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Dealing With Less Conclusive Results James Anstead, PhD DNASolutions

There are several circumstances when you might find yourself receiving and/or having to explain inconclusive or less than conclusive results to a client and it's helpful to have a plan when you enter that conversation. We often see statistically inconclusive results in second degree relative tests such as half-siblings and avuncular tests and even when tests meet the criteria for supported or not-supported there are occasions when clients may give us reason to investigate those tests further, especially if the support isn't very strong.

The first thing I always advise clients is to review the test, particularly if it's a complex family reconstruction, and determine if the relationships defined in the test are as expected. As discussed in the last issue, terms such as uncle and cousin are open to interpretation and sometimes a discrepancy between the client's description and the laboratories interpretation can emerge (a sketch of a family tree can be very helpful for this discussion). If everything is in order, then there are several approaches that can be taken to increase the discriminating power of the test (making an inconclusive result less likely). Bear in mind that you may incur additional fees for some of these options, so always discuss with this with your vendor of choice.

1. Adding additional people to the test:

This is a very effective way to increase the power of the test but not all additions are equally useful (or available) so I always advise people to discuss which would be the most effective addition with their lab. Just to give an example, suppose we had conducted a Half-sibship test where the father was in question, adding one or both of the known mothers to the test will greatly increase the likelihood of a definitive result.

2. Adding other markers, either more STRs, Y-STRs or mitochondrial markers:

Different laboratories use different numbers of autosomal STR markers (these are the standard

relationship markers). Occasionally it may be helpful to add extra markers to a test to increase its power, although many laboratories are already using the maximum number of markers they have available. In some limited circumstances sex-linked markers such as Y-markers (male lineage) and mitochondrial markers (female lineage) may provide extra information. However, these are only useful in cases where an unbroken male or female lineage links two individuals.

3. Newer testing modalities:

There are newer types of testing being utilized in forensic relationship testing such as large panels of Single Nucleotide Polymorphisms and Next Generation Sequencing – I won't go into detail in this article, but in some cases, these can be useful for more intransigent cases (consult your lab. for details).

Finally – Sometimes no matter how we try, the relationship in question may not be conclusively confirmed or denied by DNA testing. DNA testing should only be considered one part of an investigation into relatedness. Family records, oral history and other types of evidence may be just as important and always remember, whatever else it does. DNA testing doesn't define who our family might be.

Become an AABB Assessor Accreditation Staff AABB

AABB assessors reflect the AABB membership at large and are highly educated, with a minimum of a bachelor's degree, and a minimum 2 years in the field, and hail from around the world.

Many assessors perform casework at their primary job and often conduct their education and volunteer work as an assessor during non-work hours. The assessor program requires the assessor to complete a minimum of two assessments per year, as requested. Annual assessor training is provided by AABB and a Continuing Education Report, which documents 65 hours of discipline-specific continuing education, must be submitted every two years to maintain assessor status.

The AABB Accreditation Assessor Program offers accredited facilities a cost-effective solution for professional development of staff, while enhancing a facility's quality assurance program. The program provides professional training in auditing techniques, quality program and operations, and the competitive, legal and ethical issues associated with assessments. With this training assessors are able to evaluate a facility's quality and operational systems to determine whether the service they provide is appropriate and under control. The program provides both the assessors and the facility educational exposure to new ideas with regard to quality systems.



AABB's Assessor Training Program is accredited by IEEA and is one of only a few accredited assessor training programs in the world.

Qualifications

All AABB assessors must be staff members at an AABB-accredited facility and active individual members of AABB. Review the <u>assessor qualifications</u>, <u>requirements</u>, <u>responsibilities documents</u> carefully to determine if you qualify. Additional qualifications include the following:

- 1. Minimum of a BA/BS degree in biological science.
- Minimum 3 years direct (hands on) work experience (a portion of which has occurred in the last 3 years)
 OR a minimum of 3 years direct work experience, which occurred over 3 years in the past AND current
 active involvement in a related field that allows an individual to keep informed of scientific, technical, and
 accrediting requirements of the activity.
- 3. Understanding of quality programs, audit practices or other quality practices, through either actual experience or through training.

 Active participation (through either a managerial or technical position) in managing a quality program, performing assessments, or having responsibilities for quality systems, process control or other qualityrelated practices.

Commitment

New AABB assessors complete an AABB-led training program and a training assessment before becoming Assessors. All AABB assessors are required to participate in at least two facility assessments per year, as requested. They must also participate in the continuing education program.

Assessments may vary in length from 1 to 3 days. Assessors perform a desk assessment prior to the on-site assessment. During the assessment there may be some preparation required in the evenings. Travel time to and from the location may also be required.

Review the <u>financial and time commitment information</u> for more details.

Application Process

- 1. Complete the online Assessor Application.
- 2. Email your resume/Curriculum vitae and a letter of recommendation from your current employer to Accreditation@aabb.org

NOTE: As of Jan. 4, all AABB Assessors are required to be vaccinated against COVID-19 to participate in inperson assessments. Please review AABB's Vaccination Policy Update for more information.

AABB Relationship Testing is accepting inquiries from individuals who wish to volunteer for service on one of our committees. Positions of possible interest include the Relationship Testing Accreditation Committee and the Relationship Testing Standards Committee. Visit AABB's Volunteer Opportunities web page to apply.

Solicitation is also hereby made for written articles of interest, or suggestions for article topics, for the next issue of this Newsletter. Submissions may be made to Charles M. Kelly, PhD, and sent to ckelly@intelligenetics.com.

Advantages and Complications of Using Non-Autosomal Testing in Relationship Analysis: YSTR Testing Debra L. Davis, PhD DNA Diagnostics Center

In establishing paternity, direct testing of the alleged father or testing of both paternal grandparents is always the best option. These tests can either directly exclude a falsely accused alleged father or will provide a probability of paternity of greater than 99% when the alleged father is not excluded. Cases that involve indirect relationship testing, including testing with a single grandparent, avuncular, or siblingship tests will only provide a statistical likelihood that gives evidence to support the tested relationship. These are second- degree relationships and a test of this nature also will not directly exclude the relationship. However, if a sufficient number of second-degree relatives are tested (typically 3 or more) the alleged father's profile can potentially be reconstructed and the test may be just as informative as a direct paternity test. Unfortunately, a laboratory is frequently faced with only one possible relative to test. Such tests have a higher likelihood of the result being inconclusive which means, according to AABB standards, having likelihoods between 10 and 1/10. A case of this nature may be resolved by providing additional information from non-autosomal data such as from YSTR, XSTR or mitochondrial DNA testing.

If a male paternal relative of the alleged father is tested with a male child, YSTR testing may be useful. Y STR testing examines the contribution to the child of alleles on the male-specific Y chromosome. A father will pass his Y chromosome to his son unchanged (with the exception of a rare mutation). Since the Y chromosomes in any paternal line will be identical, the unrelated Alleged Paternal Relative can be excluded directly by this test. Therefore, YSTR testing is useful for excluding the unrelated male paternal relatives of a male child.

If there is a YSTR profile match between the two tested male parties, this is an indication the two male individuals may be of the same male lineage. Although non-matching YSTR profiles are sufficient to exclude two individuals from a paternal relationship, matching YSTR profiles alone cannot confirm a paternal relationship. The likelihood of a YSTR profile match can be obtained from a YSTR database. This likelihood value must then be combined with the likelihood of the relationship in question derived using autosomal STRs (used in standard paternity and kinship cases) following the appropriate calculations. Although a YSTR profile consists of multiple loci, the aggregate YSTR result must be treated as a single marker in the evidence of the relationship. Any single inconsistency between the YSTR profiles should be considered a mutation and the likelihood of the match adjusted appropriately. Thus, matching YSTR profiles may be useful to support relationship testing odds and increase the likelihood of the relationship just like any single autosomal STR marker.

However, if the YSTR result is inconsistent with the autosomal relationship result, the interpretation may not always be clear. A high combined autosomal relationship likelihood result accompanied by mis-matched YSTR profiles, may indicate a maternal relationship, an error in the described pedigree, or potentially an underlying relationship that has not been identified by the client. Thus, the parties may be closely related but not as described. Conversely, if the tested parties share common YSTR profiles but demonstrate a low likelihood of the relationship through autosomal testing, the YSTR result may indicate the parties are distantly related. However, this result could also indicate that there was just insufficient information from the autosomal test to support the relationship, but that relationship really does exist. In this scenario, the YSTR testing is a powerful tool to support a relationship that might otherwise be missed.

Interpretation of YSTR results can be tricky because YSTR profile matches often lead to high relationship index values. When combining a likelihood of a YSTR profile match with an inconclusive autosomal relationship index, the combined result may appear to provide strong evidence of the tested relationship that does not actually exist. This is particularly concerning when testing for a second- degree relationship where a more distant relationship is the more probable one, but it has not been suggested by the clients. An example might be if two paternal cousins allege themselves to be half siblings. A half-sibship test between the individuals in this scenario may yield an inconclusive likelihood of the relationship (Relationship index = 1/10 to 1). If a YSTR test is performed, the YSTR profiles will match, since the individuals share a common grandfather, and their fathers are brothers. When these likelihoods are combined, the result would appear to be conclusive and be a strong indication of paternal halfsibship relationship. However, the combined likelihood that includes the YSTR data would be an overstatement because there is a secondary underlying relationship. Therefore, the likelihoods must be evaluated separately, both with and without the YSTR result. A low or inconclusive likelihood of the relationship based on autosomal data, when combined with a YSTR match, may produce a result that supports the hypothesized relationship but there may also be other relationships that would produce a similar result. It is not the place of the testing laboratory to report on such alternative relationships unless their possibility is raised by the clients themselves. However, the report issued by the testing laboratory needs to include wording calling attention to the possibility of such unevaluated scenarios.



AABB is now the Association for the Advancement of Blood & Biotherapies.

Learn more about our new name and brand – and watch as we evolve throughout 2022.



