

Quality report clinical exome sequencing

Sample	Group	Cutoff [x]
1-1-3A	1	10
1-1-4A	1	10

Generally important metrics

Sample	Average coverage [x]	Average completeness [%]
1-1-3A	123.047	83.145
1-1-4A	123.499	82.87

Diagnostic yield

Sample	Diagnostic yield [%]
1-1-3A	46.062
1-1-4A	45.708

Gender confirmation

Sample	Gender	Average coverage [x]	
		Chromosome X	Chromosome Y
1-1-3A	male	93.073	145.447
1-1-4A	female	176.271	1.172

Explanations

General

The parts of the genome that has been analyzed is composed of all protein coding, exonic intervals referenced in the [CCDS database](#). Additionally, the data also includes splicing sites (donator and acceptor positions, two bases on either of each exon.)

Coverage: An alternative measure of sequence depth. It is defined as the ratio of bases sequenced deeper than a specified cutoff, e.g. 10x. The current cutoff is defined per sample and is found in the "Cutoff" column (first table).

Completeness: gives an overview of the success of sequencing. It's defined as the ratio of transcripts with a completeness of 100%.