

Cellmark Forensic Services

The use of DNA databases in forensic casework

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Cellmark Forensic Services

- Established in 1987 – 30 years of specialist forensic analysis
- One of the UK's largest Forensic Service Providers (FSP)
- Comprehensive range of forensic techniques
- Circa 500 staff in the UK
- Contracted by the majority of UK police forces
- Additional contracts with Military customers and with UK Government

Cellmark Forensic Services (*continued*)

- Currently Cellmark is responsible for:
 - Approximately 30-40% of crime scene DNA samples loaded to the UK National DNA Database (NDNAD)
 - Approximately 30-40% of subject reference DNA profiles loaded to the UK NDNAD

Types of DNA databases utilised at Cellmark

- Forensic Information Database Service (FINDS)
 - Custodians of UK's National DNA Database
- Nationally used allele frequency databases
- Missing Persons Database (MPDD) – managed by FINDS
- Vulnerable Persons Database (VPDD) – managed by FINDS
- Central Elimination Database (CED) – managed by FINDS
- Interpol
- Y-STRs (YHRD)
- Mitochondrial DNA (EMPOP)
 - 12S Animal Species Testing (BLAST)

Why use DNA databases?

- Crime scene DNA databases
 - Identify otherwise unknown offenders
 - Provide further evidence in relation to known offenders
 - Link crime scenes and serial offences
 - Prevention and detection of crime
 - In excess of 60 countries now have operational DNA databases*, each with varying uses/requirements
- Reference DNA databases
 - Including Y-STR database, YHRD and the mitochondrial DNA database – EMPOP
 - Provide a frequency estimate to produce a strength of evidence for the findings

FINDS - National DNA Database (NDNAD)

- Established in 1995 and operate by the Home Office. First of its type in the world
 - Police forces own the profiles but do not operate/have access to the database
 - Number of integrity checks undertaken routinely
 - Accredited by appropriate bodies
- Multiple chemistries accepted (NGM SElect, ESI-17, ESSplex SEQS, Globalfiler)



FINDS - National DNA Database (NDNAD) (*continued*)

- Holds two types of profiles
 - Crime stains
 - Subject samples from anyone suspected, cautioned or charged with a recordable offence
- As of 31/12/17, contained > 6 million profiles*
 - >5,600,000 individuals (including duplicates)
 - >555,000 crime scenes (>180,000 remain unmatched)



FINDS - National DNA Database (NDNAD) (*continued*)

- Matches can be one of three categories:
 - Scene to individual
 - Depending on the quality of the crime stain profile (full/partial/mixture), matches may require confirmation from a reporting scientist at the FSP.
 - Scene to scene
 - Individual to individual



FINDS - National DNA Database (NDNAD) (*continued*)

- Between April 2001 and March 2016 there were 611,557 matches
- Between July 17 and Dec 17 there were >13,000 matches
 - 390 homicides, 607 rapes and 440 urgents
 - **This equates to a match rate of 65.9%***
- As of 31st March 2016, NDNAD held > 180,000 unmatched crime stain profiles*



Concerns & limitations regarding DNA databases

- Ethical issues / retention legislation
 - Should samples from individuals be taken at birth and uploaded pre-emptively?
 - Potential for ‘innocent’ individual's DNA to be uploaded when no elimination samples provided
 - UK has had recent changes in legislation regarding the storage and retention of arrestee samples
- Security and data integrity
 - Could the data within the sample be used for genetic research
 - Data errors could have serious consequences

Concerns & limitations regarding DNA databases (*cont.*)

- Cost
 - Running of NDNAD cost the UK Home Office and police forces £3.7m in 2015/16*
- Adventitious matches to profiles generated using older DNA profiling systems and those crime scene profiles which are more partial in their profile information
- Of no use if awareness for potential DNA recovery from crime scenes is not present within policing community
 - Including anti-contamination routines and chain of custody
 - Only 11% of crimes in 2015/16 had a crime scene investigator sent by the police to look for evidence*
- Failure to take suspect DNA for upload

UK NDNAD – Routine crime scene uploads

- When a profile meets specific NDNAD load criteria, it can be submitted for permanent upload
- Either generates a match (scene to scene, scene to individual, individual to individual) or no match
 - Report issued to force in either instance
- ‘Positive’ report contains details of the matching profiles, sample types, DNA profiling chemistries and profile completeness (if crime stain) amongst other information
- Scene to scene matches can be useful in identifying serial offenders or those crimes which were not initially identified as potentially linked

Case example

- 2017 UK murder case
- Cigarette submitted at 10.30pm for urgent profiling and NDNAD load
 - Suspicious vehicle seen at the location
- Match reported to force by 8pm following evening
 - Force described this as a ‘major breakthrough’
- Allowed them to identify further suspects by tracing movements and known associates
 - Further exhibits/evidence were able to be rapidly identified and significantly progress the case

UK NDNAD - Speculative searches

- A one-off search of the NDNAD at that specific moment in time
 - Routinely used for samples not meeting NDNAD load criteria
 - Also used for urgents and covert/counter-terrorism cases
- Between July 2017 and December 2017, there were >9,000 speculative searches*
 - >1,500 identified a potential match
- Can be repeated monthly, quarterly, bi-annually or annually to capture new matches
- ‘DNA Boost’ – specialist speculative search function
 - Designed for more complex mixed DNA results

Case example

- UK Sexual offence case from 2015 - unknown offender
- Swabs from victim's face and neck and jeans and underwear submitted
- Clothing both gave similar results – major matching aggrieved with minors
 - Composite prominent minor contributor identified and submitted for speculative search
 - Designated as 'Unknown Male One' - no matches identified. Partner of aggrieved?

Case example (*continued*)

- Neck swabs gave a 3-person mixture
 - Were able to eliminate aggrieved and left two prominent contributors
 - Unknown Male One could be a possible contributor
 - Remaining prominent submitted for speculative search
 - Identified one individual and arrested

UK NDNAD - Familial searches

- Relies on the heredity nature of DNA
 - Half from the mother, half from the father
- DNA from a crime scene can be submitted for a familial search of the NDNAD
 - Searches for possible close relatives
- Police then use other intelligence information to investigate/eliminate those identified from the search

UK NDNAD - Familial searches (*continued*)

- Due to being resource heavy and costly, usually only carried out for most serious crimes
 - 17 carried out in 2015/16 on UK NDNAD*
- Some have raised ethical concerns over the intrusion of this process
 - Any familial searches require authorisation from FIND Strategy Board

Familial search case example

- Murder of Lynette White in 1988
 - 3 men originally convicted despite no forensic evidence
 - Convictions overturned in 1992
- Cellmark began re-investigation of case in 1999
 - Incomplete male DNA profile obtained from cellophane on a cigarette packet close to victim's head – 'Cellophane Man'
 - Scene photo's reviewed and identified bloodstaining on a skirting board
 - Scene revisited 13 years later and blood still present under paintwork
 - Full profile matching 'Cellophane Man'

Familial search case example (*continued*)

- No match on NDNAD/elimination samples/mass screen so familial search carried out in 2003
 - Initial dataset contained 600 names
 - Reduced to 70 individuals after reviewing degree of allele sharing with ‘Cellophane Man’
 - The individual identified from the Familial search was a 14-year old boy *who wasn’t even born at the time of the murder*
 - Profiled his father – close but no match....
 - Profiled his father’s 1st brother - close but still no match....
 - Profiled his father’s 2nd brother – **MATCH!**
- **First ever familial search of the NDNAD which led to the arrest of a suspect**

Allele frequency databases – evaluating database identifications

- Once a potential ‘match’ between an individual and a crime scene profile has been identified from a NDNAD search (or direct comparison), the evidential significance of that match is often requested by force
 - What is the likelihood that the crime scene sample originated from someone other than and unrelated to Mr X?
 - Could this be an adventitious match?

Allele frequency databases – evaluating database identifications

- Using databases of known alleles, we can calculate the frequency of a particular allele within the known population (and therefore the profile frequency)
 - Allele frequency databases are now standard for all UK FSPs for DNA-17 loci
 - Compiled by NDNAD Unit (with assistance from King's College London)
 - Allele frequencies recorded for main ethnic groups within UK (6 groups)
 - Agreed national F_{st} value of 3%

UK - Missing Persons Database (MPDD)

- Separate from NDNAD but operated by FINDS
- Profile records from belongings of people who have gone missing loaded
 - Or their close relatives
- When unidentified remains located and a profile obtained, it is also loaded and any potential matches relayed to the FSP
 - Assists with police investigations
 - Can provide closure for families

UK - Missing Persons Database (MPDD) (*continued*)

- As of 31/12/17*
 - 261 unidentified bodies and 42 crime stains
 - 1352 references and 295 kinship references
 - Any foreign unidentified remains, believed to be British nationals must also involve MPDD for profile transfer (usually also includes Interpol)

UK - Vulnerable Persons Database (VPDD)

- Separate from NDNAD but operated by FINDS
- Holds profiles of people who are at risk of harm (or they consider themselves at risk)
 - Domestic abuse, child sexual exploitation, trafficking etcetera
- If there is potential or significant risk of harm to an individual held on the VPDD, their profile can be loaded to the NDNAD to identify any matches
 - Assists with police investigations
 - If the VP goes missing, it is also advisable to copy the profile to the MPDD
- As of March 2016, 3261 records were on the VPDD and in 2015/16, no requests were made to compare to the NDNAD*

UK - Central Elimination Database

- Recommendation of UK Forensic Regulator and held by FINDS as a separate entity
- Key to identifying any contamination events on NDNAD from force staff
- Samples taken from new members of police staff and officers and uploaded
 - Regular, national, searches of crime stain profile records against the elimination records
 - Currently retrospective search but plan to initiate searches when sample uploaded

UK - Central Elimination Database (*continued*)

- Plan to extend the database to include records of staff who have access to the DNA supply chain and may potentially contaminate a crime scene/sample
- Cellmark Forensic Services also has an internal staff elimination database to monitor and source possible contamination

International - Interpol

- Treaty of Prüm, 2005. Enabled signatories to exchange data regarding DNA (amongst others) of persons of interest and to co-operate against terrorism and trafficking more easily
 - Adopted by a number of EU member states
- Data sharing only - no link to an individual's information on system
- Police in member countries can submit profiles (offender/crime scene/missing persons/unidentified bodies) to automated DNA database
 - Results within 15 minutes

International - Interpol

- As of 2017, contained more than 173,000 profiles from more than 84 countries*
- Access can be extended to non-member countries upon request

International - Y-STR databases

- Y-STRs are those contained on the male Y chromosome
 - Various commercial Y-STR kits available
- Y-STR profile is inherited down the paternal line
 - All close male paternal-line relatives will contain the same Y-STR profile (barring any mutation event)
- Advantages over routine STR profiling
 - Sexual assault where expecting high levels of female DNA or multiple male offenders
 - Can provide intelligence on number of individuals involved
- Also used in identification and paternity cases

International - Y-STR databases (*continued*)

- Y-STR profiles can be directly compared to one another to determine whether or not they could have originated from the same paternal line
 - The significance of that potential match can then be evaluated using YHRD: Y-Chromosome STR Haplotype Reference Database
- YHRD
 - It is not an equivalent to the NDNAD
 - Need reference sample for comparison
 - Made up of profiles submitted from worldwide forensic and academic institutions
 - No crime stains

International - Mitochondrial DNA (mtDNA)

- Mitochondria are small organelles present in many types of cells in several thousand copies
 - Their small genome has a greater degree of variability between individuals
 - This and their abundance in cells makes them ideal for DNA profiling attempts on small or degraded samples
- Genome is inherited down the maternal line
 - All close maternal-line relatives will contain the same mtDNA sequence (barring any mutation event)

International - Mitochondrial DNA (mtDNA) (*continued*)

- mtDNA sequences can be directly compared to one another to determine whether or not they could have originated from the same maternal line
 - The significance of that potential match can then be evaluated using EMPOP
- EDNAP Mitochondrial DNA population Database (EMPOP) is a non-commercial, scientific project of the Innsbruck Medical University (and scientific partners)
 - It is not an equivalent to the NDNAD
 - Need a reference sample for comparison
 - Made up of profiles submitted from worldwide forensic and academic institutions

International - Mitochondrial DNA (mtDNA) (*continued*)

- Used by Cellmark in forensic casework to generate a frequency estimate for the mtDNA haplogroup (profile) generate from samples.
 - A query search will give a breakdown of the number of matches (if any) to the entire database, then by origin and finally by metapopulation
 - A frequency calculation is also displayed, if there are any matches, within a given population group

Animal Species Identification using 12S Mitochondrial DNA

- Cellmark uses a region of the mtDNA that codes for the 12S ribosomal gene
 - Highly conserved region
- Uses can include:
 - Animal crime such as unlawful slaughter, poaching, baiting, interference
 - Wildlife forensics
 - Contaminated food products
 - Identification/confirmation of species
- Sequence obtained can be compared against an internal database – BLAST: Basic Local Alignment Search

'Creature of Metapec'

- The 'Creature of Metapec' was a small, vaguely humanoid creature which was allegedly discovered by a Mexican farmer in a trap in Metapec, Mexico.
- UFO journalist Jaime Maussan bought the dead body of the Creature from the farmer for US\$23,000, because he thought it was an alien. He paid for numerous DNA test to be carried out, but the results had been inconclusive.
- www.telegraph.co.uk/culture/tvandradio/9714984/Alien-Investigations-Channel-4-review.html



‘Creature of Metapec’ (*continued*)

- A number of years after its initial discovery a sample was submitted to Cellmark for 12S testing and a useable sequence was obtained.
- This 12S sequence was searched against BLAST
- The sequence came back as marmoset
- In 2012, this case was featured as part of a Channel 4 documentary investigating aliens



Cellmark database hosting capabilities

- Cellmark has developed a capability to maintain and operate bespoke DNA Databases for smaller international police forces without their own DNA Database infrastructure
- Crime scene and arrestee samples are received, processed, uploaded and compared according to protocols agreed with the submitting force and in compliance with the local legislation
- Potential matches issued to the force in a report format:
 - Formal match confirmations can be requested, if required
- Force SEDs are maintained and automatically compared prior to matches being issued

The use of DNA databases in forensic casework

Thank you for your attention
Any Questions?

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