

WES NOONAN SYNDROME / RASOPATHY DG 3.6

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
BRAF	139.7	100.0	100.0	613706
CBL	127.1	100.0	100.0	613563
CDC42	156.6	100.0	100.0	616737
HRAS	157.5	100.0	100.0	218040
KRAS	161.6	100.0	100.0	609942
LZTR1	134.2	100.0	100.0	616564;605275
MAP2K1	137.0	100.0	100.0	615279
MAP2K2	132.0	100.0	100.0	615280
MAPK1	130.3	100.0	100.0	619087
MRAS	135.9	100.0	100.0	618499
NF1	147.4	100.0	100.0	601321
NRAS	139.4	100.0	100.0	613224
PPP1CB	150.0	100.0	100.0	617506
PTPN11	144.8	100.0	100.0	163950
RAC1	142.0	100.0	100.0	No OMIM phenotype
RAF1	133.3	100.0	100.0	611553
RIT1	136.9	100.0	100.0	615355
RRAS	113.2	100.0	99.8	No OMIM phenotype
RRAS2	144.6	100.0	100.0	618624
RREB1	118.6	100.0	100.0	-
SHOC2	151.1	100.0	100.0	607721
SOS1	145.6	100.0	100.0	610733
SOS2	141.1	100.0	100.0	616559
SPRED1	146.7	100.0	100.0	611431
SPRED2	141.0	100.0	100.0	No OMIM phenotype

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

Median Coverage describes the average number of reads seen across 50 exomes.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with no value for coverage are non protein coding genes.

Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors