Next Generation Sequencing - targeted panel
Neurofibromatosis type 1, version 1, 1-1-2022

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Technical information

DNA was enriched using Agilent SureSelect Custom Capture (ELID 3293151) and paired-end sequenced on the Illumina platform (outsourced). Data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the genome and variant detection. The aim is to obtain a unique coverage of the coding regions (CDS) +/- 30 basepairs for the indicated transcripts of at least 20x. When unique coverage is between 10x and 20x, regions are manually checked in the BAM file. When below 10x and within the CDS +/- 10 basepairs, Sanger sequencing is deployed for those regions. The detected variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. As no technique has a 100% sensitivity, we cannot exclude that pathogenic variants remain undetected. 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 15 nucleotides and of somatic mosaic variants (all types of sequence changes).

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	Transcript	median depth	% covered >30x	% covered >50x
NF1	613113	NM_000267.3	2105,20	100,00	100,00

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 55 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±30bp flanking introns)
- % Covered 10x and 20x describes the percentage of a gene's coding sequence (±30bp flanking introns) that is covered at least 30x or 50x