Exploring Capabilities and Limitations:
Capabilities of Probabilistic Genotyping with Mixtures of Biological Family Members

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2017 SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories

- Binary statistical models typically assume unrelated individuals
  - **3.2.7** Statistical models commonly used for estimating DNA profile rarity typically involve an assumption of unrelated individuals…
    - *"The probability of randomly selecting an unrelated individual with a DNA profile matching that of the [evidence] is approximately…"*

- **4B.11** Likelihood ratios can be applied to mixtures of close biological relatives if the assumptions include the unknown contributor(s) are unrelated to the assumed contributors
  - *_____ times more likely to be observed if it originated from the VICTIM and SUSPECT than if it originated from the VICTIM and an unknown individual in the Caucasian population.*

Study Design

- 11 families with 1-4 children
  - 2 families: 1 child
  - 4 families: 2 children
  - 4 families: 3 children
  - 1 family: 4 children

- DNA adjusted to 0.1ng/ul

- PowerPlex® Fusion amplifications contained ~0.5ng input DNA

- GMIDX® used to interpret mixture profiles

- .hid files uploaded to TrueAllele®

- TrueAllele® protocol used in accordance with routine casework

Study Design (cont'd)

- Two contributor mixtures:
  - M + F 1:1; 1:4 and 4:1
  - 11 families = 33 mixtures

- TrueAllele $^®$ runs:
  - 2c as 2uk
  - 2c as 3uk
  - 2c as 1 uk + M or F (selected mixtures)
  - 3c as 1uk + M + F (selected mixtures)

Key:

M – mother

F – father

c – contributor

uk - unknown

Evaluating Results

- Runs not meeting DFS' acceptance criteria were not used.
  - GR (Gelman-Rubin) convergence value above 1.5 were not used.
  - Match scores not reproducible within 2 ban were not considered.

2c as 2uk – Selected Family
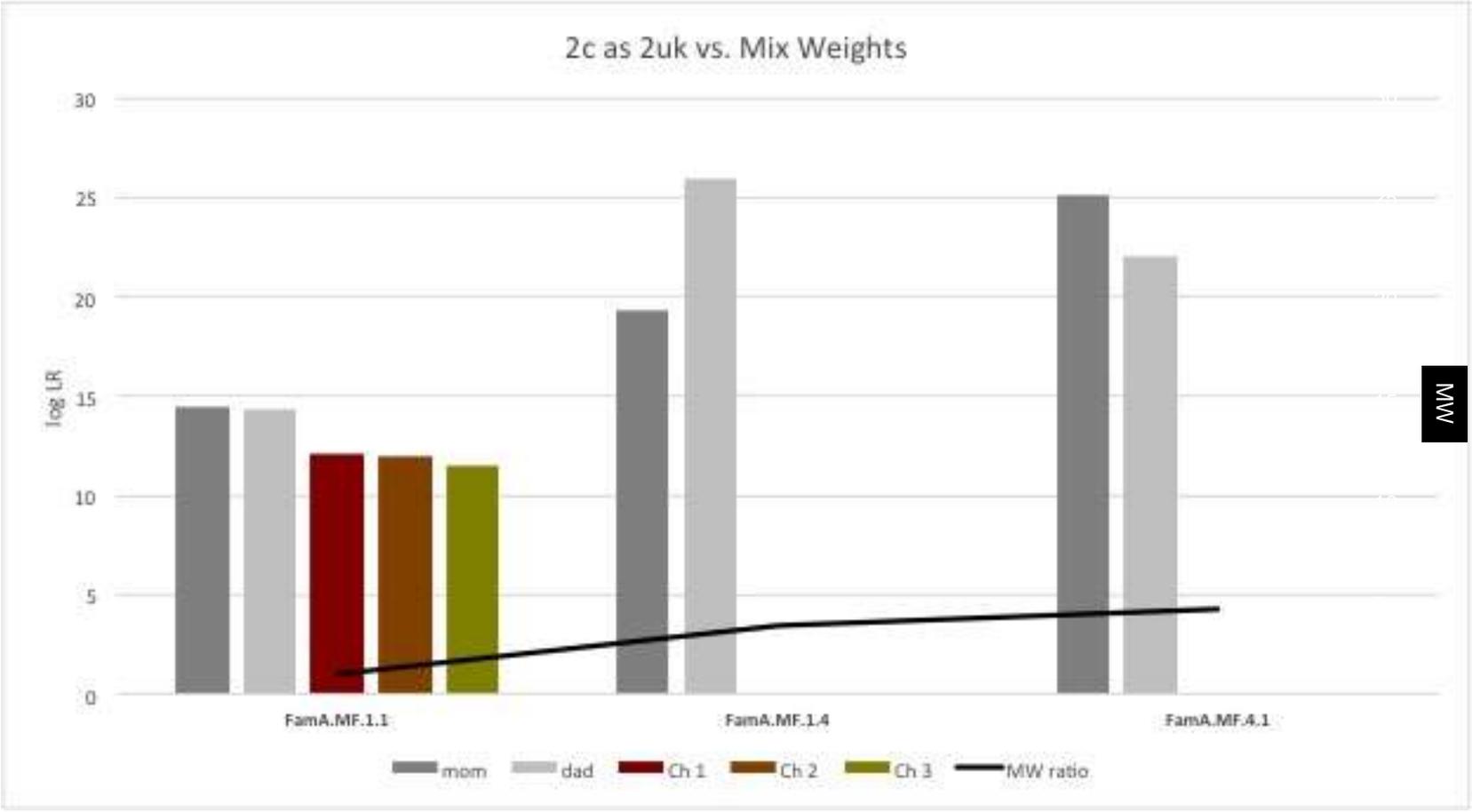
2c as 2uk w/ Mixture Ratios

2c as 2uk vs. Mix Weights
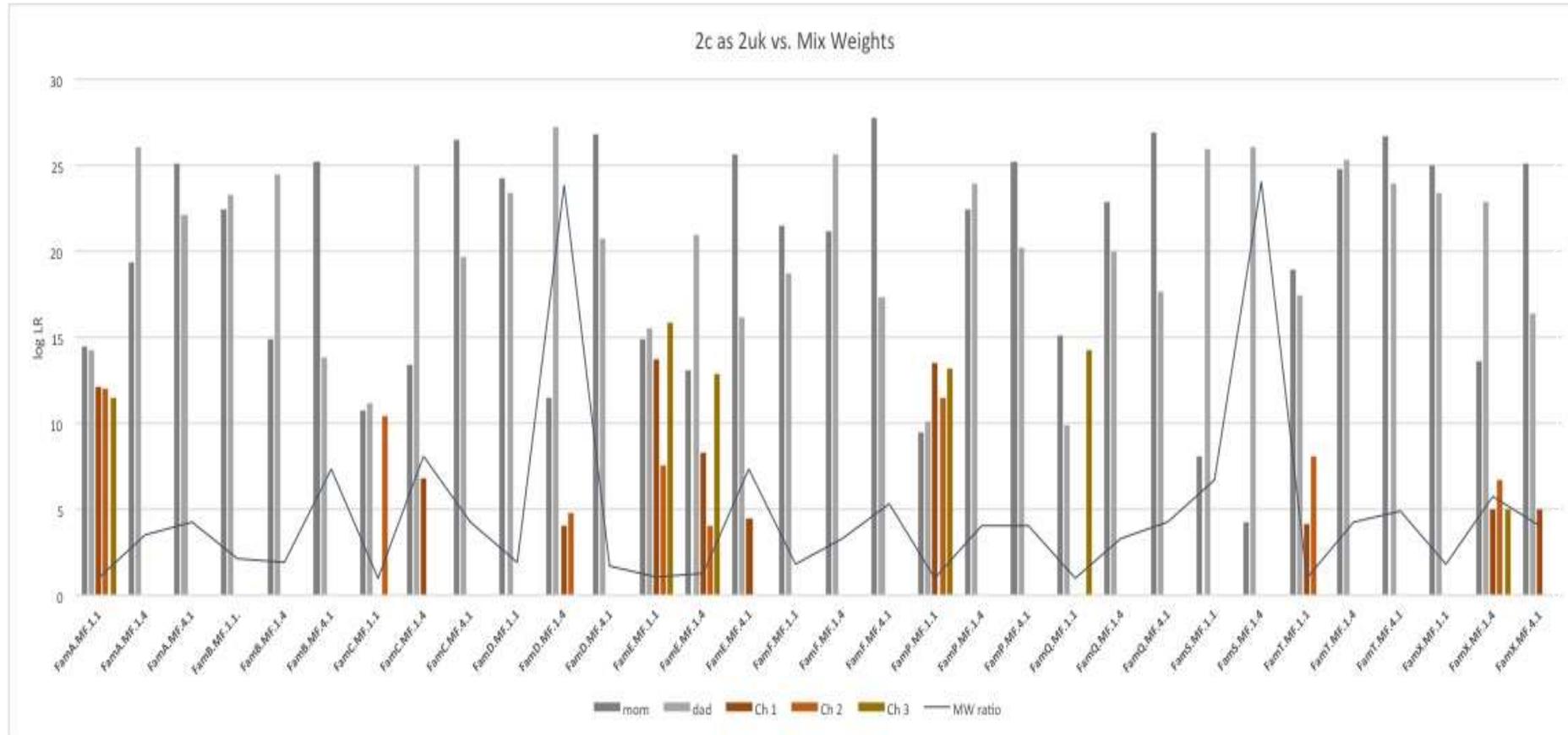
Family A ~1:1 mixture
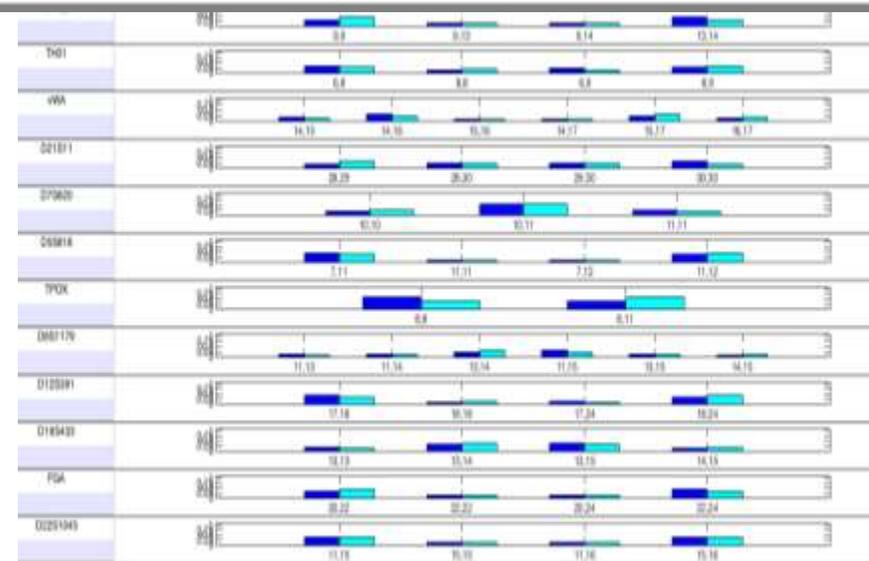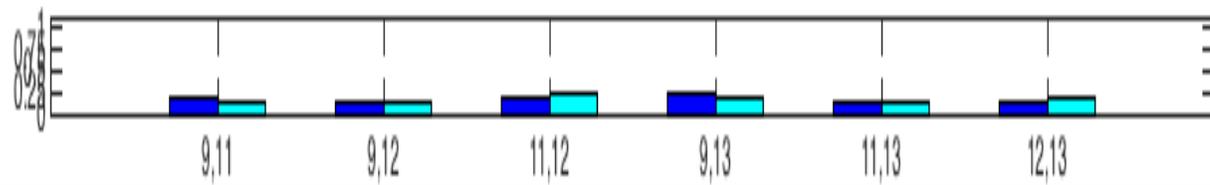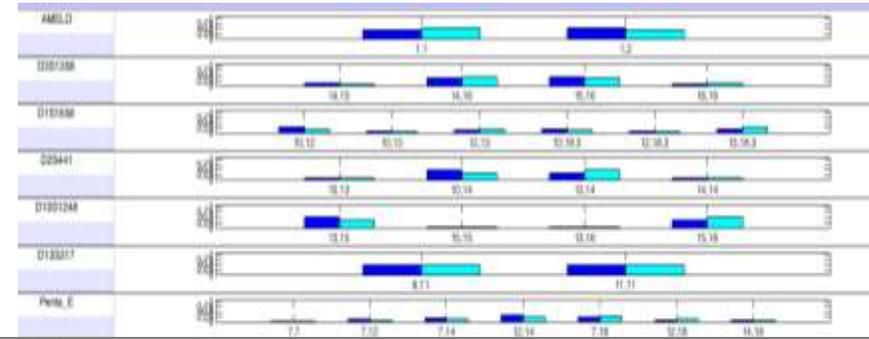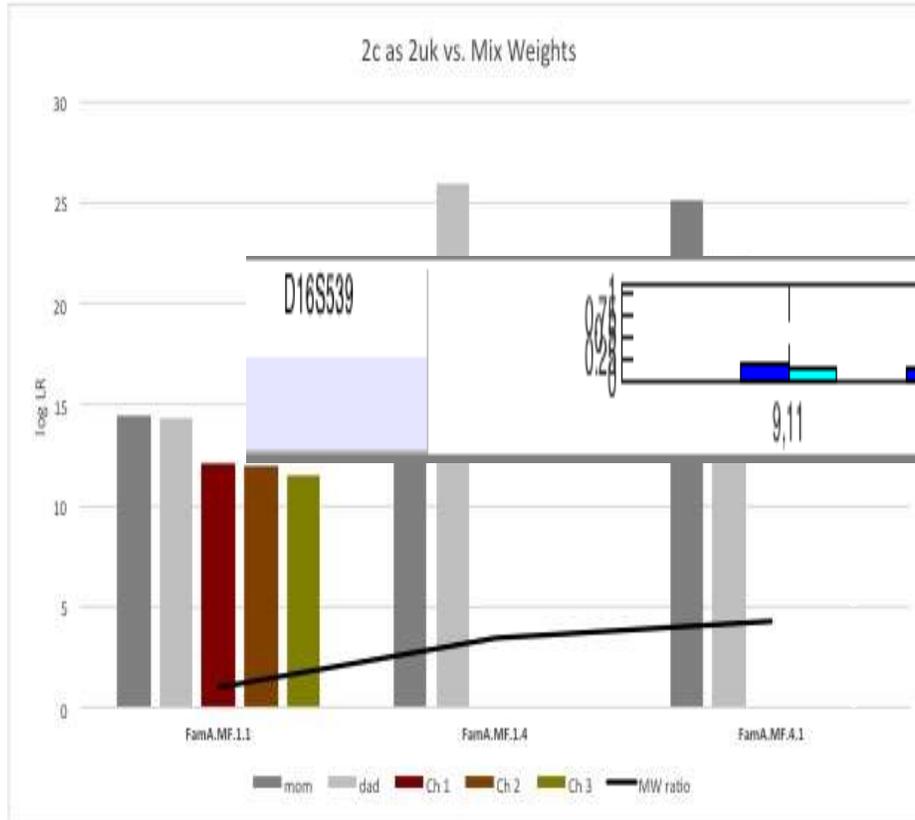
Family D ~1:24 mixture
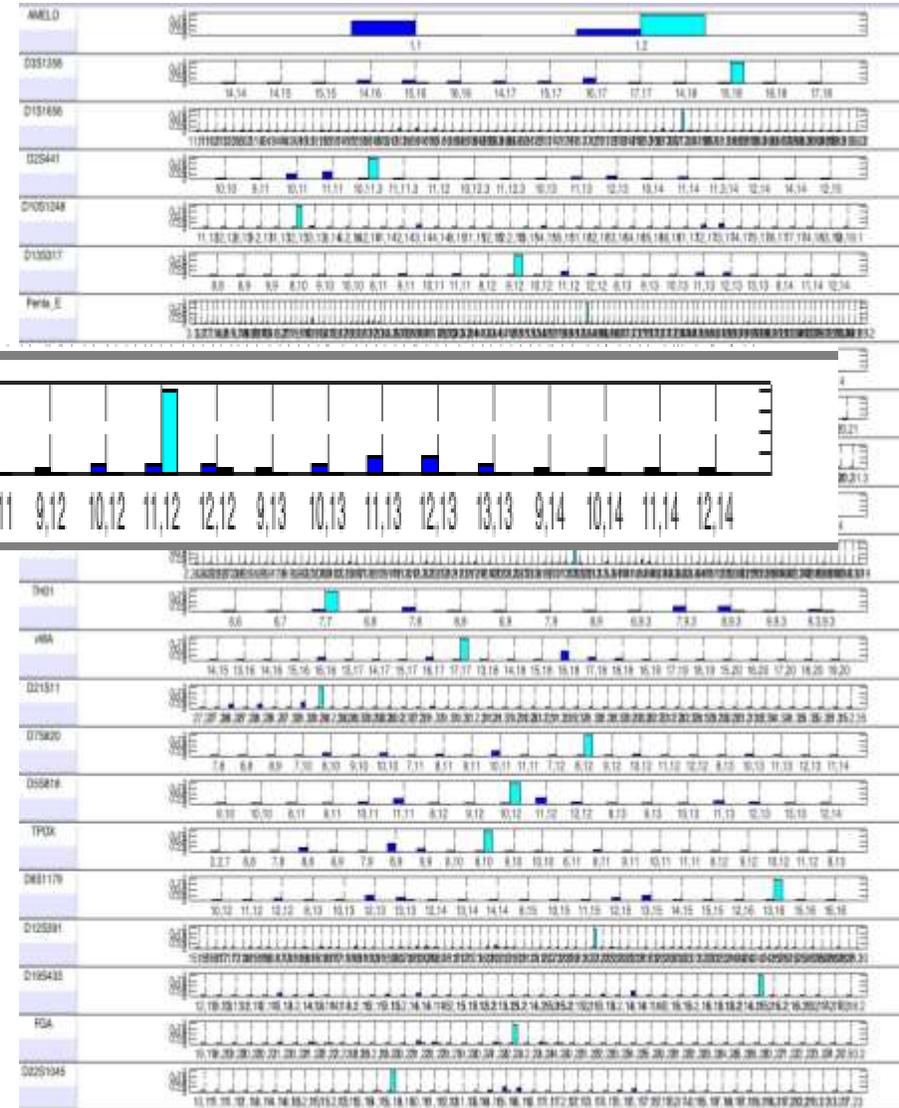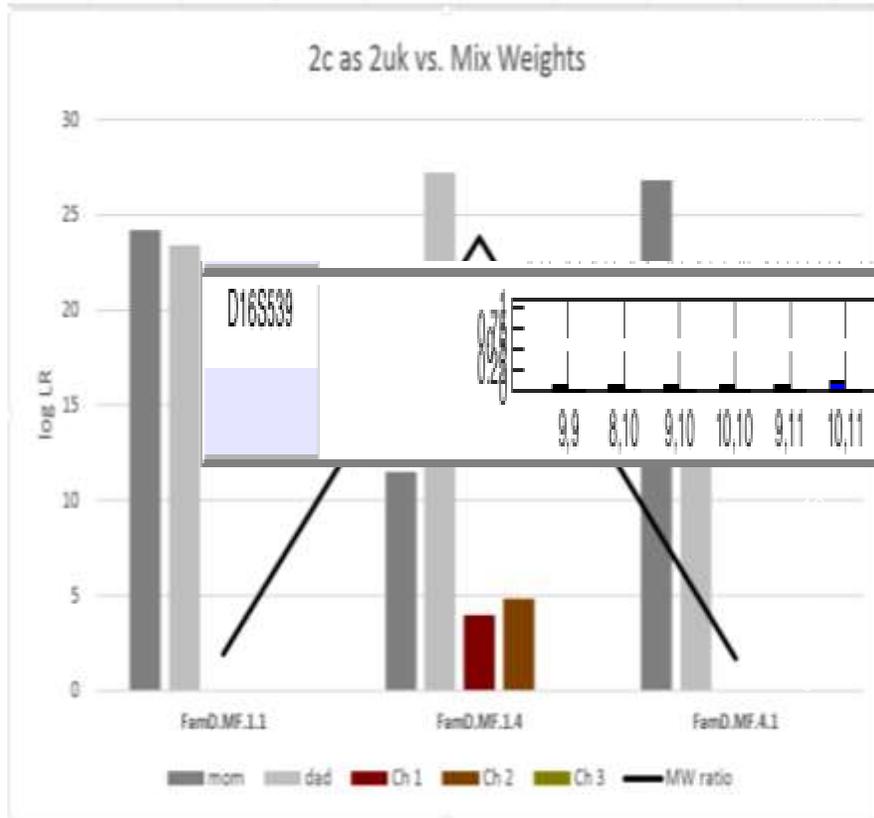
Family B mixture
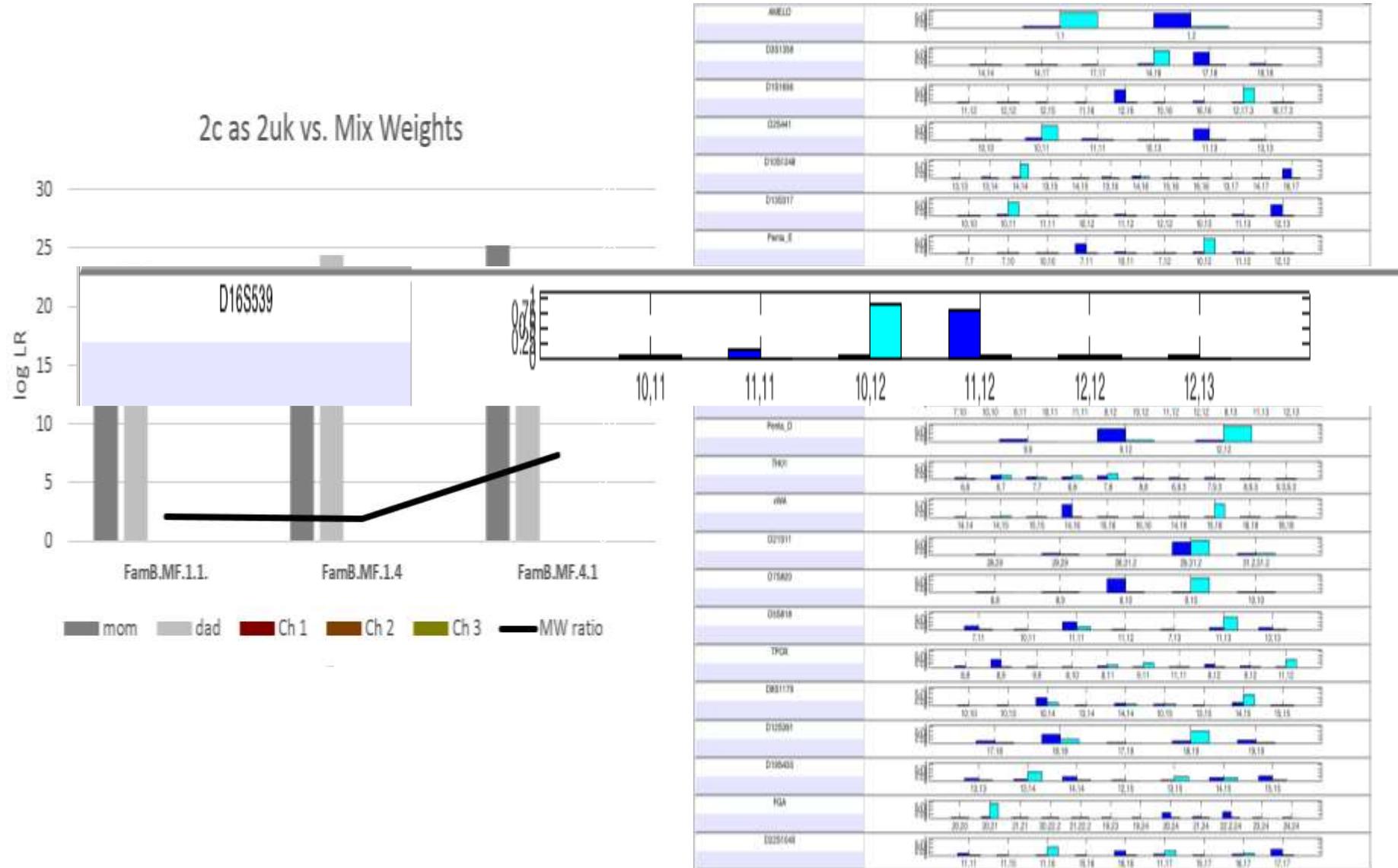
Sequentially solving with assumed known contributors

- 2c mixtures previously demonstrating adventitious matches to biological children were solved as 1uk assuming mother and 1uk assuming father

- A few mixtures needed additional runs to solve

Sequentially solving with assumed known contributors

2c as 1uk w AK
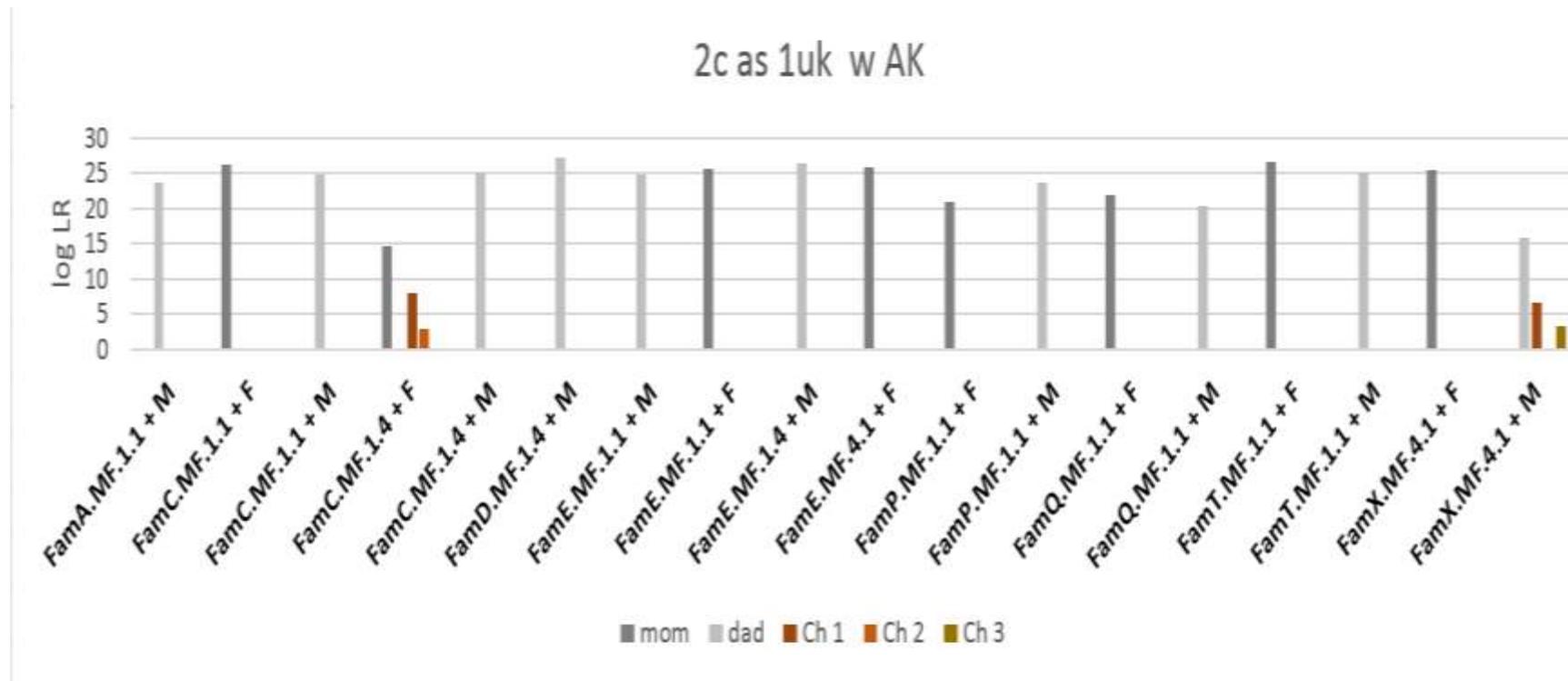
Resolution of adventitious matches using assumed knowns
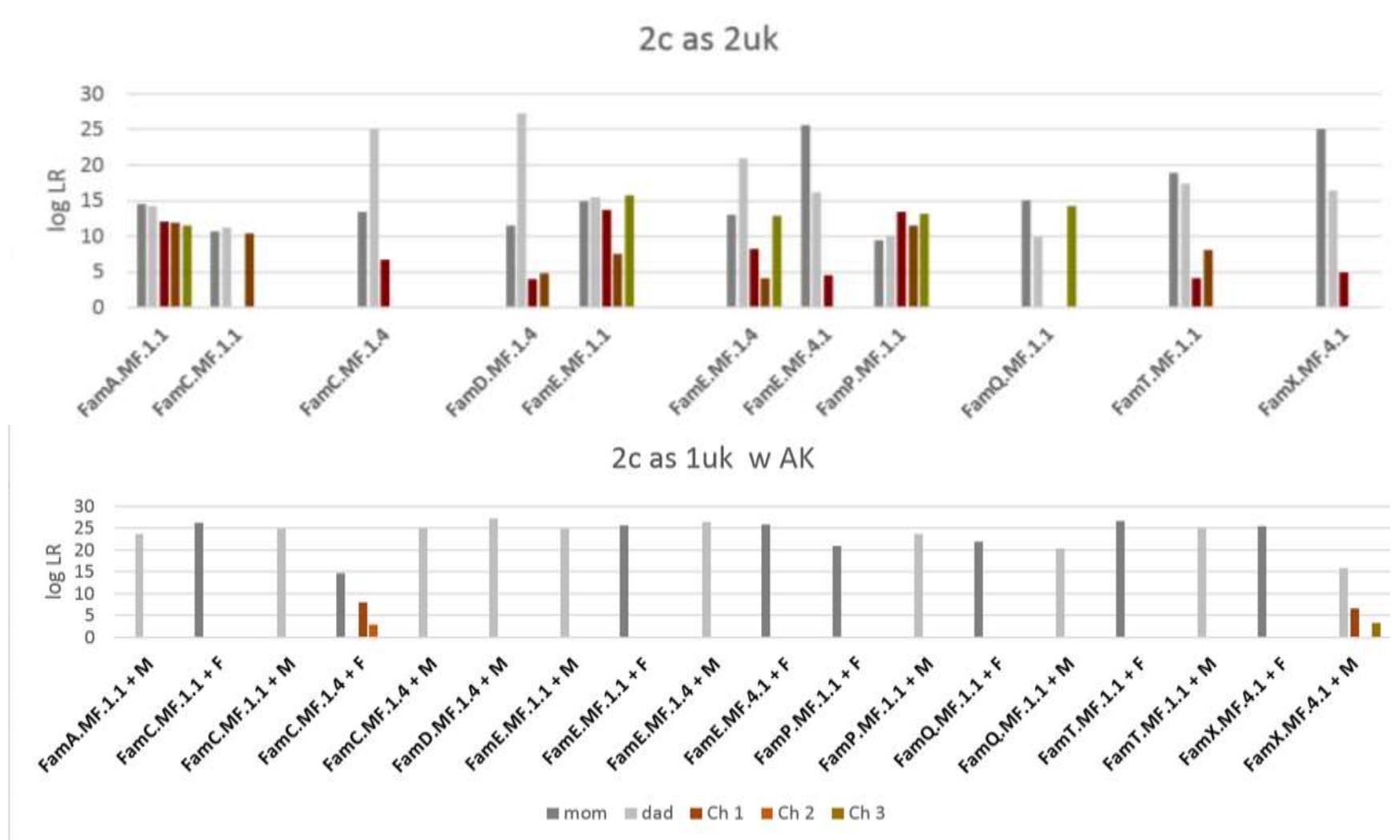
Conclusions

- Mixtures of parents can adventitiously include biological children
    - Not all mixtures will result in positive match scores to biological children

- Using a step-wise process of sequentially adding assumed references is an effective means of eliminating adventitious matches

- VA DFS Policy uses likelihood ratios generated by probabilistic genotyping when close biological relatives are included in the same DNA profile