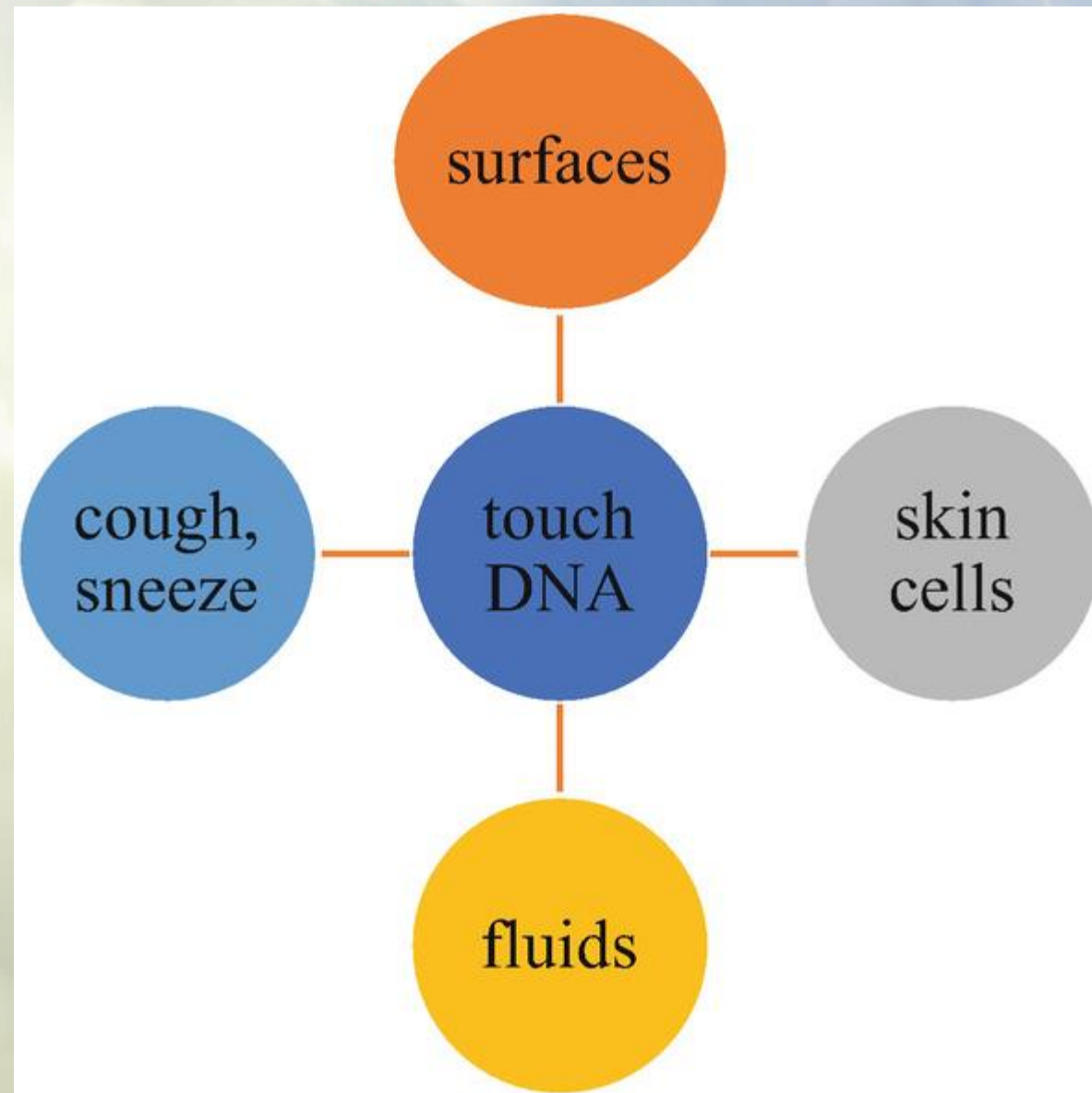


Future of DNA in Forensic Science

Gargi Deshmukh
Scientist In-charge
Genome Foundation

TOUCH DNA

- “Touch” DNA samples refer to samples where biological fluid cannot be detected, however there may be skin cells left behind due to contact with an item.



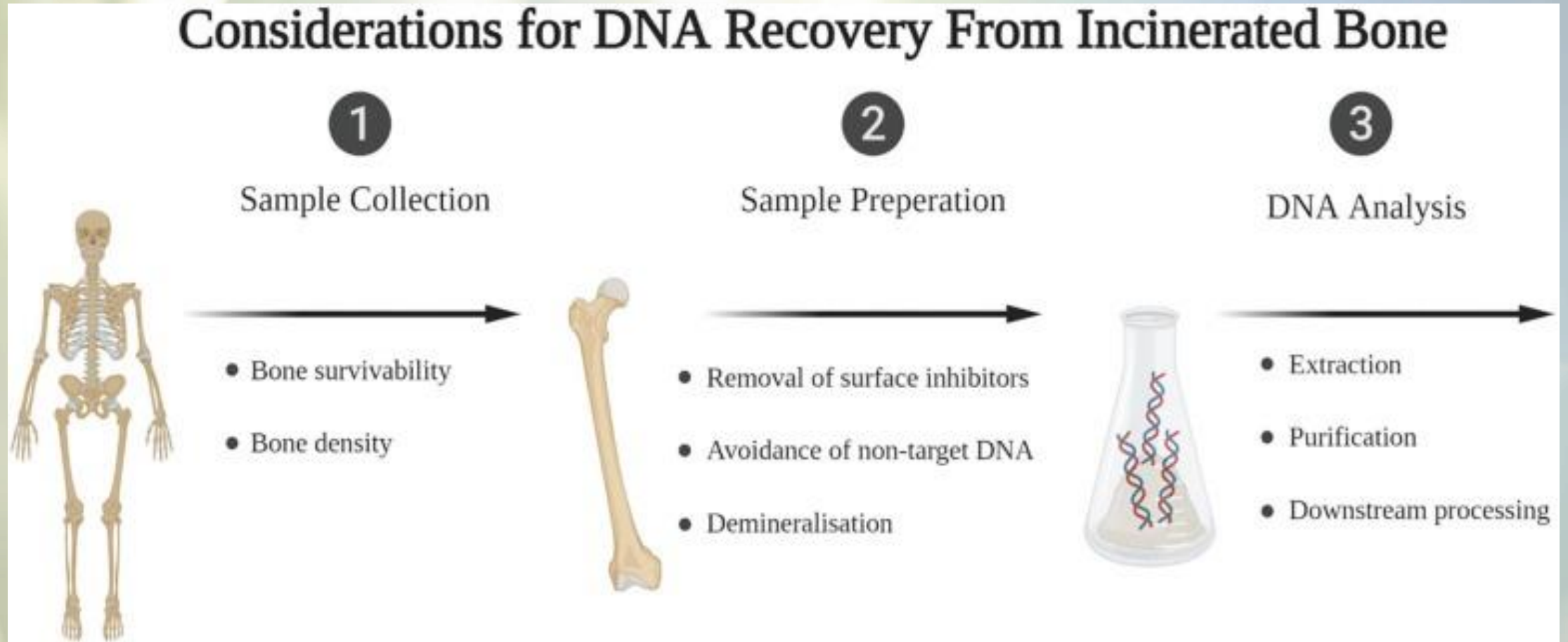
TOUCH DNA

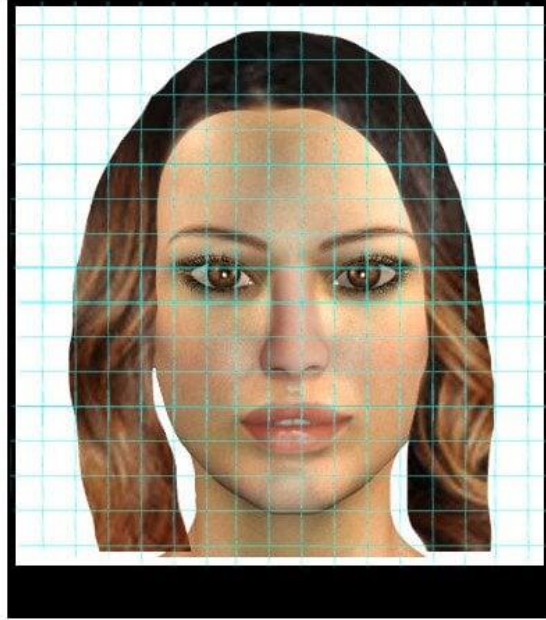
- In cases where the scientist is attempting to collect “touch” DNA for analysis, it is often difficult to collect a sufficient amount of DNA to generate a profile suitable for comparison.
- The accepted collection method has been to vigorously swab a sample with a cotton swab and forward the swab for DNA analysis.

- A few cases where touch DNA in the US has been used for getting convictions.

Item of Evidence	Type of case
Firearms, knife handles, weapon handles	any
Fired casing	any
Steering wheel and other vehicle swabs	carjacking, etc
Fingerprints on victim	any
Ligatures, hand cuffs, shoestrings	strangulation, kidnapping, rape, etc
Hand swabs from suspects	strangulation, rape
Face swabs from child victims (slapping, hitting)	child abuse
Swabs from limbs removed from animal carcasses	poaching
cell phones swabs	robbery, etc
victim neck swabs	strangulation
swabs from torn or forcibly removed clothing	rape, assault
airbag	DUI cases and others
tools	burglary
baggies	drug possession
paper demand notes	bank robberies
clothing items, hats, masks, gloves, glasses	any

DNA Recovery from Incinerated Bone





Predicted



Original

Envisaging the invisible woman!

Attempt to predict at least some features of an unknown suspect's phenotype from a small sample of DNA collected at a crime scene.

Forensic DNA Phenotyping

- “Phenotype” refers to the ensemble of all outwardly *observable* characteristics of an individual, including form and structural features, physical appearance, development and behavior.
- The genotype plays an important role in determining features of the phenotype but is not the sole contributor. Environment can also play an important role and can, for example, affect the expression of genes.

- A person's observable characteristics can be reconstructed from a small DNA sample or, in the relevant technical language, how much of a person's *phenotype* is determined from their *genotype*.
- This approach focuses not on STRs, but rather on single-nucleotide differences that occur at specific DNA locations, either within protein-coding genes or in DNA sections that affect gene expression.

Physical Features which can be Predicted

- Eye colour
- Hair colour (natural)
- Hair pattern
- Physical stature
- Skin tone/colour
- Eyebrow shape
- Male pattern baldness



Eye colour

HERC2, OCA2, SLC45A2,
TYR, IRF4, SLC24A4



Hair colour

MC1R, OCA2, SLC24A4,
HERC2, TUBB3, ECOC2,
SLC45A2, KITLG, IRF4,
TYR, PIGU, TYRP1



Skin colour

SLC24A4, SLC24A5, SLC45A2,
ASIP, ANKRD11, BNC2, DEF8,
HERC2, IRF4, KITLG, MC1R,
OCA2, PIGU, RALY, , TYR, TYRP1,
DEF8, MYEF2, CTXN2, CEP152



Hair shape

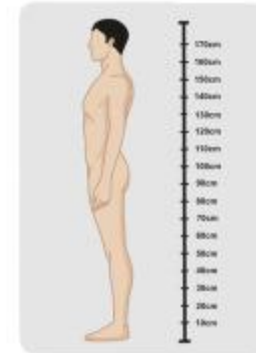
AR/EDA2R, EBF1,
HDAC9, TARDBP



Baldness

EDAR, TCHH, ERFF11,
PTK6, OFCC1,
KRTAP2-3, HOXC13,
WNT10A, KRT71, PADI3,
GATA3, PEX14, LIPH,
TGFA, HRNR, LGR4,
TCHHL1, RPTN,
S100A11, FRAS1, LCE3E,
LINC01494, PRRS53,
FLG-AS1, TRAF2, RPTN

GENETIC DETERMINANTS OF EXTERNALLY VISIBLE CHARACTERS



Body height

ACAN, DNM3, IHH,
FBXW11, EFEMP1,
GH region, GHSR,
GPR126, HHIP, HMGAI,
LCORL, MICA, NOG,
NPR3, PML, PPIF,
SDR16C5, SOCS2



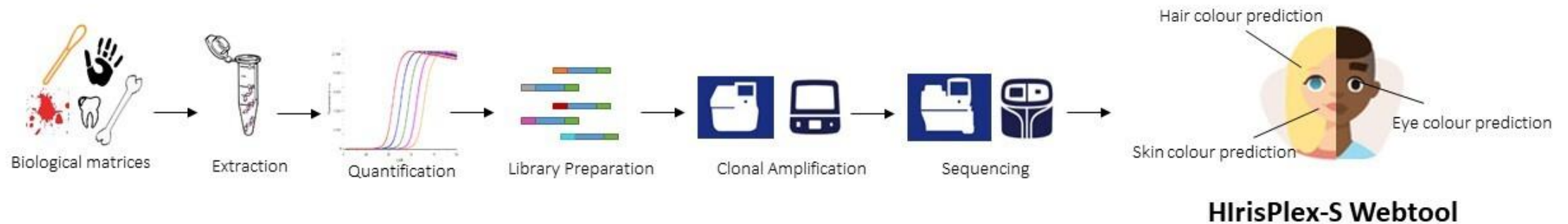
Facial features

DCHS2, PDE8A, SCHIP, ASPM, DLX6, DYNC1L1, EDAR, HOXD1-MTX2, WDR27, PAX3, TP63, PABP1-C1L2A, HADC8,
THEM163, COL17A1, PARK2, OSR1-WDR35, PAX9, SLC25A2, MIPOL1, FOXA1, MAFB, TRPC6, TBX15, RPS12, EYA4,
FREM1, HOXD cluster, PKDCC, C5orf50, EPHB3, DVL3, ZNF219, CHD8, SLC35D1, FGFR1, LRP6, SOX9, CASC17,
KCTD15, PRDM16, DHX35, PAX1, RUNX2, GSTM2, GNAI3, ALX3, SUPT3H, GL13, RAB7A, ACAD9, MBTPS1, PCDH15

- ❌ NO MATCH WITH REFERENCE PROFILES
- ❌ NO MATCH WITH GENETIC PROFILES
ENTERED IN CRIMINAL DATABASES



FORENSIC DNA PHENOTYPING



Quick



Effective



High coverage



Guess the ages!

A. 18; B. 25; C. 38; D. 50; E. 65



DNA Phenotyping used in casework

- The [Idaho Falls case](#) is worthy of note because a previous suspect had spent 20 years in jail for the crime, despite the fact that his DNA did not match that collected at the crime scene. He was finally released in 2017 after the DNA phenotyping was completed. And a new suspect identified with the aid of phenotyping was arrested, and confessed to the crime, in 2019. There were also two rape and murder cases in the 1990s in the Netherlands solved with the aid of DNA phenotyping. Snapshot composites have been released publicly in several other unsolved cases, including ones of sexual predation, in the hopes that they might lead to identification of suspects.

Pros & Cons of DNA Phenotyping

- The accuracy is not great enough to be used in court proceedings, unless an indictment aided by DNA phenotyping has produced a subject whose DNA STR profile matches that of the crime scene sample.
- The composite generated might not match exactly due to chemical alterations to the observed characteristics.
- As an investigative aid, phenotypic composites should perhaps be compared to the more traditional composite subject sketches produced from eyewitness descriptions.
- The composite generated may be used as an investigative tool to get confessions or answers from suspects/culprits.

DNA on the Go

- The vision looks like this: researchers, consumers, government employees—everyone—will be reading the DNA of their own bodies and the living things around them and streaming that data on the internet.
- DNA-reading sensors would be integrated into food production equipment, polluted waters, farms and phones. They would track the presence and movement of food pathogens and deadly viruses.



- MinION and PromethION are based on nanopore sequencing technology.
- An electrical potential is applied, causing ions to flow through nanopores. As individual strands of DNA pass through the pores, its presence causes specific changes in the currents.
- Software quickly translates the sequence of currents into the corresponding DNA components: nucleic acids represented by the letters A, T, G and C. The sequence of the four letters comprises the unique genetic code for every living thing.
- It tells users what species of microbes they've got by matching their DNA reads to the DNA sequences of known species in its database.

