

# From Forensic Genetics to Genomics – Perspectives for an Integrated Approach to the Use of Genetic Evidence in Criminal Investigations

**Peter M. Schneider**  
Institute of Legal Medicine  
University of Cologne (Germany)



..... Northumbria University Centre for  
**FORENSICS**SCIENCE

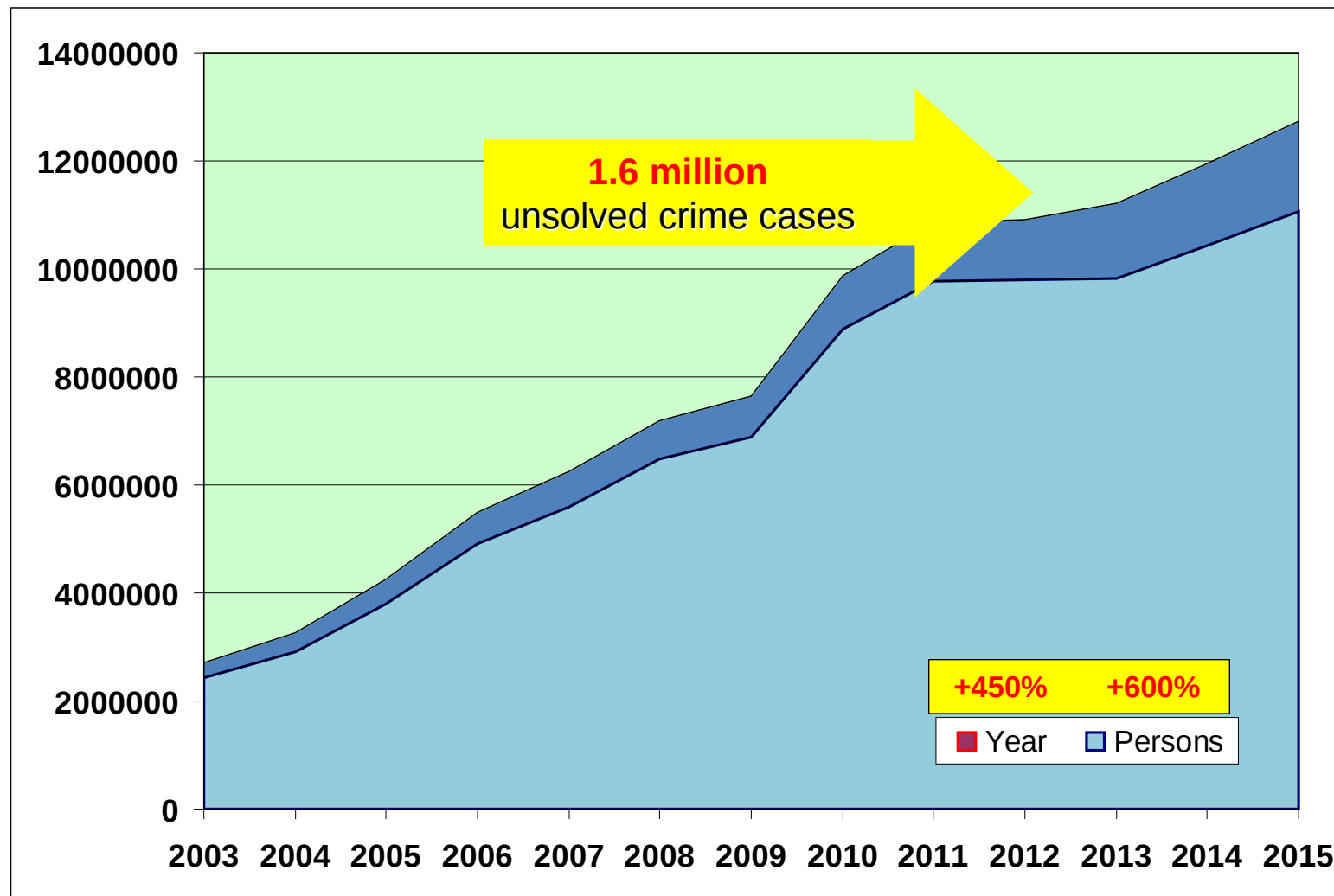


# Biological evidence – as source of abundant information at a crime scene

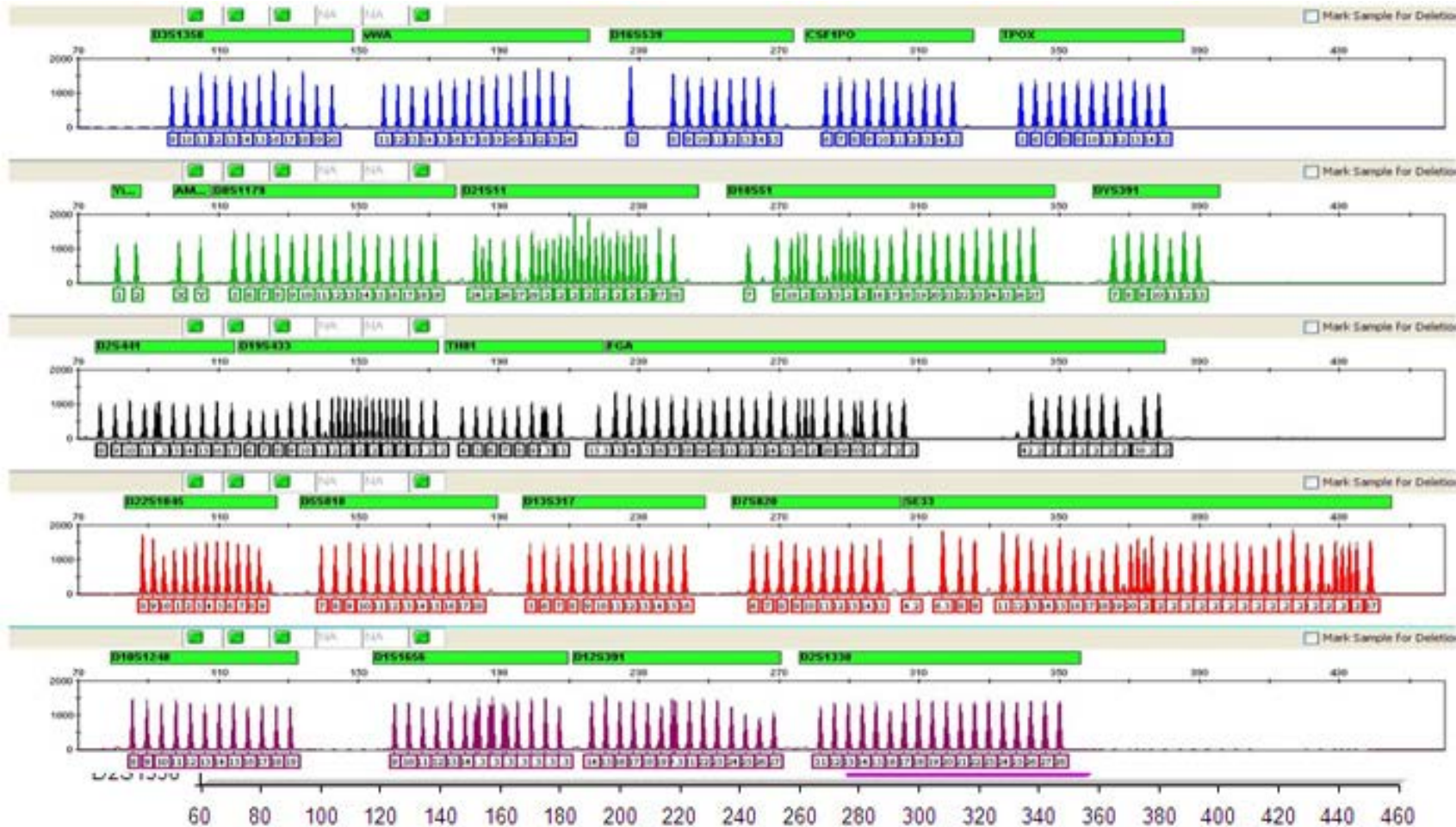


- **Body fluid stains**
  - Blood, semen, saliva, etc ...
- **Contact traces**
  - "Touch DNA" from handled items, tools, weapons etc ...
- **Skin particles or "flakes"**
  - Clothing items
  - Body surface
  - can be recovered from tape lifts that have been taken for fiber analysis
- **Target: single source DNA profiles for database searches**

# The Growth of DNA Databases in Europe 2003 – 2015



# 20 Years STR Typing by Electrophoresis



2016: 6 dyes / 24 markers

- **Standardization**

- Commonly used nomenclature of alleles and genotypes
- Allele frequency estimates based on world-wide population studies

- **Reliability**

- Very good reproducibility of results within and between laboratories
- Sensitive detection of „low level“ DNA amounts

- **Acceptance**

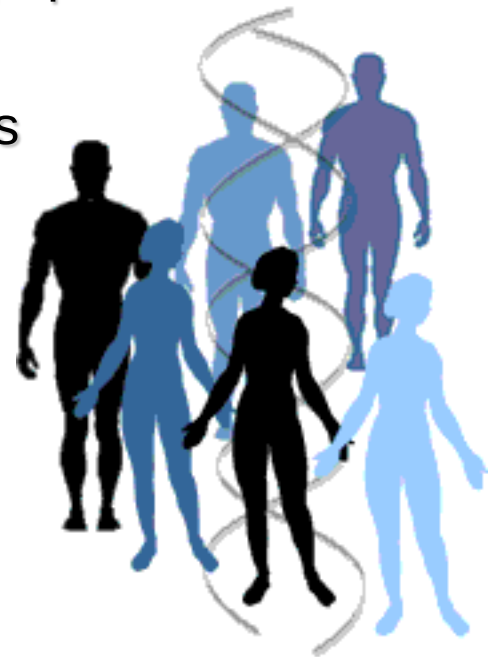
- Growing number of national DNA databases
- Non-coding STR markers: „built-in“ privacy protection
- Excellent cost/benefit ratio



- **Challenging DNA samples**
  - Loss of information due to degradation
  - Complex mixtures cannot be resolved
- **Kinship testing**
  - Not sufficiently powerful for distant family relationships (e.g. for identification of human remains)
  - Number of alleles/locus too small → allele frequencies too high
- **Technical restrictions**
  - Multiplex typing using CE restricts number of markers per assay
  - PCR/CE artifacts compromise detection sensitivity



- **Identification** of the type of human body fluid or tissue samples found at a crime scene or victim,
  - including complex mixtures such as those that can be found in sexual assault cases.
- **Information** on the biogeographic ancestry, i.e. the population genetic origin of an unknown person.
- **Prediction** of certain externally visible characteristics such as pigmentation and facial shapes.
- **Estimation** of the biological age of an unknown person at the time of the placement of a body fluid sample.



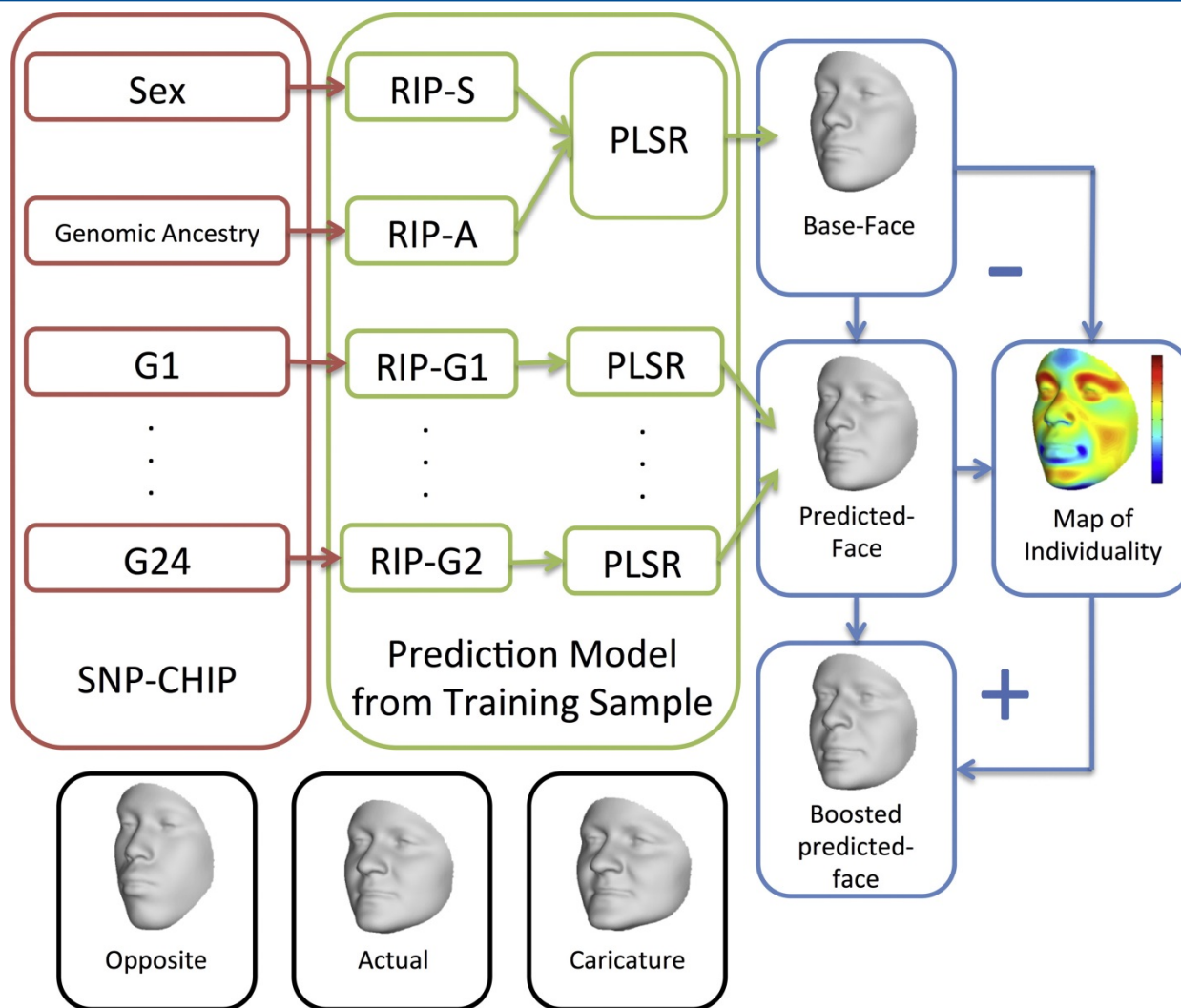
# "Next" Generation Sequencing

- The „next“ generation is already the „current“ generation  
= Massively Parallel Sequencing (MPS)
- Chip-based library sequencing of more than 2 billion base pairs and 5.5 million reads in a single run
- Up to 96 bar-coded samples can be analyzed on a single chip in one experiment
- Required overall time:
  - Library / template preparation: 1-2 days
  - Loading & sequencing: 0.5-2 days
  - Data analysis: >1 day



- **gDNA SNP profiling**
  - Identity SNPs (autosomal, non-coding)
  - Lineage SNPs (Y-Chr. / mtDNA, non-coding)
  - Ancestry informative SNPs (both coding and non-coding)
  - Phenotyping SNPs (FDP, mostly coding)
  - X-Chr. SNPs for kinship cases (non-coding)
- **DNA methylation profiling**
  - CpG islands associated with cell-specific gene expression
  - Age prediction & body fluid / tissue identification
- **mRNA profiling**
  - body fluid / tissue identification from cDNA
  - Combined with coding SNP typing → donor identification

# Forensic DNA Phenotyping: Face morphology (Claes et al., 2014)



# Individuals' faces compared with computer-generated DNA predictions



Credit: The New York Times; Images and renderings by **Mark D. Shriver/Penn State University**

# DNA SNAPSHOT



**This is not a phenotype,  
but a stereotype!**

Sex: Male ♂

Skin: Dark / Dark Olive

Eyes: Brown / Black

Hair: Brown / Black

Freckles: None

Ancestry: 92% West African  
8% NW European



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Not: Very Fair, Fair, or Light Olive 90.7

Not: Blue or Green 94.6

Not: Red or Blond 59.3

25.0 Not: Few, Some, or Many



<http://Parabon-NanoLabs.com/Snapshot>

- First genetic photofit image released in a South Carolina murder case
- Information on genetic markers (skin, hair, eye color, etc.)
- A few facial trait SNPs
- Analysis carried out by commercial company Parabon NanoLabs
- **Highly speculative!**
- **No valuable information except ancestry ...**

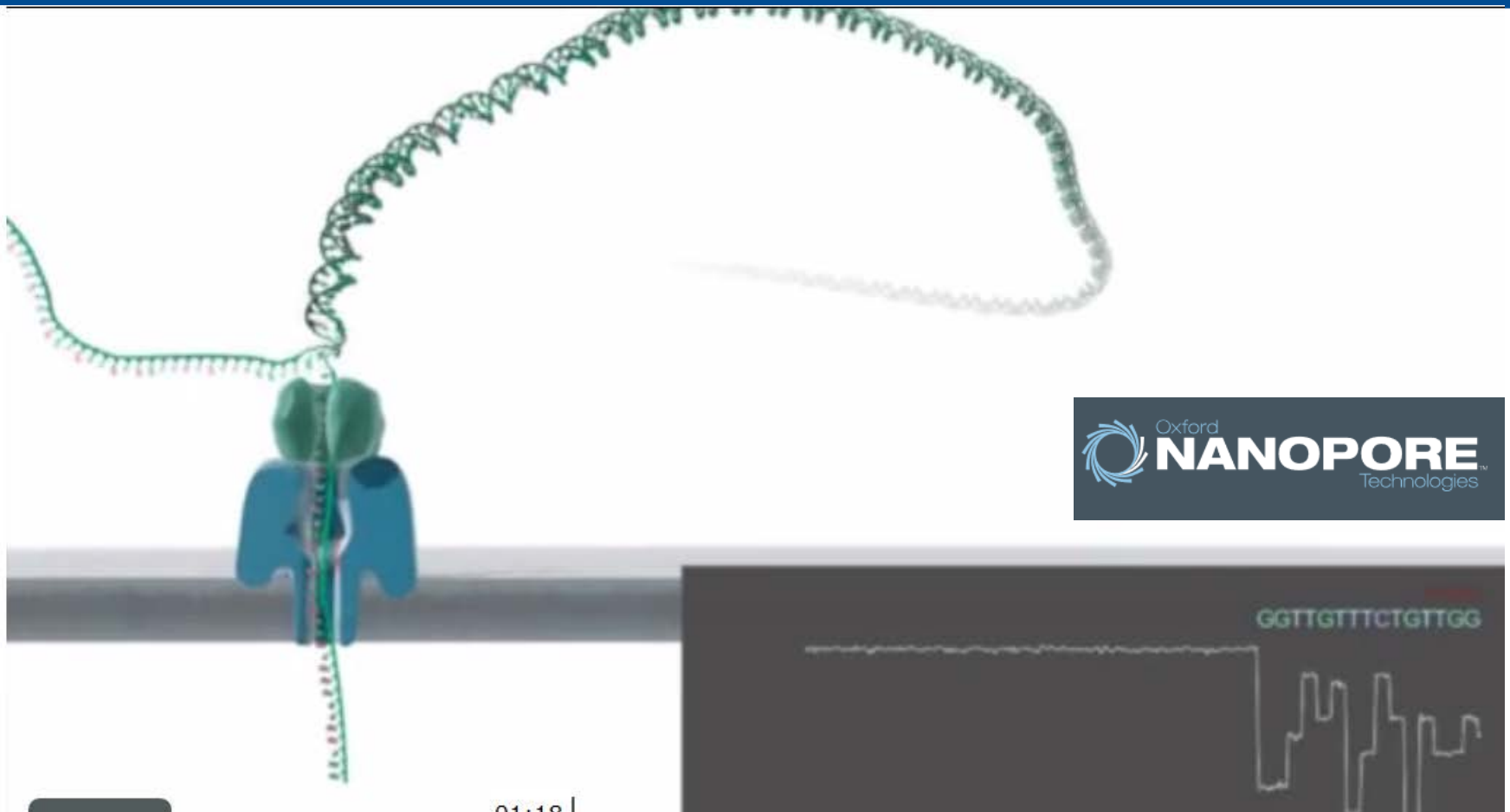
# What else is happening?

- The arrival of direct DNA sequencing
- No PCR, no library needed, just a laptop and ...



- Long read single molecule sequencing  
Oxford Nanopore MinION

# Single molecule long read sequencing



# Oxford Nanopore MinION



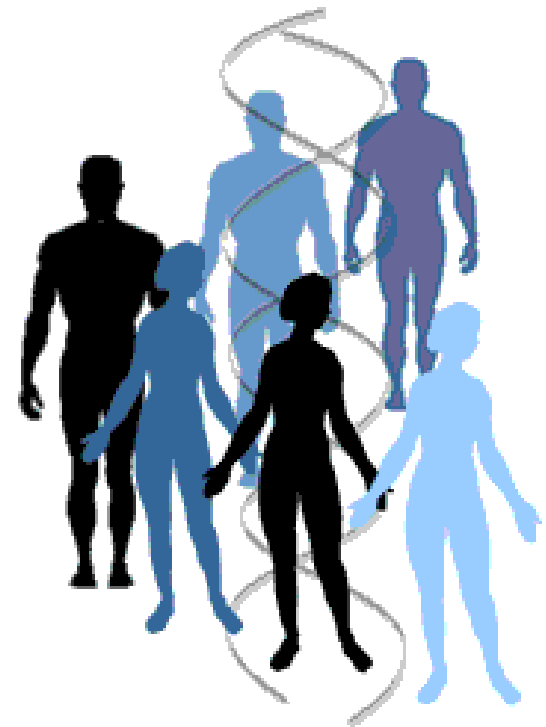
→ Has scene-of-crime testing arrived?

## STRENGTHS

- „All-inclusive“ approach for all forensic markers
- Maximal amount of data from minimal amount of sample
- Deconvolution of complex mixtures

## WEAKNESSES

- Long sample-to-data period
- Expensive (for the time being)
- Lack of standardization
- Requires new resources for bioinformatics and data storage

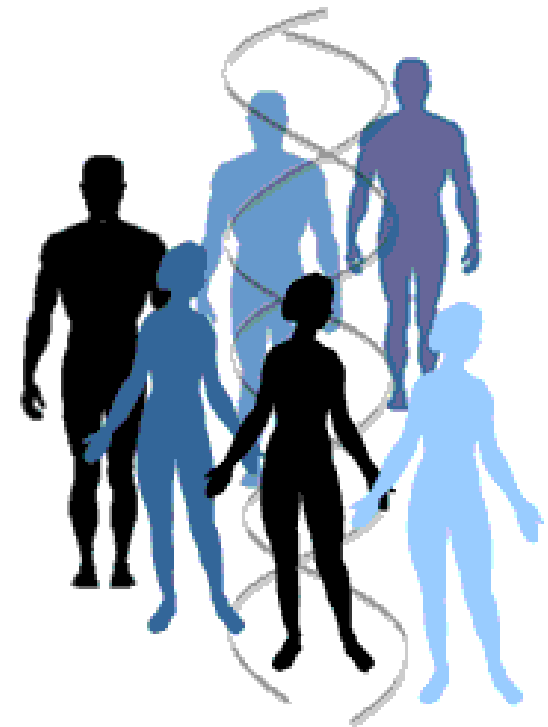


## OPPORTUNITIES

- May provide new intelligence leads on previously unsolved cases
- Allows to generate all data in advance, and irrespective of available information at the time of analysis

## THREATS

- All-inclusive typing kits may collect more information than needed
- Entering the „slippery slope“ of accumulating sensible personal data
  - *Why not sequence the entire genome?*
- How can the massive amounts of data be secured and filtered?



- **MPS is only a new technology, but**
  - ... the limitations have neither been explored nor defined
  - ... has created a demand to collect more data than needed
  - ... there is a growing interest, both from industry, to sell a new product, and from the forensic community to use and explore what is available

**The unlimited application in casework and for national DNA databases may eventually lead to a paradigm shift!**

**What can be done?**

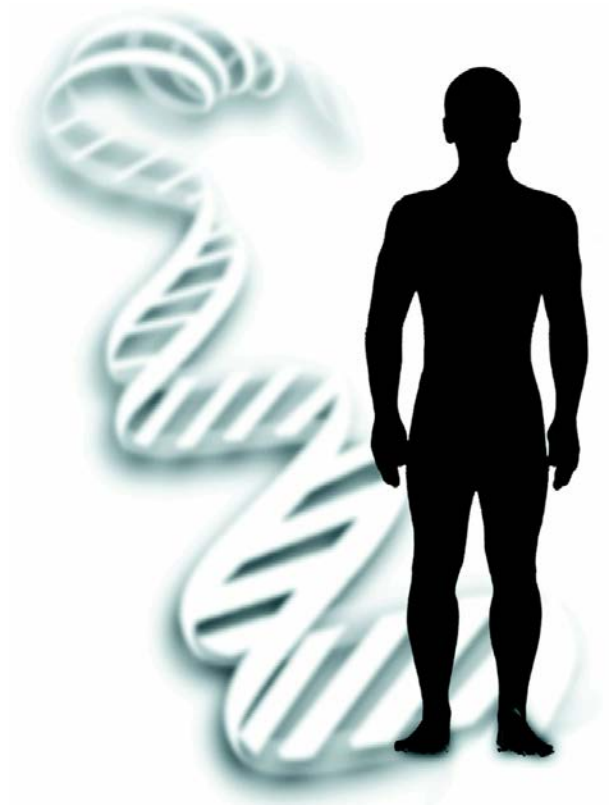
## What can be done:

- Create separate modules (primer sets) for coding / non-coding / ancestry informative markers
  - to be used alone or in combination
  - depending on depth of investigation, or the legal framework
- Keep sensible genetic data separated from national databases



## What can be done:

- Use bioinformatic filters to mask sensible personal data
- Develop a ,privacy by design‘ framework of probabilities to communicate results of FDP and ancestry typing without disclosing any DNA data to the end-user
- Ensure that DNA-based investigations are performed by informed and trained scientists





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Beigetreten ▾ Teilen Benachrichtigungen ...

Diskussion Mitglieder Veranstaltungen Fotos Dateien

Gruppe durchsuchen 🔍

Beitrag Foto/Video Umfrage Mehr

Schreib etwas ...

NEUESTE AKTIVITÄTEN

**Peter Schneider**  
 15. Juni um 00:14

Very timely: "This means that everyone in the UK is a migrant, some who moved here themselves and others who's ancestors settled years ago." Who would have guessed -- except for those who don't have a clue!



**DNA shows there is no such thing as a separate United Kingdom**

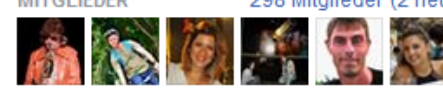
Leading University Experts have analysed the DNA from over 2,000 individuals whose grandparents all lived in the same part of the UK. The aim was to provide a fine scale genetic picture of the People of the British Isles whilst understanding how each

DNA-WORLDWIDE.COM

MITGLIEDER HINZUFÜGEN

+ Gib einen Namen oder eine E-Mail-Adresse ein ..

MITGLIEDER 298 Mitglieder (2 neu)



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Genetischer Fingerabdruck · Wissenschaft · Genetik

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