wyndham forensic group inc.
analysis & support

FORENSIC DNA ANALYSIS

Strengths and Limitations

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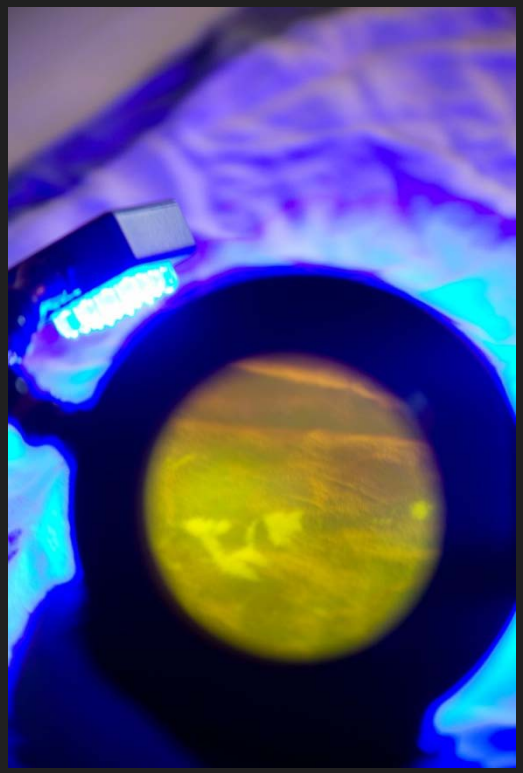
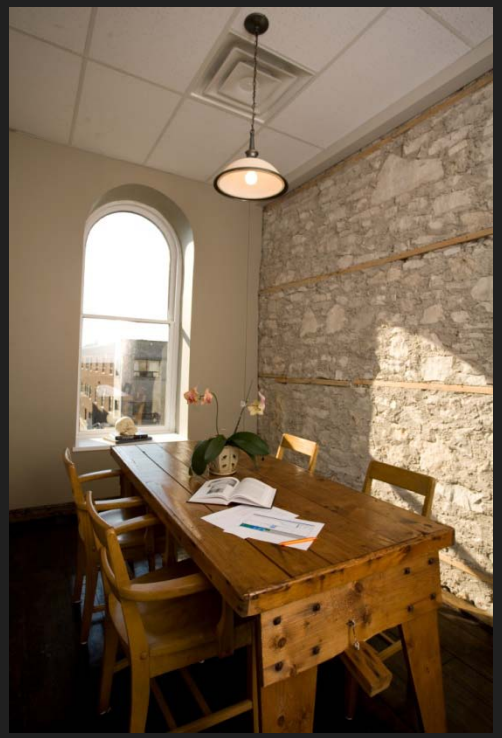
General Information Presentation
Fall 2009

OUTLINE



- History of Forensic DNA Analysis
- Current State
- Laboratory Process
- Language of DNA Profiling
- Strengths
- Limitations
- Conclusion

HISTORY



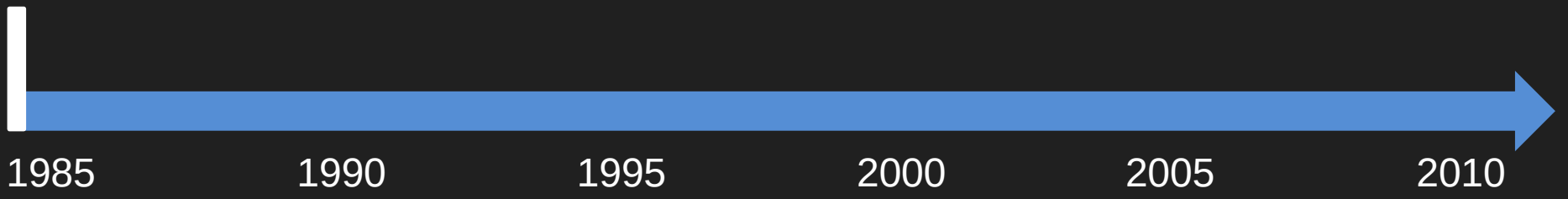
The slides that follow provide a brief overview of the evolution of forensic DNA analysis over the past 25 years. This account is not meant to be exhaustive and dates indicated in the timeline are approximate. It is intended only as a general guide.

1985 1990 1995 2000 2005 2010

HISTORY

DNA fingerprinting is developed in the United Kingdom by Sir Alec Jeffreys at Leicester University. It detects variability which exists between individuals at certain locations along their DNA.

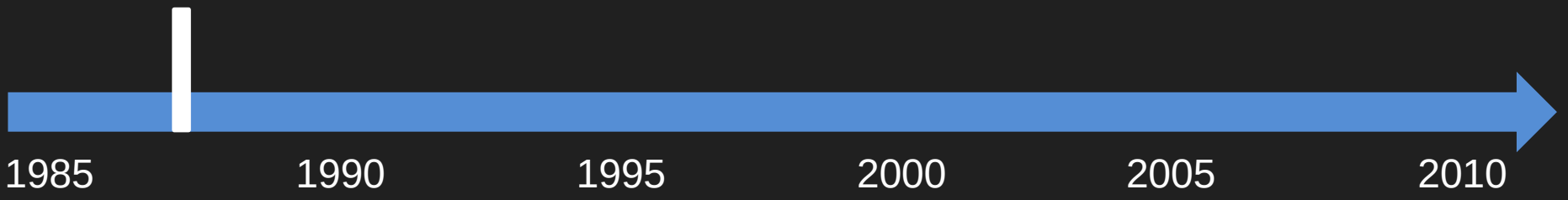
Around the same time period, a technique called *PCR* is first described which allows scientists to make copies of DNA in a test tube. It revolutionizes the field of molecular biology but a number of years will pass before it is utilized with regularity in forensic science.



HISTORY

Soon after Jeffreys' technique is developed, a DNA fingerprint is generated from a crime scene sample in a double homicide investigation in Leistershire, England. Initially, testing exonerates the prime suspect in the case.

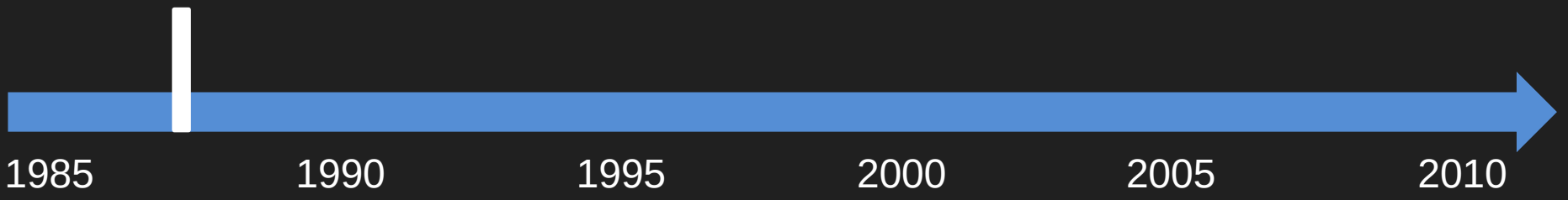
An exhaustive campaign of voluntary blood testing for males in three small villages is then undertaken to identify the perpetrator. After thousands of men respond, one comes forward to notify police that he was asked to give his blood sample under a different name – Colin Pitchfork. Eventually, the real Pitchfork is arrested and confesses to both murders.



HISTORY

Although the DNA tests developed by Jeffreys are highly discriminating, their success is dependent on the availability of large quantities of high-quality DNA. They are therefore of little to no value in the analysis of numerous crime scene samples where quantities of DNA are limited or where DNA is degraded. They are also labour-intensive and time-consuming.

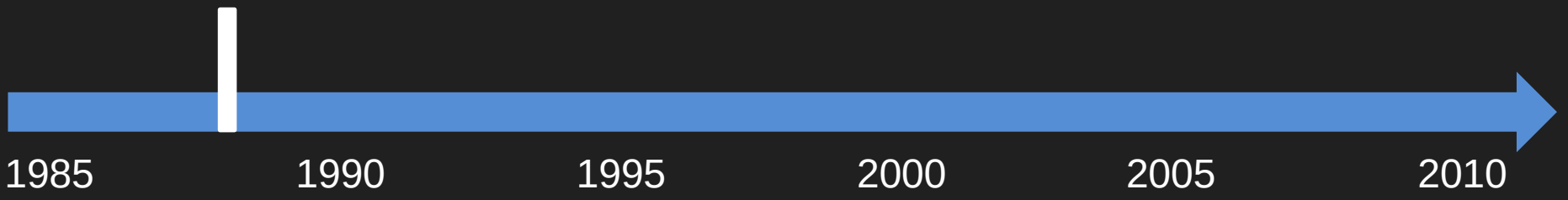
PCR-based forensic DNA analysis methods would eventually overcome most of these limitations.



HISTORY

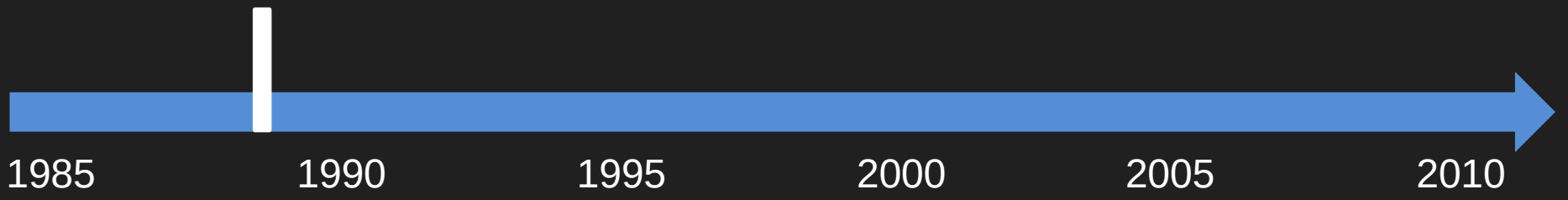
Forensic DNA testing is first introduced in the United States by private companies. A number of cases are successfully prosecuted but in the case of *New York v Castro,* a major challenge is brought forward and the DNA evidence is ruled inadmissible.

The FBI would begin forensic DNA casework shortly after it was introduced in the US, and Canadian labs would follow suit very soon after that.



HISTORY

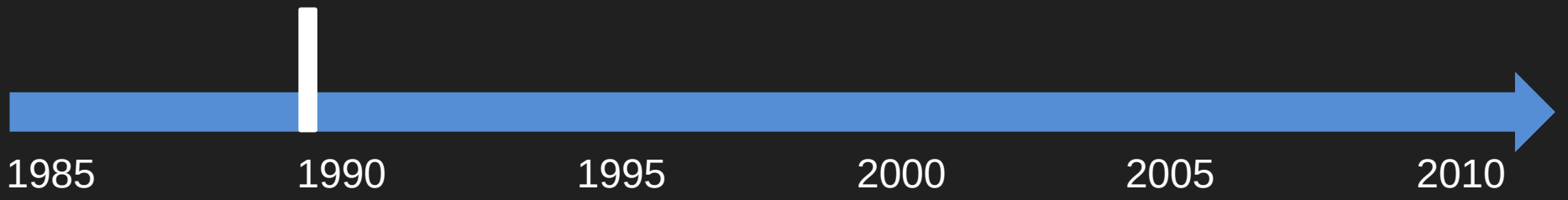
As forensic DNA methods are developed and implemented in forensic labs across North America, the Technical Working Group on DNA Analysis Methods (TWGDAM) is formed with Canadian participation, under the auspices of the FBI, as a means of bringing standardization and quality assurance to the field.



HISTORY

DNA fingerprinting methods are applied to casework in Canadian forensic laboratories.

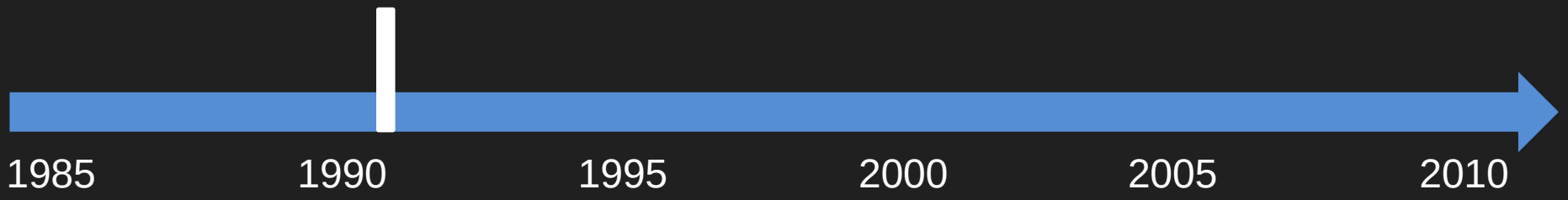
PCR-based methods are being developed for forensic use in Canada and elsewhere.



HISTORY

There is significant debate within the academic and legal communities regarding the quality and reliability of forensic DNA analysis. Initially, concern regarding laboratory procedures gives way to contention regarding how statistics are calculated when profiles match. Some have referred to this time period as the ‘DNA Wars’.

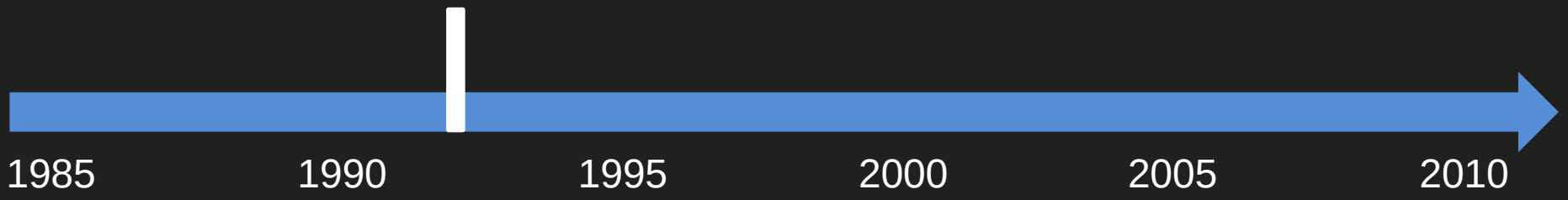
A report commissioned by the US National Research Council adds fuel to the debate and is roundly criticized.



HISTORY

PCR-based forensic DNA methods are first used by Canadian labs in casework.

Early applications of these tests demonstrate success in developing DNA profiles where Jeffreys' technique could not. They are also quicker. However, they are not as informative.

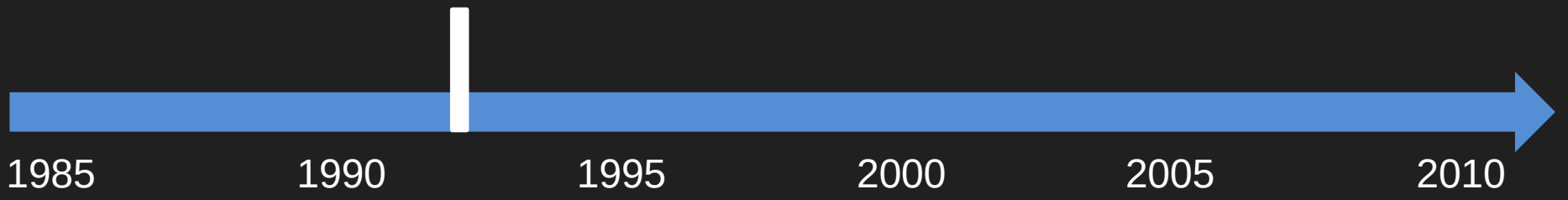


HISTORY

A specialty application for the analysis of mitochondrial DNA (mtDNA) is employed in casework in the UK.

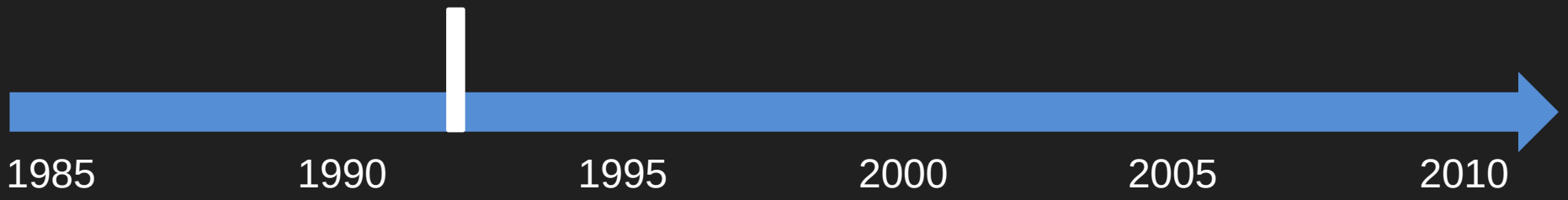
mtDNA is found in the power-generating mitochondria of cells. Unlike nuclear DNA, which is the target of traditional analyses, thousands of copies exist in every cell meaning that results can be generated with fewer cells than are required for nuclear DNA.

mtDNA is also maternally-inherited, meaning that all children share the exact same mtDNA type as their mother.



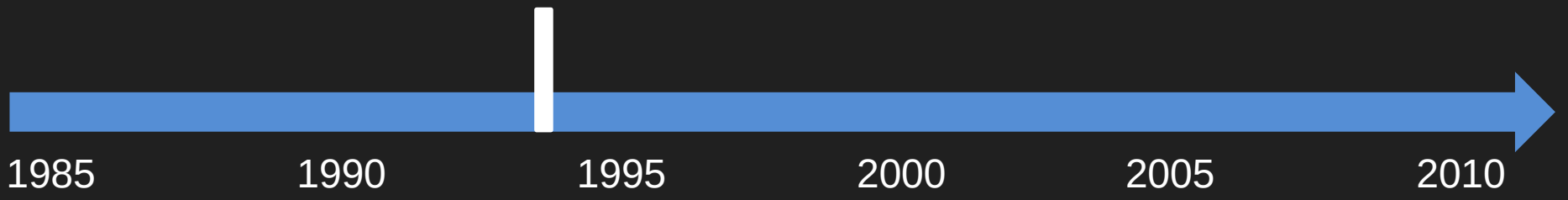
HISTORY

While the analysis of mtDNA has an important niche application to this day and can provide results in cases where no other type of analysis is possible (eg. single hair shafts and very old bones), nuclear DNA analysis remains the preferred choice of tests overall due to its much higher power of discrimination.



HISTORY

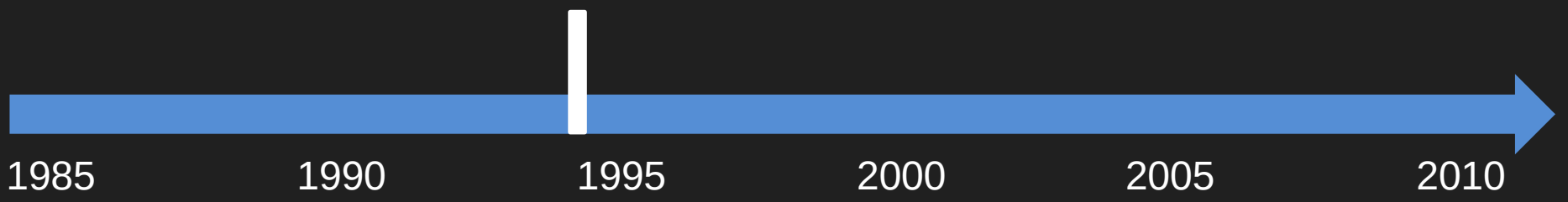
Leading proponents from each side of the ongoing DNA debate collaborate to publish a joint communication describing a consensus which effectively ends the ‘DNA Wars’.



HISTORY

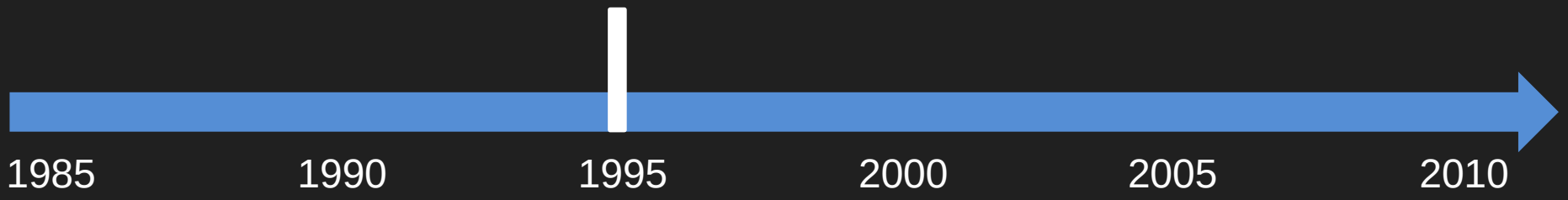
A new generation of PCR-based methods are developed and implemented in Canada. They detect variability which exists between individuals at certain locations along their DNA.

These locations, called *Short Tandem Repeats (STRs),* are similar to those targeted in the early technique pioneered by Jeffreys, except they are smaller and more likely to withstand the effects of degradation.



HISTORY

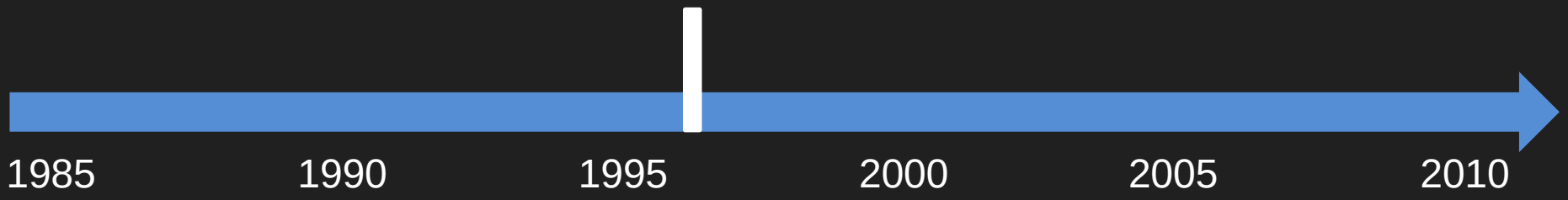
In Canada, Bill C-104 becomes law. The Bill allows judges to order a ‘DNA Warrant’ in order for police to obtain biological samples from persons so that their DNA profiles can be compared against DNA profiles developed from crime scene samples.



HISTORY

A second report is issued from a new committee of the US National Research Council pertaining to the quality of DNA analysis methods and the interpretation of DNA profiles, including population genetics and statistics.

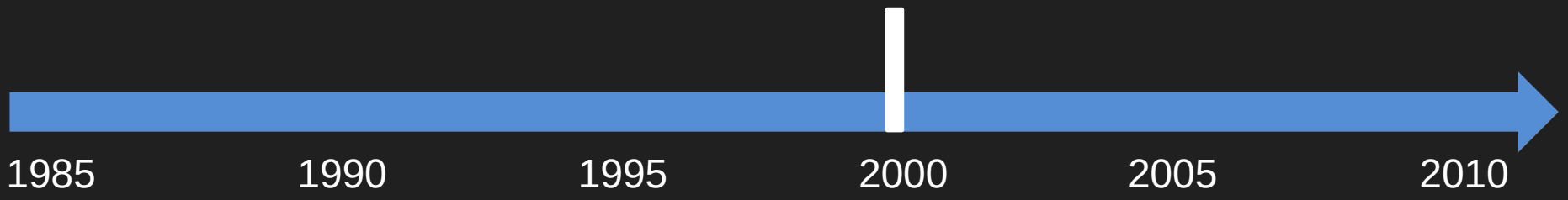
The report, colloquially known as *NRC II*, is widely accepted across the forensic community and its recommendations are followed to this day.



HISTORY

Canada implements a National DNA Databank, which compares the DNA profiles from scenes of crime to the DNA profiles obtained from persons convicted of designated offences.

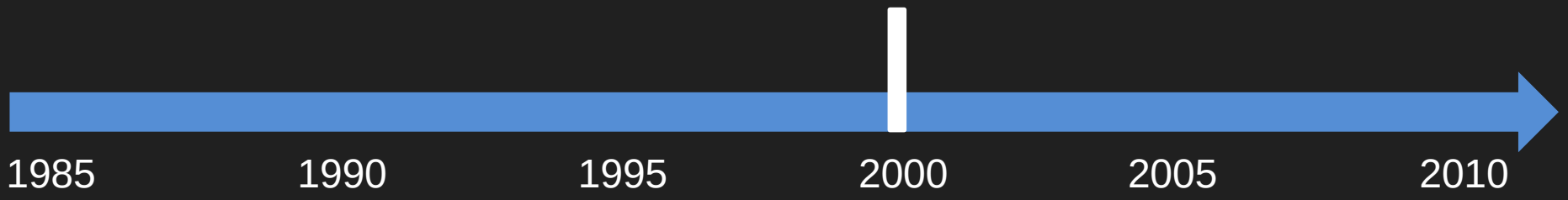
The databank requires that participating laboratories utilize the same PCR-based DNA analysis techniques so that results are comparable.



HISTORY

By now, a standard set of PCR-based tests are used by forensic laboratories throughout North America. The tests also share elements with those employed in Europe and elsewhere.

This commonality means that DNA profiles can be compared across countries, adding greater investigative value in some cases.

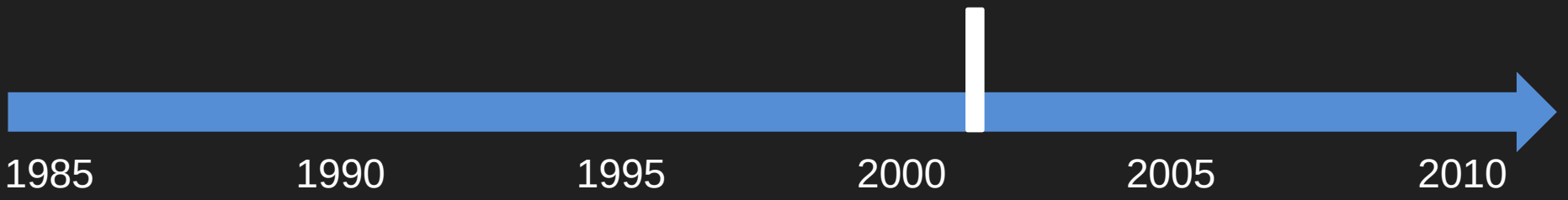


HISTORY

A specialty application for the analysis of male-specific Y-chromosome DNA is developed and implemented.

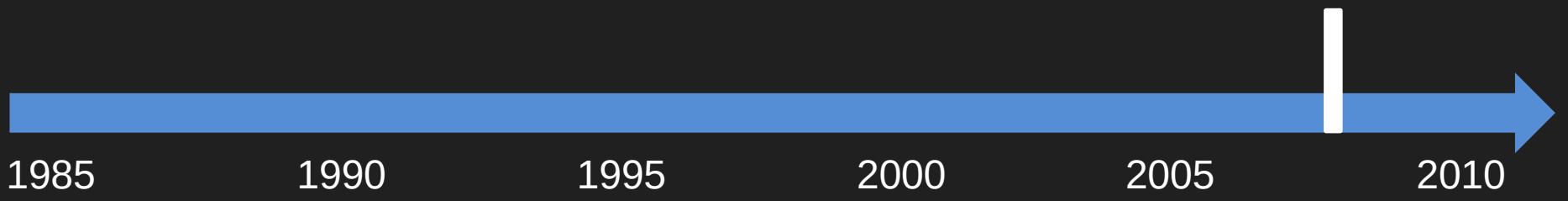
Because the test is specific for male DNA, it can be of high value in sexual assault cases where female-male mixtures prevail.

Y-STR DNA is paternally inherited by males only, meaning that all male children share the exact same Y-STR profile as their father.

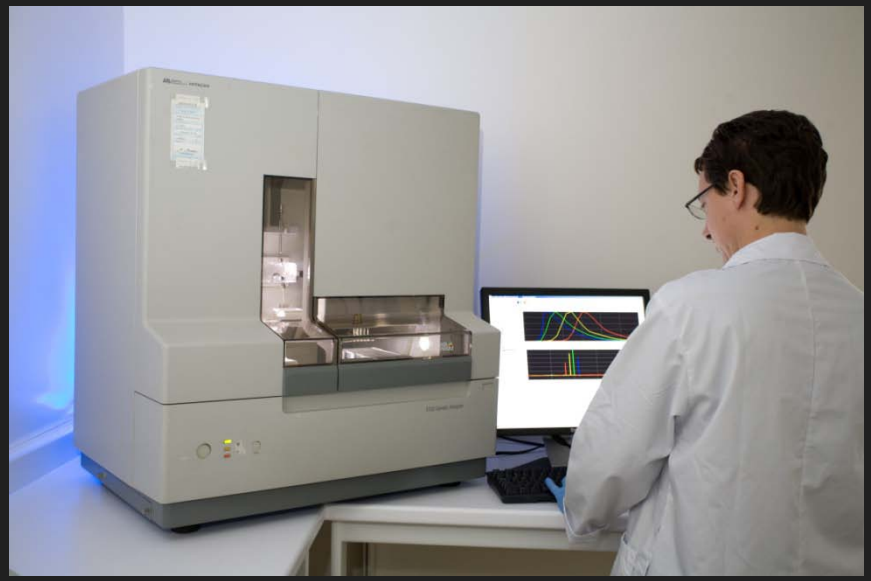


HISTORY

In Canada, Bills C-13 and C-18 (which received royal assent a few years earlier) are proclaimed and take effect. The legislation provides a number of amendments to the DNA Identification Act, including an expansion of the list of designated offences.



CURRENT STATE



- Since the advent of DNA databanks, technical advances in the field of forensic DNA analysis have shifted from the development of new tests to the development of faster ways of performing standardized tests through automation
- Throughout North America, a standard DNA analysis involves PCR testing of anywhere between 9 and 15 STR locations
- Occasionally and when warranted, male-specific Y-STR testing is utilized
- More rarely, mtDNA analysis is undertaken, though far fewer laboratories have the capacity to perform this test

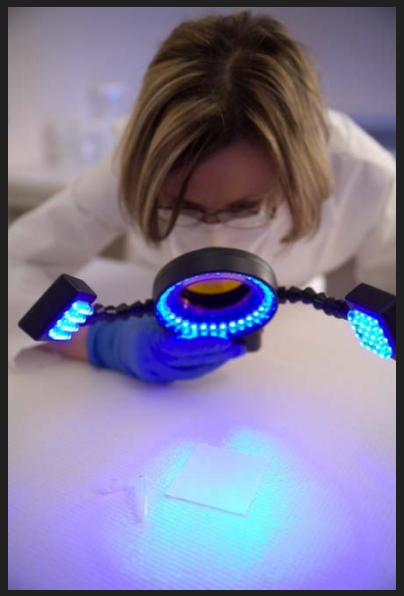
CURRENT STATE

- Forensic DNA analysis has more rigorous standards for quality than any other forensic science discipline
- Most laboratories performing forensic DNA analysis are accredited, though the process is voluntary in Canada
- Technical procedures for conducting DNA analysis are sound and generally accepted within the scientific community
- Many large laboratories have implemented robotic technology to automate and speed up portions of the analysis

CURRENT STATE

- Though errors can occur at any stage of the analysis process, the interpretation of results poses a particular risk
- Interpretation can be problematic with very low levels of DNA and/or mixed sources of DNA
- A high proportion of cases where DNA is tested have samples where one or both of these conditions prevail
- Interpretation in such cases can be subjective, and relies on key assumptions and individual opinions that may not always be correct

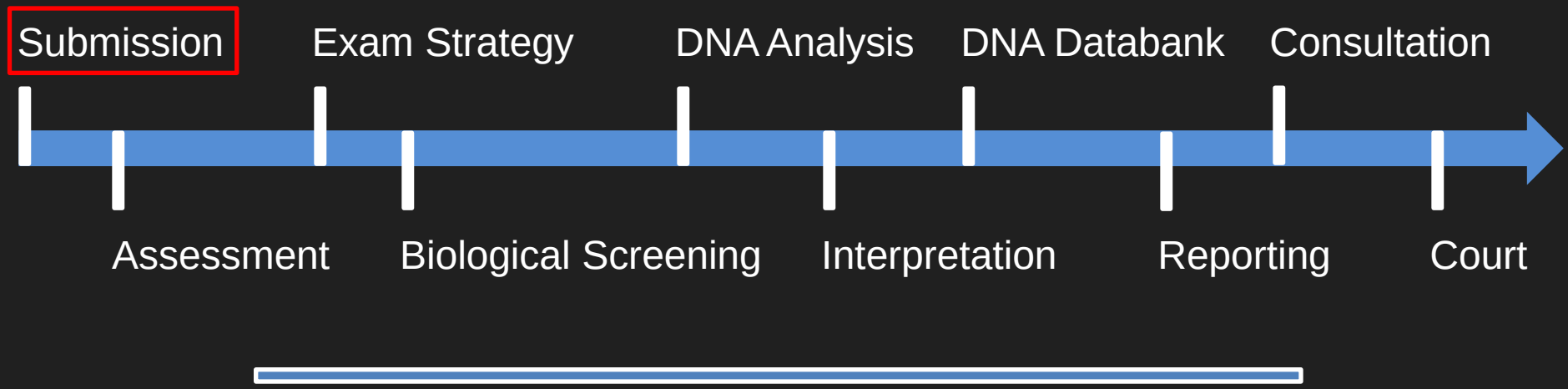
LABORATORY PROCESS



The following slides outline a general flow for a forensic laboratory process. It may be slightly different from laboratory to laboratory.

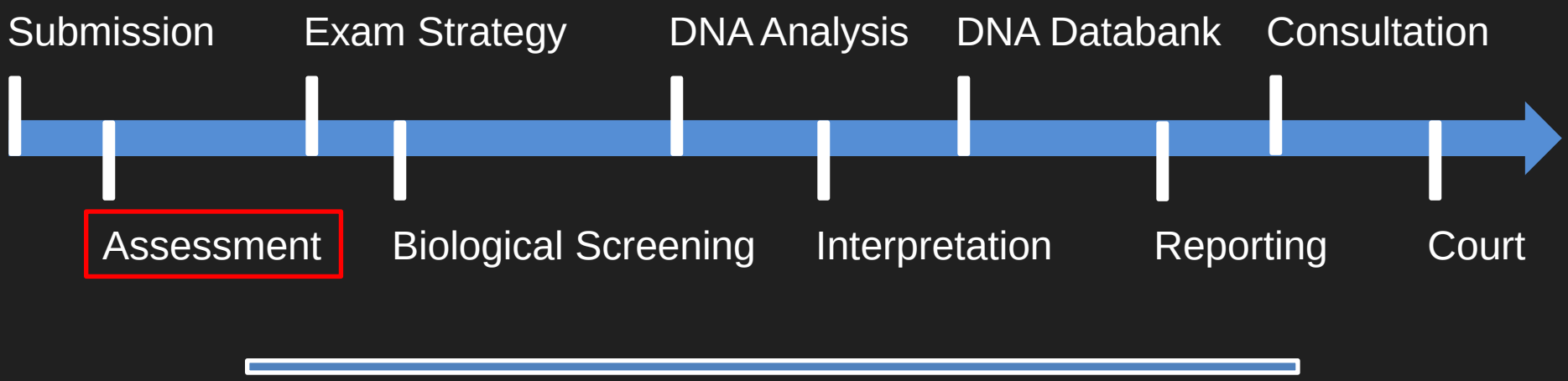
Submission
Assessment
Exam Strategy
Biological Screening
DNA Analysis
Interpretation
DNA Databank
Reporting
Consultation
Court

LABORATORY PROCESS



Delivery of physical items to the laboratory for testing. Often, only a subset of items collected in relation to a crime are submitted. In many cases, consultation between laboratory staff and submitters can assist in focusing submissions so that only the most pertinent items are sent in. However, items that have not been submitted may be very important to test in order to address alternate theories of a crime.

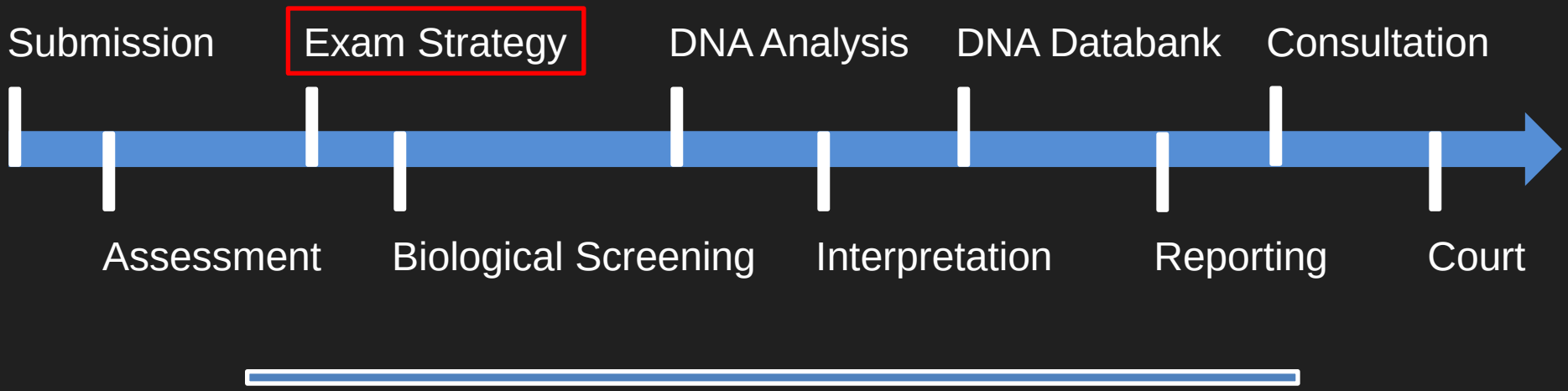
LABORATORY PROCESS



A review of the alleged circumstances of a crime, along with the list of submitted evidence items. From this review, scientists determine the most suitable item exam strategy to address the circumstances of the case.

Theories of the crime which differ from the one used in this assessment may affect the item exam strategy.

LABORATORY PROCESS

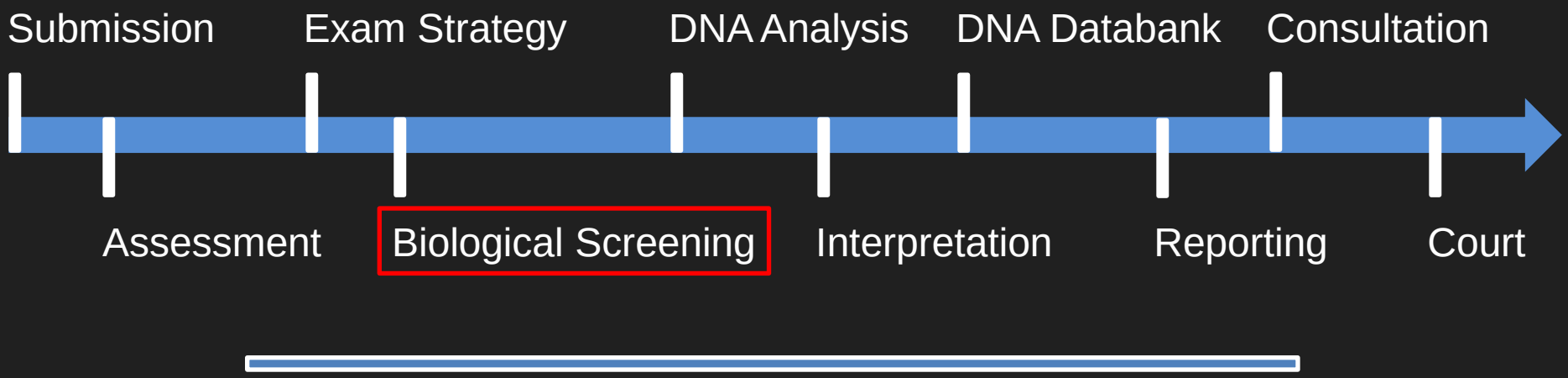


The determination as to which of the submitted items to examine, and what specific tests are to be employed in the examination.

These include tests at both the biological screening phase and the DNA analysis phase of the process. Exam strategies may be altered as the testing progresses.

The exam strategy is dependent on the assessment, which in turn is based on the account of the circumstances provided to the laboratory.

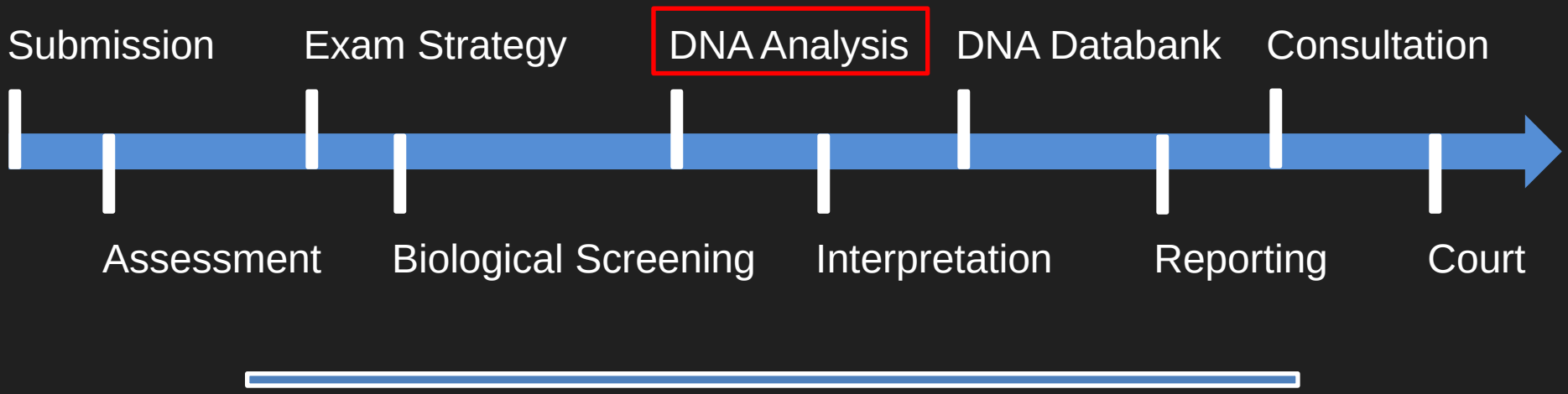
LABORATORY PROCESS



Usually, biological stains (eg. blood, semen, saliva) must be located and identified on items of evidence before they can be subjected to DNA analysis. This process involves meticulous and often labour-intensive searching of evidence items in addition to the application of simple biochemical and other tests.

It is important to not overlook the criticality of this human-driven stage in the process. If evidence is missed during the screening process, it may have grave consequences.

LABORATORY PROCESS



A series of systematic laboratory methods used to develop a DNA profile from a sample selected for analysis. Often, only subsets of samples identified during the biological screening stage are selected for DNA analysis. Decisions as to which samples to process for DNA analysis may be based on assumptions and opinions that are not always correct.

A schematic of the DNA analysis process may be found on the next slide…

DNA ANALYSIS

Cellular Material from Body Fluid Stain

Extract and Purify

DNA Extract

Quantify

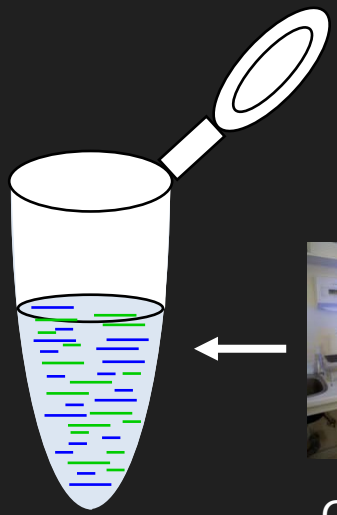
Sufficient?

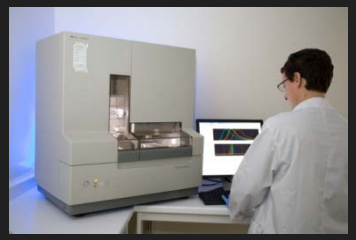
Yes

No

STOP

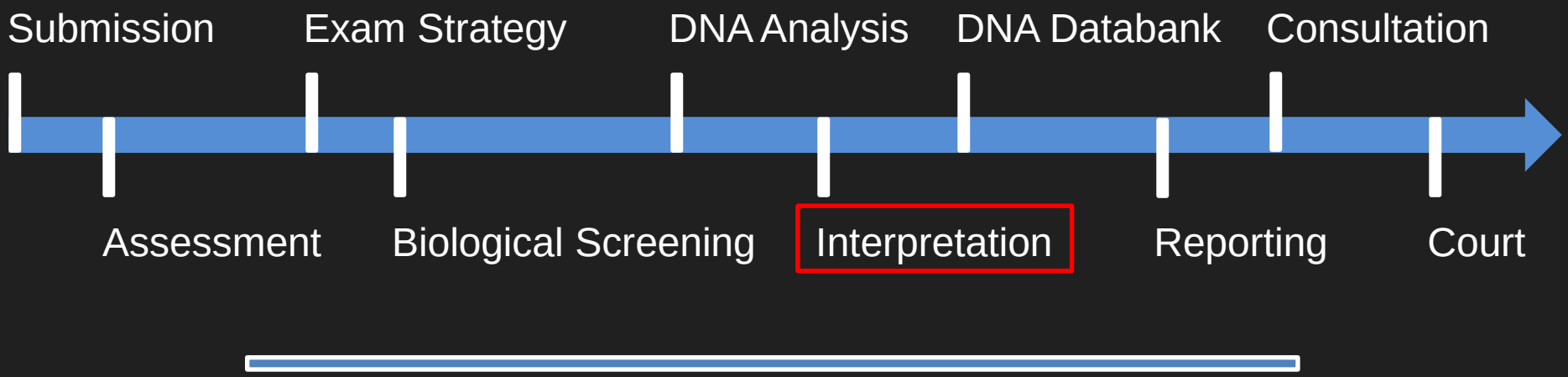
Copy Locations of Interest





Separate Copied Fragments

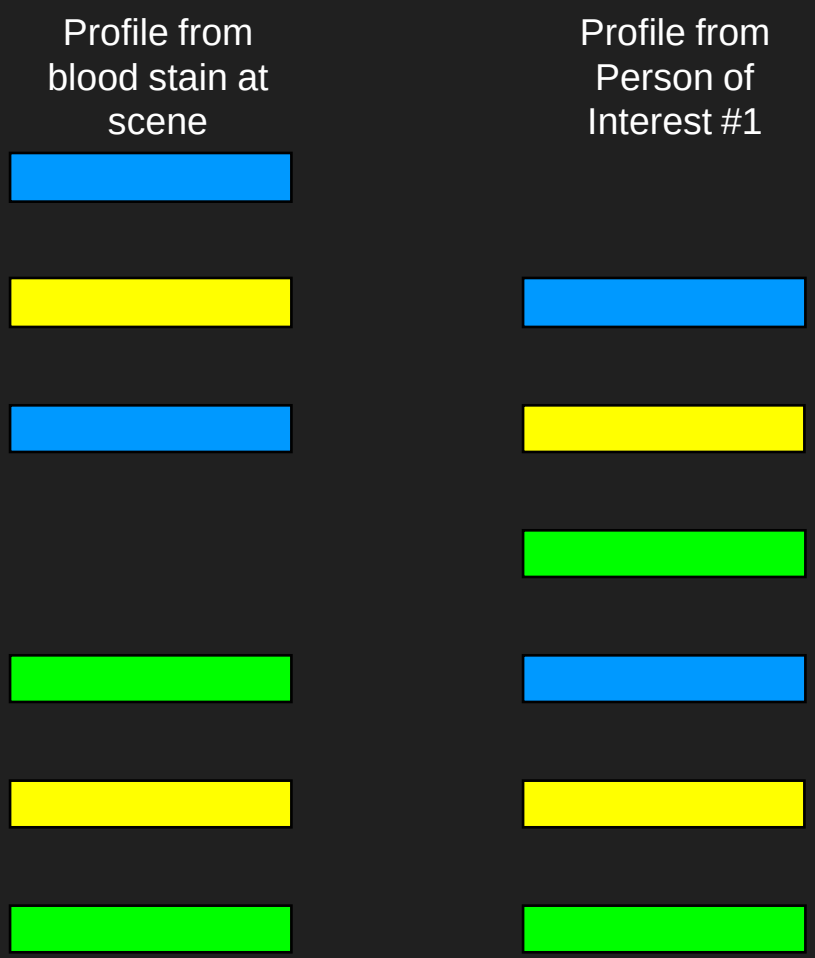
LABORATORY PROCESS



Interpretation involves analyzing raw data from the DNA analysis process, identifying artefacts, determining the genotype (or profile) associated with the results and comparing profiles from different samples, including known profiles from persons of interest.

This can be relatively straightforward for some profiles, given the appropriate training and expertise, but is far more complex when dealing with mixed profiles or profiles generated from very small amounts of DNA.

INTERPRETATION



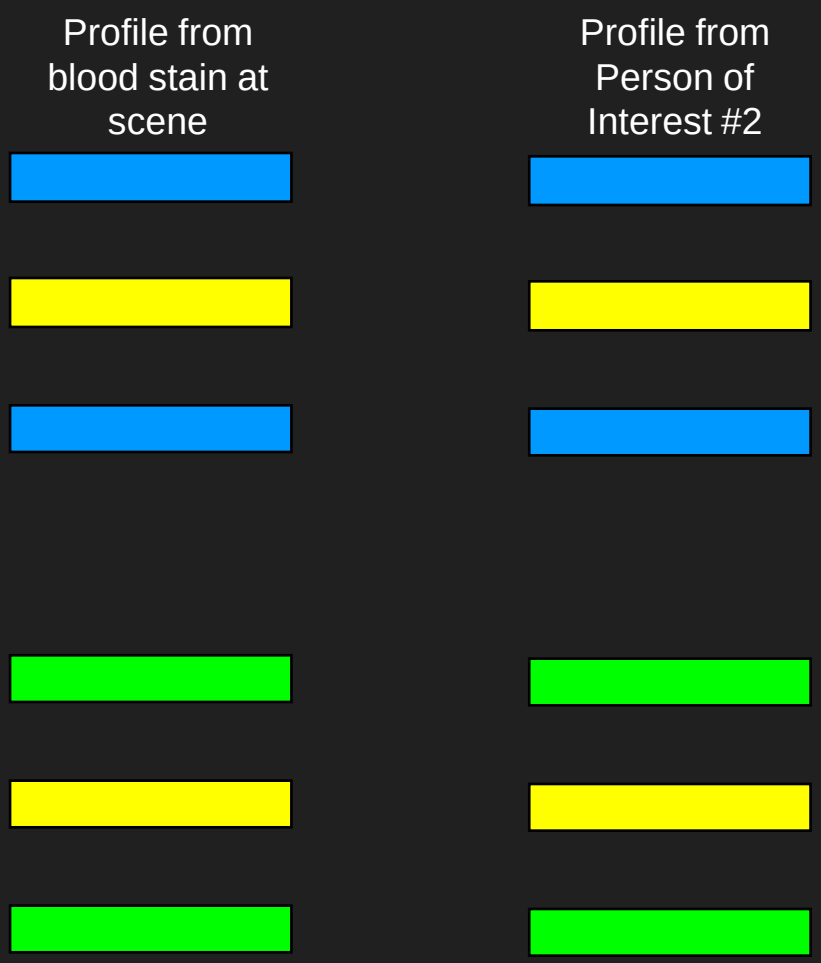
DNA profiling essentially involves pattern matching

The image to the left is a simplified way of depicting physical DNA profiles, with each column representing a profile from a different sample

From the patterns observed, it is clear that the profiles from Person of Interest #1 and the blood stain from the crime scene do not match

Therefore, Person of Interest #1 is excluded as the contributor of the profile from the blood stain

INTERPRETATION

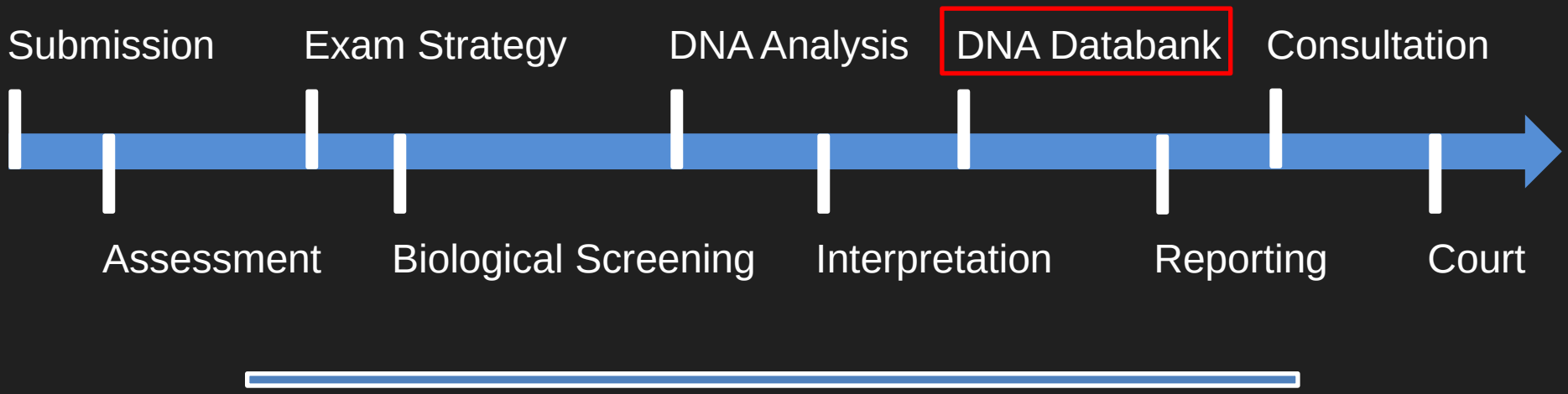


On the other hand, the profile from Person of Interest #2 clearly matches the profile from the blood stain at the scene

A scientist would therefore conclude that Person of Interest #2 cannot be excluded as the source of the blood stain at the scene

A statistical statement of weight, such as a random match probability, would normally be attached to such a statement

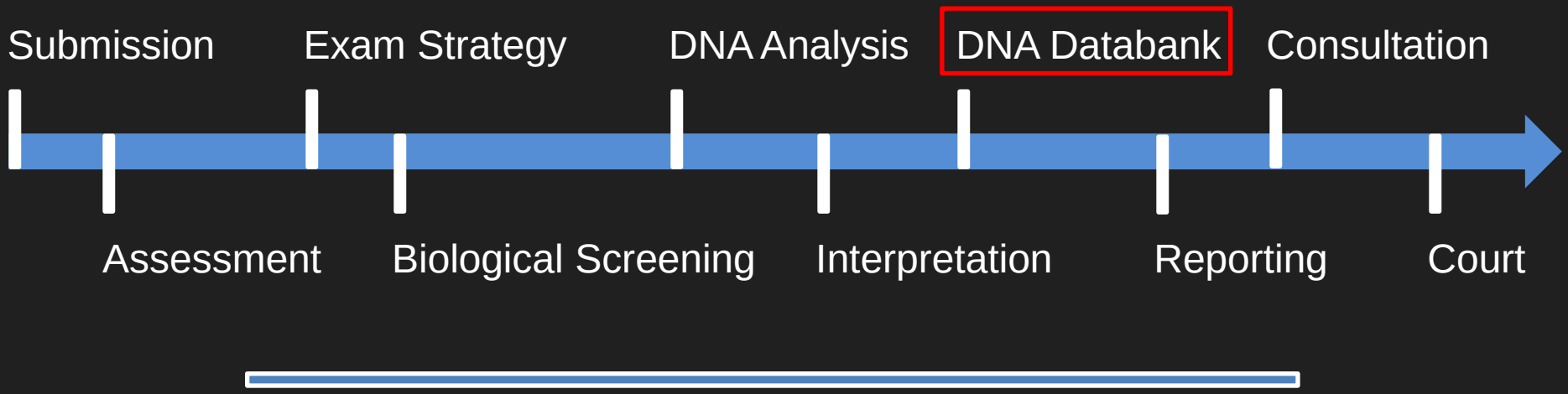
LABORATORY PROCESS



DNA profiles can be recorded in a simple numerical format (an example is provided later in the presentation), which means that they can be readily searched against other profiles using computer software.

Eligible DNA profiles from crime scene samples are entered into the DNA Databank to be searched against other crime scene profiles and to be searched against known profiles from individuals convicted of designated offences.

LABORATORY PROCESS

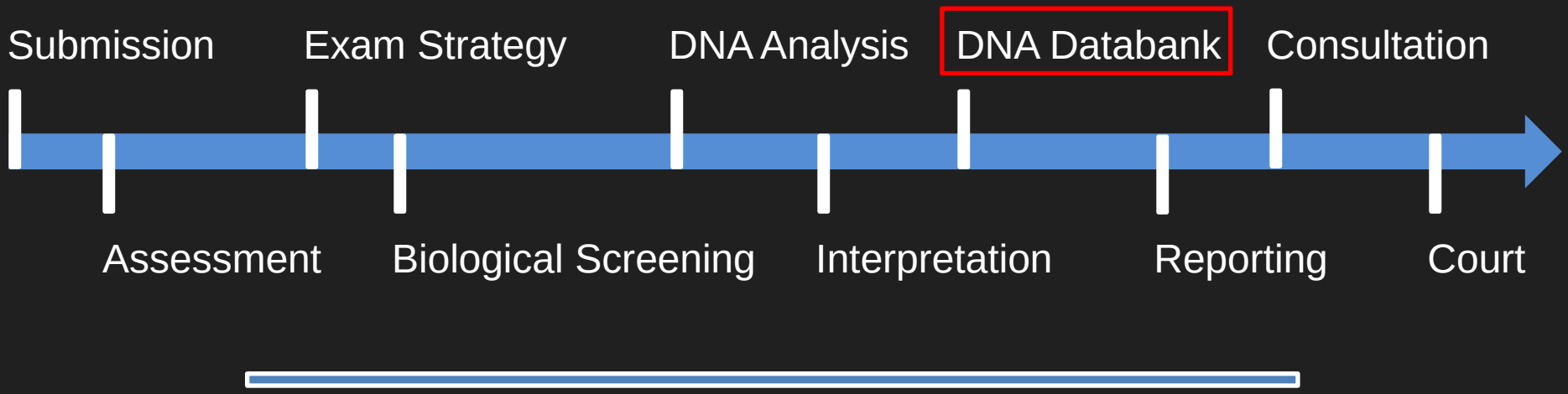


Thus, determining crime scene profiles is of value even when specific comparison samples have not been submitted to the laboratory.

Databank hits provide investigative information which is often not otherwise available.

In Canada, hits to offender profiles usually spur an application for a DNA Warrant sample which is then profiled in the originating laboratory.

LABORATORY PROCESS

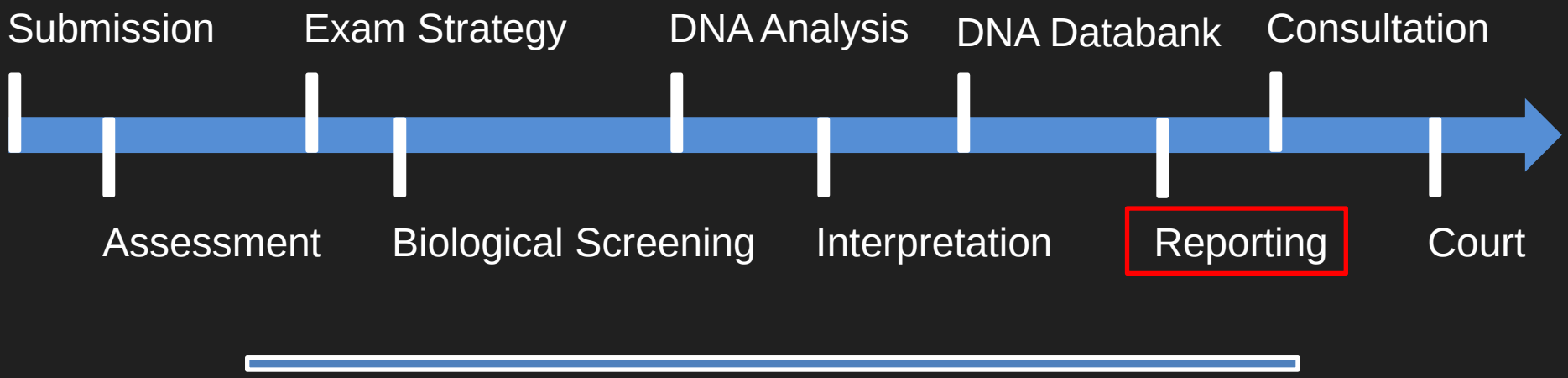


The effectiveness of databanks is dependent, in part, on the quality of the profile information included.

Misinterpreted crime scene DNA profiles may ‘hit’ to profiles and individuals with no association to the events at hand.

Partial, or degraded, crime scene profiles may ‘hit’ ’ to profiles and individuals with no association to the events at hand.

LABORATORY PROCESS

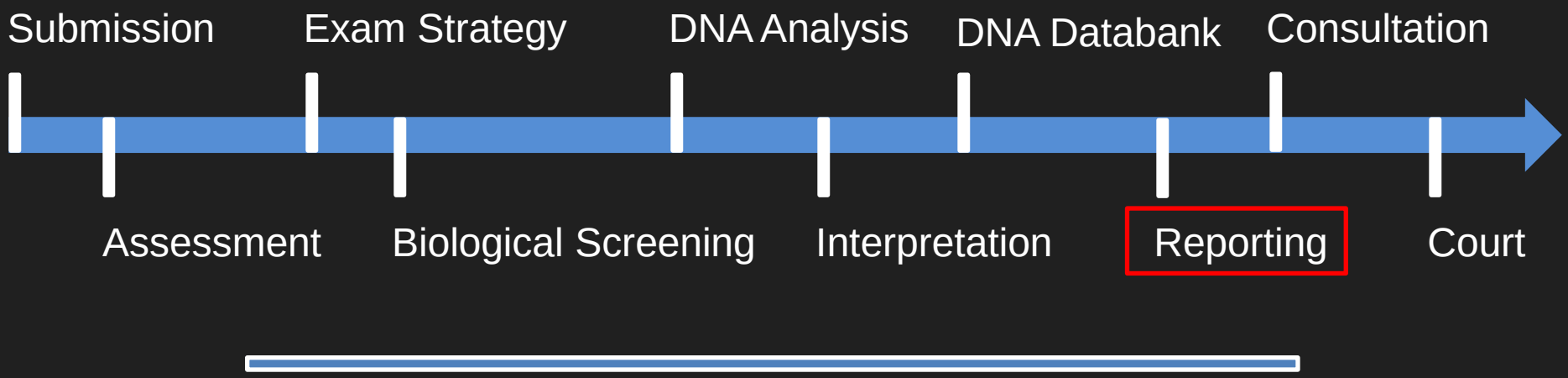


There is considerable variation from laboratory to laboratory in the way by which forensic biology/DNA results, conclusions and opinions are conveyed.

Whatever the format, language used generally has very specific meaning but can nevertheless sometimes be interpreted by different people to mean different things.

It is vital that limitations be clearly addressed in all forensic science reports.

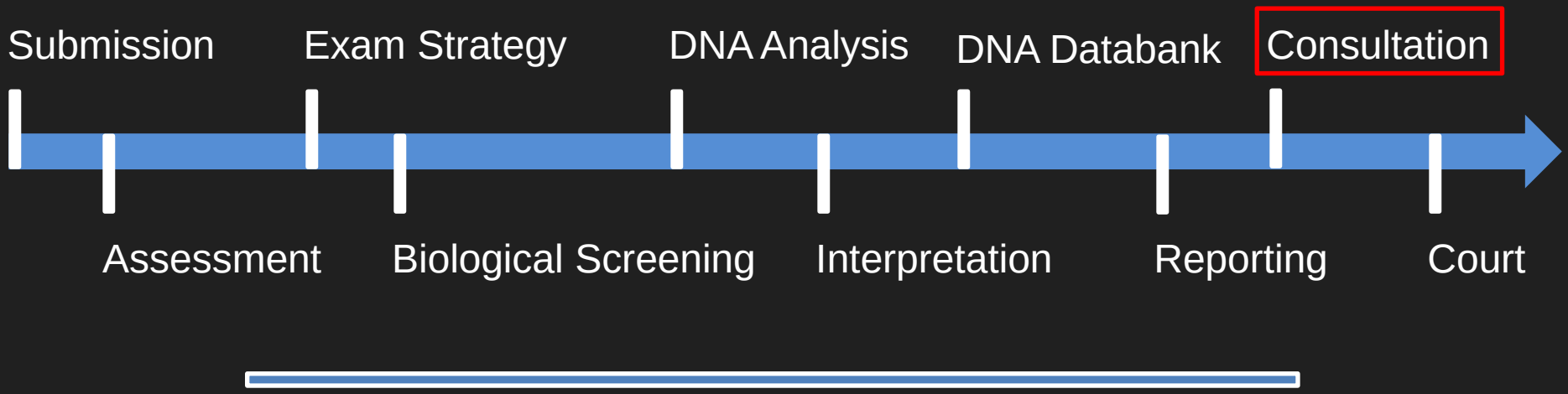
LABORATORY PROCESS



Reports may be silent on very important issues of context.

For example, a forensic DNA report may provide very definitive information regarding who a DNA profile belongs to; however, there may be numerous means by which that profile came to be deposited where it was ultimately detected, some of which could be unrelated to any crime.

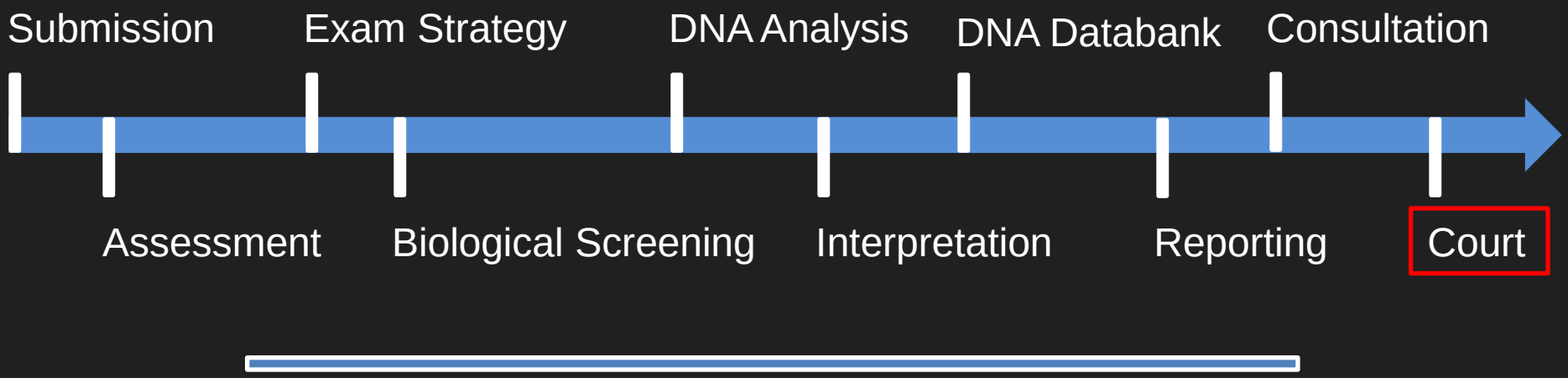
LABORATORY PROCESS



For this and other reasons, preparing for an effective presentation of expert evidence at court requires advance discussion and preparation on the part of the expert and the appropriate counsel.

Many government forensic scientists will make themselves freely available to consult with defence counsel in advance of trial, though confidentiality cannot necessarily be assured.

LABORATORY PROCESS



Experts and counsel, together, have a joint responsibility to ensure that all results and opinions are fully and fairly presented, and that evidence is provided no more or less weight than it deserves.

The format for presenting evidence in court may vary from simple oral evidence to prepared presentations, depending on the focus of the evidence and the issues at hand.

LANGUAGE OF DNA PROFILING

Amelogenin	XX
D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

The following section describes a number of terms used in DNA profiling…

The depiction on the left is of a DNA profile developed at 9 STR locations (plus the Amelogenin location which indicates the gender of the profile), recorded in numeric format.

LANGUAGE OF DNA PROFILING

LOCUS

a specific test site on the DNA

(plural = loci)

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ALLELE

variant form of DNA

A person has two alleles at each locus, one inherited from their mother, and one from their father

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GENOTYPE

the combination of alleles found at any given locus

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GENOTYPE

the combination of alleles found at any given locus

also extends to describe alleles found at all loci tested

LANGUAGE OF DNA PROFILING

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HETEROZYGOTE

a single-locus genotype with two different alleles

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D7S820	8,11

HOMOZYGOTE

a single-locus genotype with two copies of the same allele

STRENGTHS & LIMITATIONS

Forensic DNA analysis, when performed properly by qualified technical and scientific staff in an accredited facility, is rightly considered one of the most powerful tools for the individualization of bodily substances

As with any laboratory analysis, however, errors can and do happen

Even when error-free, forensic DNA analysis has a number of limitations – many of these will be case-specific but the following slides outline a number of general strengths and limitations that should always be considered

STRENGTHS & LIMITATIONS

STRENGTH

Forensic DNA analysis can be extremely discriminating. Random match probabilities are often so low as to essentially pinpoint an individual as the source of a bodily substance to the exclusion of all others.

LIMITATION

The random match probability is reported when an individual (e.g. Mr. X) cannot be excluded as the source of a DNA profile. It is the probability that a randomly selected person unrelated to Mr. X would coincidentally share the observed DNA profile from the crime scene sample.

Relatives of Mr. X will have a higher probability of coincidentally sharing the crime scene profile than randomly selected people would.

STRENGTHS & LIMITATIONS

STRENGTH

Forensic DNA analysis is extremely sensitive, meaning that DNA profiles can be successfully determined from very low quantities of DNA.

While blood, semen, and saliva are all rich sources of DNA and are commonly encountered in casework, DNA can also be profiled from touched or handled surfaces where small numbers of skin cells have been left behind.

LIMITATION

It's not uncommon to find low levels of DNA on various surfaces and objects, whether a crime has occurred or not.

A very important question when dealing with profiles from low-level DNA is whether the DNA detected was deposited during the alleged events.

Could it have been deposited prior to the alleged events? After the alleged events but prior to sample collection? Or even following sample collection prior to DNA testing?

STRENGTHS & LIMITATIONS

STRENGTH

DNA profiles can be successfully developed from bodily fluids and from other sources such as hairs and shed skin cells.

LIMITATION

Searching evidence for samples on which to perform DNA testing is challenging. The process is low-tech, meticulous, labour-intensive and subject to error. The likelihood, for instance, of finding a blood stain that is no larger than the period at the end of this sentence somewhere in the recesses of a vehicle is relatively low.

In some instances, searching is altogether impossible. There is no reliable way to localize shed skin cells. Success relies on sampling areas of expected deposition based on the item type or taking educated guesses as to the location based on the case history provided to the lab.

STRENGTHS & LIMITATIONS

STRENGTH

Multiple testing systems are available to forensic scientists for use under various circumstances, thereby increasing the likelihood of successfully generating DNA profiles.

While the standard STR testing is the preferred choice given its power of exclusion, it is not always suitable.

In some cases, mtDNA or Y-STR testing may be preferable.

LIMITATION

A failure to exclude someone using either mtDNA or Y-STR testing is different than when using standard STR testing.

mtDNA is passed exclusively from mothers to their children such that all maternal descendants share the same profile. Unrelated individuals may also share the same profile.

The Y chromosome is passed exclusively from fathers to their sons such that all paternal male descendants share the same profile. Unrelated males may also share the same profile.

STRENGTHS & LIMITATIONS

STRENGTH

Current technologies may permit individual DNA profiles to be readily determined even when mixed in samples with DNA from other individuals.

Mixtures of DNA are prevalent in forensic science.

LIMITATION

Mixtures of DNA can be very complex and are one of the most significant challenges to forensic scientists.

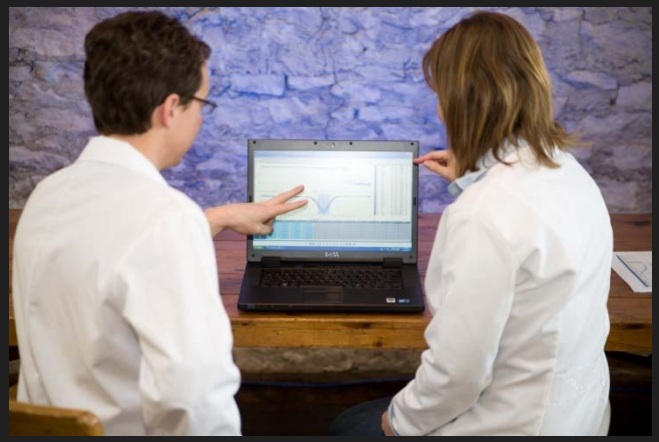
In many cases, interpretation is required to properly analyze a mixed sample. Interpretation, in turn, can be subjective and relies on assumptions and opinions which may not always be true.

Mixtures of DNA that involve low levels of DNA can pose an even greater challenge and interpretation may carry a higher risk of error.

CONCLUSION

- Forensic DNA analysis has had a tremendous positive impact in the criminal justice system, but its reliability should not be taken for granted
- Despite high standards for quality, errors can and do happen
- Moreover, there are a number of limitations to any forensic DNA analysis, even when it is properly conducted
- The context and limitations of DNA results are different in each case

ABOUT Wfg



- Privately-owned Canadian company committed to the timely delivery of forensic biology/DNA laboratory test results and opinions to all interested parties in the justice system.
- Incorporated March 2009
- Services:
  - Forensic Biology/DNA Testing (upon accreditation)
  - Forensic Biology/DNA Casework Consultation
  - Training and Professional Development
  - Laboratory Management and QA Consultation
  - Laboratory Auditing

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