International Congress Series 1288 (2006) 822–824

www.ics-elsevier.com

SampleCheck, a laboratory information management system for quality assurance of DNA-profile analysis in parentage and forensic testing

P. Reitz, M. Jung *

bj-diagnostik GmbH, Kerkrader Str. 11, 35394 Giessen, Germany

Abstract. To increase the validity of laboratory results SampleCheck performs plausibility checks on DNA profiles. The platform independent programme interacts with a database storing DNA profiles and further information. The main function of SampleCheck is to perform plausibility checks on DNA profiles to detect pipetting errors, contaminations with other DNA and if samples during collection or pipetting may have been interchanged. Checks can be performed batchwise and/or against the whole database. Every sample is monitored for correct gender by comparing expected and measured gender. At the same time a comparison of the measured profiles and the profiles of coworkers and positive controls takes place. Additionally, a single sample or the samples of a whole batch can be checked for identical profiles and profiles sharing a common allele on each marker like in father–child and mother–child relations. Allele frequency tables for different populations, mutation rates, coworkers and positive control profiles can be stored. SampleCheck enables the user to import this information from different file formats to the database. LR-values can be calculated using three different methods.

Keywords: DNA; LIMS; Quality assurance; Plausibility check; Parentage testing; Likelihood ratio; Forensic

1. Introduction

Any relatedness case contains recurrent, valuable information like gender or role of the sample's donor. This information combined with the uniqueness of measurement results (DNA profiles) and relatedness between sample donors opens the possibility for extensive plausibility monitoring (locally: batchwise and globally: in the whole database). Sample and case information (expected gender, role) can be entered manually or automatically retrieved

* Corresponding author. Tel.: +49 641 9446053; fax: +49 641 4994139.
E-mail address: michael.jung@bj-diagnostik.de (M. Jung).

0531-5131/
doi:10.1016/j.ics.2005.08.015

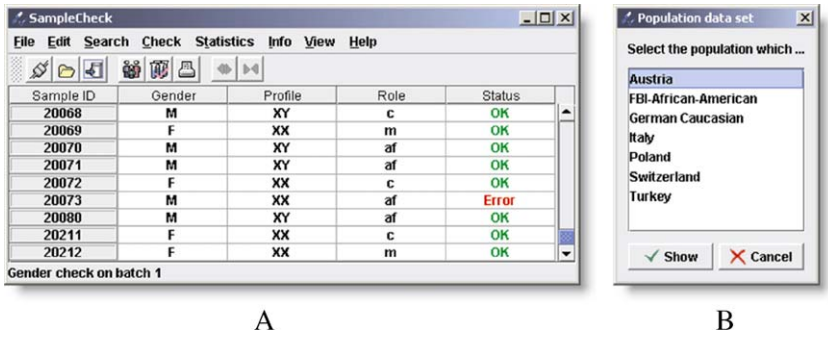

Fig. 1. Gender check result list (A) and menu for selection of population allele frequency data set (B).

from additional databases, e.g. from a LIMS (Laboratory Information Management System). SampleCheck can be adjusted to read case data from any other database. The programme is structured in two parts, plausibility checking and relatedness testing. As DNA profiles are stored and grouped in families it is self-evident to include also likelihood ratio calculation for standard cases. It is not necessary to export profiles to some other programme for LR calculation. Hence, this reduces the risk of errors during exporting and calculation [1].

2. Plausibility checks of DNA profiles from the laboratory

Paternity testing allows checks of data integrity from expected and measured values. A paternity case always gives information before the laboratory analysis begins. During the registering of samples from a new case in a LIMS the gender of a tested person and the role within the family are stored. Let us call this information expectation values. The gender of a tested person must be matched by the result obtained through the measurement. This is the first check to be done by our system, for a result see Fig. 1A. Sample 20073 shows the measured gender XX and this does not match with the registered role “alleged father” of the sample. Fig. 1B is an example from the population data set to be used for likelihood calculation. Data integrity also means that the role must match with the gender. Any

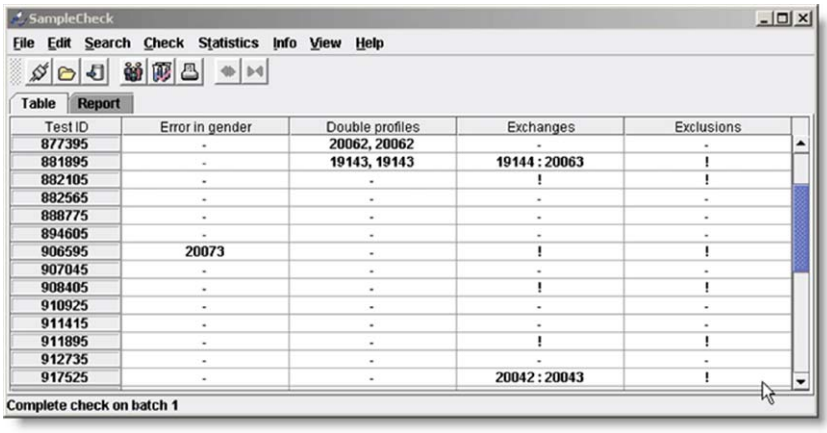

Fig. 2. Complete batch check result reveals five possible inconsistencies that must be inspected.

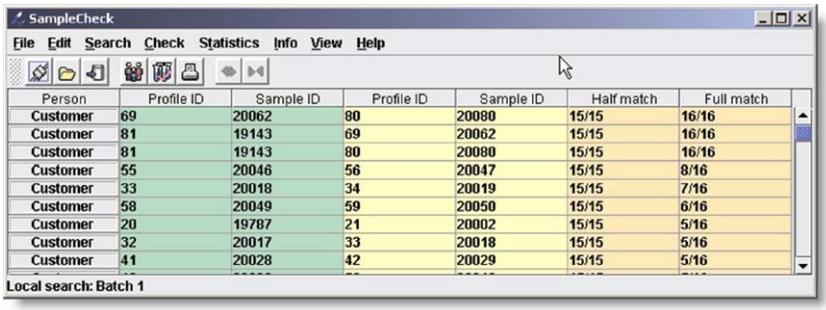

Fig. 3. Result of search for equal profiles and profiles sharing alleles on the tested markers.

inconsistencies reported by the programme help to control and verify plausibility of the laboratory work. Usually, for unknown donors of forensic samples, there are no expectation values of this nature. But still, SampleCheck is applicable in DNA profile mass screenings when the gender of a tested person is known. Technically, expectation values from a LIMS are compared against data from SampleCheck's DNA profile database. Inconsistencies may be uncovered in both databases vice versa. SampleCheck, in general, targets errors during sample collection by the customer or sample handling in the lab. Pipetting errors like sample interchange or double pipetting (a DNA profile appears more than once in a batch) and contamination with coworkers' or positive control DNA will be detected and reported (Fig. 2). Special tasks like exchanging the profiles of e.g. mother and daughter are performed automatically to test if an exclusion may change to a non-exclusion. Furthermore an exclusion rate will be calculated for every batch and for the whole database. Fig. 2 summarises gender errors, double profiles, exchanges and exclusions. Besides an exclusion, the sign (!) informs the user that not all samples of the case (test ID) were measured in a batch. We artificially introduced three errors in the batch used here: 1) the measured gender of sample 20073 was changed to XX, 2) DNA profiles for samples 20042 and 20043 were exchanged and 3) the DNA profile for sample 19143 was overwritten with the profile of sample 20062. The complete batch check then reports five problems. Error 2) results from a mix up of samples from daughter and mother leading to an apparent exclusion. The double profiles are detected and exchanging 19144 with 20063 (or vice versa) will convert exclusions to non-exclusions. Samples 19143, 19144, 19145 and 20062, 20063, 20064 belong to two families with the order father, child and mother. In both cases paternity is not excluded. Besides intra batch checks, inter batch checks can be performed using all profiles in the database. A result screen is given in Fig. 3.

3. Concluding remarks

SampleCheck is a powerful tool for quality management in DNA profile measurement. To our knowledge, SampleCheck is the first commercially available software to monitor plausibility in DNA testing labs. Further information can be requested from the authors.

Reference

[1] P. Reitz, M. Jung, www.samplecheck.com.