PREFACE

The 2006 Annual Report includes respondents from accredited laboratories in the United States, Canada and Europe. Many of the laboratories report testing a broad range of cases, including relationship tests for routine paternity testing, immigration, as well as prenatal and postmortem evaluations. Almost all of the laboratories responding to the survey performed immigration testing and reconstruction (family study) cases. Two of the reporting laboratories indicated that they sent their cases to other laboratories for testing and, as such, the data presented here are from the remaining laboratories that performed the testing.

In this report, AABB provides some commentary for laymen on common misconceptions in paternity testing. Some of the commentary is from the previous year’s report, as the commentary remains relevant to issues raised this year.

On January 1, 2008, the 8th edition of Standards for Relationship Testing Laboratories went into effect. The Relationship Testing Standards Program Unit would also like to remind readers that the Guidance for Standards for Relationship Testing Laboratories, published in 2008, discusses the Standards in detail and provides suggestions on how to comply with the standards and contains explanations of the requirements, the various types of calculations used, and addresses other issues in relationship testing.

ANNUAL VOLUME OF TESTING

The volume reported for cases tested in 2006 was 420,740. Note that, for some laboratories, only the total number of cases reported is available. The statistics in this report therefore are based on a smaller number of laboratories, as indicated above. The total volume of cases represents an increase of 21,860 cases (or 5.48% increase) from the 2005 volume. A summary of the totals of all years since 1988 is shown in Table 1 and Figure 1.

Table 1. The Number of Relationship Cases Reported for 1988-2006.

<table>
<thead>
<tr>
<th>Year</th>
<th>No. of Cases</th>
<th>Year</th>
<th>No. of Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>1988</td>
<td>77000</td>
<td>1998</td>
<td>247317</td>
</tr>
<tr>
<td>1989</td>
<td>85231</td>
<td>1999</td>
<td>280510</td>
</tr>
<tr>
<td>1990</td>
<td>120436</td>
<td>2000</td>
<td>300626</td>
</tr>
</tbody>
</table>
In this report, laboratories were asked if they were testing cases where the chain of custody did not meet the requirements of the *Standards for Relationship Testing*. These so-called “non-legal” tests are generally collected by the individuals being tested, and are not “witnessed by a competent person with no interest in the test outcome” (as required by standard 5.2.2 in the *Standards for Relationship Testing Laboratories*, 8th edition.)

The *Standards* does not prohibit accredited laboratories from performing these types of tests, but because “non-legal” tests do not meet the requirements of *Standards*, laboratories cannot claim or advertise that their “non-legal” testing is encompassed by their AABB accreditation – regardless of whether the testing of the samples conforms to *Standards*. Particularly in relationship testing, the quality of the results depends on the testing just as much as it does on the integrity of the sample collection process. Of the laboratories reporting, over half (53%) reported that they performed “non-legal” testing. Those laboratories reported 19,582 non-legal cases or 5.55% of the total cases reported. Some

<p>| | | |</p>
<table>
<thead>
<tr>
<th></th>
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<tbody>
<tr>
<td>1991</td>
<td>143459</td>
<td>2001</td>
</tr>
<tr>
<td>1992</td>
<td>161000</td>
<td>2002</td>
</tr>
<tr>
<td>1993</td>
<td>189904</td>
<td>2003</td>
</tr>
<tr>
<td>1994</td>
<td>193000</td>
<td>2004</td>
</tr>
<tr>
<td>1995</td>
<td>149100</td>
<td>2005</td>
</tr>
<tr>
<td>1996</td>
<td>172316</td>
<td>2006</td>
</tr>
<tr>
<td>1997</td>
<td>237981</td>
<td></td>
</tr>
</tbody>
</table>

**Figure 1.** Graph of the Case Volume for 1988-2006.
laboratories did not track the number of non-legal cases they evaluated, but it seems appropriate to estimate that no more than 10% of all cases were of a “non-legal” type.

Note that during 2007, the Department of Homeland Security, United States Citizenship and Immigration Service (USCIS) met with the committee to discuss this and other issues. Because the sample collection in “non-legal” tests is not controlled, there is the potential for fraud. Accordingly, “non-legal” testing is not acceptable for immigration purposes.

LABORATORIES BY SIZE

Table 2 indicates the size of the various responding laboratories by volume of cases reported. Note that this breakdown reflects individual laboratories, but that a single corporation may own several laboratories. There appears to be a decrease in the number of laboratories reporting 1-500 cases per year.

Table 2. Laboratories by the Volume of Cases Reported.

<table>
<thead>
<tr>
<th></th>
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<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>1-500</td>
<td>40</td>
<td>26</td>
<td>25</td>
<td>20</td>
<td>19</td>
<td>19</td>
<td>13</td>
<td>17</td>
<td>14</td>
<td>18</td>
<td>16</td>
<td>16</td>
<td>8</td>
</tr>
<tr>
<td>501-1,000</td>
<td>6</td>
<td>4</td>
<td>8</td>
<td>7</td>
<td>6</td>
<td>5</td>
<td>6</td>
<td>6</td>
<td>2</td>
<td>3</td>
<td>2</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>1,001-5,000</td>
<td>7</td>
<td>9</td>
<td>6</td>
<td>10</td>
<td>11</td>
<td>9</td>
<td>11</td>
<td>11</td>
<td>13</td>
<td>11</td>
<td>7</td>
<td>8</td>
<td>8</td>
</tr>
<tr>
<td>5,001-10,000</td>
<td>6</td>
<td>4</td>
<td>3</td>
<td>5</td>
<td>0</td>
<td>3</td>
<td>3</td>
<td>5</td>
<td>1</td>
<td>3</td>
<td>7</td>
<td>7</td>
<td>5</td>
</tr>
<tr>
<td>10,001-50,000</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>5</td>
<td>5</td>
<td>7</td>
<td>8</td>
<td>6</td>
<td>7</td>
<td>7</td>
<td>6</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>50,001 – 100,000</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>&gt;100,000</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Total Laboratories</td>
<td>62</td>
<td>46</td>
<td>46</td>
<td>48</td>
<td>43</td>
<td>44</td>
<td>42</td>
<td>46</td>
<td>38</td>
<td>43</td>
<td>40</td>
<td>42</td>
<td>30</td>
</tr>
</tbody>
</table>

EXCLUSION RATE

In 2006, two laboratories did not track the number of exclusions. For the laboratories tracking exclusions, there were 347,719 cases completed, and 89,890 of those (25.85%) were reported as exclusions. The average exclusion rate for the laboratories reporting exclusions is 23.13% with a standard deviation of 5.64. The median exclusion rate is 25.06% with a range of 10.99% to 31.83%. The explanation for the range of exclusion rates is complex but appears to be 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 laboratories, there were 4,579 exclusions from laboratories reporting exclusion data (total of 15,082 cases) or an exclusion rate of 30.36%, a higher percentage than the 26% seen for legal testing. The range for non-legal laboratories is 10.99 to 35.04%.

**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. This claim is incorrect. The exclusion rate includes a number of factors. One is that a case may include several men as alleged fathers because the mother 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 determines which man is the likely biological father while excluding the others. Another factor is that the unexcluded alleged father, as part of his defense, may allege that the mother had multiple sexual partners during the time of conception. These other 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 (for example, the mother was married to someone other than the biological father at the time of conception) there is a legal presumption that the husband is the father. The husband is then 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**

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 (disputed parent is missing and other relatives are used to evaluate parentage). Some laboratories reported using different CPIs for different classes of clients (private versus public contracts, or for different technologies). For these laboratories, the higher CPI was used for this report.

The results for the laboratories that responded are shown in Table 3. The most common minimum CPI for a standard trio is 100, with 55% of laboratories using this value, with a range of 100 to 100,000. For cases where the mother is not tested, the most common minimum CPI is 100 with 62% of laboratories using this value, with a range of 100 to 10,000. For the family study or reconstruction cases, 58% indicated that they report “whatever was obtained” and 81% considered a combined paternity index of 101 or less reportable. For sibling studies about 90% of the laboratories considered a combined paternity index of 101 or less reportable.
A common issue is the significance of the paternity index and the reliability of the AABB standard requiring a CPI of 100 to 1. At least one laboratory has claimed that AABB is only concerned about how the testing is performed, but not the meaning of the test. In fact, the Relationship Testing Standards Program Unit is concerned about the meaning of the tests and thus chose a CPU of 100 to 1. 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.

Another issue arises with regard to performing other relationship analyses, such as reconstruction cases, trios with genetic anomalies, and samples from exhumations, coroners, and other postmortem testing. Importantly, note that a CPI of less than 100 is not an indicator of no relationship, unless the CPI is 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, which are typically used 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, since testing is often performed in conditions that are not ideal. Another 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.

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).

<table>
<thead>
<tr>
<th>CPI</th>
<th>Trio</th>
<th>One Parent</th>
<th>Family Study (Reconstruction)</th>
<th>Full Sib</th>
<th>Half Sib</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whatever Is Obtained</td>
<td>0</td>
<td>0</td>
<td>15</td>
<td>12</td>
<td>12</td>
</tr>
<tr>
<td>10</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>3</td>
<td>1</td>
</tr>
<tr>
<td>25</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>100</td>
<td>16</td>
<td>18</td>
<td>5</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>101</td>
<td>0</td>
<td>0</td>
<td>1</td>
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<td>0</td>
</tr>
<tr>
<td>150</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>200</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td>1</td>
<td>1</td>
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<tr>
<td>400</td>
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<td>0</td>
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<td>0</td>
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<tr>
<td>500</td>
<td>2</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>1000</td>
<td>3</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>0</td>
</tr>
</tbody>
</table>
TESTING WITHOUT THE MOTHER

There is still a strong concern about the apparent increase in the number of clients submitting disputed paternity cases without the mother. Testing without the mother presents a number of problems. First, the paternity index, on average, is cut in half. 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. Obtaining a sample from the mother is also an important quality control step, because results that exclude the mother may indicate a problem with 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 who is 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, because 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 the mother’s location is unknown or she is deceased. Every effort should be made to test the mother.

TECHNOLOGY USE

In 2006, the survey showed a continued trend toward the increased use of polymerase chain reaction (PCR) technology (STR analysis) with a decrease in the use of restriction fragment length polymorphism (RFLP) methods. PCR technology was used in 98.53% of reported cases and RFLP was used in 1.12% of reported cases. This is also the first year that no cases were evaluated using serological HLA testing.

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 2006

<table>
<thead>
<tr>
<th>Technology</th>
<th>Number of Cases</th>
<th>Utilization (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>STR</td>
<td>344616</td>
<td>98.53</td>
</tr>
</tbody>
</table>
Table 1: Use of Various Technologies

<table>
<thead>
<tr>
<th>Technology</th>
<th>Count</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>RFLP</td>
<td>3906</td>
<td>1.12</td>
</tr>
<tr>
<td>HLA Class II Molecular</td>
<td>35</td>
<td>0.01</td>
</tr>
<tr>
<td>Y Chromosome</td>
<td>1079</td>
<td>0.31</td>
</tr>
<tr>
<td>HLA Class I Molecular</td>
<td>139</td>
<td>0.04</td>
</tr>
<tr>
<td>SNP</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>HLA Serology</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Red Cell Antigens</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Red Cell Enzymes/ Serum Proteins</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Allotyping</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

*Note that some cases used more than one technology. Not all laboratories responded to this question.

Figure 2 shows the use of various technologies since 1990. As indicated above, the most commonly used technologies in 1990 (red cell antigens, HLA, and red cell enzymes and serum proteins) now account for less than 1% 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 versus RFLP). Note that in some instances, multiple technologies were used in the same case.
SAMPLE SOURCE

Laboratories reported approximately 812,177 samples used for the casework in 2006. Not all laboratories reported the samples they used. Of these samples, buccal swabs accounted for 98.05% of the samples. Whole blood samples accounted for .64%. The use of blood spot cards decreased to 1.16% of samples. Various other samples were also reported (See Table 5).

Table 5. Sample Source in 2006

<table>
<thead>
<tr>
<th>Sample</th>
<th>Number</th>
<th>Percent of Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Buccal Swabs</td>
<td>812,177</td>
<td>98.0501</td>
</tr>
<tr>
<td>Blood</td>
<td>5,278</td>
<td>0.6372</td>
</tr>
<tr>
<td>Blood Spot Cards</td>
<td>9,641</td>
<td>1.1639</td>
</tr>
<tr>
<td>Amniotic Fluid</td>
<td>693</td>
<td>0.0837</td>
</tr>
</tbody>
</table>
AMELOGENIN

The amelogenin locus is now used in a number of laboratories to test for the gender of the sample. A number of males lacking the Y or X amelogenin allele have been observed. Laboratories were asked to measure the apparent X males observed in their laboratory. Like other DNA loci, amelogenin is subject to mutations. Therefore, occasionally normal males have a female amelogenin phenotype or a Y phenotype. The X male phenotype was most commonly seen in the Hispanic populations, in about 1/1392 men. The Y male phenotype was most commonly seen in the Black population in about 1/1688 Black males.

Table 6. A Summary of Data on Apparent X and Y Males Seen with ABI Primers

<table>
<thead>
<tr>
<th></th>
<th>Black</th>
<th>White</th>
<th>Hispanic</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Number X Males Observed</strong></td>
<td>6</td>
<td>12</td>
<td>16</td>
</tr>
<tr>
<td><strong>%</strong></td>
<td>0.0099</td>
<td>0.0254</td>
<td>0.0718</td>
</tr>
<tr>
<td><strong>Number Y Males Observed</strong></td>
<td>36</td>
<td>6</td>
<td>4</td>
</tr>
<tr>
<td><strong>%</strong></td>
<td>0.0592</td>
<td>0.0127</td>
<td>0.0180</td>
</tr>
</tbody>
</table>

MUTATION REPORTS

One area of concern is the number of inconsistencies necessary to render an opinion of non-paternity. The laboratories were asked if they had seen any case where, in the opinion of the expert, the inconsistencies were double or triple “mutations” and not sufficient to render an opinion of non-paternity. Twelve laboratories stated they had reported cases with double or triple mutations. Eleven laboratories did not observe any mutations. The
laboratories reported 47 cases with double mutations and no cases with triple mutations as inclusions. Most laboratories report these “double mutation” cases with the inconsistencies noted and statistically considered. This illustrates the importance of accurate assessments of potential mutations and null alleles.

**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 the mutation rate as the PI, while others, most commonly, used the mutation rate divided by the average probability of exclusion. Some laboratories used the mutation rate as a transmission frequency and some of the laboratories used Brenner’s method in looking at the repeat length difference between STR alleles.

A summary of the mutation frequencies for each STR locus is provided in Appendix 1. Note that these frequencies incorporate the indeterminate findings. The calculations are summarized at the bottom of the table for each paternal allele as shown in Appendix 2. The frequencies for changes from one allele to another are presented in Appendix 2 Appendix 3.

A continuing objective of this year’s report is to begin to collect data on STR loci to provide laboratories with frequencies to use in the mutation calculation. The guidance document for the 7th and 8th edition of standards contains a discussion of two methods that might be useful. One limitation of this data set is if the laboratory did not see any mutations, the laboratory did not provide data on the maternal and paternal meiosis. Many laboratories did not provide any data so the data presented is from a few laboratories.

If one wished to determine the specific mutation frequency at locus D3S1358 for the apparent paternal mutation event of the alleged father’s allele 16 changing to an allele 17 in the child. Using the attached table specific calculations could be made. Suppose that are 16 instances where, simply, 16 changed to 17 out of 79247 meiosis reported or a frequency of 0.000202. However there are several other opportunities for this change. If there were five instances where the alleged father’s 16 could have changed to either a 15 or 17 (child is a clone of the mother or mother was not tested). To incorporate this data one approach is to calculate the relative chance that the change was 16 to 17 rather than 16 to 15. Note the clear changes and calculate the relative chance of each change. Multiply the relative chance time the number of changes where the allele is 16 to 15 or 17, which is 5 in this data set, to obtain the relative portion attributable to a 16 to 17 change.

Table 6. Relative Chance of allele 16 changing to 15 or 17.
<table>
<thead>
<tr>
<th>Change</th>
<th>Observed</th>
<th>Relative Chance</th>
<th>Portion of 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>16 to 17</td>
<td>16</td>
<td>16/31 = 0.516</td>
<td>5 * 0.516 = 2.58</td>
</tr>
<tr>
<td>16 to 15</td>
<td>15</td>
<td>15/31 = 0.484</td>
<td>5 * 0.484 = 2.42</td>
</tr>
<tr>
<td>Total</td>
<td>31</td>
<td>1</td>
<td>5</td>
</tr>
</tbody>
</table>

From this data, add 2.56 to the 16 observed potential changes from 16 to 17 to get the total of 18.56. Similarly, there were seven observations where the alleged father has alleles 16 and 18 either of which could mutate to a 17.

**Table 7. Relative Chance of allele 16 or 18 changing 17.**

<table>
<thead>
<tr>
<th>Change</th>
<th>Observed</th>
<th>Relative Chance</th>
<th>Portion of 7</th>
</tr>
</thead>
<tbody>
<tr>
<td>16 to 17</td>
<td>16</td>
<td>16/26 = 0.615</td>
<td>7 * 0.615 = 4.305</td>
</tr>
<tr>
<td>18 to 17</td>
<td>10</td>
<td>10/26 = 0.385</td>
<td>7 * 0.385 = 2.695</td>
</tr>
<tr>
<td>Total</td>
<td>26</td>
<td>1</td>
<td>5</td>
</tr>
</tbody>
</table>

From this data, add 4.305 to the 18.56 potential changes (paragraph above) from 16 to 17 to get the total of 22.865.

Hypothetically, there were instances where the father’s alleles 16 and 19 could have change to a 17 or 18. To incorporate this data a similar approach is used.

**Table 8. Relative Chance of allele 16 or 19 changing to 17 or 18.**

<table>
<thead>
<tr>
<th>Change</th>
<th>Observed</th>
<th>Relative Chance</th>
<th>Portion of 1</th>
</tr>
</thead>
<tbody>
<tr>
<td>16 to 17</td>
<td>16</td>
<td>16/21 = 0.762</td>
<td>1 * 0.762 = 0.762</td>
</tr>
<tr>
<td>16 to 18</td>
<td>0</td>
<td>0/21 = 0</td>
<td>1*0 = 0</td>
</tr>
<tr>
<td>19 to 17</td>
<td>0</td>
<td>0/21 = 0</td>
<td>1*0 = 0</td>
</tr>
<tr>
<td>19 to 18</td>
<td>5</td>
<td>5/21 = 0.238</td>
<td>1* 0.238 = 0.238</td>
</tr>
<tr>
<td>Total</td>
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From this data, add 0.762 to the 22.865 above yielding 23.627.

Lastly, data from those cases where the mutation is either maternal or paternal may be incorporated (indeterminate). Hypothetically there were 7 instances where the mutation to a 17 could have been from a paternal 16. The approach to incorporate these data is similar to the above. First look to the data to determine the frequency of the changes.

**Table 9. Relative Chance of allele 16 changing to 17.**

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<th>Change</th>
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<th>Portion of 7</th>
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Annual Report, Page 11 of 102
<p>| | | | |</p>
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<tr>
<td>16 to 17 Maternal</td>
<td>$\frac{1}{67521} = 1.481e-5$</td>
<td>$1.481e-5 / 2.167e-4 = 0.0683$</td>
<td>$7 * 0.0683 = 0.478$</td>
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<tr>
<td>16 to 17 Paternal</td>
<td>$\frac{16}{79247} = 2.019e-4$</td>
<td>$2.019e-4 / 2.167e-4 = 0.9317$</td>
<td>$7 * 0.9317 = 6.522$</td>
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<td>$2.167e-4$</td>
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Finally add 6.522 to the 23.627 yielding 30.149. Thus for this hypothetical population the frequency of paternal mutation from a 16 to a 17 is $30.149 / 79247 = 0.00038$ as compared to the 0.000202 without incorporating all possible mutation events. The committee invites comments on alternative methods of determining the mutation frequencies.
Appendix 1. Summary of Apparent Mutations at various Loci analyzed by PCR in 2006.

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<th>Black Maternal</th>
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## Appendix 2. Paternal & Maternal Mutation Data

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**Total Paternal Meiosis by Race**

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**Relative Frequency Paternal**

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## CSF Maternal Mutations

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### D2S1338 Paternal Mutations

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### D3S1358 Maternal Mutations

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### D5S818 Paternal Mutations

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|                          | 0.0006732 | 0.0005317 | 0.0008381 |

### Frequency Maternal Indeterminate

|                          | 0.0001203 | 0.0000894 | 0.0001160 |

### Total Paternal Mutation Frequency

|                          | 0.0021447 | 0.0023605 | 0.0019594 |

### Total Maternal Mutation Frequency

|                          | 0.0003833 | 0.0003969 | 0.0002712 |
### D5S818 Maternal Mutations

| Maternal Mutation – Allele: | Caucasian | | Black | | Hispanic | | American Indian | | Asian (Oriental) |
|---------------------------|-----------|---------|--------|--------|-----------|--------|----------------|-----------------|
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| 11 | 10 | | | | | | | | |
| 11 | 12 | | | | | | | | |
| 11 | 13 | | | | | | | | |
| 12 | 10 | | | | | | | | |
| 12 | 11 | | | | | | | | |
| 12 | 13 | | | | | | | | |
| 12 | 10 or 11 | | | | | | | | |
| 12 | 11 or 13 | | | | | | | | |
| 13 | 12 | | | | | | | | |
| 13 | 14 | | | | | | | | |
| 13 | 12 or 14 | | | | | | | | |
| 14 | 13 | | | | | | | | |
| 14 | 15 | | | | | | | | |
| 15 | 14 | | | | | | | | |
| 17 | 18 | | | | | | | | |
| 10 or 12 | 11 | | | | | | | | |
| 10 or 14 | 9 or 13 | | | | | | | | |
| 11 or 12 | 13 or 14 | | | | | | | | |
| 11 or 13 | 12 | | | | | | | | |
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## Total Maternal Mutation Frequency

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D16S539 Maternal Mutations
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| From | To | Number | Frequency | | | | Number | Frequency | | | | Number | Frequency | | | | | | | | |
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| 9 | 10 | 1 | 0.0000158 | | | | | | | | | | | | | | | | | |
| 10 | 9 | | | | | | | | | | | | | | | | | | | |
| 10 | 11 | 1 | 0.0000150 | | | | | | | | | | | | | | | | | |
| 10 | 9 or 11 | | | | | | | | | | | | | | | | | | | |
| 11 | 9 | | | | | | | | | | | | | | | | | | | |
| 11 | 10 | | | | | | | | | | | | | | | | | | | |
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| 13 | 12 | 2 | 0.0000317 | | | | | | | | | | | | | | | | | |
| 13 | 14 | 1 | 0.0000158 | | | | | | | | | | | | | | | | | |
| 13 | 12 or 14 | 1 | 0.0000158 | | | | | | | | | | | | | | | | | |
| 14 | 13 | 4 | 0.0000634 | | | | | | | | | | | | | | | | | |
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| 15 | 14 | 2 | 0.0000317 | | | | | | | | | | | | | | | | | |
| 15 | 16 | | | | | | | | | | | | | | | | | | | |
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| 10 or 12 | 11 or 13 | | | | | | | | | | | | | | | | | | | |
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| 10 or 13 | 11 or 12 | 1 | 0.0000158 | | | | | | | | | | | | | | | | | |
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| Paternal Frequency | 0.0016256 | 0.0017232 | 0.0013230 | 0.0174672 |
| Maternal Frequency | 0.0013077 | 0.0013928 | 0.0016832 |
| Sum | 0.0029333 | 0.0031160 | 0.0030062 |

**Relative Frequency**

| Paternal | 0.5541821 | 0.5530104 | 0.4400862 |
| Maternal | 0.4458179 | 0.4469896 | 0.5599138 |
| Indeterminate | 0.0004938 | 0.0006174 | 0.0007656 |

**Frequency**

| Paternal Indeterminate | 0.0002737 | 0.0003414 | 0.0003369 |
| Maternal Indeterminate | 0.0002201 | 0.0002760 | 0.0004287 |

**Total Paternal Mutation Frequency**

| 0.0018992 | 0.0020646 | 0.0016599 |

**Total Maternal Mutation Frequency**

| 0.0015278 | 0.0016688 | 0.0021119 |
## D21S51 Maternal Mutations

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**Total Maternal Meiosis by Race**

| Frequency | 0.0013077 | 0.0013928 | 0.0016832 |
## FGA Paternal Mutations

<p>| Paternal Mutation – Allele: | Caucasian | | | Black | | | Hispanic | | | American Indian | | | Asian (Oriental) | |
|---------------------------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|-----------|
| From | To | Number | Frequency | From | Number | Frequency | From | Number | Frequency | From | Number | Frequency |
| 17.2 | 18.2 | | | | | | | | | | | |
| 18 | 19 | | | | | | | | | | | |
| 18.2 | 19.2 | | | | | | | | | | | |
| 19 | 17 | 1 | 0.0000188 | | | | | | | | | |
| 19 | 18 | 3 | 0.0000565 | | | | | | | | | |
| 19 | 20 | 4 | 0.0000753 | | | | | | | | | |
| 19 | 21 | 1 | 0.0000188 | | | | | | | | | |
| 19.2 | 20.2 | | | | | | | | | | | |
| 20 | 18 | 1 | 0.0000188 | | | | | | | | | |
| 20 | 19 | 4 | 0.0000753 | | | | | | | | | |
| 20 | 21 | 6 | 0.0001129 | | | | | | | | | |
| 20 | 22 | | | | | | | | | | | |
| 20 | 19 or 21 | | | | | | | | | | | |
| 20.2 | 21.2 | | | | | | | | | | | |
| 21 | 18 | | | | | | | | | | | |
| 21 | 19 | 1 | 0.0000188 | | | | | | | | | |
| 21 | 20 | 2 | 0.0000376 | | | | | | | | | |
| 21 | 20.2 | 1 | 0.0000148 | | | | | | | | | |
| 21 | 22 | 7 | 0.0001318 | | | | | | | | | |
| 21.2 | 21 | | | | | | | | | | | |
| 21.2 | 22.2 | | | | | | | | | | | |
| 22 | 20 | 1 | 0.0000188 | | | | | | | | | |
| 22 | 21 | 2 | 0.0000376 | | | | | | | | | |
| 22 | 23 | 10 | 0.0001882 | | | | | | | | | |
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**Total Paternal Meiosis by Race**

- Caucasian: 44,348
- Black: 65,369
- Hispanic: 17,554

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Frequency 0.0003202 0.0004698 0.0001362
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Indicate the number of cases: 2934 Black, 748 Hispanic, 229 American Indian, 0.0003408 Frequency.

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**FGA Mutations that are Indeterminate as to the Parental Origin**

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**Frequency**

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