Specifications Sanquin Molecular diagnostics for LAD (X069)

Targeted sequencing with the Ion Torrent System is able to identify single nucleotide variants, small insertions and small deletions. Variants in repeat sequences, large homopolymers and large insertions/deletions are not or difficult to identify.

The **LAD AmpliSeq Panel v1** (IAD69604_201) exists of 74 amplicons and is covering 19 Kbase. 100% of desired areas (exons with in general 5 base exon padding) are covered in the design that includes the following five genes: *ITGB2, SLC35C1, FERMT3, CALDAG-GEFI* and *RAC2.*
There has been described one pathogenic deletion in *CALDAG-GEFI* at c.814-16_814-5del12. This position, just outside the 5 bases of exon padding, is also included in the panel.

Coverage of the LAD Ampliseq Panel

The percentage of target bases that is covered at least 20 times is at least 96.7% for the recommended Mapped Reads of 250,000. If necessary we can achieve a 100% 20x coverage of the whole panel.

Read more about the technique and reporting in the background information on the Sanquin website.