The Future of DNA Databases

Peter M. Schneider
Institute of Legal Medicine
University of Cologne
Germany
DNA Database Topics - Overview

- Legislative issues
- The numbers game
- Expansion strategies
- Cleaning up
- Familial searching
- Forensic DNA phenotyping
- The future …
No database without standards

- 1985 – A. Jeffreys invents DNA fingerprinting
- General database requires standardized genetic typing data
- Not possible with MLP/SLP results
- 1986-1994: Scientific and technical progress
  - Polymerase chain reaction
  - Short tandem repeat systems
  - Fluorescent detection enables multiplex strategy
DNA databases – a success story

- 1995: First National DNA Database in England
- 2002: Interpol DNA Inquiry
  - 77 countries use forensic DNA typing methods
  - 41 countries have established DNA database
- 2005: Seven EU countries agree on trans-national DNA data exchange (treaty of Prüm)
Database legislation issues

- Collection of reference samples from persons
- Inclusion of suspects / convicted offenders
- Severity of offence / type of crime
- Retention of DNA samples
- Retention / removal of DNA profiles
Collection of reference samples

- Intimate vs. non-intimate samples
  - Buccal swabs considered "non-intimate" in most legislations

- Informed consent and/or court decision
  - Court decision required in many legislations, then samples can be taken without consent
  - Informed consent mandatory in some legislations
  - Collection of "abandoned" body material
Which persons in a database?

- **Suspects**
  - For any recordable offence
  - Only if formally charged with crime
  - Only when ordered by a judge
  - Never

- **Convicted offenders**
  - Only depending on seriousness of offence
  - With additional decision by judge

- **Retroactive inclusion of convicted offenders**
  - Risk assessment for future offences
Which crimes in a database?

- Capital crimes
- All sexual offences
- Serious crime, depending on the expected conviction (e.g. more than one year of prison)
- Minor offences, if repeatedly committed
- Any recordable offence
Retention of reference samples

- Offender samples
  - Retained
    - to confirm a match, before a report is sent out
    - to type more loci, if necessary
  - Destroyed
    - when corresponding profile is removed from database
    - after typing is completed to ensure protection of privacy
    - in case of a match report, a fresh sample has to be collected for verification
Retention of offender profiles

- **Suspects**
  - Unrestricted retention
  - Removed
    - after charges have been dropped
    - after acquittal
    - either immediately, or after a defined number of years

- **Convicted offenders**
  - Unrestricted retention, even after death
  - After a defined period (e.g. 10-40 years)
    - depending on seriousness of offence, or the age of the offender
    - if no further offence has been committed
    - following a case review
Changes in legislation

- Germany
  - Court decision not required any more
    - for crime scene samples
    - for samples from suspects, if provided with informed consent
  - Threshold for database entry has been lowered
    - also for minor offences, if committed repeatedly

- Netherlands, Sweden
  - Threshold for offender inclusion lowered, inclusion of suspects introduced

- Norway
  - Amendment to include suspects
"The prospect of ‘rolling back’ the NDNAD and the removal of large numbers of profiles from it seems highly unlikely. This is in part explained by the way in which the database has become represented as an essential, seemingly indispensible, technology in the contemporary armoury of crime management"

(Williams & Johnson 2008)
"S. and Marper v. the United Kingdom"

1. The Case

- S. (a juvenile offender who was not convicted) and Marper (charged with harassment, but not further prosecuted after reconciliation) asked for their samples to be destroyed.
- The police refused, the application was brought to court, and finally rejected by the Court of Appeal.
- However, the European Court on Human Rights ruled that the retention of samples is a violation of Art. 8 and 14 of the Convention of Human Rights.
"The question … remains whether such retention is proportionate and strikes a fair balance between the competing public and private interests.

In this respect, the Court is struck by the blanket and indiscriminate nature of the power of retention in England and Wales. The material may be retained irrespective of the nature or gravity of the offence with which the individual was originally suspected or of the age of the suspected offender …

The retention is not time-limited; the material is retained indefinitely whatever the nature or seriousness of the offence of which the person was suspected."
Alec Jeffreys (2008)

- "The real concern I have in the UK is what I see as a sort of 'mission creep'.
- "Now hundreds of thousands of entirely innocent people are populating that database - people who have come to the police's attention ... by being arrested or charged with a crime and subsequently released.
- "This was not the initial purpose of the database which was originally meant to hold the DNA of convicted criminals. I have real concerns about the retention of innocent individuals on the National DNA Database."
"S. and Marper v. the United Kingdom"

3. The Consequences

- Changes proposed by British government
  - Profiles of all children under 10 removed.
  - Profiles of all convicted adults and under 18s indefinitely retained.
  - Profiles of adults arrested but not convicted will be retained for 6-12 years.
  - Profiles of under 18s convicted once of less serious offences to be removed at 18.
  - Profiles of under 18s arrested but not convicted of serious offences will have profiles retained for 12 years.
  - Profiles of under 18s arrested but not convicted of less serious offences to be deleted after six years or on 18th birthday.
  - All CJ samples to be destroyed within six months of processing.
DNA Database Topics

- Legislative issues
- The numbers game
European DNA Databases 2003-2008

- Persons: +180%
- Stains: +170%
The Treaty of Prüm

- ... allows the law enforcement agencies from the participating countries to consult each other's DNA, fingerprint and motor vehicle registration databases.
- ... provides instruments for the fight against terrorism, of police cooperation and for the fight against illegal migration.
- DNA profiles from one country can be searched in a fully automated way against the DNA database of other countries on a hit/no hit-basis, i.e. only anonymized DNA profiles are compared.
- After a match, the associated information has to be exchanged using existing mutual legal assistance procedures.
The Treaty of Prüm

- The Treaty of Prüm was signed in May 2005 by the Netherlands, Austria, Germany, Belgium, Luxembourg, Spain and France.
- Finland, Italy, Portugal, Slovenia, Greece, Sweden, Bulgaria, Romania, Slovakia and Hungary have joined the treaty.
- The EU council has accepted to convert the information exchange part of the treaty into EU legislation.
- As a consequence all 27 EU countries will be able to exchange DNA profiles in the near future.
The European Standard Set (ESS)

- The ESS comprises seven STR loci
  - additional STR loci are typed, but not commonly used in all EU countries

<table>
<thead>
<tr>
<th>Locus</th>
<th>SGM plus</th>
<th>ESS</th>
<th>DAD</th>
</tr>
</thead>
<tbody>
<tr>
<td>VWA</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>THO</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D21</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>FGA</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D8</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D3</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D18</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D16</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>D21</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>D19</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>SE33</td>
<td></td>
<td></td>
<td>X</td>
</tr>
</tbody>
</table>

- Prüm matching rules require at least six fully typed loci
Adventitious matches

- Adventitious matches depend on
  - the match probability of the DNA profile
  - the size of the database
  - the number of comparisons

- The European data exchange may result in adventitious matches, if
  - large data sets are compared against each other
  - the number of common loci is not extended
Scenario for adventitious matches

- Data set from country A \( (n_A) \)
  - 1,000,000 unresolved crime scene samples
- Data set from country B \( (n_B) \)
  - 1,000,000 convicted offenders
- Match probability (MP) for ESS loci: 1 in \( 10^9 \)
- Expected number of adventitious matches
  \[
  = \text{number of searches} \times \text{database size} \times \text{MP}
  \]
  \[
  = 10^6 \times 10^6 \times 10^{-9} = 1,000
  \]
DNA Database Topics

- Legislative issues
- The numbers game
- Expansion strategies
The ESS Expansion Project

- In 2005, a decision was adopted by the ENFSI and EDNAP groups to increase the number of ESS loci.
- A recommendation was published to include more robust loci with short amplicons, rather than already established STRs which frequently fail to give results, and/or have a poor power of discrimination.
- Gill et al. (2006): “…it is unrealistic to suggest that laboratories can change by abandoning loci in favour of new ones. Rather, it is proposed that new core loci are decided and then laboratories expand their systems while retaining their existing set of STRs.”
### The ESS Expansion Project

<table>
<thead>
<tr>
<th>Locus</th>
<th>SGM plus</th>
<th>ESS</th>
<th>Germany</th>
</tr>
</thead>
<tbody>
<tr>
<td>VWA</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>THO</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D21</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>FGA</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D8</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D3</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D18</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>D16</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>D21</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>D19</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>SE33</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
</tbody>
</table>

- It was recommended to increase the common data range, until additional loci are validated and kits available.
Six new loci were recommended in 2005, and five were finally included into a possible expansion set. Kits are now (in 2009!) made available by manufacturers.
What about SNPs?

- Current STR technology is locked into existing databases
- Presently, SNPs do not offer any advantage that would justify a replacement of STRs for databasing
- SNPs can be used as adjunct for special cases, e.g. for highly degraded samples in victim identification
- Technically, SNP data can be easily stored in databases, if necessary
DNA Database Topics

- Legislative issues
- The numbers game
- Expansion strategies
- Cleaning up the databases
The hunt for the "phantom" – a critical reappraisal of DNA database reality

- In 2007, a female police officer was shot in her car, her male colleague was severely wounded
- A female DNA profile was identified in the car
- The profile generated multiple hits in the DNA databases of Germany and Austria
- The profile resurfaced 39 times in subsequent analyses of other cases (from burglaries to homicides)
A frantic search for a female perpetrator, dubbed the "phantom", began …
"Men may also carry female DNA" a police official declared.
What happened then?

- The profile reappeared this year in two more cases where the DNA source was a known male person
- Contamination was now seriously considered
- Cotton swabs were identified as potential source
- The supplier provided reference samples from the factory workers – and a retired 71-year old woman was found to match the phantom's DNA profile
It is not the science itself which is flawed but the interpretation and context in which these results may be used.
Cleaning up the DNA databases

- Add only relevant crime scene samples
- Check profiles for contamination from
  - Manufacturers
  - Crime scene officers
  - Laboratory personnel
- If possible, create elimination databases with appropriate provisions for data protection
- Check stain-to-stain hits from unrelated crimes
- Avoid partial profiles
  - to prevent adventitious matches
DNA Database Topics

- Legislative issues
- The numbers game
- Expansion strategies
- Cleaning up
- Familial searching
- Forensic DNA phenotyping
Familial searching strategies *

- First degree relatives share 50% of their genes
- STR data may be used to identify persons related to the perpetrator who has left a crime stain
- Comparable to DVI scenario
- Search strategies must include a likelihood ratio based on the rarity of shared alleles to avoid adventitious matches
- Y-haplotype and mtDNA data may help to increase strength of evidence

Familial searching in databases

- In the court, testimony can be refused if a relative is concerned
- Individuals on database may be used as "unwilling informants" about their relatives
- The use of national DNA databases for familial searching has to be legally authorized
- Familial searching should be restricted to serious crime, e.g. murder and sexual assault cases
- More loci would increase strength of evidence
Forensic DNA Phenotyping (FDP)

- Prediction of outer physical traits from crime scene stains, e.g.
  - hair, eye, skin color
  - body height and stature
- May help to provide investigative leads to identify potential suspects
- Not useful to establish a match between suspect and stain
- *FDP data do not belong in any database!*
The Future ...

- More countries will introduce DNA databases
- Databases will be continuously expanded, but the inclusion and retention criteria have to be justified and legally secured
- Privacy rights will play an more visible role, and must be respected
- A population-wide database will not become acceptable
The Future ...

- STRs will continue to be used as main source of evidence
- More loci need to be added continuously to prevent adventitious matches
- Inefficient STR loci may be phased out after new loci have been accepted
- International collaboration and data exchange will continue to grow
Acknowledgement

- I would like to thank my colleagues and friends from the following groups and institutions for sharing their data: