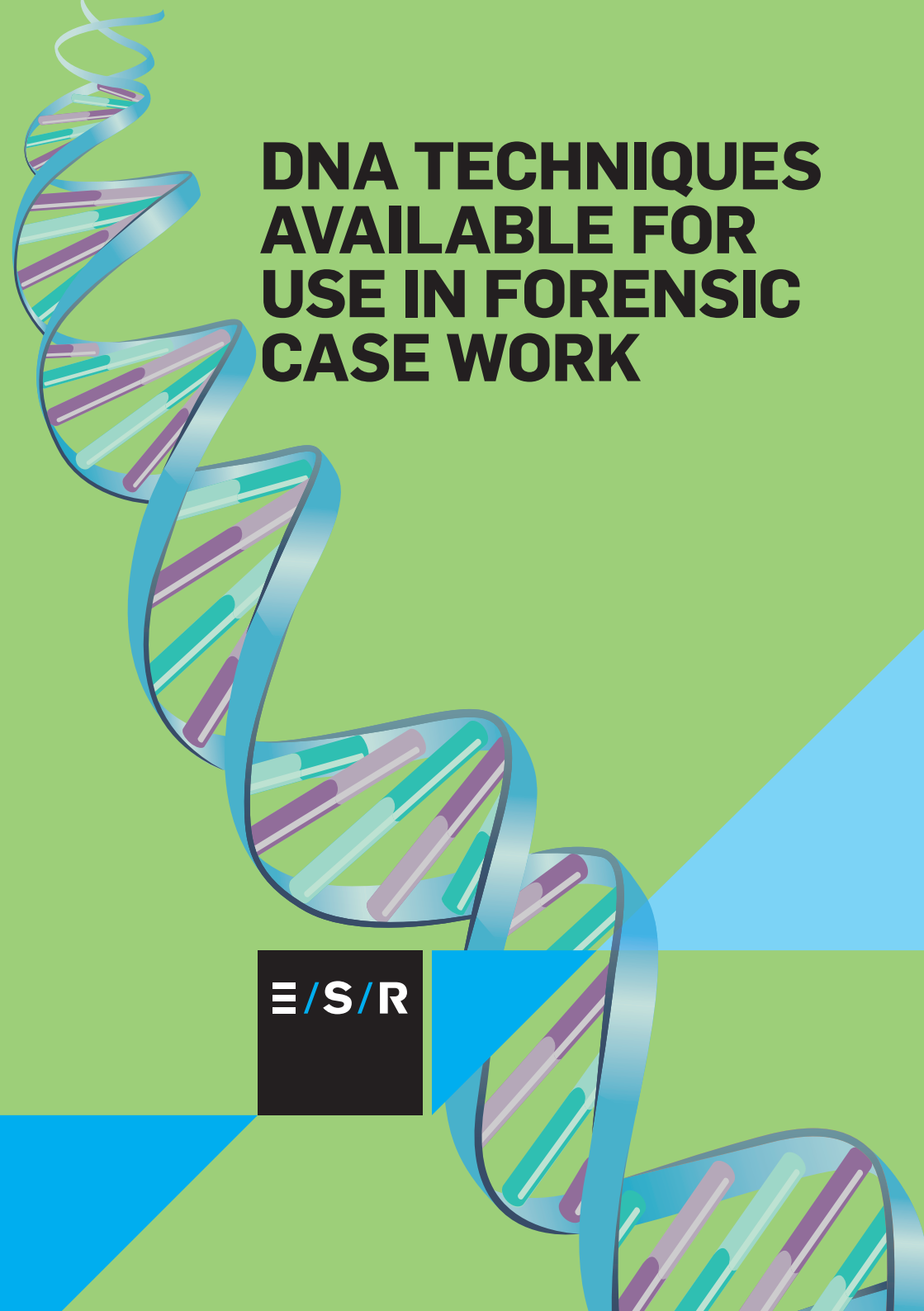
DNA TECHNIQUES AVAILABLE FOR USE IN FORENSIC CASE WORK



ESR

FOR FORENSIC ASSISTANCE
24 HOURS A DAY, 7 DAYS A WEEK
0800 FORENSIC
0800 367 367

KEY CONTACT

Dr Stephen Cordiner
Forensic Manager – Service Centres
E: stephen.cordiner@esr.cri.nz

STANDARD DNA AND SPECIALIST TECHNIQUES AVAILABLE FOR USE IN FORENSIC CASEWORK

It seems hard to believe that two decades ago, using DNA to fight crime was unheard of. The development of DNA profiling revolutionised forensic science and the investigation of crime worldwide.

In New Zealand, DNA profiling is now used routinely to:

- investigate a wide range of crimes from burglaries to homicides
- identify suspects and exclude the innocent, reducing police investigation time
- solve historic cases
- assist in the reconstruction of crimes and crime scenes
- assist in identifying human remains, including those from disaster events
- assist in forensic paternity investigations.

ESR is at the forefront of research and development internationally with a thriving research and development programme.

Our expertise is recognised throughout the world.

Here we outline the range of techniques currently offered by ESR to the criminal justice community based on DNA and its sister compound RNA.

The techniques you will find described are:

- **STANDARD DNA PROFILING** tests used to deliver highly discriminating results in most cases
- **LOW COPY NUMBER (LCN) DNA** profiling tests for highly sensitive testing of very small amounts of DNA
- **MINIFILER™ DNA** profiling used for analysing very degraded DNA
- **Y STR DNA** profiling used for selective analysis of the Y chromosome found only in males

Buccal cells

- **mRNA** analysis for the detection of body fluids
- **LASER MICRODISSECTION** for the isolation of specific cells for further testing.

All of these techniques use a method known as the Polymerase Chain Reaction (PCR) to obtain profiles. This is a standard technique used to amplify or selectively copy specific regions of DNA or RNA many times. In this way, minimal amounts of DNA or RNA isolated from small or degraded samples can be increased to a level where they are able to be detected, profiled and compared with other samples.

In addition, we are now using STRmix™ – expert forensic software that can resolve previously unresolvable mixed DNA profiles.

http://STRMIX.esr.cri.nz

For further information on testing in relation to your own case please contact a Service Centre Case Manager or the Service Centre Duty Scientist on 0800 FORENSIC (0800 367 367).

STANDARD (AUTOSOMAL) DNA PROFILING

Human DNA is packaged in 46 chromosomes: 22 autosomal pairs, and either two X chromosomes in females or one X and one Y chromosome in males. The majority of DNA profiling carried out at ESR uses systems that target autosomal DNA sites. These DNA sites are found in both males and females and are highly variable amongst individuals, thereby delivering highly discriminating results. Currently, the standard DNA profiling test is used on routine sample types such as blood, semen and saliva stains as well as body tissue, hair and bone. It includes a gender test.

These DNA profiles can be:

- compared with reference DNA profiles from suspects, complainants and those providing samples for elimination purposes
- searched against the DNA Profile Databank to identify possible suspects
- searched against the Crime Sample Database to link crimes
- used for establishing family relationships
- used for familial searching
- used in missing person enquiries and DVI (Disaster Victim Identification).

DETECTS: Currently fifteen autosomal DNA regions (sites) and a gender test.

SENSITIVITY: Good sensitivity for routine samples.

DISCRIMINATING POWER: Very discriminating. It is highly unlikely that two unrelated people will have matching DNA profiles using the current system.

DATABANK: Generated profiles can be searched against the DNA databank.

TURNAROUND TIME: Five days for volume crime. Complex cases determined by circumstances.

CASE EXAMPLES:

- Blood left at a burglary scene is linked to an individual on the DNA Profile Databank within the five day turnaround time for volume crime cases, resulting in a swift arrest.
- A DNA profile from semen found on Marie Jamieson (homicide case) undergoes familial searching leading to the apprehension of Joseph Reekers.
- DNA profiles from post mortem samples were used to identify the deceased after the Christchurch earthquake.

LOW COPY NUMBER (LCN) DNA TESTING USING SGM PLUS®

Low Copy Number (LCN) DNA profiling is used mostly on contact samples where there are very small amounts of DNA. It is the only DNA profiling test currently available that can be used on these types of samples. The sensitivity of the test requires strict procedures for the entire testing process.

It is highly recommended that ESR staff should carry out LCN sampling.

DETECTS: 10 variable DNA regions and a gender test. It is fully compatible with standard DNA profiling.

SENSITIVITY: LCN DNA profiling is the most sensitive DNA test currently available. It provides results with only trace amounts of DNA present.

DISCRIMINATING POWER: High. It is very unlikely that two unrelated people will have matching DNA profiles.

LIMITATIONS: Due to the sensitivity of the test, mixtures of DNA are frequently detected which may make it uninterpretable. Careful targeted sampling may assist in reducing potential mixed cell samples.

DATABANK: Generated profiles can be searched against the DNA databank.

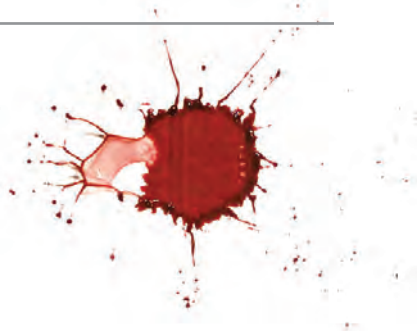
TURNAROUND TIME: Because this is complex testing turnaround times vary depending on the samples.

Markers indicate possible body fluids

Luminol highlights body fluids

CASE EXAMPLES:

- DNA recovered from a smudged hand/fingerprint mark.
- DNA recovered from a handled item such as a knife or other weapon.
- DNA recovered from trace amounts of body fluid such as blood after luminol detection.



MINIFILER™ DNA ANALYSIS

Minifiler™ DNA profiling targets smaller lengths of DNA to produce better results when testing smaller amounts of DNA sequences. This is a useful test for samples where the amount of DNA present in a sample is low, or the DNA has been degraded due to the age of the sample or because of environmental conditions.

Minifiler™ may also be useful when samples contain inhibiting substances, such as soil and fabric dyes, that could interfere with the standard DNA profiling test. Minifiler™ results can also be combined with partial DNA profiling results from standard and/or LCN DNA profiling tests to provide a more complete profile.

The Minifiler™ DNA profiling test analyses eight of the DNA sites used in the standard DNA test. Although fewer DNA sites are analysed compared to the standard DNA profiling test, the Minifiler™ DNA test is more sensitive.

DETECTS: Eight variable DNA regions, a gender test and is fully compatible with standard DNA profiling.

SENSITIVITY: The sensitivity falls between the standard DNA profiling test and the LCN DNA profiling test. Minifiler™ is not as sensitive as LCN DNA testing.

DISCRIMINATING POWER: On their own, Minifiler™ results are not as discriminating as standard testing. However, Minifiler™ results can be combined with profiling results obtained from other techniques for greater discrimination. A full Minifiler™ profile will still meet ESR's most discriminating 'extremely strong support' category.

DATABANK: Minifiler™ DNA profiling results are compatible with the DNA Profile Databank and the Crime Sample Database.

TURNAROUND TIMES: The turnaround time for Minifiler™ DNA profiling is similar to that for standard DNA profiling.

CASE EXAMPLES:

- An historic sexual assault where standard, LCN and Minifiler™ tests were used to obtain sufficient DNA profiling results to load to the Crime Sample Database resulting in a link.
- A bone washed up on a beach gave no results using the standard DNA test but sufficient results were obtained from Minifiler™ to allow comparison to reference samples.
- A swab of blood traces from a knife recovered two months after an assault gave no results using the standard test, but sufficient DNA profiling results were obtained for comparison purposes using the Minifiler™ test.

Y STR – Y CHROMOSOME DNA ANALYSIS

The Y STR profiling test targets only male DNA that may be present in a particular sample by analysing DNA sites on the Y chromosome only. Therefore, DNA from females is not detected using this technique.

The main advantage of the Y STR DNA profiling system is that it selectively targets male DNA even in the presence of large amounts of female DNA. This means that results can be obtained from very small amounts of male DNA, which were not previously possible.

Y chromosomes are passed from father to son; therefore paternally related male individuals cannot be distinguished using this current Y STR technique.

Lower genetic diversity has been found in populations of Polynesian origin due to the nature of recent population movements within these groups. In some cases this is reflected in a decrease in statistical significance of any correspondence found.

Y STR DNA profiles can be:

- compared to reference samples from male individuals
- used to support family relationships and are often used in missing persons or disaster victim identification work.

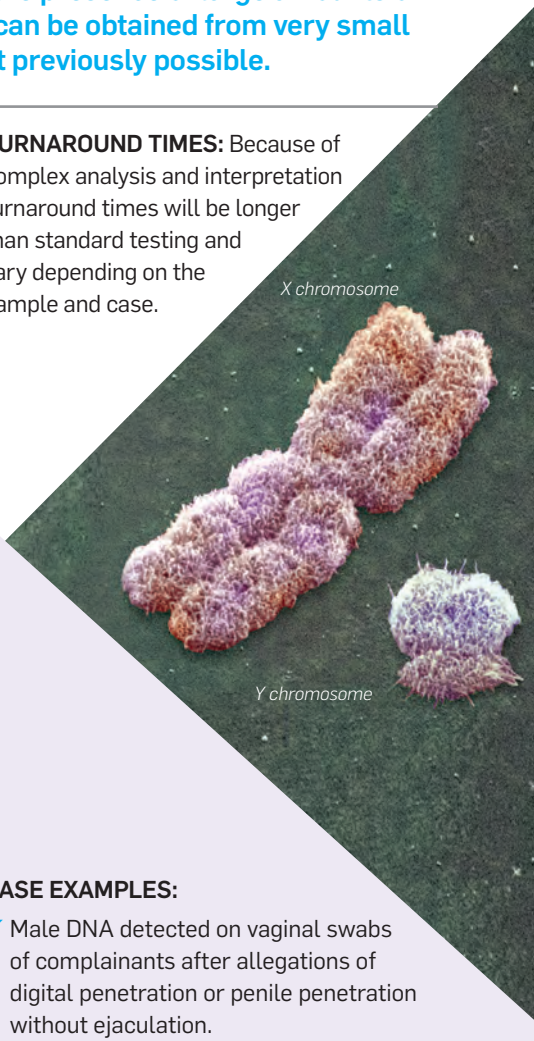
DETECTS: 25 STR loci on the Y chromosome.

SENSITIVITY: Results can be obtained from very small amounts of male DNA.

DISCRIMINATING POWER: Cannot currently discriminate between father and sons or other paternally related males.

DATABANK: Y STR DNA profiles cannot be compared to the DNA Profile Databank.

TURNAROUND TIMES: Because of complex analysis and interpretation turnaround times will be longer than standard testing and vary depending on the sample and case.



CASE EXAMPLES:

- Male DNA detected on vaginal swabs of complainants after allegations of digital penetration or penile penetration without ejaculation.
- It has been used to estimate the number of male contributors to samples containing low levels of DNA from more than one male.

mRNA ANALYSIS FOR BODY FLUID IDENTIFICATION

The identification of body fluids can be important in determining the body fluid source of a DNA profile or in corroborating different versions of events.

mRNA is an intermediary compound between DNA in the cell nucleus and the cell proteins. The mRNA profile of a cell is unique for each cell type. By exploiting these differences ESR has developed an mRNA test that can be used to detect blood, menstrual blood, vaginal fluid, saliva and semen (with or without sperm).

mRNA testing can be used when:

- regular chemical tests have not been successful in identifying the body fluid
- there is no chemical test available (vaginal material, menstrual blood).

The mRNA test uses a by-product of the DNA extraction process. It is preferable for mRNA testing to be requested (or considered) in advance of the samples being processed. However, samples suitable for mRNA analysis have been retained since July 2010 and will be kept for at least two years from receipt should mRNA testing be retrospectively required. Older cases may not have sample remains suitable for mRNA testing and re-processing of original materials such as fabric stains may be needed.

As mRNA is a more fragile molecule than DNA, and more susceptible to environmental influences, it is possible that results may not be obtained from all samples for which DNA profiles are available. Conversely, the amount of mRNA per cell can be greater than the amount of DNA therefore the opposite is also possible.

DETECTS: the body fluid source of DNA – blood, menstrual blood, vaginal fluid, saliva and semen (with or without sperm).

DATABANK: Not applicable.

TURNAROUND TIMES: Because of complex analysis and interpretation turnaround times will be longer than standard testing and vary depending on the sample and case.

CASE EXAMPLES:

- A bloodstain is found on a suspect's car seat. The complainant alleges a sexual assault and that she was menstruating. The suspect alleges the complainant had a nose bleed in his car. Menstrual blood is detected.
- A female DNA profile is obtained from the mouth of a bottle. The suspect says she drank from the bottle, the complainant alleges it was inserted into her vagina. mRNA profiles consistent with saliva are found.

Spermatozoa

LASER MICRODISECTION (LMD) DNA TESTING USING SGM PLUS®

LMD is a microscopic technique for the isolation of a particular cell type from a mixture of cells. It provides improved analysis of challenging forensic samples.

It is particularly useful in the analysis of semen stains when sperm numbers are low as it is possible to isolate the sperm and provide a useful DNA profile from these cells alone. It can be used on samples where there is a small amount of sperm mixed in with a large amount of female cells such as a vaginal swab, or to recover other cell types such as vaginal cells, or buccal cells when mixed with sperm or skin cells.

After cell recovery standard, LCN, Minifiler™ and/or Y STR DNA profiling techniques can be applied. The choice of profiling technique will depend on the number of the cells recovered, quality of the DNA recovered from the cells and the results available from other samples for comparison.

LMD can be used on samples where there is a small number of sperm mixed with a large amount of female cells, such as a semen stained vaginal or oral swab. It can also be applied to samples that contain small numbers of epithelial cells (vaginal or buccal cells) when mixed with sperm, or skin cells on samples such as penile swabs or male underclothing.

CASE EXAMPLES FOR STANDARD LMD ANALYSIS:

- Sexual assault swabs such as vaginal, cervical and oral swabs and when sperm numbers are low.
- Semen stains detected on clothing/ bedding when sperm numbers are low.
- Penile swabs where small numbers of epithelial cells are likely to be present.

Male epithelial cells showing fluorescent labelling of the X chromosome (red) and the Y chromosome (green).

INTERPRETING DNA

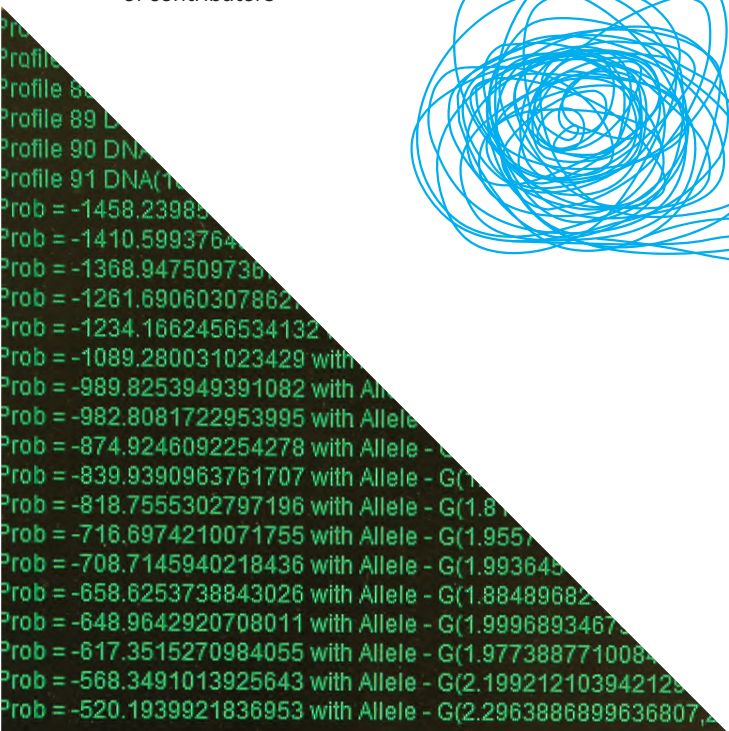
STRmix™ is expert forensic software, developed by ESR and Forensic Science South Australia (FSSA), that can resolve previously unresolvable mixed DNA profiles.

As well as improving interpretation of DNA profiles from a single source, it can also determine the contributors to complex DNA mixtures. STRmix™ has been in routine use at ESR since August 2012.

STRmix™ also includes a function that allows mixed DNA profiles to be compared directly against the National DNA Profile Database. This is a major advance for cases where there are no suspects and there is mixed DNA from individuals in one sample.

STRmix™ ENABLES ESR TO:

- interpret DNA results faster
- compare profiles against a person of interest and calculate a likelihood ratio
- resolve previously unresolvable, complex DNA mixtures with no restriction on the number of contributors
- use more of the information in a DNA profile
- search complex, mixed DNA profiles against a database.



FOR MORE INFORMATION
0800 FORENSIC
0800 367 367

INSTITUTE OF ENVIRONMENTAL SCIENCE AND RESEARCH LIMITED

Kenepuru Science Centre
34 Kenepuru Drive, Kenepuru, Porirua 5022
PO Box 50348, Porirua 5240
New Zealand
T: +64 4 914 0700 **F:** +64 4 914 0770

Mt Albert Science Centre
120 Mt Albert Road, Sandringham, Auckland 1025
Private Bag 92021, Auckland 1142
New Zealand
T: +64 9 815 3670 **F:** +64 9 849 6046

NCBID – Wallaceville
66 Ward Street, Wallaceville, Upper Hutt 5018
PO Box 40158, Upper Hutt 5140
New Zealand
T: +64 4 529 0600 **F:** +64 4 529 0601

Christchurch Science Centre
27 Creyke Road, Ilam, Christchurch 8041
PO Box 29181, Christchurch 8540
New Zealand
T: +64 3 351 6019 **F:** +64 3 351 0010

www.esr.cri.nz