23 YEARS OF FORENSIC DNA : HOW ONE LOUSY BLOT CHANGED MY LIFE

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Once upon a time there was no DNA in forensics and crime investigators had to make do with other letters (ABO, Se, PGM). Then in 1984 DNA fingerprinting arrived entirely by accident. This talk will be a personal retrospective on how DNA fingerprinting and DNA profiling of minisatellites
were developed and how they underwent the transition from an academic curiosity to being speedily implemented in casework ranging from immigration disputes to paternity cases and the first murder investigation solved by DNA. I will also discuss non-human applications.

There were significant limitations to these old Southern blot approaches to DNA typing. The solution of course came with PCR, first reported in 1985 and made user friendly in 1988 with the introduction of thermostable DNA polymerases. It was immediately obvious that rapid, sensitive PCR-based typing systems would quickly replace the much more cumbersome Southern blot
approaches, though in retrospect it is surprising how slow this transition took place, with DNA profiling remaining the system of choice well into the 1990s. The main question was which PCR platform would win. The choice was extensive: SNP systems such as HLA-DQA and PolyMarker, mtDNA, AMPFLPs, MVR-PCR and microsatellites/STRs (only discovered in 1989). Very early casework made it clear that STRs would win the race, and the rest, as they say, is history.

Multiplex STR typing systems also offered the possibility of the creation of major national criminal intelligence DNA databases, with the UK database established in April 1995 being the first of its kind. I will discuss the current usage and resounding success of such databases, plus the potentially worrisome directions in which some databases are now evolving. More recent DNA marker systems allowing geographic/ethnic classification of unknown individuals or the recovery of phenotypic information will be discussed, along with concerns about genetic privacy. Future directions such as database expansion, high speed DNA typing and technology miniaturisation will be discussed along with their implications.

Finally, the highly variable repeat DNA loci used in DNA typing have also provided a superb platform for developing new approaches to studying de novo germline mutation in humans. This work has revealed the involvement of meiotic recombination in minisatellite instability and the existence of recombination hotspots as remarkably dynamic features of the human recombination landscape. It has also provided new methods for studying the effects of environmental agents such as ionising radiation on heritable mutation.