VALIDATION OF A PROBABILISTIC GENOTYPING SOFTWARE KONGOH IN DNA MIXTURES WITH DEGRADATION OR PCR INHIBITION

Sho Manabe, Chie Morimoto, Shuntaro Fujimoto, Eriko Hirai, Yuya Hamano, Keiji Tamaki
Department of Forensic Medicine, Kyoto University Graduate School of Medicine

We developed an open source software Kongoh (the Japanese word for “mixture”) based on a continuous model to promote DNA mixture interpretation in Japan. Kongoh was validated by referring the SWGDAM guidelines for the validation of probabilistic genotyping systems. However, case-type samples of DNA degradation and PCR inhibition were not fully validated in the current version of Kongoh (v.2.0.1). In this study, we validated Kongoh v.2.0.1 to determine the limitation in DNA mixtures with degradation or PCR inhibition.

We used publicly available data sets in the Project Research Openness for Validation with Empirical Data (PROVEDIt). We selected 65 two-person, 66 three-person, and 64 four-person mixture profiles with degradation through UV damage or sonication, or with inhibition by humic acid. These profiles were typed by the Identifiler Plus kit with 28 cycles and ABI 3130xl Genetic Analyzer with an injection time of 10 s. The likelihood ratios (LRs) of the true contributors and 100 computationally generated non-contributors were calculated for each mixture. To calculate LRs, we first calculated the likelihoods of 1¬–4 contributors in both the prosecutor hypothesis (i.e., a person of interest is a contributor) and defense hypothesis (i.e., an unknown person is a contributor). We then calculated the LR from the ratio of the maximum likelihood for each hypothesis.

Most of the LR values of the true-contributors tended to be strongly supportive (i.e., LR > 10,000) for the prosecutor hypothesis. However, some LR values of true-contributors were < 1 especially in the mixtures with a high degree of degradation such as UV 105 min and 30 cycles of sonication. In these mixtures, approximately half of the loci located at the right side of the electropherogram were dropped out. We also confirmed that the LR values of the non-contributors tended to be near 1 as DNA was more damaged and the amount of DNA was smaller. These results suggest that Kongoh generates expected results for interpreting DNA mixtures with degradation or PCR inhibition.