Sequencing Report: Coverage

Samples included	141-1-2A	141-2-1U	141-2-2U

Key metrics for dbCMMS v1.0

Sample Id	Cutoff	Avg. Coverage	Avg. Completeness [%]	Diagnostic Yield [%]
141-1-2A	10	143.2874	99.4854	92.249
141-2-1U	10	210.3256	99.667	94.028
141-2-2U	10	193.0433	99.6035	92.5032

Sex Check

Sample Id	Chromosome X Coverage	Chromosome Y Coverage	Sex Prediction
141-1-2A	145.74	0.77	female
141-2-2U	198.66	0.95	female
141-2-1U	109.8	48.53	male

Missed Transcripts

Gene Id	Transcript Id	Sample	Completeness [%]
BOLA3	CCDS33224.1	141-1-2A	82.5082
BOLA3	CCDS33224.1	141-2-1U	83.7146

Further information

General

The parts of the genome that have been analysed are all protein-coding parts of exons annotated in the CCDS database, together with all splice donor and acceptor sites (two bases in both ends of all introns).

Key metrics for Gene Panel

The estimated average coverage and completeness have been made on the transcript level (vs. exon for full key metrics). This will in all likelyhood not differ much from the more accurate exon level estimate but it should be noted that they can differ.

Sex check

Depending on the coverage of X and Y chromosomes, a simple still very accurate sex prediction can be made on the samples. This is a simple control step that will yield a warning that samples can have been mixed upstream.

Missed transcripts

A missed transcript is defined as having less than or equal to 50% completeness. The idea is to set the `MISSED_TRANSCRIPT_CUTOFF` setting to a value where finding a variant in the remaining (uncovered) bases is so unlikely that it can be discarded.

Note also that this 'risk' will be a factor both of the the `MISSED_TRANSCRIPT_CUTOFF` and the original completeness cutoff (default: 10x).