GeneMarker®HID

by SoftGenetics

Automatic Allele Conflict Identification and Pedigree Drawing Tool with GeneMarker®HID

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Introduction

Relationship testing involves testing known genotypes in a familial relationship for simple Mendelian inheritance laws. Since the short tandem repeat (STR) core loci selected for forensic genotyping demonstrate the Mendelian characteristics of segregation and independent assortment, the Relationship Testing tool in GeneMarker HID is an excellent choice to determine genetic relationships between family members. Children inherit one allele from each parent; sharing one allele identity by descent (IBD) with each parent. Siblings may have 0, 1 or 2 alleles IBD. GeneMarkerHID has software tools that automatically draw pedigree tree trios; accurate and rapid comparison of STR profiles from family members in a pedigree tree; and allele conflicts linked to sample traces. Features of the program include: user friendly linked navigation, management control and tracking, project comparison; individual peaks are quality flagged including quality reasons, supervisor review module, exportable CODIS and LIMS reports, and bulk printing capabilities.

Procedure

1. File→Open data→Project→Run→to make allele calls (See Chapter 2, GM HID User Manual) or open previously saved GM HID project
2. Select Relationship Testing
3. Select Family Group Tool
4. Match files by section, fixed position or group order
5. Select 'Match' and then 'Okay'
6. Use Main Tool Bar toggle to select 'conflict with parents' or 'conflict with siblings'
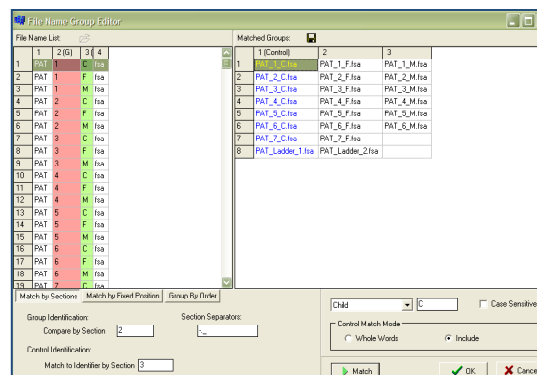7. Place the cursor over a red node to display allele conflicts

Figure 1: Using the Family Group Tool provides automated drawing of trio pedigree trees.

Results

In **figure 2** eleven allele conflicts between the child and father are automatically displayed by red type in the genotype of the pedigree tree and a drop-down list in the pedigree tree. Single left-click the Marker name and the family members' traces will appear in the Charts tab on the right.
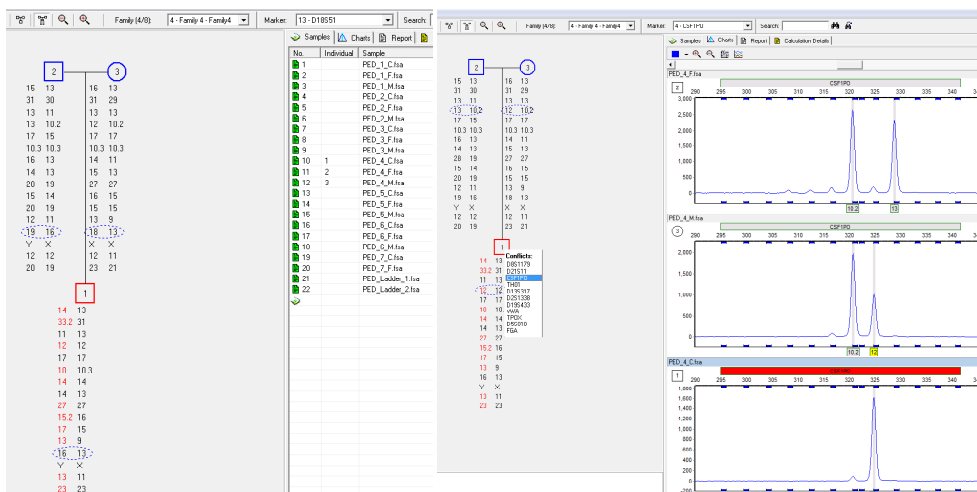
Once a family is created in the Pedigree Tree, GeneMarkerHID automatically identifies any Mendelian inheritance conflicts. In the example in figure 2, the node of a child with allele conflicts is outlined in red. The conflict alleles are in red and the alleles that are in agreement with Mendelian inheritance patterns are in black. The pedigree tree is linked to the sample list and the sample traces. Place the cursor over the red node to activate a dropdown list of the conflicting alleles. Click on each marker name in the list to link to the section of the sample traces and circle these alleles in the genotypes of the pedigree tree. Extended family members can be added to increase the generations represented in the pedigree tree. Right click on the node of an individual for options to add a mate or a child.

Discussion

Traditional analysis of family traces for allele conflicts requires manual side-by-side screening of each set of traces. With GeneMarker HID, the automated identification of allele conflicts between parent/child or between siblings is a great time-saver for analysis.

GeneMarkerHID Relationship Testing has all of the strengths of GeneMarker including; unique pattern recognition and sizing technology providing >99% accuracy, easy linked navigation, management control and tracking, project comparison, exportable CODIS and LIMS reports, bulk printing capabilities, instrument compatibility with ABI, MegaBACE and Beckman-Coulter and compatibility with STR kits (including: Cofiler®, Profiler®, Identifiler®, Minifiler®, SGMPlus®, PowerPlex®)

Acknowledgements

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