

Whole Exome Sequencing

Gene package Movement disorders, version 1, 27-8-2015



Dept. Clinical Genetics



Technical information

After DNA enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Hiseq platform (101bp paired-end, Illumina). The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina Software CASAVA. Reads are mapped to the genome using BWA (reference: <http://bio-bwa.sourceforge.net/>). Variant detection is performed by Genome Analysis Toolkit (reference: <http://www.broadinstitute.org/gatk/>). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. Additionally, MPLA analysis was performed for *APTX*, *FXN* and *SETX* (SALSA P316 Recessive Ataxias probemix; MRC Holland) and for several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). For *ATN1*, *ATXN1*, *ATXN2*, *ATXN3*, *ATXN7*, *ATXN10*, *FMR1*, *PPP2R2B*, *C9ORF72* and *TBP* a repeat expansion test was performed. For *FXN* and *CACNA1A* a repeat expansion test was performed in addition to either MPLA and/or exome sequencing. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

ABCB7	COMT	GOSR2	PDYN	SPAST
ABCD1	COQ2	GPR56	PEX10	SPG11
ABHD12	COQ9	GRM1	PEX7	SPG20
ACTB	CP	GRN	PHYH	SPG21
ADCK3	CSTB	HEXB	PIK3R5	SPG7
AFG3L2	CYP27A1	HPRT1	PINK1	SPR
ALDH3A2	CYP2U1	HSPD1	PLA2G6	SPTBN2
ANO10	CYP7B1	HTRA2	PLP1	STUB1

ANO3	DBT	IFRD1	PMM2	SUOX
AP4B1	DCAF17	ITPR1	PNKD	SYNE1
AP4E1	DCTN1	KCNA1	PNPLA6	SYNJ1
AP4M1	DDC	KCNC3	POLG	SYT14
AP4S1	DDHD1	KCND3	PPP2R2B	TAF1
AP5Z1	DDHD2	KCNJ10	PRKCG	TARDBP
APTX	DLAT	KCNMA1	PRKRA	TBP
ARSA	DLD	KIAA0196	PRRT2	TDP1
ARX	DNAJC13	KIF1A	PSEN1	TECPR2
ASPA	DNAJC6	KIF1C	RAB29	TGM6
ATCAY	DNMT1	KIF5A	REEP1	TH
ATL1	EEF2	L1CAM	RNASEH2A	THAP1
ATM	EIF2B1	LRRK2	RNASEH2B	TIMM8A
ATN1	EIF2B2	MAPT	RNASEH2C	TMEM240
ATP13A2	EIF2B3	MARS2	RNF170	TMEM67
ATP1A3	EIF2B4	MECP2	RNF216	TOR1A
ATP2B3	EIF2B5	MMADHC	RTN2	TREX1
ATP6AP2	EIF4G1	MRE11A	SACS	TTBK2
ATP7B	ERLIN2	MTHFR	SAMHD1	TTC19
ATXN1	FA2H	MTPAP	SCN8A	TTPA
ATXN2	FBXO7	NIPA1	SERAC1	TUBB4A
ATXN3	FGF14	NKX2-1	SETX	UCHL1
ATXN7	FLVCR1	NOL3	SGCE	VAMP1
ATXN10	FMR1	NPC1	SIL1	VCP
B4GALNT1	FTL	NPC2	SLC16A2	VLDLR
BCKDHA	FXN	NUP62	SLC18A2	VPS13A
BCKDHB	GALC	OPA1	SLC19A3	VPS35
BSCL2	GAN	PANK2	SLC1A3	VPS37A
C9ORF72	GBA	PARK2	SLC20A2	WDR45
C10orf2	GBA2	PARK7	SLC25A15	WDR81
C19ORF12	GCDH	PAX6	SLC2A1	WWOX
CA8	GCH1	PDE8B	SLC30A10	ZFYVE26
CACNA1A	GFAP	PDGFB	SLC33A1	ZFYVE27

CACNB4	GIGYF2	PDGFRB	SLC52A2	ZNF592
CCDC88C	GJC2	PDHA1	SLC6A3	
CCT5	GLB1	PDSS1	SMPD1	
CIZ1	GNAL	PDSS2	SNCA	