



## In This Issue

- Chimerism Chaos in a Paternity Test
- Direct Sibling to Sibling DNA Testing for USCIS Cases
- Free AABB Workshop @ ISHI27
- RT Collector Training Flyer
- Great Resources
- Volunteer Opportunities

Do you have an interesting case or question you would like to share? Email us at

[nikkib@aabb.org](mailto:nikkib@aabb.org)

## Chimerism Chaos in a Paternity Test

A thirty-something infertile couple chose to undergo assisted reproductive technology (ART). The woman delivered a single healthy child, but the couple noted that the child's ABO blood group was AB while their own groups were both A, causing them to question the origin of the B allele. A home DNA paternity test indicated an exclusion of the male parent as the biologic father of the child, raising the possibility that the wrong sperm sample had been used during the insemination procedure at the fertility clinic. The paternity test was repeated using STR genetic tests at an AABB-accredited laboratory. Test results also indicated non-paternity.

The fertility clinic launched an internal investigation, and provided records that the male partner was the only Caucasian donor on the day of the insemination procedure. The couple and their child all appeared to be Caucasian, but the man had "cutaneous mosaicism" (patches of skin with two different tones) with a linear pattern of pigmentation that followed the embryonic lines of Blaschko. At the advice of coauthor DBS, an ancestry study was pursued. Its study of many SNP loci suggested that the excluded man bore an avuncular (uncle/nephew) relationship to the child. Since the man had no brother living in close proximity, and considering the man's unusual bi-toned skin, he was suspected of being a chimera. To confirm this hypothesis, follow up testing arranged by coauthor KMS at DDC Laboratories, overseen by coauthor MLB. Semen collection and STR tests revealed evidence of chimerism when more than 2 alleles per locus were seen at loci on multiple chromosomes. The proportion of each twin's seminal DNA was estimated from the two different heights of fluorescent allele peaks after PCR, inferring the twin that sired the child appeared to contribute 10% of seminal spermatozoa.

**Chimerism** is a condition in which an individual's body cells are derived from two zygotes with different genomes. In essence, a "natural" chimera is a composite of dizygotic (DZ) twins. A chimera is thought to arise by fusion of four gametes (2 ova + 2 sperm) into one early zygote (a "tetragametic" chimera) or by migration of embryonic cells from one DZ twin to the other. Migration may be via the twins' mother and uni- or bi-directional. In experimental mammals, migrating cells remain multi-potent (differentiate into >1 kind of tissue) until the blastula stage of embryogenesis. Human chimera usually remain unnoticed. Some have been suspected clinically when: 1) the fused DZ twins are of different sex and show evidence of ovotesticular disorder of sexual development (formerly termed true hermaphroditism and true intersex), 2) a transfusion service finds "mixed field" agglutination of erythrocytes during pre-transfusion serologic testing, 3) a karyotype shows more than 2 heteromorphic banding patterns per

chromosome, 4) there is skin pigment (or eye color) mosaicism, 5) a highly heterozygous locus (e.g. HLA) reveals more than 2 alleles, and 6) evidence of non-maternity turns up unexpectedly (because true non-maternity is extremely rare). On the other hand, non-paternity is frequent among civil paternity cases so that false exclusions of alleged fathers are problematic. In fact, the paternity case reported here is the first documented one with an alleged father with confirmed natural chimerism. (*Post-transplant* chimeras have been seen.) If the possibility of a chimera arises in a contentious paternity case, a test of the alleged father's (AF's) semen might provide the evidence of his paternity, but the absence of evidence of paternity cannot rule out that the AF is a *microchimera* with too few sperm from one twin to reach the signal threshold of the lab's DNA method.

The frequency of natural human chimerism is unknown. Estimates as high as 8-10% have been proposed, but there is no evidence yet that supports those figures. The frequency of chimerism may increase with greater use of assisted reproductive technology because ART procedures increase DZ twinning. When two cell populations coexist in one corpus, their tissue distribution is uneven and sampled tissues may contain the cells of only one twin. Furthermore, one tissue may not be representative of another. (Commonly sampled buccal cells have never shown evidence of natural human chimerism in hundreds of thousands of paternity cases using PCR, but dozens of chimera have been found in blood group tests using less sensitive serologic methods.) Finally, the number of cells of one twin may be too few to detect by the usual lab methods.

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\*Details of the case summarized here are to be published in a peer-reviewed journal. A forthcoming AABB audio conference will address the subject of chimerism more extensively.

## Direct Sibling to Sibling DNA Testing for USCIS Cases

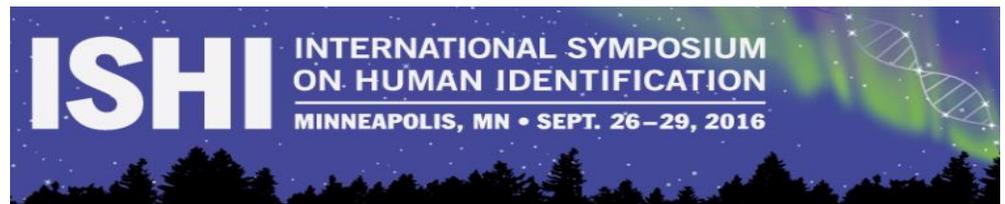
On March 29<sup>th</sup>, the U.S. Department of Justice Board of Immigration Appeals decided on the acceptance of sibling testing; “Direct sibling-to-sibling DNA test results reflecting a 99.5 percent degree of certainty or higher that a full sibling biological relationship exists should be accepted and considered to be probative evidence of the relationship.” The USCIS is drafting a new policy on direct sibling to sibling testing which we will forward to the accredited labs when released. More information concerning this ruling can be found on the following websites.

<https://www.justice.gov/sites/default/files/pages/attachments/2016/03/29/3860.pdf>

<http://www.aila.org/infonet/amicus-brief-matter-of-ruzku>

The current laws and the Foreign Affairs Manual can be found at:

<https://travel.state.gov/content/visas/en/law-and-policy/laws-and-regulations.html>



### *Free AABB Workshop at ISHI*

**Sunday, September 25<sup>th</sup> 8:00 am to Noon**

**This workshop is presented compliments of AABB  
Advance registration required. Register at [ishinews.com](http://ishinews.com)**

Workshop topics to include:

- Accreditation Program Updates
- Common Nonconformances of the 12th Edition
- CAP Proficiency Testing
- Complex Calculations
- Validation of Brutus Software for Complex Calculations
- USCIS and State Department Q & A





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**Registration Fees\***

December 23, 2015 – December 31, 2016: \$99

January 1, 2017 – June 30, 2017: \$59

July 1, 2017 – December 31, 2017: \$29

## The AABB Relationship Testing Annual Report Summary

To obtain a FREE  
copy

**JUST CLICK:**

<http://www.aabb.org/sa/facilities/Pages/RTestAccrFac.aspx>

## GREAT RESOURCES

- ✚ Access FREE Recording of the previously held (Nov. 17<sup>th</sup>) audio conference titled Changes from the 11<sup>th</sup> Edition to the 12<sup>th</sup> Edition of Standards at the following link:  
<https://www.youtube.com/watch?v=oF7dYzXiB7g&feature=youtu.be>
- ✚ AABB has launched the 2<sup>nd</sup> Edition of the Collector Training certificate course for individuals who would like to be recognized as AABB-trained sample collectors for relationship testing purposes. Find more information at the following link:  
<http://marketplace.aabb.org/EbusPPROD/Default.aspx?TabID=55&productId=12970612&>
- ✚ Standards for Relationship Testing Laboratories for 12<sup>th</sup> Edition is now available. You can purchase a copy at:  
<http://marketplace.aabb.org/ebuspprod/Marketplace/AllProducts/ProductDetail.aspx?productId=12878771>

### **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 at the following link:  
<http://www.aabb.org/membership/governance/Pages/default.aspx>

## **WANTED** **Audio Conference Content**

Would you like to repurpose your old talks or presentations?

Share your content as part of AABB's 2016-17 RT Audio Conference Series.

Please let us know if you have given a talk or presentation in the last 2-3 years on a topic that you think may be of interest to the relationship testing community. Topics of interest may include but are not limited to calculations, new technologies, expert systems, court room basics, forensics, DNA etc. If you decide to submit your content, you can choose to moderate the audio conference or we can assign a speaker for you.

For more information or to submit your content, contact Nikki Bass at [nikkib@aabb.org](mailto:nikkib@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](mailto:nikkib@aabb.org)

### **RTAPU or RTSPU Member**

- ❖ Are you interested in ensuring that assessment/audit procedures are in consistent with AABB policies established by the AABB Accreditation Program Committee?
- ❖ Are you interested in working with U.S. Citizenship and Immigration Service and/or the Dept. of State as it relates to RT?

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 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. To get involved, please contact Nikki Bass at the AABB National Office at [nikkib@aabb.org](mailto:nikkib@aabb.org)

#### **2015-2016 RTAPU Members**

**Robert Wenk, MD Chair**

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