Quality report clinical exome sequencing

Sample	Group	Cutoff [x]
P575-101E	P575-coriell	10
P575-105E	P575-coriell	10
P575-106E	P575-coriell	10

Generally important metrics

Sample	Average coverage [x]	Average completeness [%]
P575-101E	91.997	97.51
P575-105E	102.294	97.764
P575-106E	75.942	97.205

Diagnostic yield

Sample	Diagnostic yield [%]
P575-101E	77.379
P575-105E	82.267
P575-106E	73.226

Gender confirmation

Sample	Gender	Average coverage [x]	
		Chromosome X	Chromosome Y
P575-101E	male	50.194	22.008
P575-105E	male	55.432	30.832
P575-106E	female	77.08	0.378

Explanations

General

The parts of the genome that has been analyzsed is composed of all protein coding, exonic intervals referenced in the CCDS database . Additionally, the data also includes spicing 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 i 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 of100%.