



Specifications Sanquin Molecular diagnostics for UBA1 (X084)

Design UBA1 Panel

The Ion AmpliSeq UBA1 panel (IAD244649) exists of 43 amplicons and is covering 100% of submitted areas (all coding regions (exons)) and is able to analyze variants in *UBA1*. Indicated exons (Table 1) include flanking intronic regions based on 10 base exon padding.

Table 1. Design UBA1 panel

Gene	Chromosome	NCBI Transcript	Exon	Coverage %
UBA1	chrX	NM_003334.4	1-26 (full)	100

Coverage of the NGS UBA1 Panel

Coverage is the number of times a base is sequenced. The deeper the coverage of each base the greater the reliability and sensitivity of the sequencing assay. The minimum depth of coverage required for detection of somatic variants with the UBA1 Panel is 500X. The percentage of Target Base coverage (%Base500x) is the percentage of target bases in a panel that is covered at least 500 times.

Coverage for the NGS UBA1 panel is in silico validated and is 100% for all amplicons. The percentage of target bases that is covered at least 500 times (%Base500x) is 100% at 225.000 Mapped Reads and no amplicons are encountered below 500x coverage.

Reporting: addition hematological malignancies variants

This test does not distinguish between somatic and germ line alterations in analyzed gene regions, particularly when variant allele frequencies (VAF) are near 50% or 100%. If nucleotide alterations with a VAF near 50% or 100% are present and there is also a strong clinical suspicion or family history of spinal muscular atrophy, appropriate genetic counselling may be indicated.

Correlation with clinical, histopathologic and additional laboratory findings is required for final interpretation of the results. The final interpretation of results for clinical management of the patient is the responsibility of the managing physician.

NGS data are interpreted with the current knowledge concerning variants in relation to disease or as explanation of a phenotype. For reporting variants, the following guidelines will be followed: 'Best Practice Guidelines for Reporting Molecular Genetics results' written by R.J.L. Treacy and D.O. Robinson. The authorization of the results is done by a recognized Clinical molecular geneticist. All variants are annotated and reported as designated by the Human Genome Variation Society (HGVS) nomenclature, as described at [their website](#).

Field Code Changed