Whole Exome Sequencing
Gene package Noonan syndrome/RASopathies, prenatal
version 2, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
A2ML1	610627	100	99.96	99.39	97.99
BRAF	164757	95.04	94.58	93.81	88.27
CBL	165360	100	100	100	99.03
HRAS	190020	100	100	100	100
KRAS	190070	100	100	100	100
LZTR1	600574	100	100	100	98.04
MAP2K1	176872	100	100	100	97.96
MAP2K2	601263	100	100	98.27	87.68
NRAS	164790	100	100	100	99.54
PPP1CB	600590	100	100	100	100
PTPN11	176876	98.37	98.37	98.37	98.37
RAF1	164760	100	100	100	96.29
RIT1	609591	100	100	100	100
RRAS	165090	100	99.61	94.38	83.53
RRAS2	600098	100	100	100	94.74
SHOC2	602775	100	100	100	98.53
SOS1	182530	100	99.96	97.71	96.03
SOS2	601247	99.61	98.32	97.02	91.85
SPRED1	609291	100	100	100	100

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 104 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x