



## In This Issue

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Do you have an interesting case or question you would like to share? Email us at [nikkib@aabb.org](mailto:nikkib@aabb.org)

### Standard 6.3.1.2

Standard 6.3.1.2, reads “These genetic inconsistencies shall be reported and incorporated appropriately into the calculations.” The Guidance for this Standard indicates that the method used to make a mutation calculation should not underestimate the rarity of the mutation. Given the data that has accumulated in the last several annual reports, which substantiates previous studies regarding the relative rarity of mutations that depart from the paternal obligate allele by more than one repeat, laboratories may want to re-evaluate their method for mutation calculations. According to the 2008 annual report, 65% of laboratories are still using the mutation rate divided by the probability of exclusion. Among those laboratories, a few modify this value when the child’s allele changes by more than a single STR repeat (i.e., dividing that value by ten, although the justification for using 10 in this formula has not been shown on a locus by locus basis). Those laboratories that do not modify this calculation in some way for those situations where the child’s allele differs by more than a single repeat are probably underestimating the rarity of mutations of the father’s allele significantly when the mutation differs by two or more repeats.

This can obviously have a significant effect on the CPI calculated for a case. However, the impact is probably low in cases where the mother, child and father are tested because the residual CPI is may be high enough to give a CPI that is above a CPI of 100. However, in cases where the mother is not tested, the resulting CPI is substantially lower. In some cases

with a relatively low residual CPI a possible two (or more) step mutation could potentially change the case conclusion. The rarity of these mutations is important to take into account to prevent false inclusions that otherwise might be detected by testing the mother. In motherless cases with a suspected two step or more mutation, the lab should have a procedure for evaluating these occurrences. The procedure might include obtaining a sample from the mother or testing additional loci to reduce the possibility of mistakenly including a close relative or randomly matching man. Other situations where a two step mutation could have an impact include: immigration cases, where the alternative hypothesis may actually be a comparison to a related man (or woman), and double mutations. Each of these situations warrants the use of a calculation that does not underestimate the rarity of these events.

For laboratories using the mutation rate over the probability of exclusion to account for mutations in the CPI, it is critical to make sure the mutation frequency is used, rather than the mutation rate as a percentage (i.e.  $0.001/PE$ , rather than  $0.1/PE$ ). If the calculation is performed using the mutation rate expressed as the percent value then the rarity of the mutation will be underestimated by one hundred fold. The impact of this can be tested by calculating a triple or even quadruple mutation using this formula. If the mutation rate is underestimated, one may achieve a respectable CPI in certain cases even with four inconsistencies calculated as a quadruple mutation. If calculated using an appropriate mutation rate, an acceptable CPI would probably not be achieved.

Laboratories may wish to consider abandoning the mutation rate over the probability of exclusion in favor of either the mutation calculation method of Brenner or, once data is available Fimmers. The method of

Fimmer's would be particularly useful as it uses measured mutation rates from one particular allele to another and thus takes into account any number of repeat differences between the two alleles thought to be involved in the mutation. Other problems with mutation rate / PE are the inability to incorporate situations where the mutation event could have been either maternal or paternal in origin.

Another problem, in that good frequency data does not exist, is silent or null alleles. These null alleles will also cause the appearance of inconsistencies in relationship testing. The formula for calculating an apparent null allele is different than that used for mutations.

Lastly, as a reminder that if your laboratory is using the mutation rate over the probability of exclusion (PE) be sure you are using the correct formula for calculating the PE. Unfortunately the forensic community used the term PE for calculating the chance of a mismatch in case work profiles that is not the same as the older PE used in calculating a mismatch in relationship testing. The laboratories properly using the relationship version of the PE should indicate in their procedures if they are using the average PE (what the original derivation uses) or the actual PE for the case in their mutation calculation. Regardless, what the laboratory is using for mutation calculations should be detailed in their procedure manuals.

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## Use of Y STRs in Relationship Testing

There is sometimes confusion on when Y-Chromosome STR testing alone is acceptable and how it should be reported. AABB does not accredit genealogical testing and does not have standards for genealogical testing. However where relationship tests are performed and are under the umbrella of AABB accreditation, then certain standards apply. In a standard paternity test, the laboratory must test multiple genetic systems. A Y-STR haplotype counts as a single system, so a Y-STR profile is not acceptable as the sole basis of a paternity test. The laboratory must also include autosomal testing. The same applies when other first degree relationships are tested and the relationship is defined (for example, is the tested man the brother of this child or an unrelated man). Where the conclusion is based in part on Y-STR testing, the Y-STR results must be incorporated into the calculation. Typically, this is accomplished for Y-STRs by using one divided by the Y-STR haplotype frequency and incorporating it into the combined likelihood ratio for the tested relationship. If Y-STR testing is used solely as verification of other testing and is not reported, then it need not be incorporated into the calculation. For relationships that are more distant than a first degree relationship, a statement of no relationship or of relationship may be rendered on findings from Y Chromosome or mitochondrial loci alone. A Y Chromosome case is effectively a male lineage test and must be reported appropriately. When Y STRs are tested alone it is not correct to report a particular relationship, for example stating the testing indicates that two tested persons have an uncle / nephew relationship is not accurate. It would be accurate to state that there may be a common paternal ancestor of these two individuals.

See Std. 5.3 #1), #2) and #3), Std. 6.3.2, and the associated Guidance that applies to those standards.



**IF YOU MISSED THE MEETING IN 2009\*\* YOU CAN STILL ORDER HANDOUTS OR STREAMING AUDIO WITH POWERPOINT SLIDES OF ALL THE SESSIONS HELD DURING THE 2007 ANNUAL MEETING BY VISITING**

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**\*\*CONTENT FROM 2002- 2009 SESSIONS ARE STILL AVAILABLE**

**Session: Relationship Testing SIG I:  
Forensic Science  
Current Topics and Case Studies**

**Held: Saturday, Oct. 9, 2010  
10:30 am – 12:00 pm**

**Director/Moderator:** Mary Mount, MT(ASCP)  
**Speakers:** Robert Bever, PhD; Todd Bille; Nathan Himes  
**Intended Audience:** Physicians, Scientists, Technologists, Nurses, Managers/Supervisors

*Event Description:*

This program will highlight forensic applications being used in the battle field, in the identification of human remains and in other interesting forensic cases.

**Event Level:** Intermediate

**Objectives:**

- Explain current topics of interest in human identification.
- Identify the latest technologies that aid in the solution of difficult forensic problems.
- Discuss approaches currently being used to solve forensic cases.

**Session: Relationship Testing SIG II:**

**Held: Saturday, Oct. 9, 2010  
2:00 pm – 5:30 pm**

**Director/Moderator:** Mary Mount, MT(ASCP)

**Speakers:** George C. Maha, JD, PhD, MT(ASCP); George Riley, PhD; Michael Baird, PhD

**Intended Audience:** Physicians, Scientists, Technologists, Nurses, Managers/Supervisors

**Event Description:**

This session covers administrative actions affecting Relationship Testing Laboratories. It will include a discussion of the process used to develop the next edition of Standards for Relationship Testing Laboratories. It will include a review of 2009 Annual Report and previous CAP PAR/PARF Surveys. The progress being made in the determination of linkage between FESFPS and Penta E will be presented. Non-conformances issued since the 2009 AABB Annual Meeting will also be discussed.

**Event Level:** Basic to Intermediate

**Objectives:**

- Describe process for developing the next addition of Standards for Relationship Testing Laboratories.
- Review 2009 AABB Relationship Testing Laboratory Annual Report and CAP PAR/PARF Survey results.
- Discuss the progress made in the determination of linkage between FESFPS and Penta E.
- Discuss non-conformances issued since the last Annual Meeting and what corrective action might be taken to address the non-conformances.

**The AABB Relationship  
Testing Annual Report  
Summary**

**To obtain a FREE copy**

**JUST CLICK:**

[http://www.aabb.org/Content/Accreditation/Parentage\\_Testing\\_Accreditation\\_Program/ptpr og.htm](http://www.aabb.org/Content/Accreditation/Parentage_Testing_Accreditation_Program/ptpr og.htm)

# GREAT RESOURCES

- ✚ The AABB Career Link is a good way to bring together great job opportunities and great candidates. To find out more information visit the following link:  
[http://www.aabb.org/Content/Professional\\_Development/CareerLink/careerlink.htm](http://www.aabb.org/Content/Professional_Development/CareerLink/careerlink.htm)

- ✚ You can obtain a list of Accredited Relationship Testing Laboratories at the following link:  
[http://www.aabb.org/Content/Accreditation/Parentage\\_Testing\\_Accreditation\\_Program/AABB\\_Accredited\\_Parentage\\_Testing\\_Laboratories/aboutptlabs.htm](http://www.aabb.org/Content/Accreditation/Parentage_Testing_Accreditation_Program/AABB_Accredited_Parentage_Testing_Laboratories/aboutptlabs.htm)

## Did you know ?

1) Additional questions or uncertainties regarding any standard can be submitted to the Relationship Testing Accreditation Program Unit for review as a topic for the newsletter and/or educational topic at the National AABB Meeting. Forward topic suggestions to [nikkib@aabb.org](mailto:nikkib@aabb.org)

2) Questions encountered during an onsite assessment can be addressed immediately by calling 301.215.6492.

## Misleading Claims of Accreditation and Logo Misuse

With the explosion of advertising on the internet, there has been increasing misuse of AABB's trademarked logos and misleading claims of AABB accreditation. We are renewing our efforts to stop such practices and are actively searching out these organizations so that we can address this problem on a more global scale. These efforts benefit accredited laboratories by preserving the strong value of AABB accreditation and by ensuring that customer attention is focused on laboratories that actually *are* accredited. Our facilities work hard to achieve and maintain accreditation and deserve the maximum benefit of that accreditation. Increased vigilance will also benefit laboratories' customers by ensuring that they get the accredited-laboratory test that they have paid for. You can aid these efforts by bringing to our attention instances of logo misuse or misleading statements regarding accreditation. Please advise AABB's Accreditation Department ([accreditation@aabb.org](mailto:accreditation@aabb.org)) by providing the offending Web site and briefly describing the issue. It would be particularly helpful if you copy and email the actual link from your browser's address bar, as some offending organizations maintain multiple Web sites. The AABB Trademark Usage Guideline can found on the AABB Web site under About the AABB/Governance and Policies.

# WANTED

## RTAPU or RTSPU Member

Are you currently an assessor? Would you like to be involved in planning for sessions at the AABB Annual Meeting? Would you like to review corrective action plans for process non-conformances? Would you like to be involved in the newsletter? If these issues are of interest to you, the **Relationship Testing Accreditation Program Unit** would like to have you as a member.

Are you currently an AABB Member? Would you like to be involved in creating and revising the Relationship Testing Standards? Would you like to review the requests for variance from the Standards? Would you like to be involved in creating and revising the Guidance for the Standards? If these issues are of interest to you, the **Relationship Testing Standards Program Unit** would like to have you as a member.

Please contact Nikki Bass at the AABB National Office at [nikkib@aabb.org](mailto:nikkib@aabb.org).

### 2009-2010 RTAPU Members

Mary Mount, MT(ASCP)  
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Karen S. Miller, MT(ASCP)  
Lloyd Osborne, PhD  
Michael L. Baird, PhD  
George Maha, JD, PhD  
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Mary Mount, MT(ASCP)  
David Einum, PhD, BS  
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Neils Morling, MD, DSc  
Dara Hofman, Esq  
Board Representative:  
Thomas Lane, MD

## Articles

Do you have an interesting case or question you would like to share through this newsletter? Or is there a topic or issue you would like us to write about? Email us at [nikkib@aabb.org](mailto:nikkib@aabb.org)

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