Inherited Disease Panel
Gene panel CHA* version 1, 18-2-2018

Leids Universitair Medisch Centrum

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
ABCD1	NM_000033	300371	105	100	100	99	98
*APP***	NM_000484	104760	137	100	99	98	98
AUH	NM_001698	600529	132	100	100	100	100
CBS	NM_000071	613381	122	100	100	100	100
CLCN2	NM_001171087	600570	122	100	100	100	100
COL4A1	NM_001845	120130	121	100	99	99	99
COL4A2	NM_001846	120090	120	100	100	100	98
CSF1R	NM_001288705	164770	109	100	100	100	100
CST3	NM_001288614	604312	87	100	100	100	96
CTSA	NM_001127695	613111	139	100	100	100	100
CYP27A1	NM_000784	606530	187	100	100	100	100
DARS2	NM_018122	610956	157	100	100	100	100
GBE1	NM_000158	607839	158	100	100	100	100
GFAP	NM_002055	137780	102	100	100	100	100
GLA	NM_000169	300644	119	100	100	100	100
GSN	NM_001127662	137350	110	100	100	100	98
HTRA1	NM_002775	602194	126	97	93	92	92
ITM2B	NM_021999	603904	148	100	100	100	100
LMNB1	NM_001198557	150340	152	100	100	100	100
MMACHC	NM_015506	609831	149	100	100	100	100
*NOTCH3***	NM_000435	600276	118	100	98	97	94
TREM2	NM_001271821	605086	109	100	100	100	100
*TREX1***	NM_007248	606609	352	100	100	100	100
TTR	NM_000371	176300	219	100	100	100	100
TYMP	NM_001113755	131222	103	100	100	100	99
TYROBP	NM_001173514	604142	110	100	100	100	100

*CADASIL and cerebral angiopathies / adult-onset leukoencephalopathies

** Core genes (if necessary additional Sanger sequencing will be performed for 100% coverage)