aaBB Accreditation Relationship Testing News

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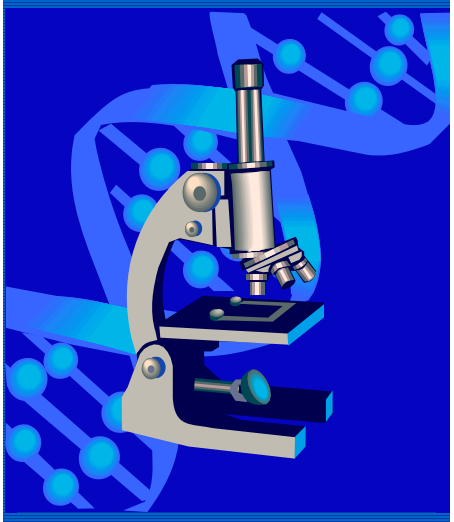

Maternal Contamination of Buccal Swabs Collected from Breast-fed Infants

Melissa D. Kahsar, PhD - DNA Diagnostics Center

Sample contamination, resulting in a mixed DNA profile, is sometimes an issue in paternity testing. When it occurs, analysis of the mixed DNA profile can be difficult, if not impossible. A common source of contamination is maternal cells, occasionally seen in maternal contamination of an amniotic fluid or chorionic villi sample. In this situation, the DNA from the prenatal sample generates a mixed STR profile containing both alleles of the mother in addition to the one allele inherited from the father.

Another cause of maternal contamination of a child's DNA sample is possible when a buccal swab is collected from a breast-fed infant. We now report a case of a mixed DNA profile in the mucosal cells of a breast-fed infant from a sample submitted for paternity testing. Visual analysis of the peak heights suggested that the proportion of mother to child DNA was approximately 50%. Subsequent conversation with the mother indicated that she did indeed breastfeed her infant immediately prior to entering the collection facility. The child was recollected and from the new sample was generated a single DNA profile.

Babovic-Vuksanovic et al. (1999) found maternal cells present in approximately 48% of buccal samples collected from infants within 5 minutes of breastfeeding. The number of contaminated samples decreased with time. Approximately 17% of buccal samples at 30 minutes after breastfeeding and 4% of buccal samples at 60 minutes or more after breastfeeding contained maternal cells. Furthermore, wiping the inside of the cheek of a breast-fed infant with a cotton swab significantly reduced the number of maternally contaminated buccal samples collected 5 minutes after breastfeeding (48% vs. 21%), although did not eliminate them. Guidelines for buccal swab collection in breast-fed infants were proposed. These include waiting at least 60 minutes after breastfeeding and cleaning the buccal mucosa to decrease the number of maternal cells before collecting a buccal sample.

In This Issue

Do you have an interesting case or question you would like to share? Email us at nikkib@aabb.org

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In our experience, maternal contamination of a child's buccal swab sample collected for paternity testing rarely occurs, likely due to sufficient time lapse between breastfeeding and sample collection. Therefore, changing collection protocols to include queries about potential breastfeeding may not be advisable. However, when maternal contamination is present in the sample of an infant, the subject of breastfeeding might be discussed with the mother prior to redraw in order to avoid a recurrence.

Reference

Babovic-Vuksanovic, D., V.V. Michels, M.E. Law, R. Bailey, W.A. Wyatt, N.M. Lindor, S.M. Jalal. 1999. Guidelines for buccal smear collection in breast-fed infants. *Am. J. Med. Genet.* 84: 357-60.

Clarification of AABB Relationship Testing Standards 5.1.2.4 and 5.1.2.5

There has been some confusion regarding the difference between Standards 5.1.2.4 and 5.1.2.5, and in which situations they apply. Both standards address situations where some or all of the genetic systems (loci) used for a test have no formal graded proficiency testing available. Standard 5.1.2.4 applies when there is no formal graded proficiency test available for <u>any</u> of the loci used to report the test results. Standard 5.1.2.5 applies when there is no formal graded proficiency test available for only <u>some</u> of the loci used to report the test results, but there is formal graded proficiency testing available for the other loci used. In both cases the laboratory must choose one of three options to demonstrate proficiency for the loci for which formal graded proficiency testing is not available: 1) test on a monthly basis known samples from when graded proficiency testing was available, 2) test on a monthly basis a standard trio of samples developed from persons of an undisputed relationship, or 3) participate three times a year in a sample exchange program. In the case of Standard 5.1.2.4, where there is no formal graded proficiency test available for <u>any</u> of the loci used to report the test results, Standard 5.1.8.1 also applies, which requires the laboratory to store the samples tested (if available) for as long as the records of that case are kept. The point is to ensure that it would be possible to confirm test results obtained when external proficiency testing is not available for any of the genetic systems used to report results.

MONTRÉAL 2008
ANNUAL MEETING & TXPO
OCTOBER 4–7

ANNUAL MEETING RECAP

- ✓ Presentations were given by Dr. Michael Baird about the use of additional loci to resolve difficult relationship cases.

One set of additional loci examined a multiplex of 12 autosomal markers. Cases were presented where these additional loci were able to provide genetic inconsistencies in order to exclude an alleged biological relationship or increase the combined paternity index in order provide support of a biological relationship. Another set of genetic markers examined a multiplex of 8 loci found on the X-Chromosome. These non-autosomal markers were divided into 4 linkage groups; two loci per linkage group. Cases were presented where the genetic inheritance of these X-Chromosome markers were helpful in determining whether a biological relationship existed.

- ✓ Dr. Anthony Carter, Laboratory Director at Sorenson Genomics/Identigene gave an overview of Genetic Ancestry Testing.

Ancestry is the second largest hobby behind gardening. People have always been interested in their origin of identity and many have pushed the field forward leaps and bounds in the last few years. Before genetic testing became available, recreating ancestral roots was only possible by using physical records and building family trees was entirely dependent upon the accuracy and availability of supporting documentation. Today there are a number of extended genetic analyses that can be performed to support the evidence of ancestry using the Y chromosome (STRs), mitochondrial DNA (sequence) as well as autosomal (SNP, STR testing and mobile elements). These tests enable individuals to not only build family trees but also perform global searches for other unknown relatives through the use of large genetic databases. With the use of surnames plus genetic profiles, family groups can become aware of each other even when there have been decades and miles of separation.

Profile matches are not the only information that is gained through ancestral marker testing. Ancestral origins dating back many hundreds of years can be uncovered through the designation of haplogroups. Y and mitochondrial haplogroup information is also important to the academic community because it gives weight to theories involving human migration. Novel tools of ancestry are now being developed at a rapid rate which affords anyone who is interested, the opportunity to discover more about their ancestral past than ever before in human history.

IF YOU MISSED THE MEETING IN 2008 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- 2007 SESSIONS ARE STILL AVAILABLE**

- ✓ Mary Mount, as Chair of the Relationship Testing Accreditation Program Unit (RTAPU), gave a presentation discussing CAP Survey Results for 2007-2008.

The CAP survey has had great participation. The number of labs performing STRs, both auSTRs and Y-STRs has nearly replaced all other testing. The child's filter paper sample was contaminated with mother's samples for PARF-C 2007. The vendor's procedure has been modified to prevent this from happening in the future. One lab reported HLA-A and HLA-B results for PARF-A 2008. The overall outcome of the 3 surveys can be found in the tables below.

PARF-C 2007 (93 Labs)

	Included AF	**Excluded AF**
Correct Results	100%	98.9%
		1 lab did not test this AF

PARF-A 2008 (97 Labs)

	Included AF	**Excluded AF**
Correct Results	100%	100%

PARF-B 2008 (96 Labs)

	Included AF	**Excluded AF**
Correct Results	99%	100%
	1 lab stated more testing needed	

The published response to the paper challenge for PARF-A 2008 was questioned by a number of people based on the calculated likelihood rations published in PAE/PARF-A Summary 2008 for D5S818, vWA and D18S51. The paper challenge scenario is as follows: A mother has 3 children. The father acknowledges a daughter and a son but the 3rd child is in questions and the father is deceased. The DNA profiles for the mother and 2 uncontested children yield a reconstructed profile that can be used to determine if the 3rd child could be the deceased father's child. The calculations submitted by CAP do not follow the calculations published in Appendix 7 starting on page 121 of the Guidance for Standards for Relationship Testing Laboratories, 8th Edition. Dr. George Maha wrote to CAP and submitted calculations based on Appendix 7. The calculations for D5S818, vWA and D18S51 are listed below.

	Wife	**Son**	**Daughter**	**Alleged Child**
D5S818	**11,12**	**11,12**	**11**	**11,12**
	p = 11 = 0.312		**q = 12 = 0.290**	**r = 1-p-q**
	$\text{Correct LR} = \frac{(3p+3q+1)}{2(p+q)(p+q+1)} = 1.4548$			$\text{CAP LR} = \frac{1+p+q}{2(p+q)} = 1.3305$
vWA	**16,17**	**17,18**	**16,18**	**16**
	$\text{Correct LR} = \frac{1}{2(r+1)} = 0.4496$			$\text{CAP LR} = \frac{p}{2p} = \frac{1}{2}$
D18S51	**12,14**	**12,14**	**12,14**	**14,22**
	$\text{Correct LR} = \frac{1}{2(p+q+1)} = 0.3771$			$\text{CAP LR} = \frac{r}{2r} = \frac{1}{2}$

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**

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

GREAT RESOURCES

- The Relationship Testing Standards Committee has completed the proposed changes for the 9th Edition of Standards for Relationship Testing Laboratories that will become effective January 1, 2010. The proposed Standards are currently available for public comment on the AABB web site at http://www.aabb.org/. Now is the time to voice your opinion. Once completed, the 9th Edition will be available at the AABB Annual Meeting in New Orleans along with the Guidance Document and the updated assessment tool.

- 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

- The Quality Systems Subcommitee works with the Accreditation Program Committee to fully implement quality principles into AABB's Accreditation Program and to provide quality tools for AABB institutional members. Currently the committee has been tasked with developing a resource, a pocket guide, to be used as a guide to help with developing, reviewing, updating, and assessing a facility's Quality Program. It will provide helpful hints for both a new facility seeking initial accreditation as well as information for the long-time member facility. The project is in the initial stages so watch for further updates in future newsletters.

- 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

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 (accrediation@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.

Assessor Continuing Education

If you are an AABB Assessor please take time to check on your continuing education. Per Accreditation Program Policy 5.1.1.f, (appendix 5.1.1.f.B), all assessors are required to submit a biennial continuing education report. Approved assessors with odd numbered membership IDs are required to submit continuing education credit hours by March 31, 2009. If your membership ID is an even number and you have not already done so, please submit your CE report as soon as possible as it was due November 2008. Specific information on the continuing education program and the policy can be found in the AIM (www.aabb.org > Members Area > Accreditation > Accreditation Information Manual > 5.1.1.f.B Assessor CE). A copy of the CE form is also on the AABB Web Site under the assessor section. Please be aware that failure to submit the required information may result in removal from the assessor program.

Please note that you can receive credit for viewing to the sessions presented at previous annual meetings through the AABB Live Learning Center. As an assessor, you will not be charged a fee to access the sessions.

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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 Pam Lubel at the AABB National Office at plubel@aabb.org.

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