<a href="#">608517</a>	NM_032578.3	LRG_410p1	23246
NEXN	<a href="#">613121</a>	NM_144573.3	LRG_442p1	29557
NKX2.5	<a href="#">600584</a>	NM_004387.4	LRG_671p1	2488
NODAL	<a href="#">601265</a>	NM_018055.5	-	7865
NOTCH1	<a href="#">190198</a>	NM_017617.3	LRG_1122p1	7881
NPPA	<a href="#">108780</a>	NM_006172.4	LRG_751p1	7939
NRAS	<a href="#">164790</a>	NM_002524.4	LRG_92p1	7989
PCSK9	<a href="#">607786</a>	NM_174936.4	LRG_275p1	20001
PDLIM3	<a href="#">605889</a>	NM_014476.5	LRG_752p1	20767
PKP2	<a href="#">602861</a>	NM_004572.3	LRG_398p1	9024
PLN	<a href="#">172405</a>	NM_002667.3	LRG_390p1	9080
PRDM16	<a href="#">605557</a>	NM_022114.3	-	14000
PRKAG2	<a href="#">602743</a>	NM_016203.3	LRG_430p1	9386
PRKAR1A	<a href="#">188830</a>	NM_002734.5	LRG_514p1	9388
PTPN11	<a href="#">176876</a>	NM_002834.3	LRG_614p1	9644
RAF1	<a href="#">164760</a>	NM_001354689.3	LRG_413p2	9829
RANGRF	<a href="#">607954</a>	NM_016492.4	-	17679
RBM20	<a href="#">613171</a>	NM_001134363.2	LRG_382p1	27424
RYR1	<a href="#">180901</a>	NM_000540.3	LRG_766p1	10483
RYR2	<a href="#">180902</a>	NM_001035.2	LRG_402p1	10484
SALL4	<a href="#">607343</a>	NM_020436.5	LRG_675p1	15924
SCN1B	<a href="#">600235</a>	NM_001037.4	LRG_420p1	10586
SCN2B	<a href="#">601327</a>	NM_004588.4	-	10589
SCN3B	<a href="#">608214</a>	NM_018400.3	LRG_421p1	20665
SCN4B	<a href="#">608256</a>	NM_174934.3	LRG_330p1	10592
SCN5A	<a href="#">600163</a>	NM_198056.2	LRG_289p1	10593
SCO2	<a href="#">604272</a>	NM_005138.3	-	10604
SDHA	<a href="#">600857</a>	NM_004168.3	LRG_315p1	10680
SEPN1	<a href="#">602771</a>	NM_206926.2	LRG_857p1	15999
SGCB	<a href="#">600900</a>	NM_000232.5	LRG_204p1	10806
SGCD	<a href="#">601411</a>	NM_000337.5	LRG_205p1	10807
SGCG	<a href="#">608896</a>	NM_000231.3	LRG_207p1	10809
SHOC2	<a href="#">602775</a>	NM_007373.4	LRG_753p1	15454
SLC25A4	<a href="#">103220</a>	NM_001151.4	LRG_441p1	10990
SLC2A10	<a href="#">606145</a>	NM_030777.3	-	13444
SMAD3	<a href="#">603109</a>	NM_005902.4	-	6769
SMAD4	<a href="#">600993</a>	NM_005359.6	LRG_318p1	6770
SNTA1	<a href="#">601017</a>	NM_003098.2	LRG_332p1	11167
SOS1	<a href="#">182530</a>	NM_005633.3	LRG_754p1	11187

Gen	OMIM	LRG Transkript ID	LRG Protein ID	HGNC-ID
SREBF2	<a href="#">600481</a>	NM_004599.4	-	11290
TAZ	<a href="#">300394</a>	NM_000116.4	LRG_131p1	11577
TBX20	<a href="#">606061</a>	NM_001077653.2	LRG_755p1	11598
TBX3	<a href="#">601621</a>	NM_016569.4	-	11602
TBX5	<a href="#">601620</a>	NM_000192.3	LRG_670p1	11604
TCAP	<a href="#">604488</a>	NM_003673.3	LRG_210p1	11610
TGFB2	<a href="#">190220</a>	NM_001135599.3	-	11768
TGFB3	<a href="#">190230</a>	NM_003239.2	LRG_399p1	11769
TGFBR1	<a href="#">190181</a>	NM_004612.2	-	11772
TGFBR2	<a href="#">190182</a>	NM_001024847.2	LRG_779p1	11773
TMEM43	<a href="#">612048</a>	NM_024334.2	LRG_435p1	28472
TMPO	<a href="#">188380</a>	NM_003276.2	LRG_443p2	11875
TNNC1	<a href="#">191040</a>	NM_003280.2	LRG_378p1	11943
TNNI3	<a href="#">191044</a>	NM_000363.4	LRG_432p1	11947
TNNT2	<a href="#">191045</a>	NM_001001430.2	LRG_431p1	11949
TPM1	<a href="#">191010</a>	NM_001018005.1	LRG_387p1	12010
TRDN	<a href="#">603283</a>	NM_006073.3	-	12261
TRIM63	<a href="#">606131</a>	NM_032588.4	LRG_757p1	16007
TRPM4	<a href="#">606936</a>	NM_017636.3	-	17993
TTN	<a href="#">188840</a>	NM_001256850.1	LRG_391p1	12403
TTR	<a href="#">176300</a>	NM_000371.3	LRG_416p1	12405
TXNRD2	<a href="#">606448</a>	NM_006440.4	LRG_417p1	18155
VCL	<a href="#">193065</a>	NM_014000.2	LRG_383p1	12665
ZBTB17	<a href="#">604084</a>	NM_003443.2	-	12936
ZHX3	<a href="#">609598</a>	NM_015035.4	-	15935
ZIC3	<a href="#">300265</a>	NM_003413.4	-	12874