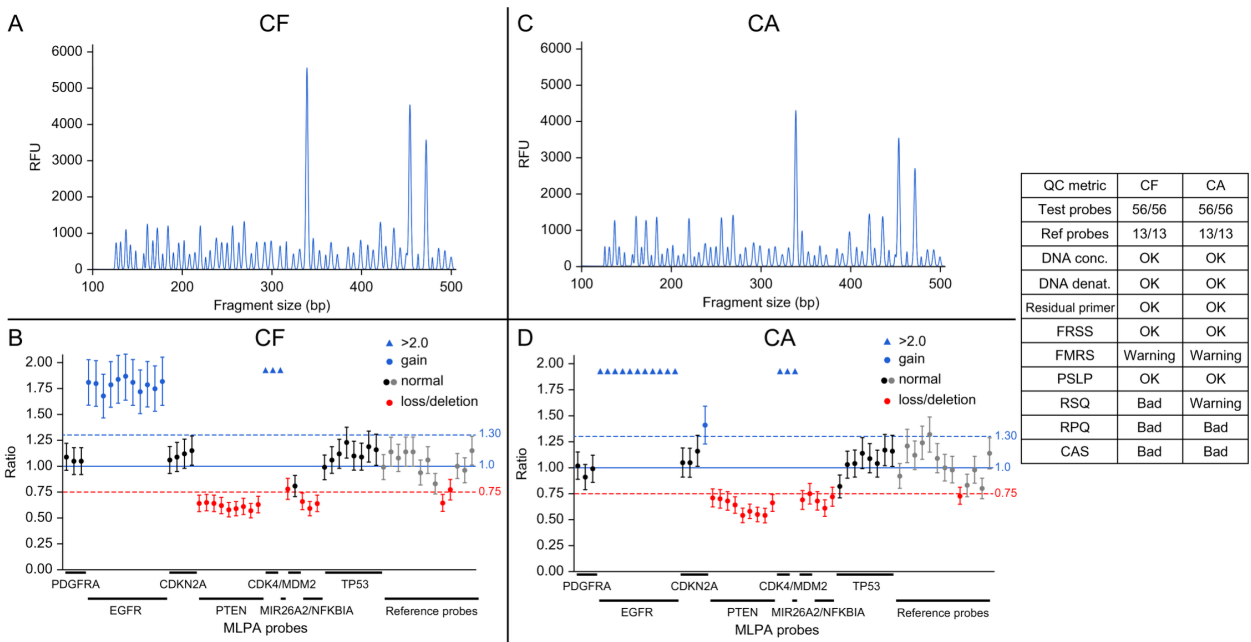


Online Resource 2. Representative discordant MLPA case



Representative discordant MLPA case. Representative MLPA findings are shown from paired CF and CA specimens. Electropherograms, normalized copy number ratio plots, and specimen-level quality-control metrics are presented. Reduced assay quality was observed in this representative discordant pair. The result should therefore be interpreted cautiously, although a contribution of spatial differences between the adjacent but non-identical specimens cannot be excluded. CF, conventional forceps specimen; CA, CUSA aspirate specimen; MLPA, multiplex ligation-dependent probe amplification; QS, quality score calculated by Coffalyser.Net software; RFU, relative fluorescence unit

Supplementary methods for MLPA quality-control metrics

For the representative discordant pair shown in Online Resource 2, the accompanying quality-control summary was included to document the suboptimal internal analytical quality associated with the discordant result. Quality control

metrics were exported from the MLPA software and reported in their abbreviated form to preserve consistency with the original software output. The abbreviations are defined as follows: FRSS, fragment run sample score; FMRS, fragment median relative signal; PSLP, probe signal ligation performance; RSQ, relative sample quality; RPQ, relative probe quality; and CAS, overall case assessment score. These metrics were used descriptively to summarize internal analytical performance. In this study, they were not analyzed as independent study endpoints but were used to support the classification of selected paired cases as quality-limited when the interpretation of copy number profiles was reduced by suboptimal assay performance.