OSIRIS VERSION 2.2: INTELLIGENT CASEWORK AND CODIS STR ANALYSIS

George Riley[1], Robert Goor[1], Douglas Hoffman[1], Tim Kalafut[2], Charity Noreuil[3], Taylor Scott[3], Joel Sutton[2], Stephen Sherry[1]
[1]National Center for Biotechnology Information, National Library of Medicine, NIH
[2]U.S. Army Criminal Investigation Laboratory/DFSC
[3]Illinois State Police, DNA Indexing Laboratory

OSIRIS, the Open Source Independent Review and Interpretation System, is freely available software for the analysis of STR profiles produced using Applied Biosystems 310, 31xx, 3500 and 3700 series Genetic Analyzers, and can be downloaded at http://www.ncbi.nlm.nih.gov/projects/SNP/osiris/. OSIRIS analyzes both .fsa and .hid format files produced by the collection software, using an independently derived mathematically-based sizing algorithm (Goor et al., 2011). OSIRIS supports numerous commercially available marker kits including CODIS-compliant kits as well as other kits favored by forensic, biomedical and relationship testing laboratories. Supported kits include GlobalFiler™, Fusion™ and PowerPlex Y23™, and OSIRIS includes sophisticated Y-STR analysis and dynamic baselining.

OSIRIS identifies peaks by iteratively fitting expected parametric data signatures to the observed data, with matches generally having correlations in excess of 0.999. Parametric peak locations are determined as time and transformed into base pair coordinates. Rather than the global Southern method, OSIRIS instead uses 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, fitting the most appropriate ladder to each sample. This integration of the ladder with the sample permits a straightforward and accurate comparison of sample peaks with ladder locus peaks. This method provides two additional peak quality measures: fit level and sizing residual (a measure of peak shift).

OSIRIS is designed to be flexible for ease of integration with the laboratory's own workflow, including its LIMS and other software, reducing manual intervention in both analysis and data transfer. It increases the efficiency and accuracy of analysis by categorizing artifacts either as critical, requiring human review, or non-critical, dependent on user-designated parameters. Analysis is fast, typically under 30 seconds per 96 well plate. For poor quality samples, OSIRIS can intelligently predict required rework conditions. An audit trail of editing by analysts and reviewers is preserved, and laboratory review procedures can be software enforced. Reporting and data export are very flexible, allowing laboratories to export tables, CODIS compatible data, LIMS-formatted data, and other formats of the laboratory's own design, and can be automated to export subsets of data directly upon analysis. OSIRIS also has quality metrics that allow it to be used as a lab process quality assurance tool.

OSIRIS version 2.2, developed in collaboration with the U.S. Army Criminal Investigation Laboratory and the Illinois State Police DNA Indexing Laboratory specifically addresses casework analysis, low level DNA analysis and editing efficiency. OSIRIS v. 2.2 decreases the amount of analyst editing of artifacts, and improves low-threshold analysis using dynamic baselining in the identification of low level peaks and artifacts.

In addition to OSIRIS having been validated for forensic casework and as an NDIS-approved expert system for CODIS testing, it has been validated for clinical testing of bone marrow engraftment, as an expert system for relationship testing and for research cell line verification in laboratories in the U.S. and internationally. Additionally, OSIRIS is used for STR analysis training and data review.

Goor RM, Forman Neall L, Hoffman D, Sherry ST, (2011). A mathematical approach to the analysis of multiplex DNA profiles. Bulletin of Mathematical Biology 73(8):1909-31. Epub 2010 Nov 20

Work at NCBI is supported by the Intramural Research Program of the NIH and the National Library of Medicine