

MLPA assay QC checklist

QC1. Electrophoresis data	QC2. QC Fragments check	QC3. Control samples	QC4. Data quality check in Coffalyzer.Net
Strength of capillary electrophoresis (CE) signal: PASSED	Benchmark fragment at 92nt: DETECTED	A. Positive control (SD024) <small>(Not applicable to P239: BRCA1 up- and downstream probes)</small>	Maximum FRSS, FMRS and CAS quality scores: PASSED
Size standard and CE quality: PASSED	Four DNA quantity fragments: DETECTED and ABOVE THRESHOLD	Artificial duplication in P002:BRCA1 and BRCA2:P090: DETECTED	No missing probe: PASSED
Regularity, distribution and shape of peaks: PASSED	Two DNA denaturation fragments: DETECTED and ABOVE THRESHOLD	Two chromosome X-specific fragment: DETECTED	Bin size correct and no peak overlapping: PASSED
MLPA peaks fall into the optimal range of our CE platform: PASSED	Chromosome X-specific fragment (For sex checking): MATCHED	One chromosome Y-specific fragment: DETECTED	Lot number of the probes was accurately shown: PASSED
	Chromosome Y-specific fragment (For sex checking): MATCHED	B. No DNA control Only five major peaks <121nt: DETECTED	
		C. Reference samples Three normal references: INCLUDED	
		Variation of individual probe among three references (SD<=0.10): PASSED	
		Normal BRCA1/2 copy number: DETECTED	
		D. Non-BRCA specific reference probes Variation of reference probes (SD<=0.10): PASSED	
		Final ratio of each reference probe fall within normal copy number range: PASSED	

P002:BRCA1 (**QC PASSED**)
P090:BRCA2 (**QC PASSED**)
P239:BRCA1 up- and downstreams (**QC PASSED**)