OPEN SOURCE INDEPENDENT REVIEW AND INTERPRETATION SYSTEM (OSIRIS) AS AN EXPERT SYSTEM FOR CONVICTED OFFENDER ANALYSIS

Charity Noreuil, Taylor Scott, Rebecca Wheeler, Donald Parker, Illinois State Police, DNA Indexing Laboratory

Our laboratory currently employs Applied Biosystems' (AB) GeneMapper ID (GMID) v3.2.1, Promega's PowerPlex 16 Amplification Kit, and AB's Data Collection Software 3.0 as our expert system for convicted offender analysis. GMID v3.2.1 as the expert system software functions acceptably; however, the user-interface is awkward and the software is limited in its capabilities. The Illinois State Police (ISP) has had the opportunity to collaborate on the Open Source Independent Review and Interpretation System (OSIRIS) software (Forman Neall et al. 2008, 2009), a novel expert system software program that has been developed by the National Center for Biotechnology Information (NCBI), a division of the U.S. National Institutes of Health (NIH) (Goor et al. 2011). This software solves many of the usability issues we have with GMID v3.2.1 as well as provides several analytical enhancements. In addition, the Federal Bureau of Investigation (FBI) recently notified the ISP of its acceptance of OSIRIS as a National DNA Index System (NDIS) approved expert system.

OSIRIS is a public domain quality assurance software package freely available by download from the NCBI website: www.ncbi.nlm.nih.gov/projects/SNP/osiris/. This software analyzes .fsa and .hid files generated from the data collection software on AB's capillary electrophoresis genetic analyzers without the need for any additional analysis software. It offers a unique opportunity to laboratories that are searching for allele calling software or an expert system because it is a cost-free alternative that is analytically precise and accurate. In addition, the software is easy to use with a simple workflow and provides rapid throughput while using minimal processor resources.

The OSIRIS software mathematically evaluates peak data by applying a sizing algorithm that was independently derived by a team at NCBI. The team created a cubic spline transformation that uses all of the internal lane standard (ILS) peaks instead of utilizing the popular local Southern method that uses two ILS peaks above and below the analyzed peak. Furthermore, the OSIRIS algorithm builds on a time scale rather than base pair size to evaluate and compare peaks between the ladders and sample profiles. OSIRIS maps the times of the ILS peaks of the ladder into the ILS peaks of the sample profile by using a coordinate transformation. This transformation will most closely align the ILS peaks onto each other and create the closest possible coordinates to a straight line. This minimizes size and shape distortion while retaining and applying all of the data in the ILS range.

The laboratory conducted a precision study to assess the software's ability to consistently determine fragment sizes of alleles. This precision study was conducted using 10 ladders and the ILS peaks 60-500 base pairs. The calculated (standard deviation)*3 values obtained for each allele were not greater than +/- 0.5 base pairs, demonstrating the software will repeatedly produce reliable results within a one base pair size difference.

OSIRIS will identify more than one positive or negative control as long as their names and profiles are added to the list of controls to validate. Positive controls are verified by comparing the DNA profiles between the obtained profile and the expected profile. For negative controls, OSIRIS will confirm the presence of primer dimer peaks and will notify the user of peaks that exist above the analytical threshold as well as the sub-analytical threshold. Numerous positive controls, orientation controls, and negative controls were analyzed without issue. OSIRIS identified and correctly assessed all of the controls.

Comparisons were conducted on approximately 4,000 profiles generated from OSIRIS and profiles obtained from manual reviews for the reproducibility study. In all instances, OSIRIS demonstrated its ability to correctly label alleles. Additionally, the export file function in OSIRIS may be used to export acceptable profiles from OSIRIS to a LIMS system or to CODIS. All of the export files generated from these 4,000 samples did not export insufficient or incorrect data. OSIRIS demonstrated its ability to correctly assess the acceptance of a DNA profile.