4. DNA Sampling – Science & Technology

Investigators are using DNA profiling to determine the suspect of the recent deadly car explosion outside the Red Fort in New Delhi, highlighting how genetic analysis plays a crucial role in forensic investigations

DNA Profiling

DNA profiling refers to a set of molecular techniques that identify individuals using unique patterns in their genetic material. It is considered the most accurate and dependable method of establishing identity, especially in - Explosions, fires, or mass disasters where bodies are charred or dismembered. Cases of missing persons, sexual assault, and homicide. Every human carries a highly unique DNA signature, except identical twins, making DNA profiling a cornerstone of modern forensics.

Understanding DNA (Deoxyribonucleic Acid)

Structure and Properties - DNA is a double-stranded helix made of nucleotides arranged in base pairs -

1. Adenine (A) pairs with Thymine (T).
2. Cytosine (C) pairs with Guanine (G).

These four bases form sequences that encode genetic instructions for -

1. Development
2. Growth
3. Cellular functioning
4. Reproduction

The order of bases (the genetic code) determines traits and biological identity.

Principles of DNA Profiling

Short Tandem Repeats (STRs) – Core of Forensic Profiling - STRs are short repetitive sequences of DNA. Each individual has distinct repeat numbers at multiple STR loci.

Inheritance Pattern - Half the STR markers come from the father and half from the mother. STR matching between mother and unidentified remains can offer extremely high confidence in identity.

Single Nucleotide Polymorphisms (SNPs) - SNPs represent single-base variations across the genome. Useful for -

1. Detecting ancestral lineage.
2. Identifying disease risk variants.
3. High-resolution forensic investigations where STR data is limited.

Mitochondrial DNA (mtDNA) - Inherited only from the mother. Highly stable and survives extreme heat, decomposition, and environmental damage. Used when nuclear DNA is degraded, but cannot uniquely identify individuals—only maternal lineage.

Y-Chromosome Markers - Passed from father to son, useful for -

1. Male lineage tracing
2. Sexual assault cases with degraded male DNA

Cannot uniquely identify individuals within the same paternal line.

Techniques Used in DNA Profiling

Polymerase Chain Reaction (PCR) - Amplifies even very small or degraded DNA samples. Allows accurate testing from minute biological remains.

Capillary Electrophoresis - Separates amplified DNA fragments based on size. Produces electropherograms—visual graphs used to determine STR profiles.

Next-Generation Sequencing (NGS) - Revolutionises forensics by examining -

1. STR sequence variations
2. SNP patterns
3. mtDNA genomes

Provides deeper resolution for -

1. Complex kinship cases
2. Population genetics
3. Ancestry mapping
4. Mixed DNA samples

Applications of DNA Profiling in Genetic Analysis

Forensic Applications

1. Crime scene identification
2. Sexual assault investigations
3. Mass disaster victim identification
4. Missing persons tracking
5. Paternity and kinship resolution

Medical Genetics

1. SNP-based testing for hereditary disorders
2. Identifying mutation carriers
3. Early detection of genetic diseases

Pharmacogenomics

1. Predicts how individuals respond to drugs.
2. Enables personalised medicine by matching drug dosages to genetic profiles.

Evolutionary & Population Genetics

Studies -

1. Human migrations
2. Founder effects
3. Genetic drift

Assesses genetic diversity in populations.

Ancestry & Genealogy

Uses autosomal SNP arrays and haplogroup analysis to -

1. Trace ethnic origins
2. Identify genetic relatives
3. Build genealogical trees

Conservation Biology

1. Detects wildlife poaching using DNA from ivory, skin, hair, bones.
2. Helps identify species/subspecies.
3. Used in animal tracking, breeding programs, and ecological monitoring.

How Genetic Analysis Establishes Identity

DNA Extraction

Samples taken from -

1. Blood
2. Bone fragments
3. Teeth
4. Hair follicles
5. Tissue/soft tissue remains

PCR Amplification - STR loci or SNP regions are amplified for comparison.
STR Profiling - Unique patterns are identified and matched across samples.
Kinship Matching - Statistical analysis compares -

1. Suspect DNA with victim
2. Child with parents

3. Unidentified remains with relatives

Uses likelihood ratios to quantify confidence of biological relatedness.

Challenges in DNA Identification

Degraded DNA - Fire, explosion, water immersion, and chemicals cause fragmentation.
Contamination - Poor handling can introduce foreign DNA, compromising results.
Insufficient DNA - Occurs in skeletal remains or very old samples.
Mixed DNA Profiles - Common in mass disasters or crime scenes with multiple individuals. Deconvolution requires advanced bioinformatics.

India and DNA Technology - Legal & Regulatory Framework

DNA Technology (Use and Application) Regulation Bill, 2019 - Aimed to create a comprehensive regulatory structure -

1. Establish DNA Regulatory Board
2. Accredit DNA laboratories
3. Standardise data storage
4. Protect privacy of individuals

Not passed, but forms the foundation for future frameworks.
Criminal Procedure (Identification) Act, 2022 - Allows collection of DNA samples from -

1. Arrestees
2. Undertrials
3. Convicts

Enables broader forensic investigation but raises privacy concerns.
Role of DBT - Department of Biotechnology supports -

1. National biobanks
2. Genome India Project
3. Advanced sequencing technologies
4. Forensic DNA labs nationwide

Ethical, Legal, and Social Concerns (ELSI)

Privacy & Genetic Data Protection - DNA reveals sensitive health and ancestry information.
Genetic Discrimination - Potential misuse in employment, insurance, or social contexts.
Debate on National DNA Databases - Balances -

1. Crime-solving efficiency
2. Individual liberty and data rights

Informed Consent - Essential for -

1. Clinical testing
2. Population genetics
3. Consumer genomic platforms (e.g., ancestry tests)

Data Ownership - Who owns DNA data - individual, laboratory, state, or corporation? Clear policies still evolving globally.

Other Methods of Genetic Analysis

A. Mendelian (Classical) Genetics - Segregation & Independent Assortment. Linkage & Recombination Mapping for locating genes on chromosomes.

B. Cytogenetic Methods

Karyotyping — detects aneuploidies (e.g., Down syndrome).
FISH — identifies specific DNA sequences using fluorescent probes.
CGH — detects chromosomal gains/losses without cell culture.

C. Functional Genetic Analysis

CRISPR, TALENs, ZFN — editing genes to study function.

RNAi — silencing genes.
Overexpression Systems — increasing expression to test effects.

D. Epigenetic Analysis

Bisulfite Sequencing — maps methylation patterns.
ChIP-seq — studies DNA–protein interactions.
ATAC-seq — measures chromatin accessibility.
Source - https - //indianexpress.com/article/cities/delhi/red-fort-blast-aiims-dna-pulwama-parents-key-case-10361012/