PROBABILISTIC GENOTYPING IN THE COURTROOM: ADMISSIBILITY, FAMILIES, SECONDARY TRANSFER AND COMPETING STATISTICS

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DIFFERENCE IN STATISTICS

RMP

Random Match Probability (RMP) – the chance that you pick a random, unrelated person from the population and they match an evidence DNA profile

Report example: The DNA profile obtained from the swab indicates one male contributor. John Doe cannot be excluded as a contributor to this DNA profile. The chance that an unrelated person, chosen at random from the general population, would be included as a contributor to this DNA profile is approximately 1 in every 7 trillion individuals.

RMP – PUT IT INTO PERSPECTIVE

- The population of Earth is about 7 billion people. It would take approximately 1000 planet Earths with that same population in order to expect to see this DNA profile once.

CPI

Combined Probability of Inclusion (CPI) – the chance that you pick a random, unrelated person from the population and they could be included as a contributor to a mixed DNA profile

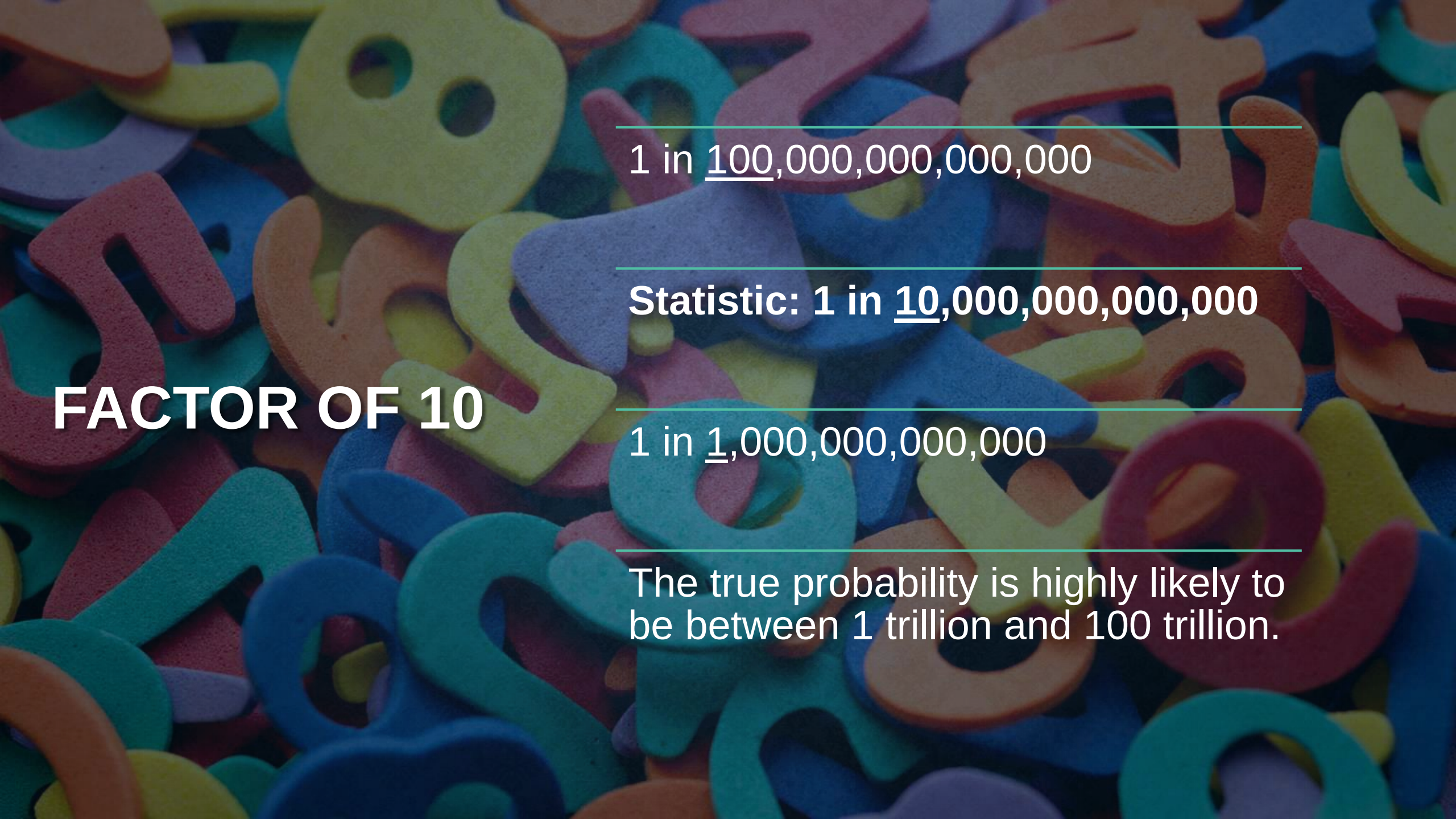
Report example: The DNA profile obtained from the swab indicates a mixture of two individuals with at least one male contributor. Jane Doe cannot be excluded as a contributor to this mixed DNA profile. The chance that an unrelated person, chosen at random from the general population, would be included as a contributor to this mixed DNA profile is approximately 1 in every 20 million individuals.

CPI – PUT IT INTO PERSPECTIVE

- For every 20 million random, unrelated people, you would anticipate that approximately one of them would be included as a contributor to that mixture

FACTOR OF 10

- Per the second National Research Council Report on forensic DNA evidence (NRC II), it is recommended that profile frequencies are assigned a confidence interval of a factor of 10
- Applies to RMP and CPI



FACTOR OF 10

1 in 100,000,000,000,000

Statistic: 1 in 10,000,000,000,000

1 in 1,000,000,000,000

The true probability is highly likely to be between 1 trillion and 100 trillion.

LR

Likelihood Ratio (LR) – a ratio of two probabilities which show a strength of support for one scenario over the other

Report example: The DNA profile obtained from the swab is approximately 12 trillion times more probable if the sample originated from John Doe and two unknown persons than if it originated from three unknown persons. Therefore, there is extremely strong support that John Doe and two unknown persons contributed to this mixed DNA profile, rather than three unknown persons.

EARTH ANALOGY CANNOT BE USED WITH LRS

RECOMMENDATIONS

OF THE

SWGDAM AD HOC WORKING GROUP ON

GENOTYPING RESULTS REPORTED AS LIKELIHOOD RATIOS

Given the increasing usage and interest in probabilistic genotyping among forensic DNA testing laboratories, the Scientific Working Group on DNA Analysis Methods (SWGDAM) empaneled an Ad Hoc Working Group to inform on matters relating to the reporting of likelihood ratios (*LR*s). This group was comprised of experts in the application of statistical principles to forensic evidence and forensic practitioners with expertise in the interpretation of mixed DNA specimens

***LR* for H_p Support and 1/*LR* for H_d Support**	**Verbal Qualifier**
1	Uninformative
2 – 99	Limited Support
100 – 9,999	Moderate Support
10,000 – 999,999	Strong Support
≥1,000,000	Very Strong Support

VERBAL SCALE

LR– PUT IT INTO PERSPECTIVE

- A likelihood ratio is a ratio of two probabilities giving a numerical value that shows strength of support for one scenario over another. In the case of DNA, in the simplest of terms, it is evaluating whether it is more likely to observe the DNA profile if it originated from Jane Doe than if it originated from an unknown individual.
- Think of a scale with two sides, the more weight there is, the further down the heavier side will go.
- The same is said for a likelihood ratio, the more weight for one scenario, the less likely the other scenario becomes.

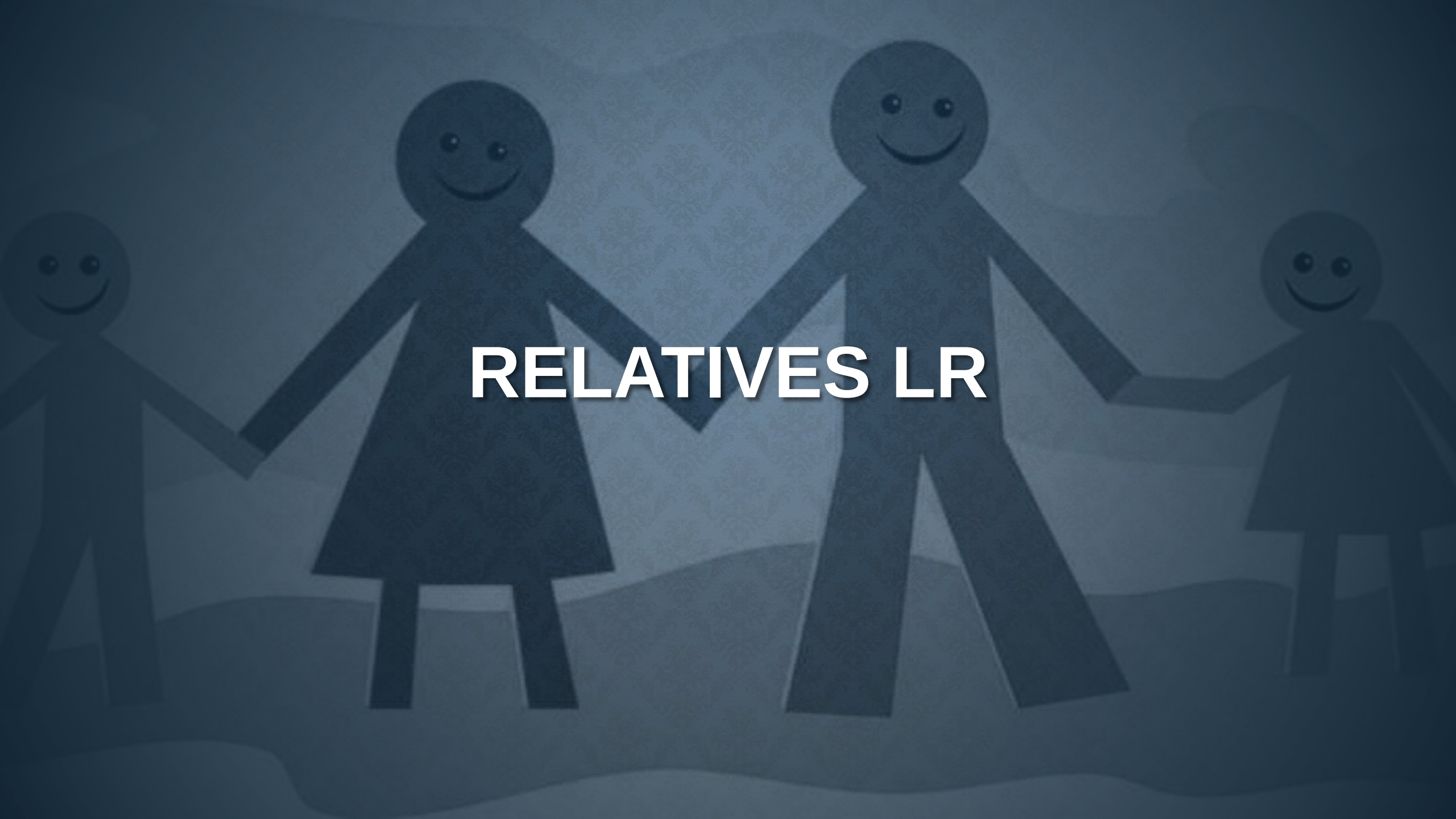
- LR = 12 trillion
- Very Strong Support

- Imagine the weight for the first scenario is on the ground

- LR = 300
- Moderate Support

- The weights of the two scenarios are almost even.

- Think about your own non-contributor study. If the highest LR seen was 231, this number is barely above that and should be addressed in testimony.


RELATIVES LR

RELATIVES

STRmix™ includes likelihood ratio (LR) propositions that consider relatives of the person of interest (POI); these LRs are in turn incorporated with the Unrelated LR into the Unified LR that is reported by DNA Labs International.

The software can resolve mixtures composed of family members and, through validation, ratios can be established to flag the possible presence of a relative of the POI in the mixture.

STRATIFIED LIKELIHOOD RATIO (LR)

SUMMARY OF LR

LR (population proportion)	DLI_GlobalFiler_AfAm_FBIextended.csv (0.14)	DLI_GlobalFiler_Cauc_FBIextended.csv (0.69)	DLI_GlobalFiler_SEHISP_FBIextended.csv (0.06)	DLI_GlobalFiler_SWHISP_FBIextended.csv (0.11)	Stratified
Total LR	**2.75E19**	9.23E22	2.82E22	6.08E21	3.65E19
Sibling	**6.45E7**	2.82E8	2.46E8	1.39E8	7.28E7
Parent/Child	**4.29E11**	1.57E13	9.99E12	2.92E12	5.26E11
Half sibs	**5.91E14**	6.75E16	3.57E16	9.49E15	7.47E14
Grandparent / Grandchild	**5.91E14**	6.75E16	3.57E16	9.49E15	7.47E14
Uncle or Aunt /Niece or Nephew	**5.91E14**	6.75E16	3.57E16	9.49E15	7.47E14
First Cousin	**8.50E16**	2.92E19	1.30E19	3.23E18	1.08E17
Unified	**2.51E15**	5.56E16	1.67E15	1.76E15	2.74E15

Report stratified, unified LR

Stratified: Provides a single LR across all populations chosen

Unified: Takes into account that unknown contributors are made up of both relatives AND unrelated people

FAMILIAL LRS

SUMMARY OF LR

TABLE 1 OF 2

Sub-source LR. 99% 1-sided lower HPD interval calculated from 1000 iterations, MCMC uncertainty on, Allele frequency uncertainty on.

LR	NIST1036_AFAM	NIST1036_ASIAN	NIST1036_CAUC
PROPORTION	0.25	0.25	0.25
Children per family	0	0	0
Population size	0	0	0
Relation of unknown in Hd to POI			
Unrelated	1.1765E24	1.8773E22	1.2367E22
Sibling	1.7961E8	2.2527E7	5.1259E7
Parent/Child	2.9818E13	1.0723E12	1.4997E12
Half Sibling	2.0270E17	4.5505E15	6.0165E15
Grandparent/Grandchild	2.0270E17	4.5505E15	6.0165E15
Uncle or Aunt/Niece or Nephew	2.0270E17	4.5505E15	6.0165E15
Cousin	1.3431E20	2.3273E18	2.7068E18

FAMILIAL RELATIONSHIPS – SIBLING LR EXAMPLE

$$\frac{Hp}{Hd} = \frac{\text{The DNA profile originated from } \textbf{John Doe} \text{ and an unknown, unrelated person}}{\text{The DNA profile originated from a } \textbf{sibling of John Doe} \text{ and an unknown, unrelated person}}$$

REPORTED UNIFIED LR:

- The DNA profile obtained from the sample is approximately **130 million** times more probable if the sample originated from John Doe and an unknown person than if it originated from two unknown persons. Therefore, there is **extremely strong support** that John Doe and an unknown person contributed to this DNA profile, rather than two unknown persons.

SIBLING LR:

- The DNA profile obtained from the sample is approximately **18** times more probable if the sample originated from John Doe and an unknown, unrelated person than if it originated from a sibling of John Doe and an unknown, unrelated person.

REPORTED LR VS SIBLING LR

FAMILIAL LRS – TESTIMONY FOR LABS NOT REPORTING UNIFIED LR

- It was not validated for use, and as such, it can not be reported at this time.
- Note: the number of children is set to 0, so the calculated familial LRs does not reflect familial proportions.

LR	NIST1036_AFAM
PROPORTION	0.25
Children per family	0
Population size	0
Relation of unknown in Hd to POI	
Unrelated	1.1765E24
Sibling	1.7961E8
Parent/Child	2.9818E13

WHY EVALUATE FAMILIAL RELATIONSHPS?

SUMMARY OF LR

LR (population proportion)	DLI_GlobalFiler_AfAm_FBIextended.csv (0.14)	DLI_GlobalFiler_Cauc_FBIextended.csv (0.69)	DLI_GlobalFiler_SEHISP_FBIextended.csv (0.06)	DLI_GlobalFiler_SWHISP_FBIextended.csv (0.11)	Stratified
Total LR	4.40E9	**3.74E8**	1.10E9	1.10E10	4.03E8
Sibling	3.23E1	**1.70E1**	2.41E1	4.78E1	1.81E1
Parent/Child	2.85E2	**9.15E1**	1.44E2	4.08E2	9.44E1
Half sibs	1.31E5	**3.56E4**	6.73E4	2.63E5	3.86E4
Grandparent / Grandchild	1.31E5	**3.56E4**	6.73E4	2.63E5	3.86E4
Uncle or Aunt /Niece or Nephew	1.31E5	**3.56E4**	6.73E4	2.63E5	3.86E4
First Cousin	9.89E6	**2.16E6**	4.73E6	2.30E7	2.22E6
Unified	8.18E8	**3.25E8**	1.05E8	4.54E8	1.30E8

Sibling LR = 18
P/C LR = 94

Reported LR = 130 million – highest verbal scale equivalent

QUESTIONS TO ASK

- Is the contributor assignment intuitive? How do you know?
- Is the likelihood ratio reflective of what you would expect to see in this type of profile? Why?
- Were there any other diagnostic issues identified with this profile?

PROPOSITIONS

PROPOSITION FORMULATION

- Do additional propositions need to be requested?
- How will this change the statistic?
- Have you checked with your analysts?
- Does it make sense for your case to request the additional propositions?
- Does the laboratory allow for additional propositions to be requested?
- Will additional information presented change what propositions should be logically considered?

PROPOSITIONS CONTINUED

- *Should any additional information become available it may be necessary to reconsider these interpretations. Additional propositions may be considered upon request if instructed to do so prior to testimony.*

LR COMPARISON

Suspect + 2 Unknowns **3 Unknowns**	**1.78E16**
Victim + 2 Unknowns **3 Unknowns**	**8.77E16**
Suspect + Victim + Unknown **3 Unknowns**	**1.54E55**
Victim + Suspect + Unknown **Victim + 2 Unknowns**	**2.61E17**

Owner + 3 unknowns/ 4 unknowns	21 septillion	Stratified
Owner + 3 unknowns/ 4 unknowns	45 quadrillion	Stratified, Unified
Suspect + 3 unknowns/ 4 unknowns	1/LR=48	Stratified, Unified
Owner + Suspect + 2 unrelated unknowns/ 4 unrelated unknowns	630 sextillion	Stratified
Owner + Suspect + 2 unknowns/ Owner + 3 unknowns	1/LR=98	Stratified, Unified

LR COMPARISON PT 2

* A cautionary tale!

SECONDARY TRANSFER

THINGS TO CONSIDER

- Sample type; i.e. touch vs body vs body fluid
- Intimate sample? i.e. cervix vs labia
- Location, i.e. sight, trigger or firearm overall
- Input/profile, i.e. 75% of a robust vs. low-level profile
- Number of contributors
- Contributor assignment
- Conditions of evidence
- Number of samples involved

QUESTIONS TO ASK

How much DNA was input into this profile?

Is that the optimal input of DNA for this type of testing?

Approximate amount of the DNA profile accounted for by the POI?

Identify other contributors? Determined/ Searched?

ADMISSIBILITY HEARINGS

STRmix™ IN THE COURT ROOM - ADMISSIBILITY

- Numerous admissibility hearings denied through motion responses
- Consider requesting an affidavit in support of your motion from the DNA analyst associated with the case, the laboratory associated with the case or the defense expert
- What are the facts at hand?
- Request internal validation and discovery in case file

ADMISSIBILITY HEARINGS

- Pre-hearing Motions
- Affidavits
- Supporting Evidence
  - Literature lists
  - Previous rulings
  - Internal validation
  - Training records

CASE CONSIDERATIONS

- What standard needs to be met?
- What evidence needs to be shown?
- Does the existing evidence demonstrate these facts?
- Does additional testing need to be conducted?
- Does additional evidence need to be tested?
- What does the case law say?
- Is your expert qualified to testify to these standards and facts?

- Consider options for other experts
- Validation Manager or TL?
- Statistician
- Other experts in the field
- John Buckleton

WHO ME?

The following is a response to the Motion to Preclude Probabilistic Genotyping Pursuant to Rule 702 and Daubert. The page numbers referenced in the points below refer to the page number in the Motion to Preclude.

1. Samantha O. Wandzek is the DNA Analyst for this case. She has worked at DNA Labs International (DLI) since 2015 and currently serves as Validation Manager/Analyst Group Supervisor/Senior DNA Analyst. Prior to DLI, Samantha worked in New York City's Office of Chief Medical Examiner in the Department of Forensic Biology for seven years, with her last position being that of a Criminalist III, which is the equivalent of a Senior DNA Analyst. Samantha holds the following degrees: BA, Chemistry (University of Colorado at Boulder) and MS, Forensic Science (Pace University).
2. Rachel H. Oefelein performed the majority of the work for the STRmix™ internal validations conducted at DLI. She has worked at DLI since 2014 and currently serves as Quality Assurance Manager/Senior DNA Analyst. Prior to DLI, Rachel worked at the Armed Forces DNA Identification Laboratory for over four years, with her last position being that of a Nuclear DNA Analyst. Rachel holds the following degrees: BS, Criminal Justice with a Forensic Science Minor (Loyola University of New Orleans) and MSc, Forensic Science (University of Strathclyde).
3. Alicia M. Cadenas reviewed the STRmix™ validation and approved it for use in casework. She has worked at DLI since 2014 and currently serves as DNA Technical Leader/Laboratory Supervisor. Prior to DLI, Alicia worked for the Virginia Department of Forensic Science in the Forensic Biology Section for over six years, with her last position being that of a Forensic Scientist. Alicia holds the following degrees: BS, Biology (Nova Southeastern University) and MSFS, Forensic Science (Florida International University).
4. DNA Labs International was founded in 2004 and has been accredited for forensic DNA analysis since 2005. Since 2005, the laboratory has been audited annually against the Federal Bureau of Investigation's (FBI) Quality Assurance Standards and the International Organization for Standardization ISO/IEC 17025:2005 by the external accrediting body now known as ANSI-ASQ National Accreditation Board (ANAB). We successfully earned our reaccreditation under these standards in October of 2017.

AFFIDAVIT RESPONSES

7. On page five, foundational validity has been established for STRmix™ probabilistic genotyping software through the developmental validation and subsequent internal validations conducted by multiple laboratories. The Scientific Working Group on DNA Analysis Methods (SWGDAM) defines a developmental validation as "the acquisition of test data and determination of conditions and limitations of a new or novel DNA methodology for use on forensic and/or casework reference samples". The developers of the software, Forensic Science South Australia (FSSA) and the Institute of Environmental Science and Research (ESR), performed an extensive developmental validation of the software (J.A. Bright et al. / Forensic Science International: Genetics 23 (2016) 226–239). In addition, before laboratories can use the software themselves, internal validations must be performed. SWGDAM defines an internal validation as "the accumulation of test data within the laboratory to demonstrate that established methods and procedures perform as expected in the laboratory". Internal validations have been performed by the Federal Bureau of Investigation (FBI), DLI and all of the other thirty plus laboratories that have STRmix™ online for casework. DLI has fulfilled the SWGDAM guidelines for validation of probabilistic genotyping systems as well as satisfied the validation requirements set forth by the laboratory's accrediting body, which have been memorialized by ANAB.
8. On page six, the method has been shown to be reliable as applied. Refer to *State of Florida v. Dwayne Cummings Frye/Daubert Ruling, State of Florida vs. Marc Regisme Daubert Ruling, Michigan vs. Larry David Smith Daubert, People v. Vincent Bullard-Daniel, State of Florida vs. Bidjoury Jean Louis Frye Ruling.*
9. On page seven, the motion mentions that the President's Council of Advisors on Science and Technology (PCAST) found subjective analysis has not been established for DNA mixtures; however, he contradicts that statement on page nine stating that "probabilistic genotyping approaches can reduce subjectivity in the analysis of DNA typing results". Multiple agencies have responded to PCAST including the Department of Justice (DCJ) that affirmed "the report does not mention numerous published research studies which seem to meet PCAST's criteria for appropriately designed studies providing support for foundational validity. That omission discredits the PCAST report as a thorough evaluation of scientific validity."

AFFIDAVIT RESPONSES CONTINUED

AFFIDAVIT RESPONSES CONTINUED

- Consult with other Attorneys
- Provide general information about laboratory and personnel signing the affidavit
- Signers can include; writing analyst, TL, technical reviewer, managers
- Address the points argued in the motion
- Provide both explanation and evidence

CHALLENGE PREP FOR THE EXPERT

- Provide exhibits prior to hearing that they can reference
- Review previous motions/rulings
- Review transcripts from past hearings for sample questions and answers.
- Review training and competency materials
- Maintain records of pertinent rulings

CHALLENGE PREP FOR THE EXPERT CONTINUED…

- Schedule pre-hearing meeting with the expert
- Consider issuing an affidavit in support of software
- Consider recommending an additional expert
- Know your current events (STRmix™ website or John Buckleton's website)
- Request your expert to be present during opposing expert testimony for both depositions and hearing

CHALLENGE PREP FOR THE EXPERT CONTINUED…

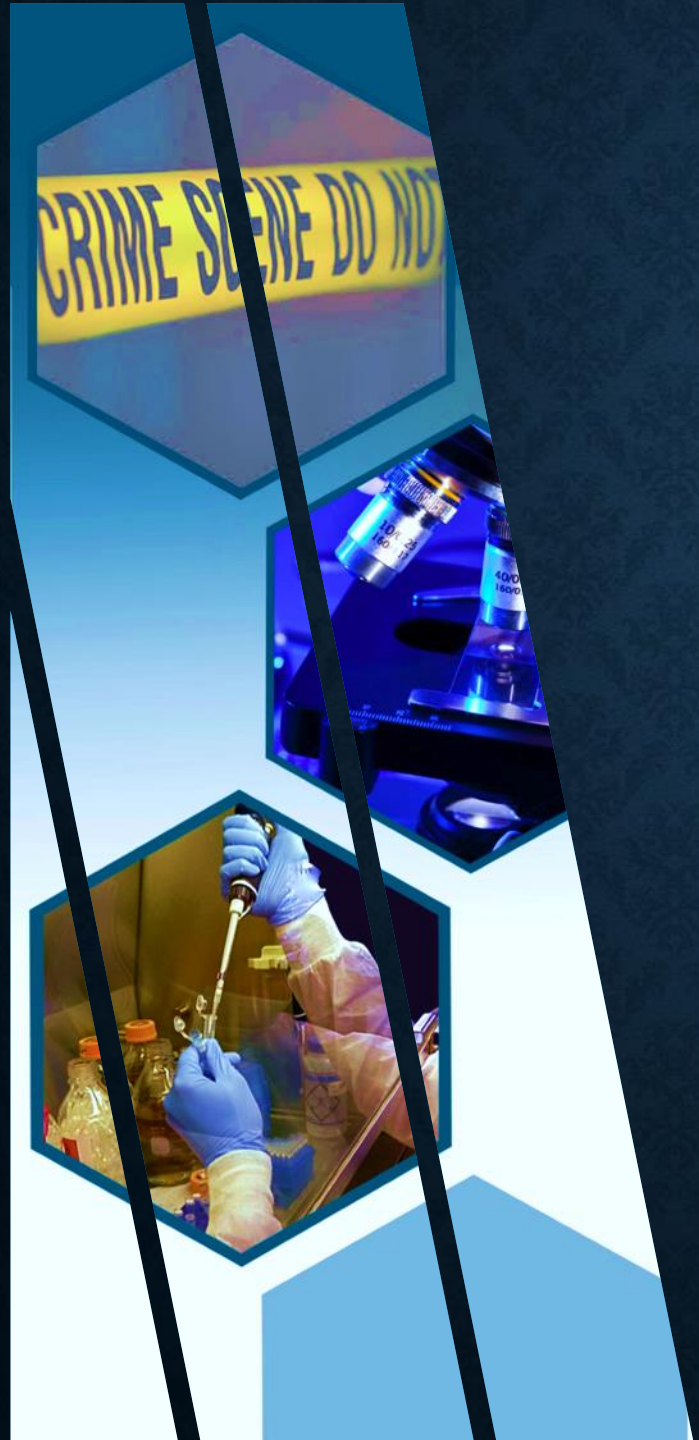
- What is being argued? Review the motions.
- What standards need to be satisfied for the court?
- Review any previous deposition transcripts on the case.
- Know real facts: internal validation studies, number of publications, actual publications to support evidence, SOP, what other laboratories use the same technology.

- Why did the admissibility get denied or affirmed?
- Has admissibility been covered in another similar case since?
- Is there evidence to refute the ruling?
- Don’t guess, it’s okay to say I don’t recall.
- If asked to review a decision/publication on the stand, it is okay to say you would need time to review it.

ADVISE YOUR EXPERT - KNOW YOUR FACTS… OR DON’T?

RESOURCES

- John Buckleton’s Wordpress Site: https://johnbuckleton.wordpress.com/strmix/
- NFSTC DNA Training: https://www.nfstc.org/service/forensics-training/dna-training/
- STRmix™ Website https://www.strmix.com/



Thank You

American Academy of Forensic Sciences

&

DNA Labs International

Team

Questions?

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