

WES NOONAN SYNDROME / RASOPATHY DG 3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
BRAF	174.2	100.0	100.0	613706
CBL	197.3	100.0	100.0	613563
CDC42	137.2	100.0	100.0	616737
HRAS	273.1	100.0	100.0	218040
KRAS	158.7	100.0	100.0	609942
LZTR1	182.8	100.0	100.0	616564;605275
MAP2K1	183.0	100.0	100.0	615279
MAP2K2	232.8	100.0	100.0	615280
MAPK1	163.0	100.0	100.0	619087
MRAS	184.7	100.0	100.0	618499
NRAS	184.4	100.0	100.0	613224
PPP1CB	155.8	100.0	100.0	617506
PTPN11	185.6	100.0	100.0	163950
RAF1	181.9	100.0	100.0	611553
RIT1	176.5	100.0	100.0	615355
RRAS	153.9	100.0	100.0	No OMIM phenotype
RRAS2	133.7	100.0	100.0	618624
RREB1	222.3	100.0	100.0	-
SHOC2	151.5	100.0	100.0	607721
SOS1	144.0	100.0	100.0	610733
SOS2	153.7	100.0	100.0	616559
SPRED1	170.6	100.0	100.0	611431
SPRED2	231.3	100.0	100.0	No OMIM phenotype

Gene symbols used follow HGCN 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