ANNUAL REPORT SUMMARY FOR TESTING IN 2010
Prepared by the Relationship Testing Program Unit

PREFACE

The annual survey provides information on the state of the relationship testing community and tries to ask questions that may be of interest and track trends in testing. As a reminder evaluation of these data is anonymous. None of the members of the Relationship Testing Standards Program Unit is aware of which laboratories submitted data. Presentation of the most current data occurs every year at the AABB annual meeting. Many of the laboratories report testing a broad range of cases, including relationship tests for routine paternity testing, immigration, prenatal evaluations, and post-mortem evaluations. Almost all of the laboratories reporting performed immigration testing, reconstruction (family study) cases.

As in the past this report AABB provides some commentary for laymen on common misconceptions in paternity testing. Some of the commentary is from previous year's report, as the commentary remains relevant to issues raised during the year. The Relationship Testing Standards Program Unit (RTSPU) would also like to remind readers that the *Guidance for Standards for Relationship Testing Laboratories*, discusses the *Standards* in some detail and provides suggestions on how to comply with the standards and contains explanations of the standards, various calculations used, and addresses other issues in relationship testing. The 10th edition of standards will go into effect on January 1, 2012 and the guidance document will be on a CD in the back of the standards. All accredited laboratories will receive a copy of the standards with the attached guidance CD. The RTSPU encourages all laboratories read the guidance document.

Sadly, many laboratories did not cooperate with this anonymous survey and either did not participate at all or refused to provide basic data. The annual report serves as an important tool to track trends and changes in the paternity testing community and is used read by government officials, reporters, the general public, and others seeking basic information on paternity testing. As such volume numbers in this report reflect opinions and data from about 60% of the paternity laboratories.

ANNUAL VOLUME OF TESTING

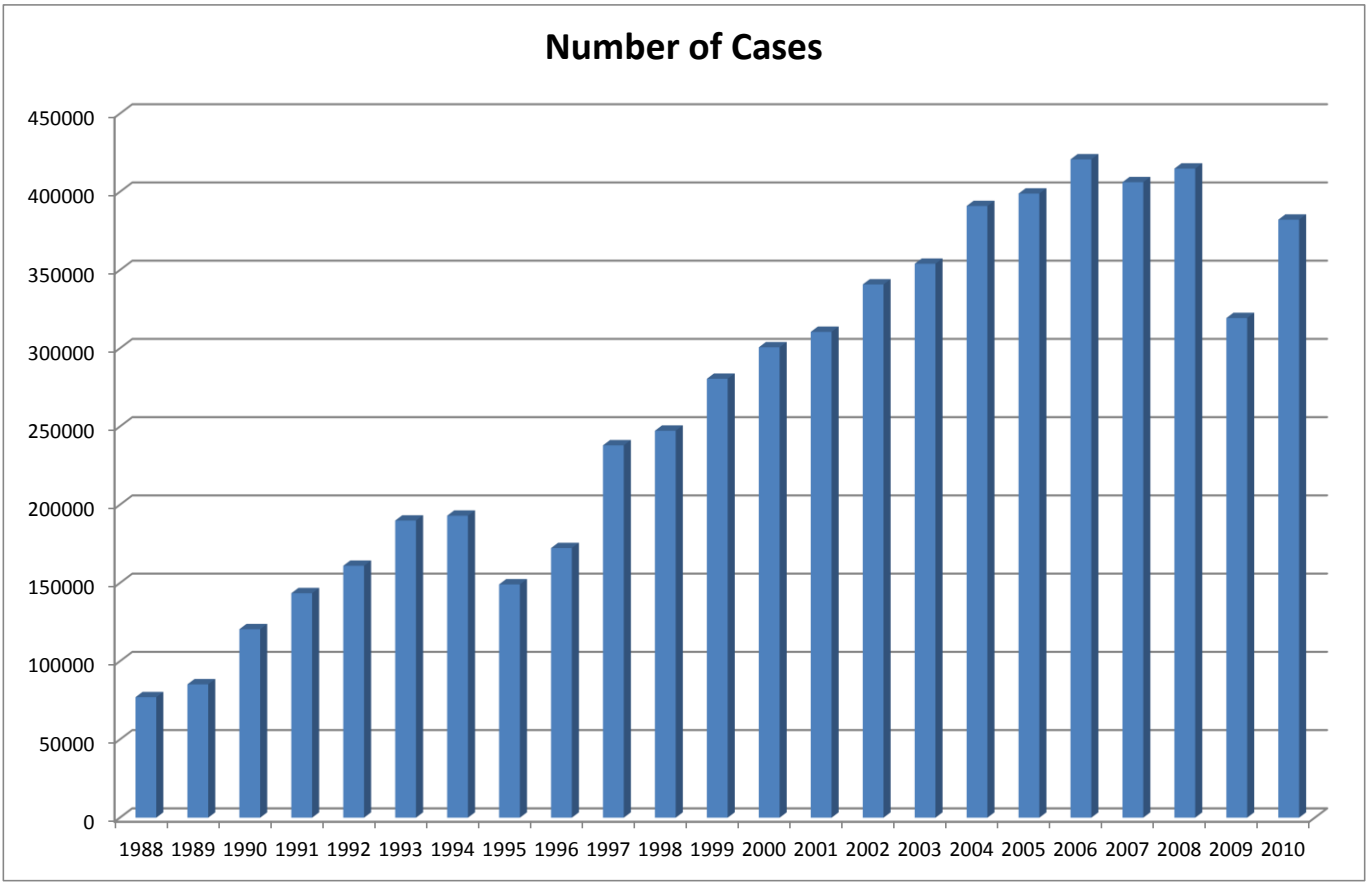
The volume reported for cases tested in 2010 was 382,199. As indicated many laboratories did not cooperate and this is an underestimate of the actual number of cases tested by AABB accredited laboratories. A summary of the total cases reported since 1988 is shown in Table 1 and Figure 1.

Table 1. The Number of Relationship Cases Reported for 1988-2010.

Year	No. of Cases	Year	No. of Cases
1988	77000	2001	310490
1989	85231	2002	340798
1990	120436	2003	354011
1991	143459	2004	390928
1992	161000	2005	398880
1993	189904	2006	420740
1994	193000	2007	406147
1995	149100	2008	414843
1996	172316	2009	319320

1997	237981	2010	382199
1998	247317		
1999	280510		
2000	300626		

Figure 1. Graph of the Case Volume for 1988-2010.



As in the past, laboratories were asked if they tested cases where the chain of custody did not meet the requirements of the *Standards for Relationship Testing*. The tested individuals, without a proper witness (see Standards), generally collect these so called "non-legal" tests. AABB has taken the position that it cannot prohibit accredited laboratories from performing these types of tests, but reminds laboratories that they cannot claim or advertise that their "non-legal" testing meets AABB standards. This includes reports that state the "testing" meets the standards and only the chain of custody is lacking. Laboratories can only conform in all aspects and cannot choose standards to which they will adhere. Of the laboratories reporting 54% reported that they performed testing of this type. Those laboratories reported 5,610 non-legal cases or 1.68% of the total cases reported. However some laboratories did not track the number of non-legal cases they evaluated or refused to provide the information. Of the laboratories performing non-legal testing, these tests account for 4.06% of their total volume.

LABORATORIES BY SIZE

Table 2 indicates the size of the various responding laboratories by volume of cases reported. Not all of the responding laboratories provided total volumes, and only 26 laboratories out of over 40 accredited laboratories. Note that this breakdown is by each laboratory, but a single corporation may own several laboratories.

Table 2. Laboratories by the Volume of Cases Reported.

Case Volumes	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
1-500	20	19	19	13	17	14	18	16	16	15	15	5	8	9
501-1,000	7	6	5	6	6	2	3	2	4	4	6	1	2	1
1,001-5,000	10	11	9	11	11	13	11	7	8	11	11	7	8	9
5,001-10,000	5	0	3	3	5	1	3	7	7	6	6	2	3	2
10,001-50,000	5	5	7	8	6	7	7	6	5	5	4	4	3	2
50,001 – 100,000	1	2	1	1	1	0	0	1	1	1	1	3	1	1
>100,000	0	0	0	0	0	1	1	1	1	1	1	1	1	2
Total Laboratories	48	43	44	42	46	38	43	40	42	43	44	23	26	26

EXCLUSION RATE

For 2010 some laboratories did not track the number of exclusions. For the laboratories tracking exclusions there were 364,587 cases completed and 90,656 (24.87%) were reported as exclusions. The average exclusion rate for the laboratories reporting exclusions is 20.44% with a standard deviation of 6.62. The median exclusion rate is 21.53% with a range of 8.52% to 30.73%. The explanation for the range of exclusion rates is complex but appears related to the laboratory's volume and client base. Anecdotal explanations for the various exclusion rates include differences with the type of case (private verses public contracts), and the geographic source of the case (rural versus metropolitan areas). For the non-legal testing, there were 838 exclusions from laboratories reporting exclusion data (total of 2,548 cases) or an exclusion rate of 32.89%, a higher percentage than the 23.07% seen for legal testing. The range for non-legal testing is 20.90% to 47.62%. For the legal tests the laboratories averaged 19.28% exclusions and for non-legal tests the laboratories averaged 35.03% exclusions.

MISCONCEPTIONS IN PATERNITY TESTING – EXCLUSION RATE

AABB has seen the exclusion rate misused by several organizations trying to claim that 30% of men are misled into believing they are biological fathers of children when the mother knows this not to be true. This view is incorrect. The exclusion rate includes a number of factors. One is a woman may allege several men as possible fathers because she was sexually active with these individuals. These are not men who were misled into believing they were fathers and then later discover they are not. The testing merely sorts out which man is the biological father and excludes the others. Another factor is that the unexcluded

alleged father, as part of his defense, will allege the mother had multiple sexual partners during the time of conception. These men are subsequently tested. Sometimes testing of a man is required because of a legal presumption. This is when the mother properly names the correct biological father but because the child is the product of a marriage (she is (was) married to someone other than the biological father) there is a legal presumption the husband is the father. The husband is tested to rebut the legal presumption even though no one believes he is the biological father of the child. There is no evidence that a large number of the men excluded in the testing were misled into believing they are the biological father of a given child.

COMBINED PATERNITY INDEX (COMBINED LIKELIHOOD RATIO)

The laboratories were asked to indicate what combined paternity index (CPI) they considered acceptable for cases with a standard trio (mother, child, father), single parent cases (mother (or father) not tested cases), and reconstruction cases (cases where the disputed parent is missing and other relatives are used to evaluate parentage). Some laboratories reported using different CPIs for different classes of clients (private verses public contracts, or for different technologies).

The results for the laboratories that responded are shown in Table 3. The most common minimum CPI for a standard trio is 100 with 48.15% of laboratories using this value, with a range of 100 to 10,000. For mother not tested cases the most common minimum CPI is 100 with 55.56% of laboratories using this value, with a range of 100 to 10,000. For the family study or reconstruction cases, 47.83% indicated that they report "whatever was obtained" and the majority considered a combined likelihood ratio of 101 or less reportable. Almost all laboratories considered a likelihood ratio of 100 or less as acceptable for sibling studies.

Table 3. The Number of Laboratories Using Various minimum Combined Likelihood Ratios for Standard Trios, One Parent (Mother (or father) not Tested (MNT)) and Reconstruction Cases (Note: not all laboratories indicated a CPI for each type of case).

	Choice of Minimum Likelihood Ratios Used by Type of Case (% Using)				
	Trio	**One Parent**	**Reconstruction**	**Full Sibling v. Unrelated**	**Half Sibling v. Unrelated**
What Ever is Obtained	**0.00**	**0.00**	**50.00**	**78.26**	**78.26**
1	**0.00**	**0.00**	**0.00**	**4.35**	**4.35**
5	**0.00**	**0.00**	**4.17**	**4.35**	**4.35**
10	**0.00**	**0.00**	**4.17**	**4.35**	**8.70**
25	**0.00**	**0.00**	**0.00**	**0.00**	**0.00**
100	**46.43**	**53.57**	**16.67**	**8.70**	**4.35**
101	**0.00**	**0.00**	**4.17**	**0.00**	**0.00**
150	**7.14**	**7.14**	**0.00**	**0.00**	**0.00**
200	**3.57**	**7.14**	**4.17**	**0.00**	**0.00**
500	**3.57**	**0.00**	**4.17**	**0.00**	**0.00**
1000	**14.29**	**14.29**	**8.33**	**0.00**	**0.00**
1001	**0.00**	**3.57**	**0.00**	**0.00**	**0.00**
2500	**3.57**	**7.14**	**4.17**	**0.00**	**0.00**

10,000	**21.43**	**7.14**	**0.00**	**0.00**	**0.00**

A common issue is the significance of the paternity index and the reliability of the AABB standard requiring a CPI of 100 to 1. The Relationship Testing Standards Program Unit is concerned about the meaning of the tests and thus the choice of the 100 to 1 for a reasonable level of significance. First and foremost, this level was chosen because it provides reasonable evidence of paternity in a standard case where a trio is tested. Generally, when a laboratory tests a case, if the disputed person is not excluded and does not reach the laboratory's minimum value, additional testing is performed to evaluate this person. This additional testing may result in non-exclusion, exclusion, or inconclusive reports. The view that AABB is only concerned with the performance of the testing, but not the meaning of the test, is incorrect.

Another issue arises with regard to performing other relationship analyses such as reconstruction cases, trios with genetic anomalies, and samples from exhumations, coroners, and postmortem testing. Importantly, note that a CPI of less than 100 is not an indicator of no relationship, unless 0 (or much less than 1), and may still in fact be a strong indicator of a relationship. Practical difficulties exist with the ability to obtain results from degraded samples, as happens in postmortem testing, and in the mathematical analysis of the relationships in reconstruction cases. Understanding this is particularly important for legislators who establish presumption levels based on paternity calculations, and contract administrators, who need to differentiate between reasonable science and what might be achieved under ideal conditions. The other important concept is that a laboratory's minimum combined paternity index, which may reflect scientific reality, is not necessarily the laboratory's testing goal or median combined paternity index. Most importantly laboratories using high minimums should not conclude that values under their minimum(s) are inconclusive, this is not acceptable as very low combined paternity indices are meaningful (see the AABB Guidance for Relationship Testing Laboratories for further discussion).

SIBLING CALCUALTIONS

Human identity labs are often called upon to help identify familial relationships in the absence of parental DNA, that is sibship testing. Sibship analyses, when submitting only two individuals for analysis, can be more demanding than parentage testing in that there are no obligatory alleles between siblings that make it possible to conclusively include or exclude the tested biological relationship. In addition, full siblings are as likely to share two alleles, identical by descent from common ancestors, as they are to share zero alleles at a given locus due to genetics. Thus a lack of shared alleles at any particular locus does not exclude a sibling relationship between two individuals. Many times additional loci will not necessarily help resolve a case.

The results for sibship analysis are expressed as a likelihood ratio and are often converted to a probability of sibship using an appropriate prior probability. Several small publications address the issues of sibship analysis and provide empirical data on the range of combined sibling-ship indices (likelihood ratios) encountered with individual pairs that are known full siblings, half-siblings, and unrelated.[1,2,3] In one study combined likelihood ratios for known full siblings ranged from 4.6 to over 1 billion and for random,

[1] Valentin, J. 1983. Positive Evidence of Paternity Calculated According to Essen-Moller: The Bayesian Approach. In Inclusion Probabilities in Parentage Testing. Ed. Richard H. Walker, M.D., pp 63-75.

[2] Reid, T.M., Wolf, C.A., Kraemer, C.M., Lee, S.C., Baird, M.L., and Lee, R.F. Specificity of sibship determination using the ABI Identifiler multiplex system. J. Forensic Science, 49: 1262-1264, 2004.

[3] Fu, J., Allen, R.W., Reid, T.M., and Baird, M. Considerations for the interpretation of STR results in cases of questioned half-sibship. Transfusion, 47: 515-519, 2007.

unrelated individuals from 0.000000045 to 0.12. There was no overlap between the group of true siblings and the group of unrelated individuals.[2] In a study of known half-siblings the combined likelihood ratio for known half-siblings ranged from 0.1 to 3763 with a median likelihood ratio of 24. The combined half-sibling indices for the unrelated pairs ranged from 0.0001 to 42 with a median likelihood ratio of 0.13. There is little overlap between the known half-siblings and unrelated pairs.[3] If a prior probability of 0.5 is correct, then a likelihood ratio of 10 to 1 (90% probability of a sibling relationship) may be considered reasonable evidence of either a full or half sibling relationship. There is need for further study.

TESTING WITHOUT THE MOTHER

There is still a strong concern about submitting disputed paternity cases without the mother. Testing without the mother presents a number of problems. First, the paternity index is, on average, cut in half. On average it appears that with mother not tested cases the combined paternity index is about one tenth that seen when the mother is tested. This also greatly reduces the ability to detect a falsely accused man, and in some cases, such as incest can easily produce false inclusions. When an apparent inconsistency (mutation) is present, it may not be possible to render an opinion of paternity without obtaining a sample from the mother. The mother is also an important QC step. If the mother is excluded it may indicate a problem in the testing. The testing of the mother may also allow for the detection of fraud, such as welfare fraud on the part of the mother or cases where the alleged father brings a child he knows is his, but in not the child of the mother. Thus, the testing of the mother, even if maternity is not disputed, is important in evaluating the questioned relationship, it improves the chance of obtaining clear results and is a quality control check for both the scientific and legal community. Testing without the mother should only be done when mother's location is unknown or she is deceased. Every effort should be made to test the mother.

TECHNOLOGY USE

In 2010 the survey showed that PCR based technologies now the technology of choice. Y Chromosome analysis was used in only about 0.12% of cases. Note that starting with the 9th Edition of Relationship Testing standards, standards for serologically tested red cell antigens, HLA serology, red cell enzymes, serum proteins, allotyping, and RFLP methods are no longer provided. These were dropped because of the lack of use or little use in the industry. Proficiency testing may be difficult to obtain and finding any laboratory to do comparison testing may be problematic. However, if a laboratory wishes to use these methods the laboratory can refer to the appropriate testing standards in the 8th edition. Note that the laboratories cannot used standards that have been superseded in newer editions, and the use of old standards applies only to technology no longer covered by the standard. Proficiency testing would need to meet the requirement of the current edition of standards.

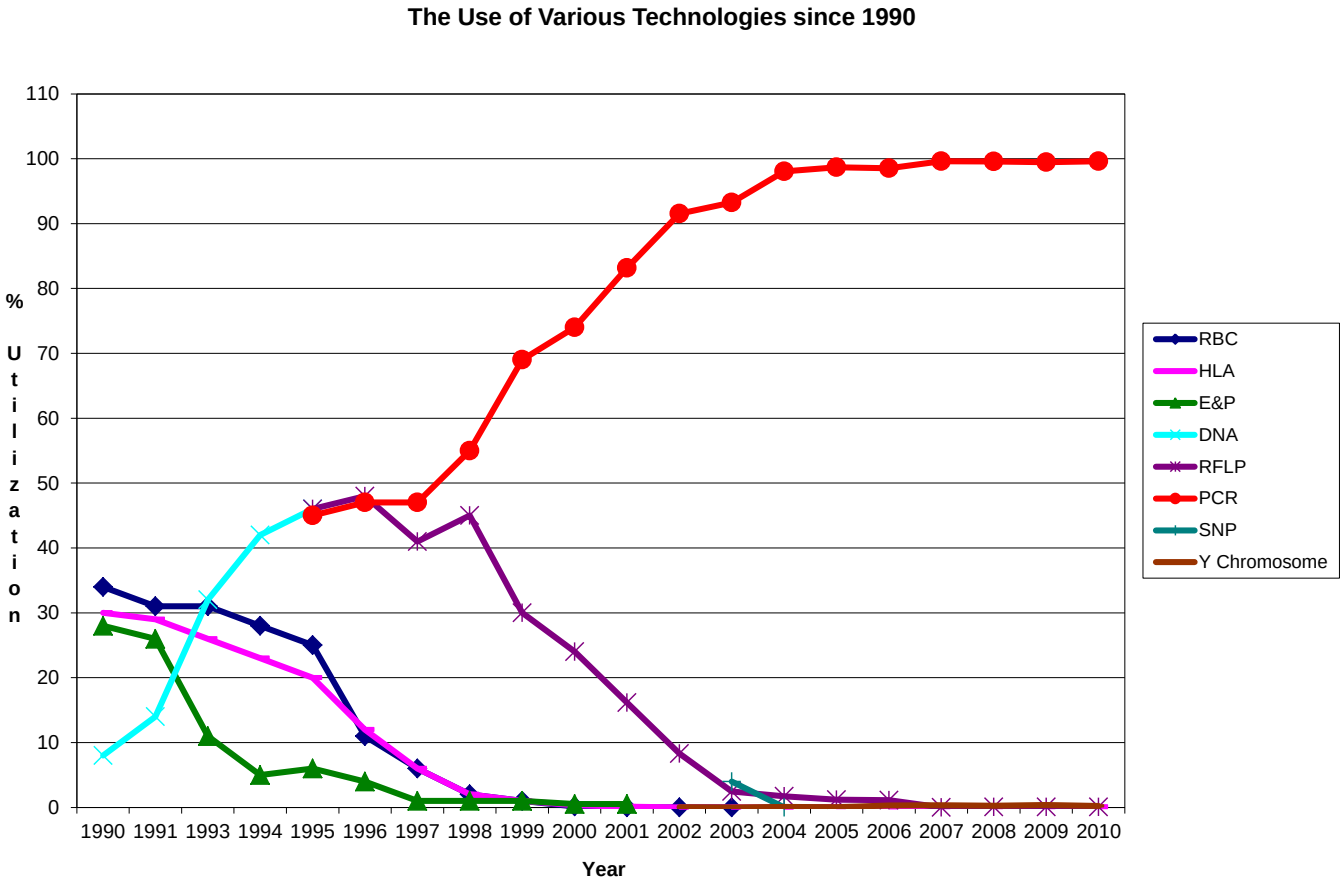
Table 4 provides a breakdown of the technology used to resolve the reported paternity cases. The three laboratories using HLA molecular methods were asked to identify the source of the frequencies. Laboratories using HLA molecular for Class I HLA methods reported using serologic tables for calculating paternity indices.

Table 4. The Technology Used in Cases Reported in 2010

Technology	Number of Cases	Utilization (%)
Red Cell Antigens	**0**	**0**
HLA Serology	**0**	**0**
HLA Class 1 Molecular	**157**	**0.04**
HLA Class II Molecular	**1**	**0.00026**
Red Cell Enzymes/Serum Proteins	**0**	**0**
Allotyping	**0**	**0**
RFLP	**307**	**0.08**
STR	**378272**	**99.76**
SNP	**0**	**0**
Y Chromosome	**457**	**0.12**
Total of All Technologies	**379194**	**100**

*Note that some cases used more than one technology.

Figure 2 shows the use of various technologies since 1990. As indicated above, the many commonly used technologies in 1995 (red cell antigens, HLA, RFLP, red cell enzymes and serum proteins) now account for 0.12% of all casework. The change in DNA technologies from RFLP to PCR technology is also obvious. Prior to 1995 the survey only asked about the use of DNA testing but not about which DNA technology was used (PCR verses RFLP). Note that in some cases multiple technologies were used in the same case.



SAMPLE SOURCE

Laboratories reported approximately 948,788 samples used for the casework in 2010. Not all laboratories reported the samples they used. Of these samples, buccal swabs account for 99% of the samples. Whole blood samples accounted for 0.23%. Various other samples were also reported (See Table 5).

Table 5. Sample Source in 2010.

Sample	Number	Percent
Buccal Swabs	948788	99.2839
Blood	2155	0.2255
Blood Spot Cards	2622	0.2744
Amniotic Fluid	655	0.0685

Misc. Tissues	122	0.0128
Paraffin Blocks	53	0.0055
Hair	67	0.0070
CVS	218	0.0228
Products of Conception	15	0.0016
Bone	183	0.0191
Teeth	1	0.0001
Total	955631	100

MUTATION CALCULATION AND FREQUENCIES

Single inconsistencies are routinely seen in the testing of paternity cases. If a laboratory comes to the conclusion that the inconsistency is a mutation, then the mutation result must be incorporated into the reported results. Laboratories were asked how they calculated the paternity index (PI) for these loci. The laboratories all appear to be using one of several calculation methods. Some laboratories are using, most commonly, use the mutation rate divided by the average probability of exclusion (62.69%) and some laboratories used Brenner's method (25.93%).

COMMON MISCONCEPTION – HOW MANY INCONSISTENCIES ARE NEEDED TO EXCLUDE A RELATIONSHIP?

The AABB standards indicate that laboratories may not exclude with one inconsistency without supporting evidence. Many laboratories have interpreted this to mean there is hard rule that if two inconsistencies are observed, the relationship is excluded. This is not correct. The guidance document cautions laboratories that double or even triple mutation can occur. The laboratory should be cautious in issuing a report as exclusionary with only two inconsistencies. There are also exceptions where one inconsistency may be sufficient. An example would be a reconstruction case where a single inconsistency is observed along with low residual likelihood ratio (low is generally considered < 1.0). The low residual likelihood ratio would be supporting evidence of no relationship. If a laboratory is uncertain about the status of a case, issuing an inconclusive report is an acceptable option. The reason the case is inconclusive should be stated in the report. See guidance document for further discussion.