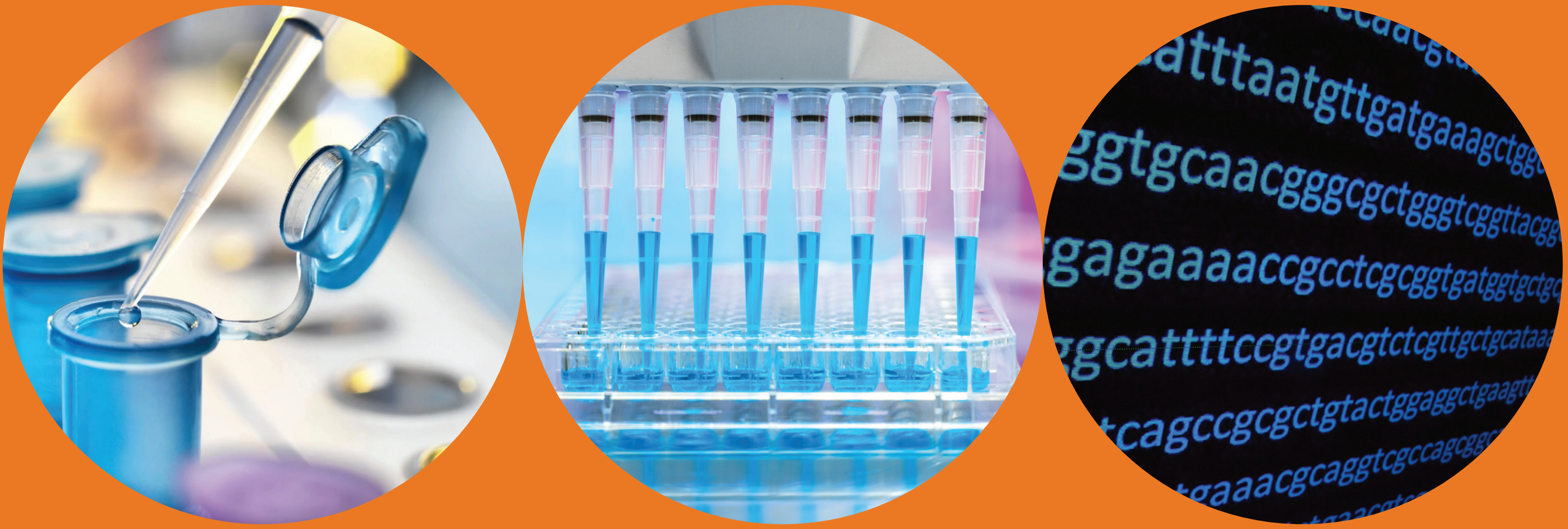
Relationship Testing

TECHNICAL REPORT FOR TESTING IN 2024

As compared to reported data from 2021, 2022 and 2023



ABSTRACT

AABB surveyed facilities accredited for Relationship Testing (RT) activities regarding data of interest to the RT community. Data were collected for the calendar year 2024 and compared to information collected from the three prior calendar years. There were 347,314 cases tested and reported in 2024. Of those tests, 54.9% were legal chain of custody cases for non-immigration purposes, with an exclusion rate of 20.4%. Of the total case volume, 7.0% were for immigration, visa, passport, or citizenship cases with an exclusion rate of 5.0%, and 38.1% were unaccredited cases lacking a chain of custody (tested for curiosity) that showed an exclusion rate of 28.7%. Of all samples collected, more than 97.2% were buccal swabs. DNA analysis of autosomal short tandem repeats comprised more than 97.6% of the tests performed. X- chromosome analysis was performed in addition to autosomal analyses on 26.6% of the cases. DNA Next Generational Sequencing (NGS) was performed on 2.4% of cases, and a small number of cases also received Y Chromosome or mitochondrial analysis. Of the laboratories surveyed, 57.9% incorporate apparent mutations into the combined likelihood ratio by dividing the mutation rate by the average probability of exclusion. Mutation data were collected from the surveyed laboratories and combined with the 2022 and 2023 data. Mutation frequencies for 48 loci are presented, including 3 loci for which mutation rates have not been previously reported.

INTRODUCTION

AABB is an international, not-for-profit association representing institutions and individuals involved in relationship testing. The AABB Relationship Testing Laboratories Accreditation Program is based on member-developed standards and provides for the assessment and accreditation of facilities performing relationship testing activities, including methods for forensic investigative genetic genealogy DNA analysis. The Accreditation Program assesses the quality and operational systems within a facility to verify compliance with applicable standards and offer objective, independent feedback to affirm sound practices and provide guidance on areas of improvement.

PREFACE

This survey provides information on the state of the relationship testing community, tries to ask questions that may be of interest, and tracks trends in testing. Evaluation of the data was anonymous. AABB scientific staff reviewed the raw data and provided only anonymized aggregate data and tables for review by the Relationship Testing Standards Committee (RTSC) and the Relationship Testing Accreditation Committee (RTAC).

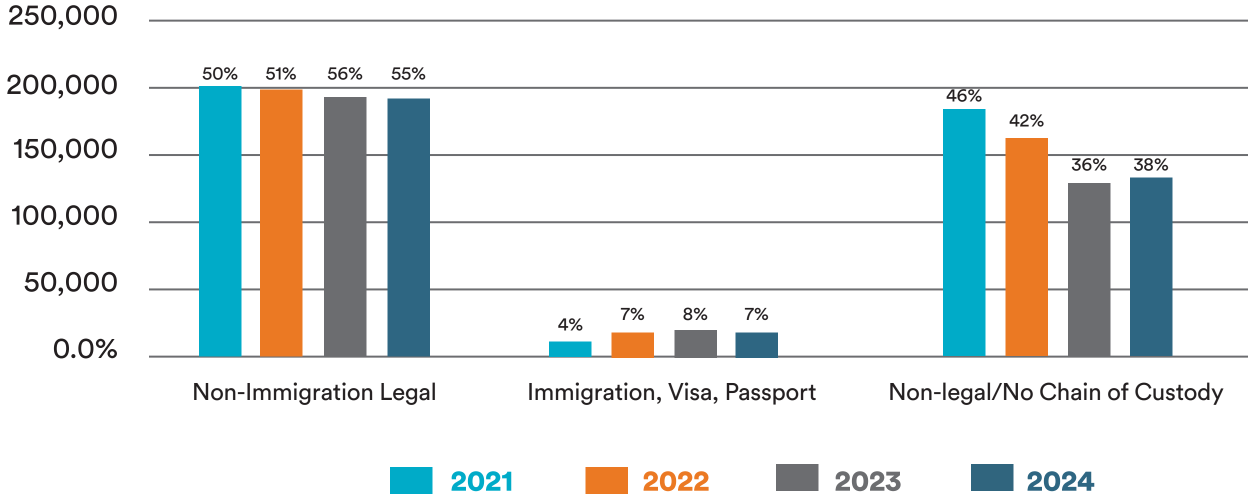
In addition to full-service laboratories, AABB accredits facilities that only perform sample collection and report verification activities. These facilities are required to send their collected samples to an AABB-accredited laboratory for testing. The testing laboratories include these sample counts in their reported data. Any data submitted by AABB-accredited facilities that perform only collection and report verification activities are excluded from counts to avoid duplication of data submitted by the testing laboratory.

ANNUAL VOLUME OF TESTING

The total volume reported for cases tested in 2024 was 347,314. There are three general categories of testing: legal cases where samples are collected with a documented chain-of-custody, legal cases intended to support a petition for U. S. immigration, and “non-legal” cases lacking a chain-of-custody generally performed for curiosity. A small decrease in immigration testing was reported over the prior year.

In addition to the volume of accredited tests, laboratories were asked if they tested cases where the chain of custody did not meet the requirements of the *Standards for Relationship Testing Laboratories (Standards).* The tested individuals, without a proper witness, generally self-collect these “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 must conform to all aspects of the *Standards* and cannot selectively choose the requirements to follow. Figure 1 indicates the volumes of cases reported by case type.

FIGURE 1. CASES REPORTED BY TYPE



LABORATORIES BY SIZE

Table 1 indicates size by the volume of cases reported for the 19 total responding laboratories.

TABLE 1. LABORATORY SIZE BY THE VOLUME OF CASES REPORTED.

Number of Cases Reported	Percentage of RT Laboratories			
	2021	2022	2023	2024
<100	26.0%	9.5%	5.6%	5.3%
100-1,000	26.0%	42.9%	44.4%	52.6%
1,001-10,000	26.0%	28.6%	33.3%	31.6%
10,001-100,000	11.0%	9.5%	5.6%	5.3%
>100,000	11.0%	9.5%	11.1%	5.3%

EXCLUSION RATE

We asked laboratories to report parentage exclusions, or hypotheses not supported for non-parentage cases, and the number of cases for which a conclusion could not be reached, separately by case type. The observed rate of exclusion varies significantly depending on the type of case, as shown in Table 2.

TABLE 2. EXCLUSIONS REPORTED BY CASE TYPE FOR 2024.

Exclusions (or hypothesis not supported)	Non-Immigration Legal	Immigration, Visa, Passport	Non-legal / No Chain of Custody
Average Exclusion Rate	20.43%	4.95%	28.73%
Inconclusive	0.30%	0.05%	0.60%

MISCONCEPTIONS IN PATERNITY TESTING – EXCLUSION RATE

AABB has observed misinterpretation of data reported for exclusion rates in previous reports. It is important to clarify what the exclusion rate does not represent. An exclusion rate of 30% does not mean that 30% of fathers in the general population are raising children that are not biologically theirs. From the data, we can only conclude that, of the people who needed a relationship test, some percentage of those tests either exclude or do not support the tested relationship. There are many situations in which the presence or absence of a relationship was never in question, but a DNA test was necessary to provide proof of a relationship or no relationship for legal reasons.

COMBINED LIKELIHOOD RATIO

The laboratories were asked to indicate what combined likelihood ratio (CLR) they considered acceptable for cases with a standard trio (mother, child, alleged father), single parent cases (mother (or father) not tested cases), and family study / reconstruction cases of more than two tested parties (cases where the disputed parent is missing, and other relatives are used to evaluate parentage).

The AABB *Standards* set the minimum CLR for parentage cases at 100. An index of 100 is meaningful, but indices of higher values can be obtained using current methods. There has been a tendency for laboratories to set much higher values as a minimum likelihood ratio, such as 10,000 to 1 and 100,000 to 1 for some of their tested hypotheses, but not all (such as family study/reconstruction cases). When setting higher thresholds for internal use, it is inappropriate to claim that lower values are not meaningful. The minimum acceptable CLR for parentage cases, by policy, exceeds the AABB *Standards* for 50% of the laboratories.

Data were not collected on minimum CLR for two-party comparisons of full siblings, half-siblings, avuncular, and single grandparentage likelihood ratios. Beginning with the 13th Edition of the *AABB Standards for Relationship Testing Laboratories*, minimum CLR is defined in the Standards for two-party non-parentage comparisons. See Table 3 for Laboratories’ minimum CLR.

TABLE 3. LABORATORIES' MINIMUM COMBINED LIKELIHOOD RATIOS (% OF LABORATORIES USING A CLR AS THEIR MINIMUM) FOR STANDARD TRIOS, ONE PARENT (MOTHER OR FATHER NOT TESTED), FAMILY STUDIES >2 PARTIES.

Minimum Combined Likelihood Ratios	Trio	One Parent	Family Study >2 parties
10			36.8%
>10.1			5.3%
50			5.3%
100	57.9%	57.9%	
175	5.3%	5.3%	
200	5.3%	5.3%	
1,000	10.5%	10.5%	5.3%
2,500	5.3%	5.3%	
10,000	5.3%	10.5%	5.3%
≥10,000	5.3%	5.3%	
20,000	5.3%		

*Note: For family studies, the sum of percentages is less than 100%, as some labs limit their testing to parentage only.

TECHNOLOGY USE

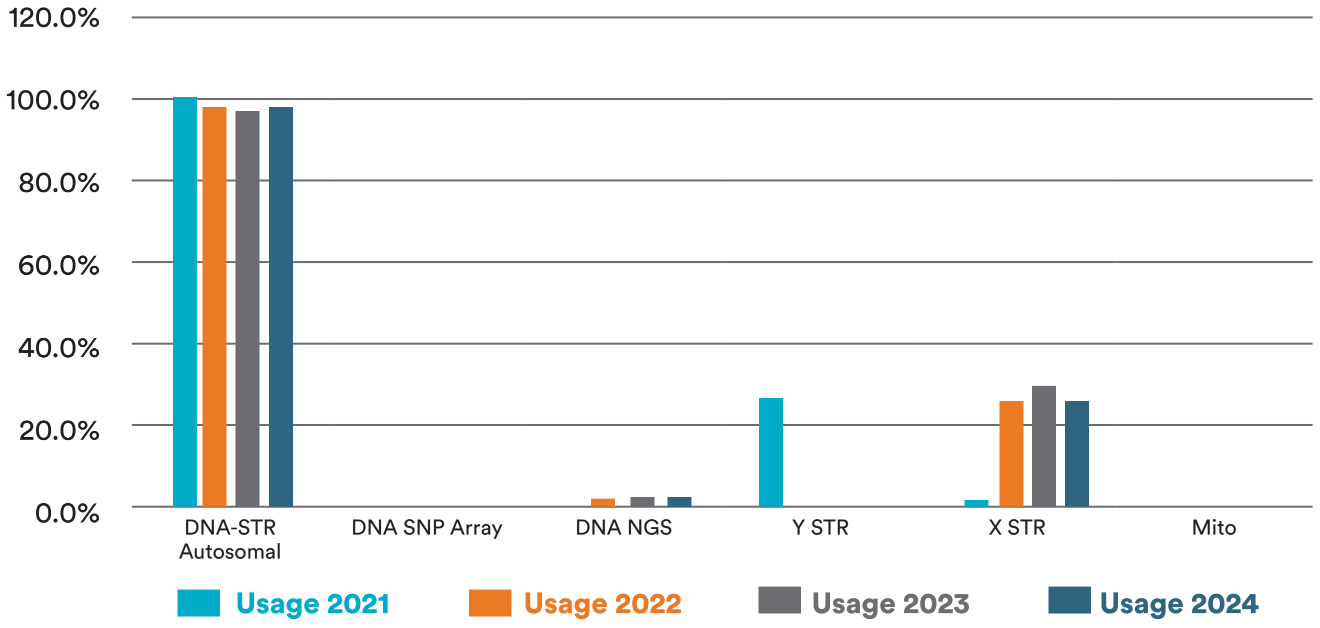
Laboratories reported the technologies used to determine relationship in cases tested. Short Tandem Repeat (STR) testing is still the primary method used across all laboratories. An increase in additional technologies used for testing was observed in 2024. More than one technology may be used to resolve an individual case. Table 4 shows the technology used to resolve the reported cases. Figure 2 demonstrates the use of technology since 2021.

TABLE 4. THE TECHNOLOGY USED IN CASES REPORTED

Technology / Method	Usage (2021)	Usage (2022)	Usage (2023)	Usage (2024)
DNA-STR Autosomal	99.83%	97.79%	97.56%	97.60%
DNA SNP Array	4.86%	none	none	none
DNA NGS	none	2.21%	2.44%	2.40%
Y STR	1.12%	0.35%	0.39%	0.42%
X STR	26.92%	26.34%	30.08%	26.62%
Mito	1.81%	0.0002%	0.01%	0.01%

*Note: Commercial DNA STR kits may contain one or more sex chromosome loci (e.g. DYS392). The tracking here is in reference to stand alone kits that test multiple Y chromosome loci.
**Note: At least one laboratory is testing multiple X chromosome loci as part of their routine testing on all samples.

FIGURE 2. THE TECHNOLOGY USED IN CASES REPORTED



SAMPLE SOURCE

Cases reported for 2024 include non-legal cases and samples collected without a chain of custody. Any of the following sample types lacking a chain of custody or not meeting the requirements for identification in the *Standards* would not be appropriate for an AABB-accredited legal relationship test. Buccal swabs account for >97% of the samples. Various other samples were also reported (See Table 5).

TABLE 5. SAMPLE SOURCE

Sample Type	Percentage Total
Buccal Swabs	97.242%
Liquid Blood	1.909%
Dried Blood Spots	0.493%
Amniotic fluid	0.013%
Chorionic villi	0.001%
Bone	0.014%
Hair root	0.090%
Paraffin block	0.010%
Tissue	0.006%
DNA extracts	0.001%
Other (Urine, Fingernails, Contact article, Toothbrush, Ear Swab, Semen, Cigarette Butts, Umbilical Cord, misc.)	0.222%

MUTATION CALCULATION

Single inconsistencies are routinely seen in the testing of parentage cases. If a laboratory concludes that the inconsistency is a mutation, then the mutation result must be incorporated into the reported results. Laboratories were asked how they calculated the parentage index (PI) for these loci. The *Standards* do not specify the method for calculation when an apparent mutation is observed, although guidance is given on commonly accepted methods of calculation. Some labs reported using more than one method. Table 6 shows the different calculation methods used for mutations.

TABLE 6. REPORTED MUTATION CALCULATION METHODS

Mutation calculation method	Usage (2021)	Usage (2022)	Usage (2023)	Usage (2024)
Mutation rate/average probability of exclusion	47.4%	65.0%	50.0%	57.9%
Using a stepwise mutation model – Brenner's Method	52.6%	40.0%	55.6%	26.3%
Standard PI using the mutation rate as the disputed parent's transmission chance	15.8%	15.0%	22.2%	21.1%
Using a stepwise mutation model – Familias	5.3%	5.0%	5.6%	5.3%
Fimmer's Method	5.3%	none	none	none
Use the mutation rate as the PI	none	none	none	5.3%
Extended Stepwise (Unstationary)	none	none	none	5.3%

Note: the sum of percentages is greater than 100%, as some labs use more than one method.

MUTATION FREQUENCIES

For the 2024 survey, laboratories were asked to provide counts of apparent mutations for trio cases tested with the mother, child, and alleged father. Data were requested in the 2024 survey to calculate separate maternal and paternal rates of mutation. Combined mutation rates additionally include data where the source of the mutation is undetermined. Null alleles were excluded from the total counts. Table 7a presents the combined 2022, 2023, and 2024 mutation rates of 48 loci, arranged in alphanumeric order for convenient reference. Table 7b sorts the data by combined mutation rate from largest to smallest.

The mutation rates are calculated using pooled data from many population groups. Since mutation events are relatively rare, there are insufficient data collected from this survey to allow the calculation of mutation rates for specific population groups. The mutation rates presented may be applied generally to any calculation involving an apparent mutation event.

TABLE 7a. MUTATION RATES (2022, 2023, AND 2024 COMBINED, DATA SORTED ALPHANUMERICALLY)

2022 -2024 Summary Mutation Rates											
	Maternal			Paternal			Undetermined	Combined			Sum of Markers Reported in Trio Inclusionary Cases
Locus	Apparent Mutations Observed	Maternal Meioses	Mutation Rate = Mutations/ Maternal Meioses	Apparent Mutations Observed	Paternal Meioses	Mutation Rate = Mutations/ Paternal Meioses	(mutation could be maternal or paternal)	Total Apparent Mutations Observed	Total Meioses	Mutation Rate = Total Mutations/ Total Meioses	
CSF1PO	64	748671	0.000085	635	748671	0.000848	192	891	1497726	0.000595	748863
D10S1248	88	746057	0.000118	329	746057	0.000441	99	516	1492312	0.000346	746156
D12S391	97	176558	0.000549	731	176558	0.00414	84	912	353284	0.002581	176642
D13S317	117	764588	0.000153	590	764588	0.000772	175	882	1529526	0.000577	764763
D14S1434	2	8334	0.00024	4	8334	0.00048	0	6	16668	0.00036	8334
D16S539	111	762881	0.000146	401	762881	0.000526	101	613	1525964	0.000402	762982
D17S1301	4	8214	0.000487	4	8214	0.000487	4	12	16436	0.00073	8218
D18S51	181	761379	0.000238	884	761379	0.001161	108	1173	1522974	0.00077	761487
D19S433	154	761070	0.000202	324	761070	0.000426	60	538	1522260	0.000353	761130
D1S1656	81	748121	0.000108	440	748121	0.000588	70	591	1496382	0.000395	748191
D21S11	350	760146	0.00046	560	760146	0.000737	113	1023	1520518	0.000673	760259
D22S1045	23	755739	0.00003	79	755739	0.000105	34	136	1511546	0.00009	755773
D2S1338	62	755756	0.000082	624	755756	0.000826	59	745	1511630	0.000493	755815

TABLE 7a. MUTATION RATES (2022, 2023, AND 2024 COMBINED, DATA SORTED ALPHANUMERICALLY) CONTINUED

2022 and 2023 Summary Mutation Rates										
	Maternal			Paternal			Undetermined	Combined		Sum of Markers Reported in Trio Inclusionary Cases
Locus	Apparent Mutations Observed	Maternal Meioses	Mutation Rate = Mutations/ Maternal Meioses	Apparent Mutations Observed	Paternal Meioses	Mutation Rate = Mutations/ Paternal Meioses	(mutation could be maternal or paternal)	Total Apparent Mutations Observed	Total Meioses	Mutation Rate = Total Mutations/ Total Meioses	
D2S441	47	749272	0.000063	206	749272	0.000275	53	306	1498650	0.000204	749325
D3S1358	51	765166	0.000067	564	765166	0.000737	152	767	1530636	0.000501	765318
D3S4529	1	8272	0.000121	2	8272	0.000242	0	3	16544	0.000181	8272
D5S818	80	764725	0.000105	453	764725	0.000592	170	703	1529790	0.00046	764895
D6S1043	51	161447	0.000316	252	161447	0.001561	37	340	322968	0.001053	161484
D6S474	15	8302	0.001807	23	8302	0.00277	1	39	16606	0.002349	8303
D7S820	55	750967	0.000073	419	750967	0.000558	98	572	1502130	0.000381	751065
D8S1115	1	7487	0.000134	3	7487	0.000401	0	4	14974	0.000267	7487
D8S1179	91	762693	0.000119	677	762693	0.000888	133	901	1525652	0.000591	762826
D9S1122	2	8325	0.00024	3	8325	0.00036	1	6	16652	0.00036	8326
D9S2157	1	2972	0.000336	0	2972	0	0	1	5944	0.000168	2972
DXS101	32	78983	0.000405	131	78983	0.001659	17	180	158000	0.001139	79000
DXS6810	20	68730	0.000291	123	68730	0.00179	53	196	137566	0.001425	68783
DXS9895	24	71055	0.000338	61	71055	0.000858	22	107	142154	0.000753	71077
DYS385AB	0	131	0	1	131	0	0	1	262	0.003817	131
DYS391	0	1898	0	2	1898	0.001054	0	2	3796	0.000527	1898
DYS392	0	76490	0	28	76490	0.000366	0	28	152980	0.000183	76490
DYS439	0	131	0	1	131	0	0	1	262	0.003817	131
DYS458	0	132	0	2	132	0	0	2	264	0.007576	132
DYS570	0	83	0	1	83	0	0	1	166	0.006024	83
DYS576	0	47	0	1	47	0.021277	0	1	94	0.010638	47
F13A01	0	4942	0	2	4942	0	0	2	9884	0.000202	4942
F13B	2	2078	0.000962	1	2078	0.000481	0	3	4156	0.000722	2078
FESFPS	0	605	0	1	605	0.001653	0	1	1210	0.000826	605
FGA	167	760043	0.00022	1348	760043	0.001774	185	1700	1520456	0.001118	760228
HPRTB	17	74966	0.000227	233	74966	0.003108	46	296	150024	0.001973	75012
LPL	0	5098	0	3	5098	0	1	4	10198	0.000392	5099
Penta C	0	5098	0	1	5098	0	0	1	10196	0.000098	5098
Penta D	8	29027	0.000276	31	29027	0.001068	2	41	58058	0.000706	29029
Penta E	14	28996	0.000483	83	28996	0.002862	1	98	57994	0.00169	28997
SE33	310	565313	0.000548	1484	565313	0.002625	73	1867	1130772	0.001651	565386
TH01	13	763048	0.000017	45	763048	0.000059	6	64	1526108	0.000042	763054
TPOX	22	754512	0.000029	79	754512	0.000105	19	120	1509062	0.00008	754531
vWA	114	758263	0.00015	1012	758263	0.001335	208	1334	1516942	0.000879	758471
YGATAH4	0	132	0	1	132	0	0	1	264	0.003788	132
▨	New mutations reported									

TABLE 7b. COMBINED MUTATION RATES (2022, 2023, AND 2024 SORTED BY MUTATION RATE)

Locus	Mutation Rate = Total Mutations/ Total Meioses
DYS570	0.013158
DYS576	0.010417
DYS458	0.007463
DYS385AB	0.003788
DYS439	0.003788
YGATAH4	0.003759
D12S391	0.002569
D6S474	0.002338
HPRTB	0.001966
Penta E	0.001684
SE33	0.001646
DXS6810	0.001421
DXS101	0.001137
FGA	0.001116
D6S1043	0.001051
vWA	0.000878
FESFPS	0.000825
D18S51	0.000769
DXS9895	0.000752
D17S1301	0.000729
F13B	0.000721
Penta D	0.000705
D21S11	0.000672
CSF1PO	0.000594
D8S1179	0.00059
D13S317	0.000576
DYS391	0.000526
D3S1358	0.000501
D2S1338	0.000492
D5S818	0.000459
D16S539	0.000401
D1S1656	0.000395
LPL	0.000392
D7S820	0.000381
D9S1122	0.00036
D14S1434	0.00036
D19S433	0.000353
D10S1248	0.000346
D8S1115	0.000267
D2S441	0.000204
F13A01	0.000202
DYS392	0.000183
D3S4529	0.000181
D9S2157	0.000168
Penta C	0.000098
D22S1045	0.00009
TPOX	0.00008
TH01	0.000042

Four laboratories reported confirmed double mutations (i.e., two tested loci out of a minimum of 20 exhibited alleles with mutations between parent and offspring). Two laboratories observed a triple mutation (see Table 8).

TABLE 8. MULTIPLE MUTATIONS OBSERVED FROM 2024 DATA

	number observed	% total testing volume
Double mutations	192	0.0553%
Triple mutations	13	0.0037%

Laboratories reported testing at 17 loci for which no mutations were observed (see Table 9). If an apparent mutation is observed for which the mutation rate is not yet known, it may be estimated. One estimate may be the average mutation rate for other autosomal loci evaluated with similar methods.

TABLE 9. TESTED LOCI WITH NO OBSERVED MUTATIONS (2022, 2023, AND 2024 COMBINED)

Locus / Test	Total Meioses
Yindel	<100
YGATAH4	<100
DYS19	<100
DYS389I	100-1,000
DYS389II	100-1,000
DYS390	100-1,000
DYS393	100-1,000
DYS437	<100
DYS438	<100
DYS448	<100
DYS456	<100
DYS481	<100
DYS533	<100
DYS549	<100
DYS635	<100
DYS643	<100
HLA AB	<100

CONCLUSION

AABB surveyed facilities accredited for Relationship Testing activities for data of interest to the RT community. A notable trend in the data is the decreased volume of testing for immigration, visa, and passport cases compared to 2023. For the 2024 report, we provided separate maternal and paternal mutation rates for many loci in addition to a combined mutation rate. Apparent mutations were observed for the previously unreported loci, D9S2157, DYS576, and F13B, and the mutation rates are now provided. The 2024 STR mutation rates have been combined with the 2022 and 2023 mutation rates to obtain a more precise and comprehensive mutation rate.

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