OSIRIS Version 2.0: INTELLIGENT DNA PROFILE ANALYSIS AND QUALITY ASSURANCE SOFTWARE

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Stand-alone Open Source Software

Installs automatically when downloaded from:
http://www.ncbi.nlm.nih.gov/projects/SNP/osiris/

OSIRIS Uses

- DNA Profile Analysis
- DNA Profile QC
- Automating Sample Reanalysis
- Process monitoring
- Training

Software Architecture

Core program	State Message Book
Compiled C++ Signal processing	XML compiles on execution Encapsulates knowledge base

- New conceptual framework
- Easily add new detection parameters and reanalysis points without modification of core program

- Lab can customize messages to its own wording and SOP
- Enhances program's flexibility and stability

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

Expanded Laboratory Settings

Laboratories settings include:
A. Artifact RFU thresholds default & by locus
B. Minimum number of analyst and reviewers required for export
C. Analysis thresholds for samples, ladders, ILS
D. Sample QC settings and Sample/Batch QA settings (to track process quality)
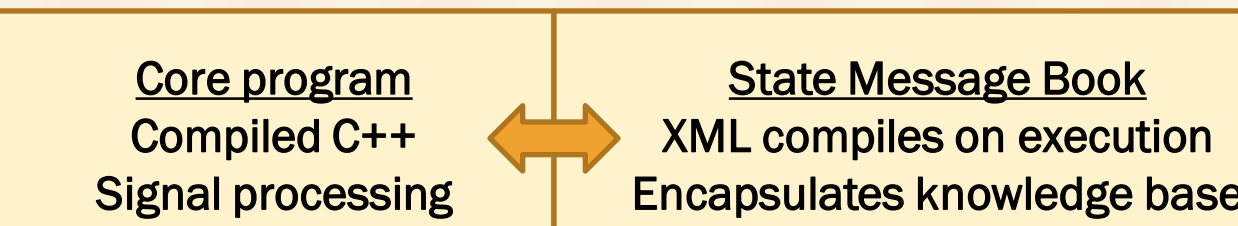E. Laboratory accepted off ladder alleles, lab positive control alleles and triallele

Expanded Artifact Recognition

Recognizes 150 artifact signatures - makes artifact identification sensitive and specific (underlined artifacts are new)

Peaks (including ILS and Ladder)
- Poor peak morphology
- Adenylation
- Pull-up
- Spike
- Crater
- Blob
- Peak shifting - exceeds threshold
- Interlocus peak
- Peak height above max. threshold
- Peak height below min threshold
- Off scale data – (Laser off scale)

Locus
- Stutter
- Heterozygous peak imbalance (incl. high stutter)
- Off-ladder alleles
- Triallele
- Allele dropout
- Single peak below homozygote threshold
- Peaks between detection/analysis thresholds
- Locus dropout
- Amelogenin extraneous peaks
- Amelogenin X allele missing
- More than three alleles identified - mixture

Channel
- Peaks between loci
- Raised baseline
- Excessive baseline noise

Sample
- Sample may be a mixture
- Locus dropouts
- Multiple trialleles - mixture

Batch
- Default lab settings are overridden
- Kit positive control missing
- Lab custom positive control(s) missing
- Negative control missing
- Ladder requires review

Control-specific

ILS
- Incorrect number of peaks
- Incorrect spacing

Ladder
- Incorrect number of alleles
- Incorrect spacing

Negative control
- Primer peaks missing
- Contamination – unexpected peaks
- Negative control requires analyst review

Positive control
- Kit positive not found
- Custom lab positive not found
- Positive control allele mismatch

Intelligent Sample Reanalysis

Intelligent Reanalysis Prediction
- Uses knowledge base to determine appropriate reanalysis of samples that do not pass QC
- Too little DNA
- Too much DNA
- Degraded DNA vs. Too much DNA
- Allows analyst to override automated reanalysis recommendations

Automated Reanalysis Recommendations
- Recommend Reamp Less
- Recommend Reamp More

Analyst-Selected Reanalysis Decisions
- Reamp Sample Regular
- Reamp Sample More
- Reamp Sample Less
- Reinject Sample
- Reextract Sample
- Do Not Rework Sample
- Verification Sample (do not reanalyze if meets minimum number of acceptable loci)

Artifact Identification

Graph View

Table View - Locus dropout

In vWA
- Pull-up
- Off scale data (Laser off scale)
- Recommend Reamp less

Graph View
Hover over artifact

Table View
Hover over artifact

In D2S1338
- Homozygote too short
- Edited and annotated
- Reamp more by analyst

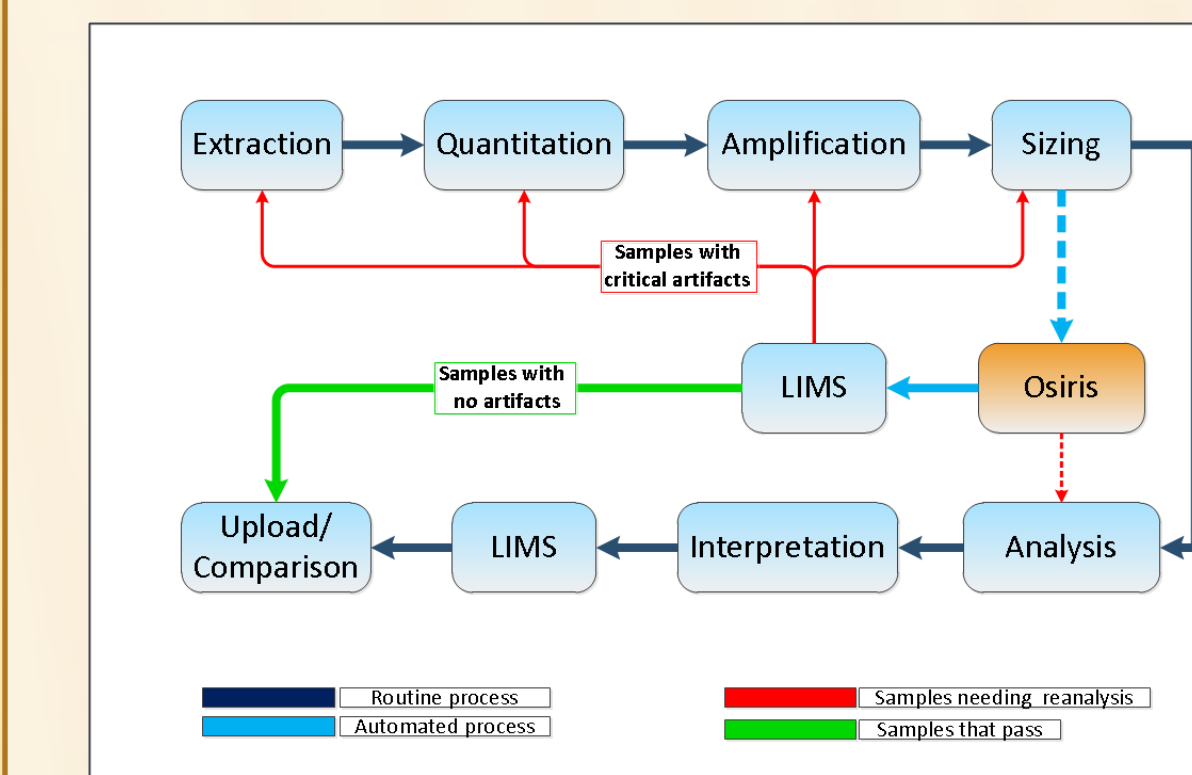Spike in D5S818 and TH01

Data Export

Data Export Formats
- LIMS import
- CODIS CMF
- Discovery project files
- Spreadsheet
- Sample artifact lists
- Artifactual sample project files
- User defined text, XML or HTML

Export to LIMS
- Exports file formatted for direct LIMS import
- Checks for presence of un-reviewed artifacts (artifact-priority driven)
- Checks for presence and quality of positive/negative controls
- Formats allele calls for alleles above/below the ladder
- Exports OSIRIS and analyst reanalysis decisions
- Can choose between different injections of the same plate for successful samples
- Takes into account:
  - Trialleles
  - QC/verification samples that do not require all loci to amplify successfully
- Export can be automatic on analysis (if no artifacts present)

Analysis and Reanalysis

OSIRIS can be used to automate process flow by sample reanalysis making intelligent reanalysis decisions and exporting them to LIMS to requeue failed samples.

LIMS Import File

UD1 & UD2 = LIMS instructions   UD3 = LIMS comment
The comment field values depends on the priority of the artifacts for the overall sample

OSIRIS Features

- Table/Graph view and classical graphic view
- Sort by sample or by artifact severity
- Flexible display - user preferences retained
- Artifacts and peaks can be edited
- Change artifacts to peaks and vice versa
- Annotate/Document editing changes
- Analysis and review history is maintained
- Analysis parameters secured for forensic use
- Analysis/Review functions software enforced

Acknowledgements