OSIRIS QUALITY ASSURANCE SOFTWARE FOR MULTIPLEX STR PROFILES

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The National DNA Index System (NDIS) currently contains nearly 6 million DNA profiles from convicted offenders and is credited with resolving tens of thousands of unsolved crimes. Given nationwide mandates to collect DNA profiles for all felony convictions and the increasing trend towards arrestee collections for certain crimes, these numbers will continue to climb. Beyond its use in the criminal justice system, high throughput DNA profiling is often the most effective means of victim identification in mass fatalities, which can require analyses of hundreds of thousands of samples.

State and Federal oversight requirements for data generated in these situations compel laboratories to find new resources for data review and reinforce the need for broadly available quality management tools. The increasing capacity challenges on forensic identification systems now provide multiple opportunities to deploy the next generation of management tools for quality assessment.

OSIRIS (Open Source, Independent Review & Interpretation System) is a public domain quality assurance software package that facilitates the assessment of multiplex STR DNA profiles based on laboratory-specified protocols. OSIRIS evaluates the raw electrophoresis data contained in any .fsa file using a new mathematical approach that assures its independence from other microsatellite genotype analysis software.

The algorithm iteratively fits expected parametric data signatures to the observed data to identify peaks, usually achieving matches with correlations in excess of 0.999. Parametric peak locations are determined with sub-second accuracy and transformed to base pair coordinates. Sizing methods traditionally rely on either the local or global Southern methods (1-3) to interpolate the ILS into base pair estimates. OSIRIS departs from this approach, using instead the correspondence between a sample's ILS and an associated allelic ladder to map the time scale of the ladder into that of the sample. This allows effective integration of the ladder with the sample for a straightforward and accurate comparison (typically within 0.1 of a base pair) of sample peaks with ladder locus peaks. Thus, in addition to extremely sensitive peak analysis, OSIRIS offers two new peak quality measures – fit level and sizing residual. These new measures can enhance quality metrics currently available to assess STR DNA profiles.

OSIRIS accommodates laboratory-specific signatures, including adjusted sensitivity to background noise, customized naming conventions and internal laboratory controls. When used in complement with other analysis methods, OSIRIS provides an independent review to assure data concordance. With appropriate forensic validation and NDIS approval, OSIRIS may alleviate the need for visual review of passing profiles.

The National Center for Biotechnology Information (NCBI) developed OSIRIS in collaboration with state, local, and federal forensic laboratories and the National Institute of Standards and Technology (NIST). This freely available, object-oriented software is written in C++ to facilitate the development of add-on applications the private and public sectors. NCBI performs internal quality assurance on its programs and will maintain OSIRIS at http://www.ncbi.nlm.nih.gov/projects/SNP/osiris as part of its extensive public domain toolkit for exploring and managing genetic data.

References:

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