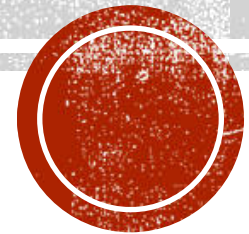


DNA PROFILING TECHNIQUES AND INTERPRETATION OF RESULTS

Gargi Deshmukh
Scientist In-charge
Genome Foundation



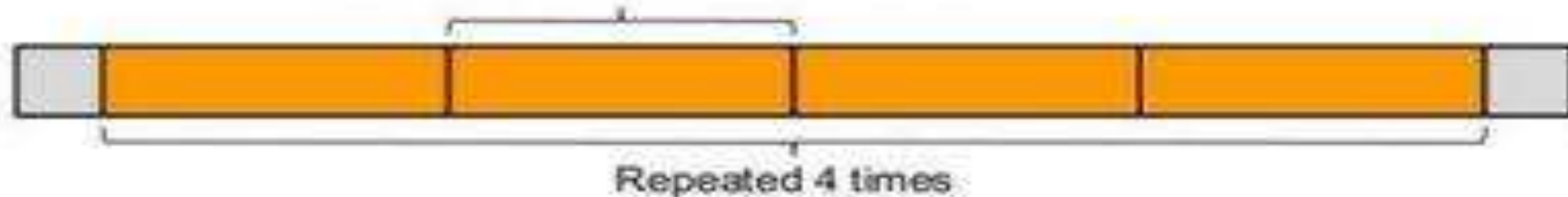
- **Repetitive DNA** is the patterns of nucleic acids that occur in multiple copies throughout the genome.
- **Tandem repeats** are one of the major types of repetitive DNA that copy repetitive sequence units that lie adjacent to each other, forming a nucleotide block.
- **VNTR** (variable number tandem repeats) and **STR** (short tandem repeats) are two types of tandem repeats found in the eukaryotic genome.



Tandem Repeat elements

Minisatellite: Variable Number Tandem Repeats (VNTR)

Repeat unit size = hundreds base pairs

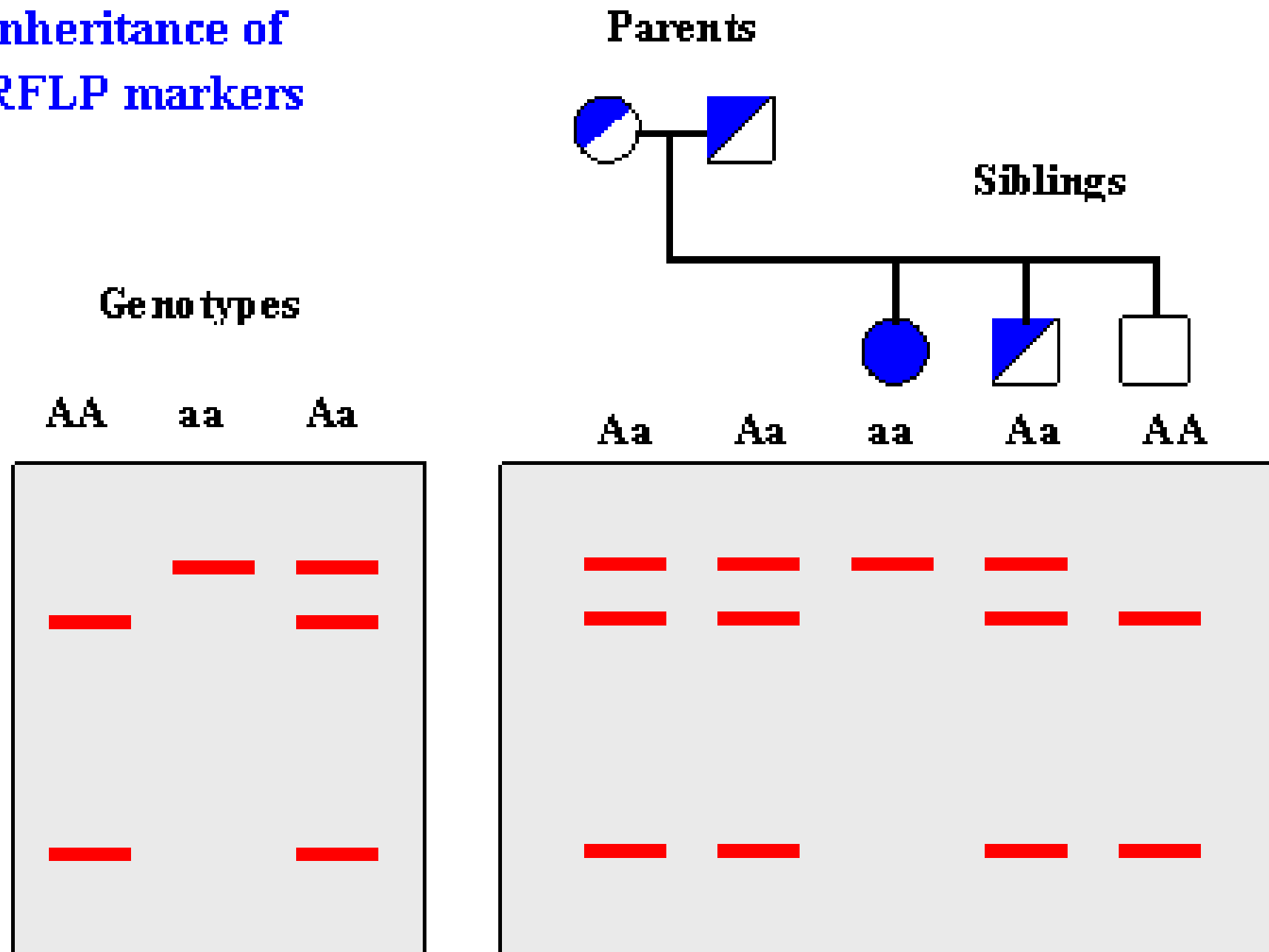


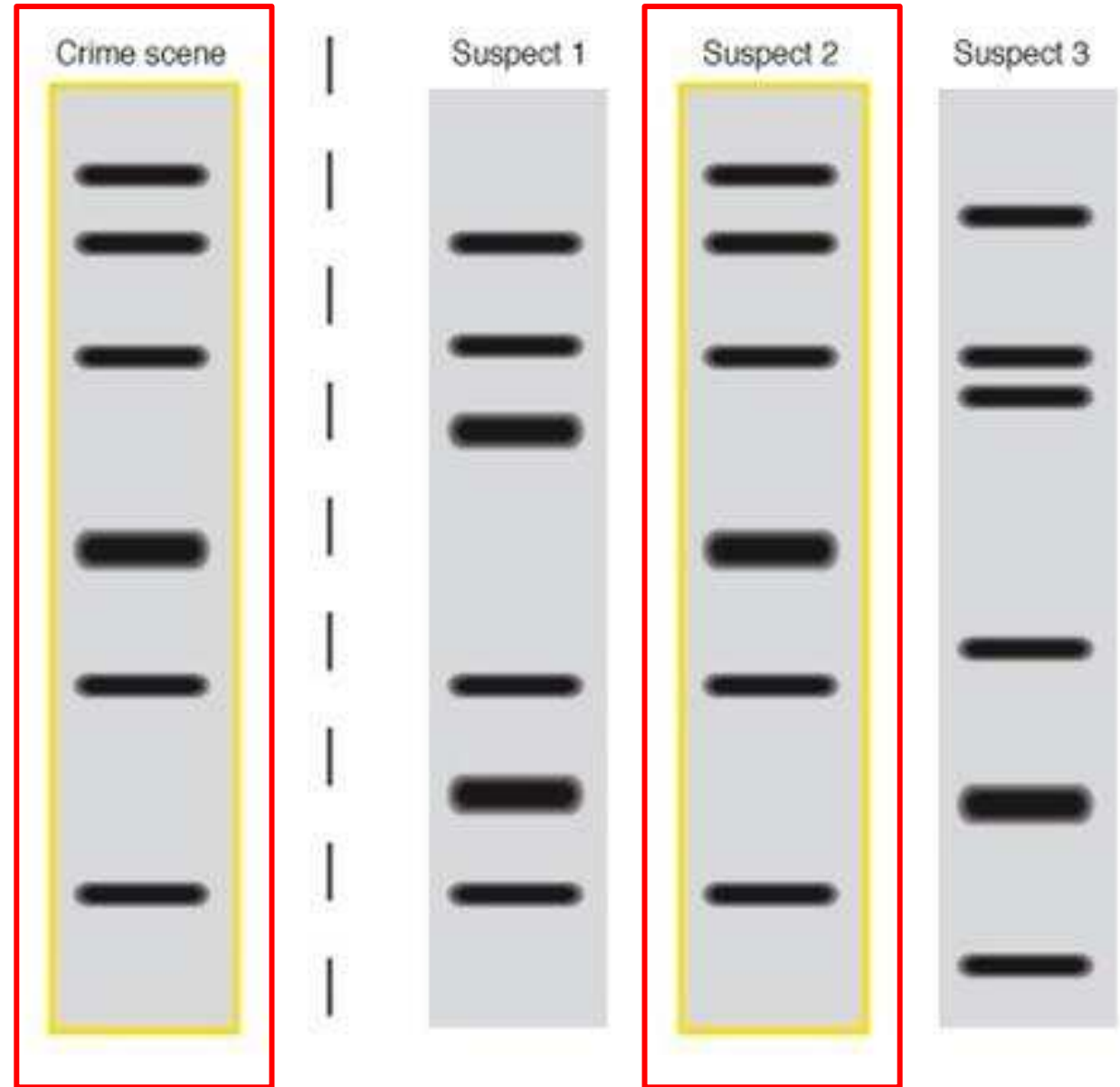
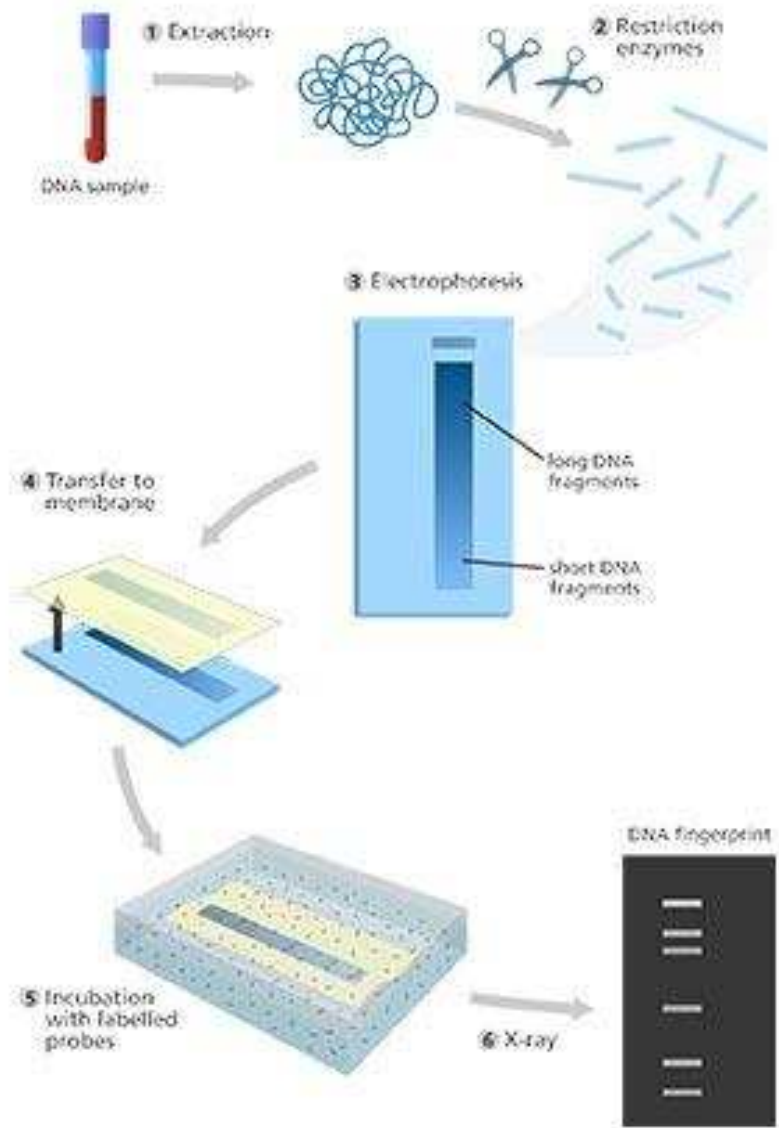
Microsatellite: Short Tandem Repeats (STR) – Simple Sequence Repeats (SSR)

Repeat unit size = 2 - 6 base pairs



Inheritance of RFLP markers





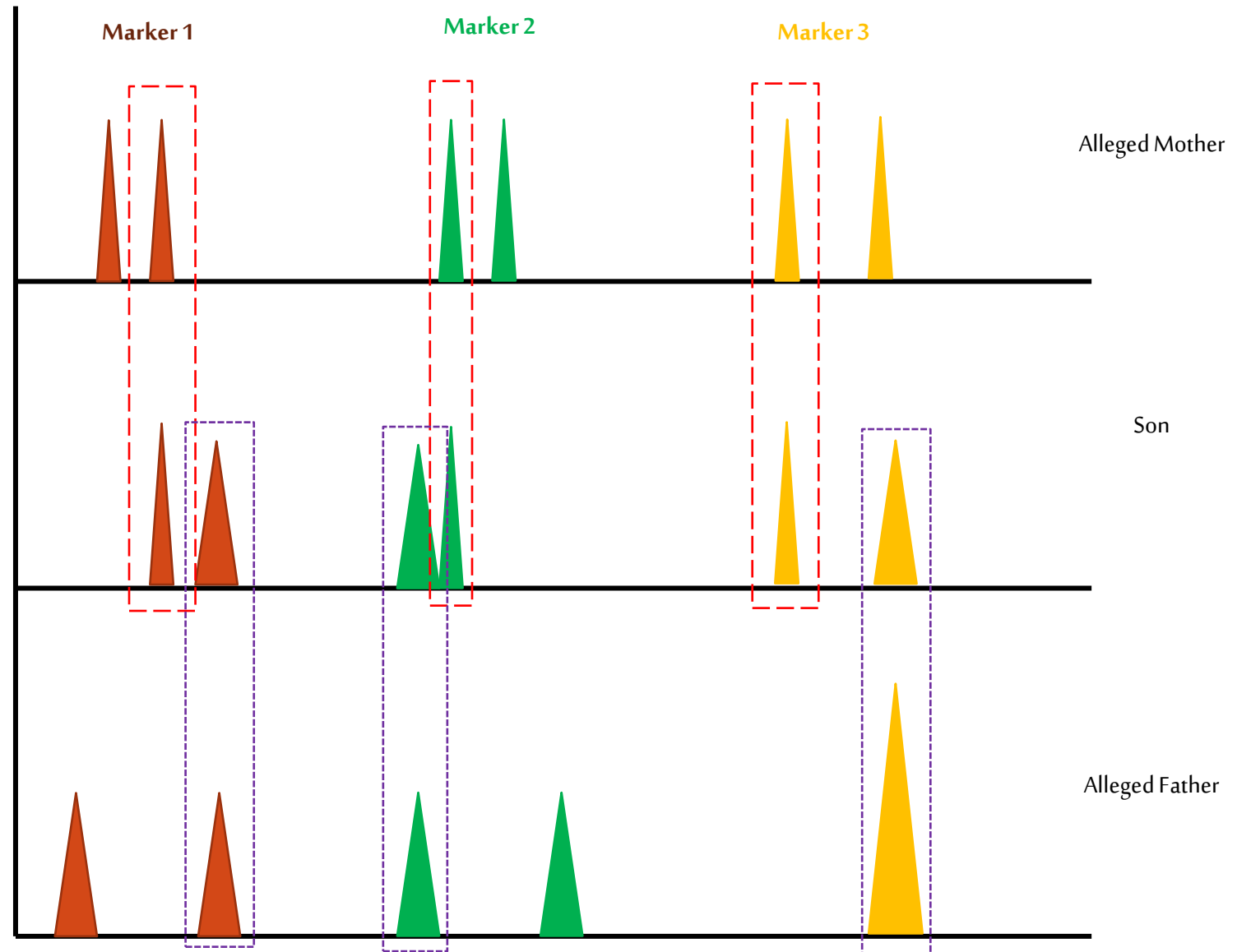
Example of Paternity & Maternity

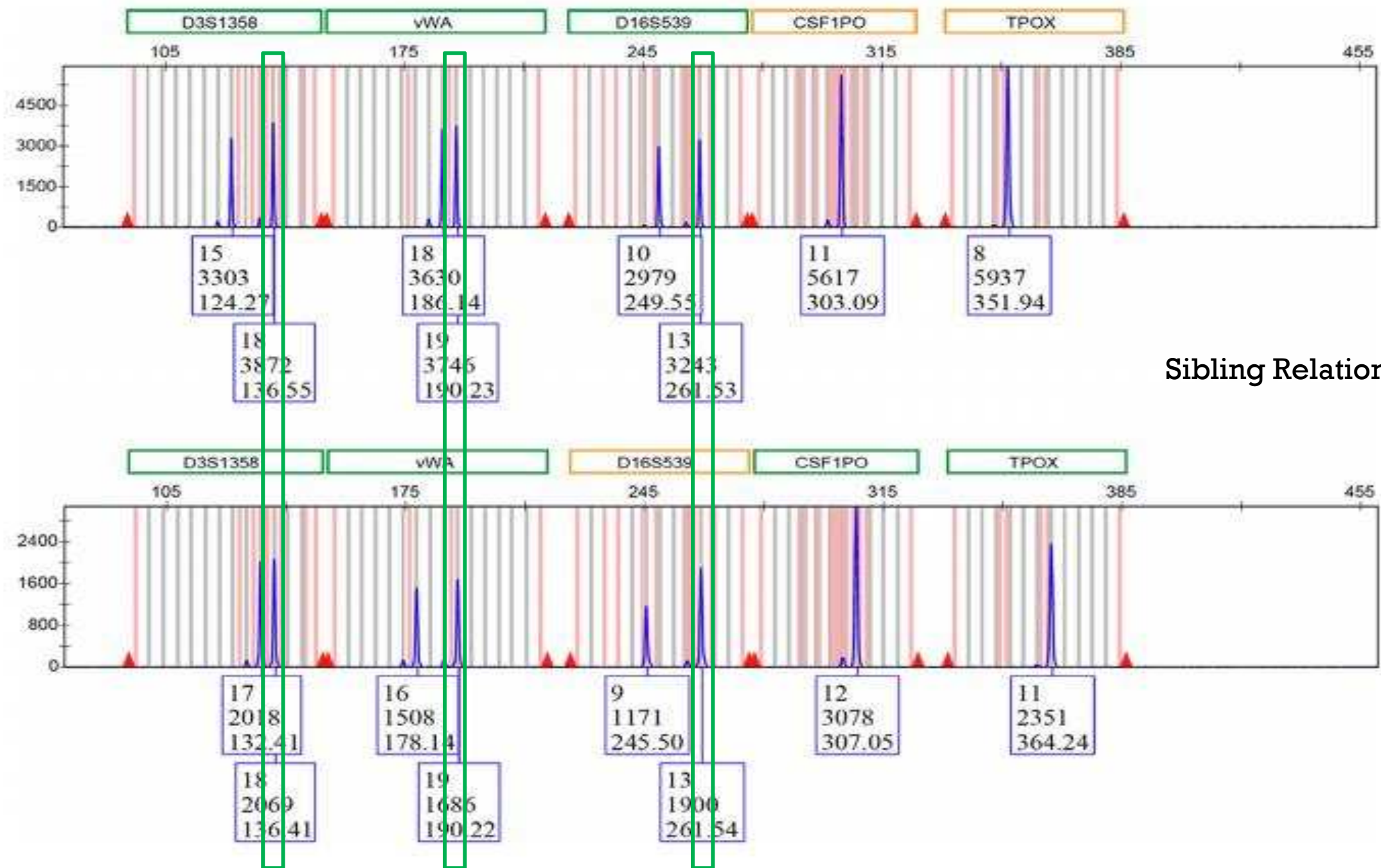
- Used to identify a child's parents
- Each child inherits one set of chromosomes from each parent
- Only the repeats present in the parents can be passed on to the offsprings

DNA Marker	Father	Mother	Daughter	Son
D7S820	9, 10	10, 11	9, 11	10, 10
D3S1358	16, 17.2	15, 15.2	15, 16	15, 16
D16S539	11, 11	9, 11	11, 11	9, 11
D8S1179	13, 15	15, 16	13, 16	13, 16
CSF1PO	10, 12	10, 10	10, 12	10, 12
SE33	25, 31.2	27, 33	31.2, 33	25, 33
TPOX	8, 12	9, 13	9, 12	9, 12
Amelogenin	X, Y	X, X	X, X	X, Y



- Paternity match – alleged parents for a minor child (15 year old).





Sibling Relationship





Victim's Reference	Victim's clothing	Accused's Reference	Accused's clothing

- RFLP comparison in a case of sexual assault.
- Classic example of Locard's Exchange Principle.



- DNA from alleged driver, airbag cuttings and dashboard.
- Comparison revealed no-matches.



Alleged Driver



Airbag Cuttings



Swab from the dashboard

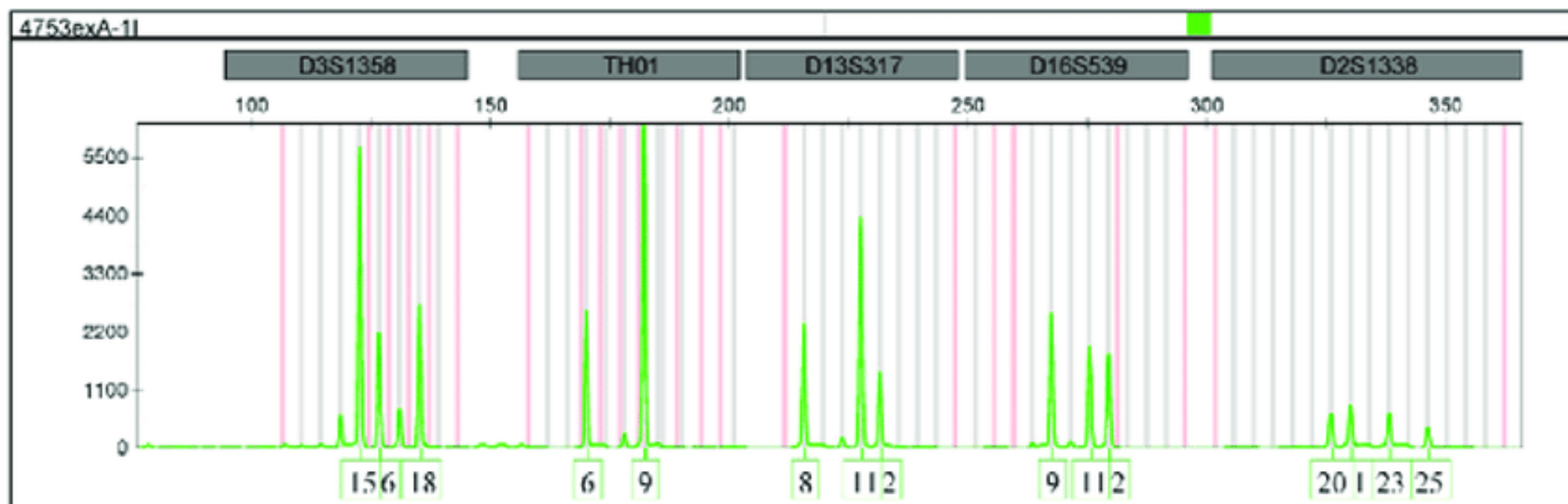
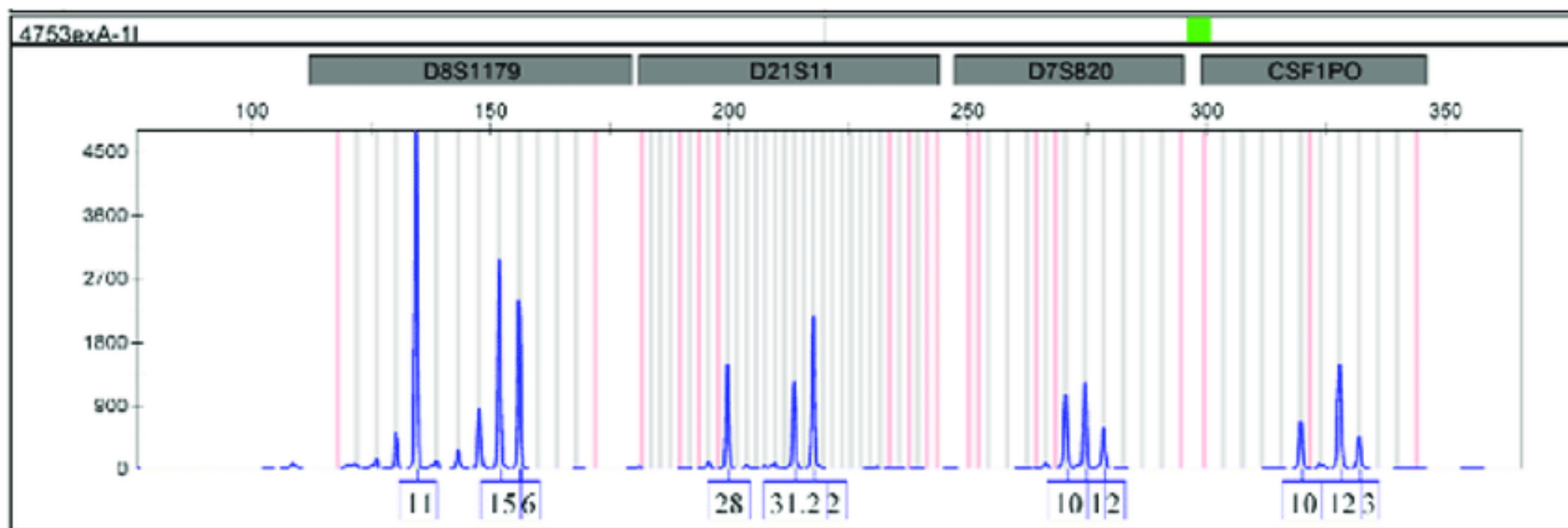


Example of Crime Scene Evidence

DNA Marker	Used Condom (from crime scene)	Suspect A	Suspect B	Suspect C	Suspect D
D7S820	10, 12	9, 9	7, 7	10, 12	8, 12
D3S1358	15, 18	13, 17	15, 15.3	15, 18	13, 18
D16S539	11, 14	11, 12	9, 11	11, 14	10, 14
D8S1179	13, 13	13, 13	15, 15	13, 13	10, 12
CSF1PO	12, 12	8, 8	10, 12	12, 12	8, 11
SE33	22, 29.2	31.2, 33	14.3, 28	22, 29.2	17, 20
TPOX	7, 9.3	9, 9	9, 12	7, 9.3	8, 9
Amelogenin	X, Y	X, Y	X, Y	X, Y	X, Y



Mixture Sample
--- DNA Profile



Marker Names	Victim (Call Girl)	Condom from the crime scene	Suspect 1	Suspect 2
Marker 1	10, 11	8, 10, 11, 15	9, 14	8, 15
Marker 2	13, 16	13, 16, 19	17, 17	19, 19
Marker 3	12.2, 15.3	11, 12.2, 15, 15.3	17, 17	11, 15
Marker 4	16, 21	12, 16, 17, 21	11, 18	12, 17
Marker 5	7, 7	7, 8, 9	5.3, 6.3	8, 9
Marker 6	18, 21	18, 21, 22.3	16.3, 20	18, 22.3
Sex Marker	X, X	X, X, X, Y	X, Y	X, Y





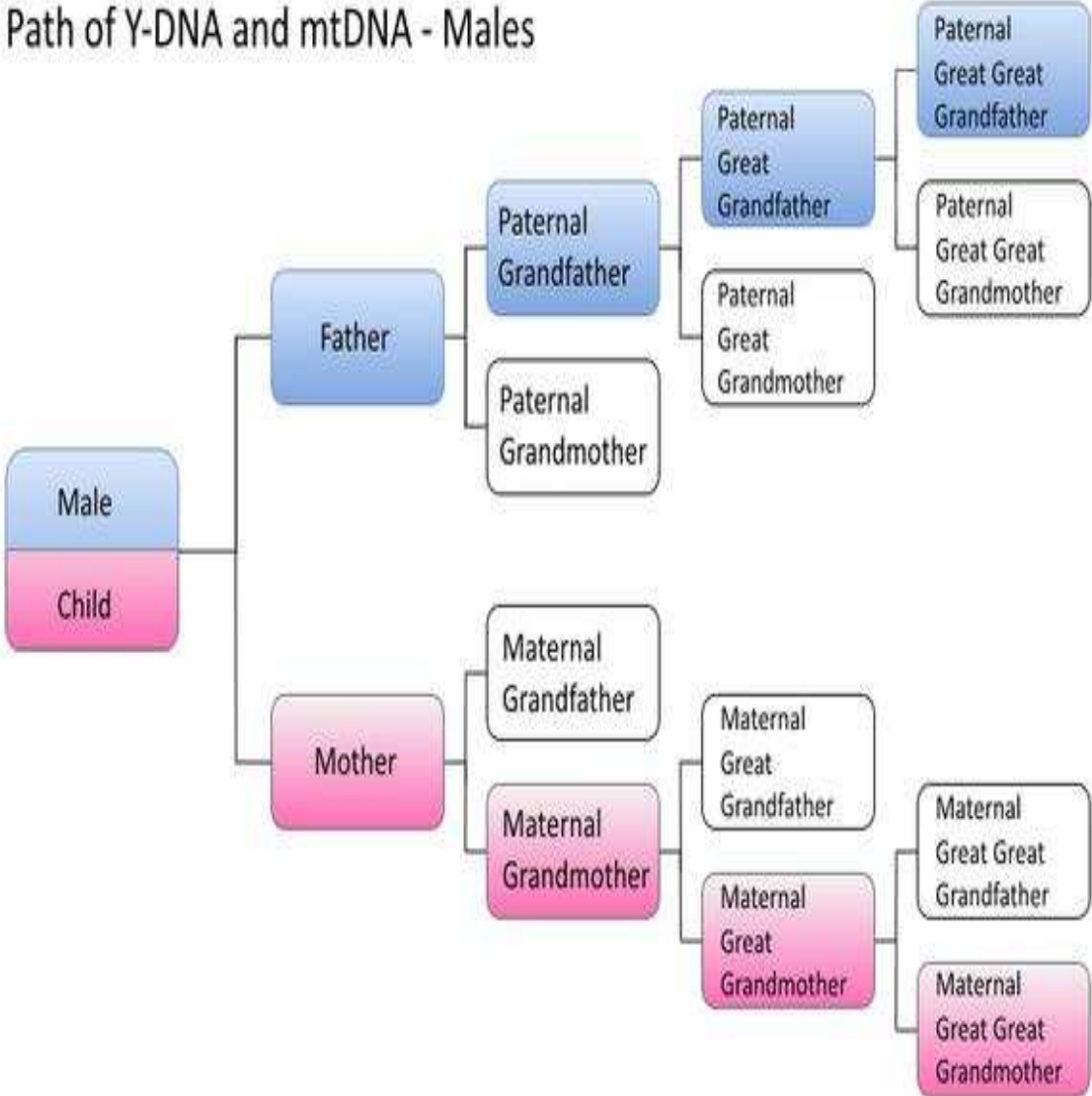
Marker Names	Deceased (Young man)	Suspected murder weapon (machete)
Marker 1	7, 9	7, 9
Marker 2	13.2, 15.2	13.2, 15.2
Marker 3	10, 15	10, 15
Marker 4	9, 16	9, 16
Marker 5	18, 23.2	18, 23.2
Marker 6	21, 21	21, 21

IDENTICAL

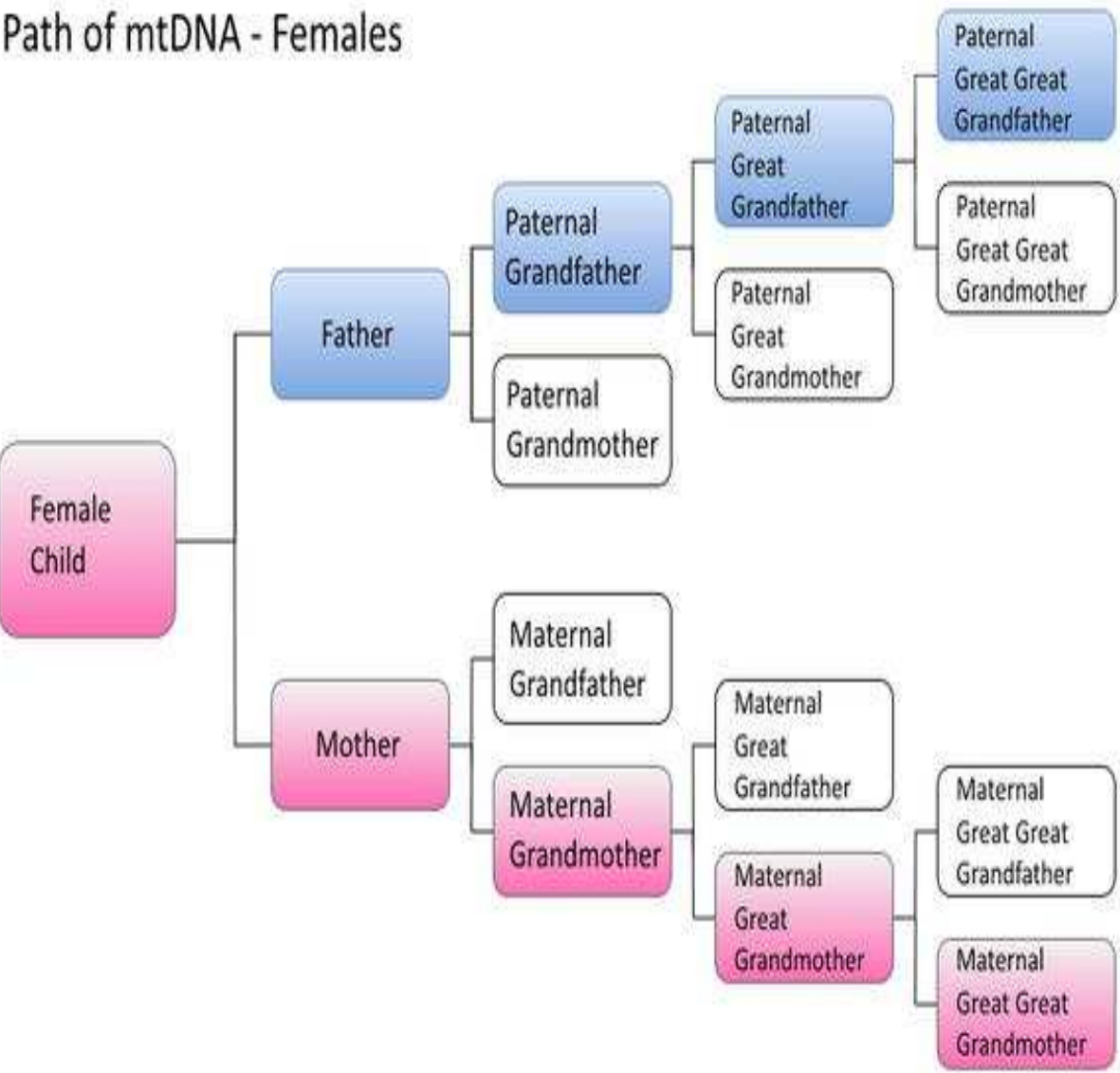
- Weapons identification – this is an integral part of getting a firm conviction.



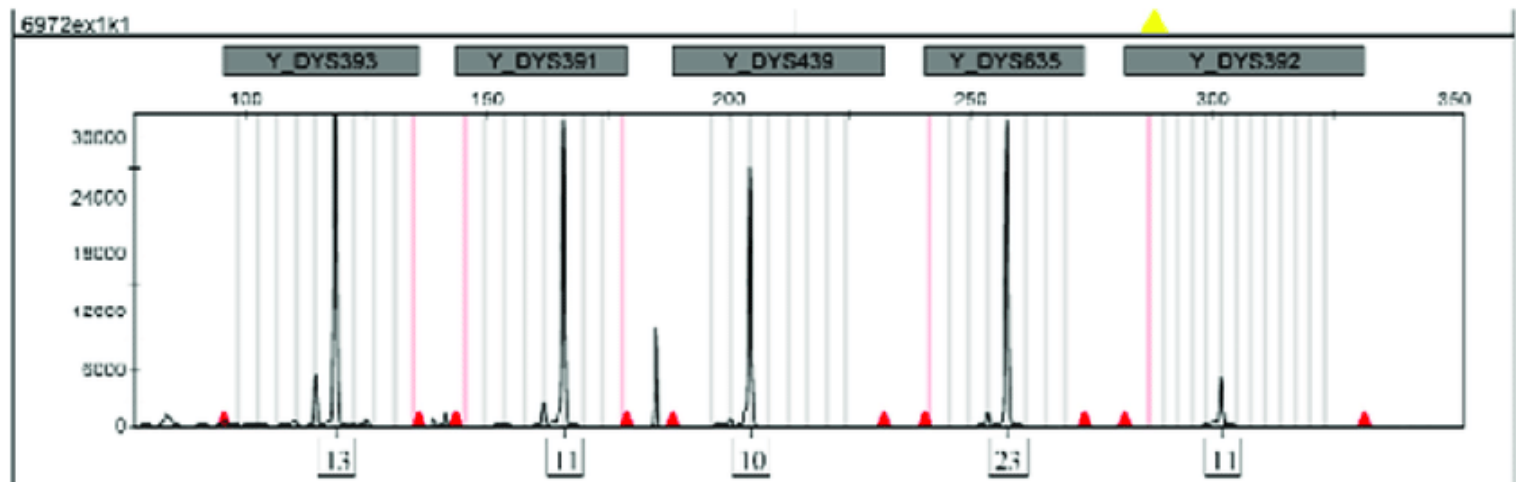
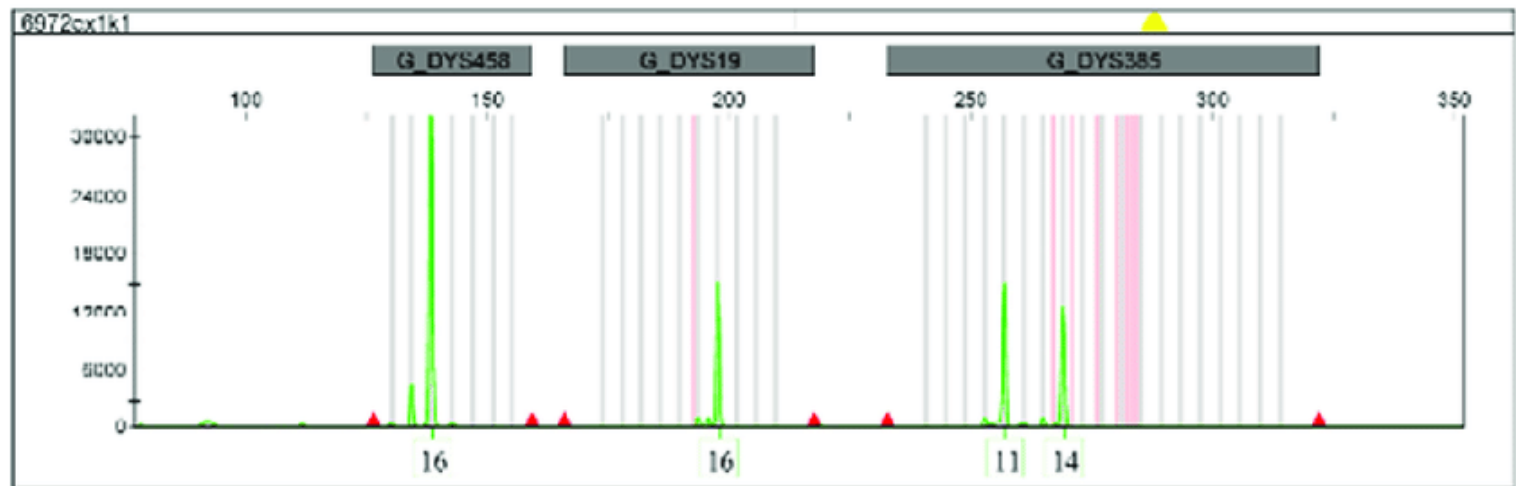
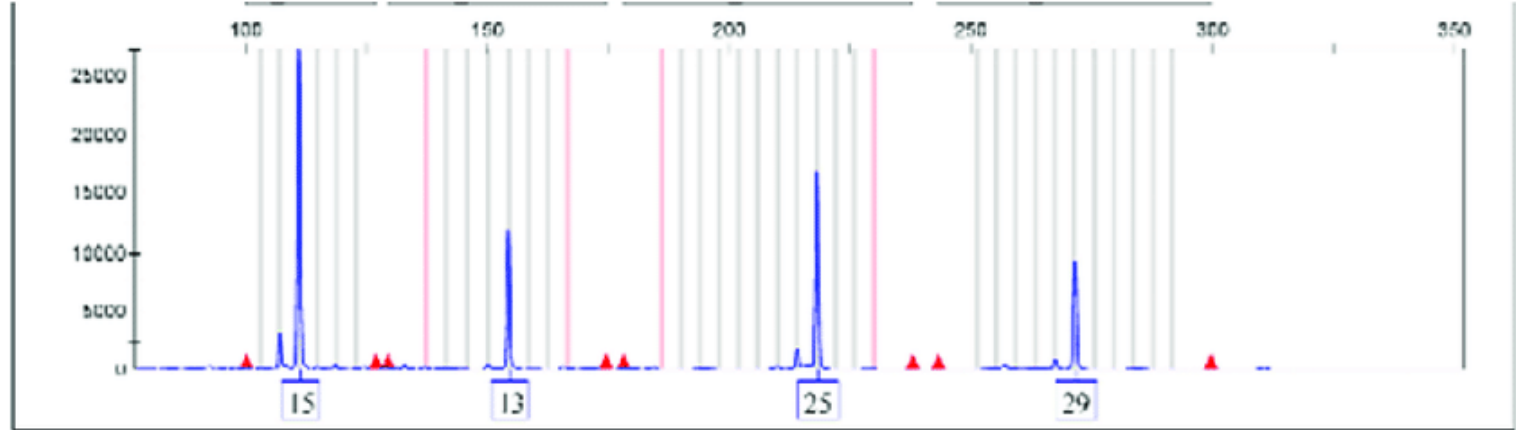
Path of Y-DNA and mtDNA - Males



Path of mtDNA - Females



Y STR PROFILE

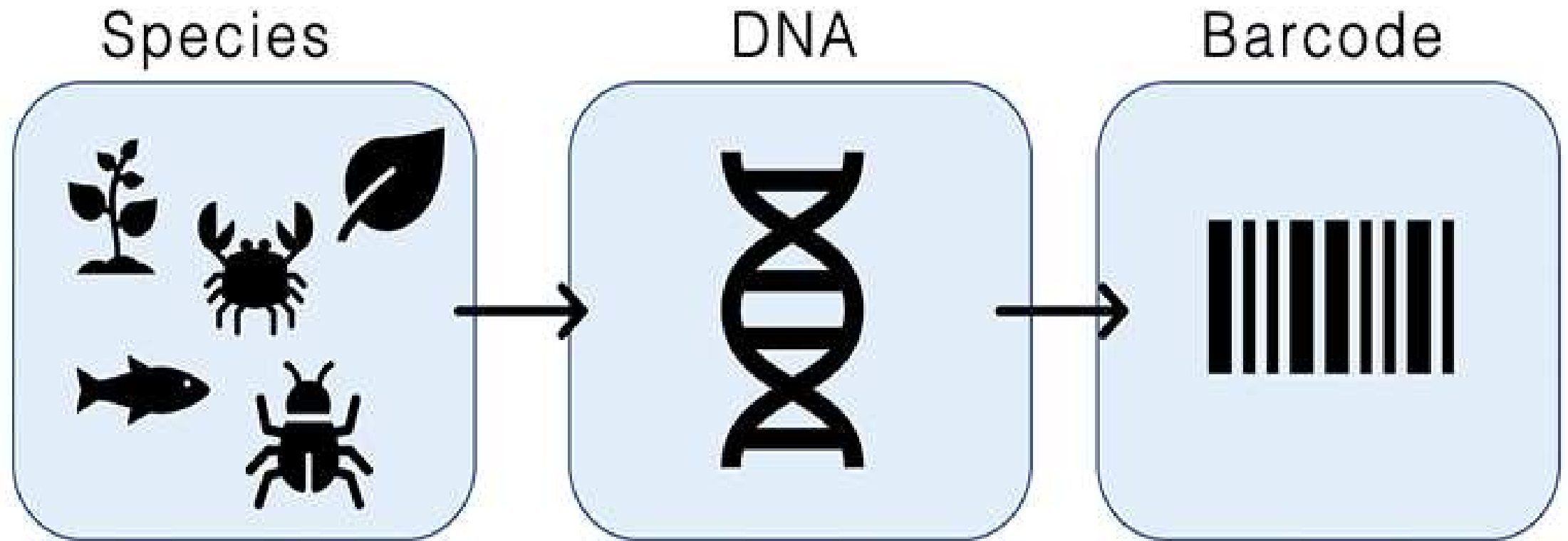


Mitochondrial Markers	Recipient	Donor
HVR Marker 1	+++++	+++++
HVR Marker 2	+++++	+++++
HVR Marker 3	----	----
HVR Marker 4	----	----
HVR Marker 5	+++++	+++++
HVR Marker 6	+++++	+++++
HVR Marker 7	----	----

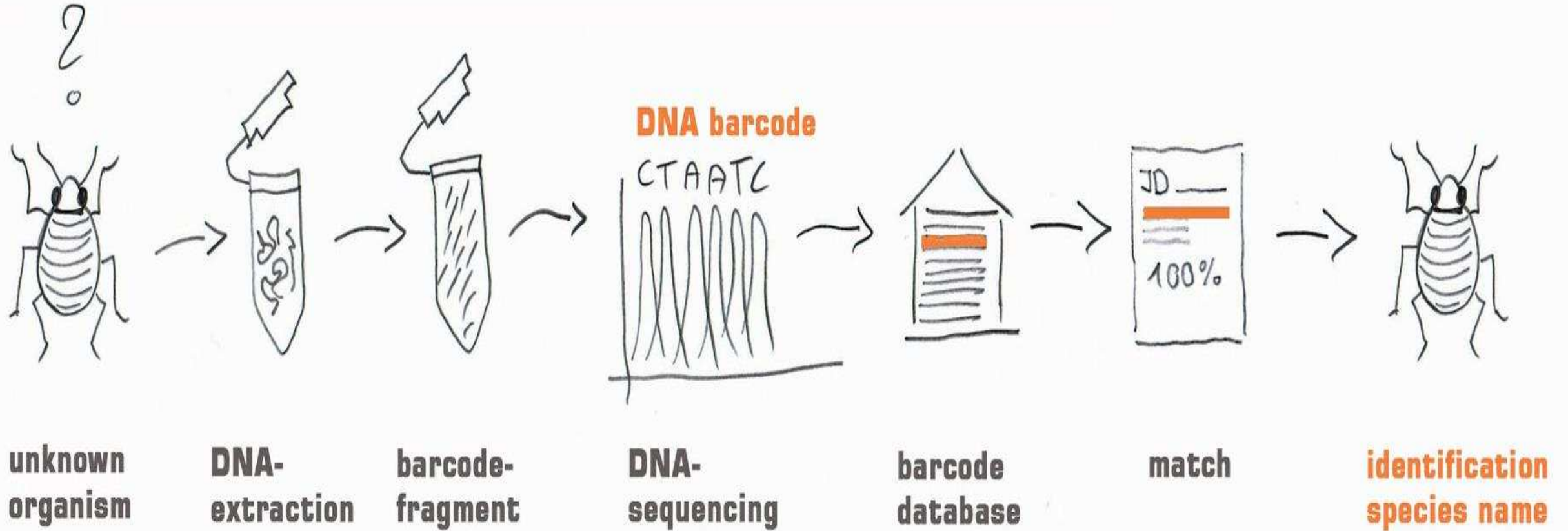
- *Identical nucleotide sequences were found in both the siblings, indicating they are maternally (biologically) related.*



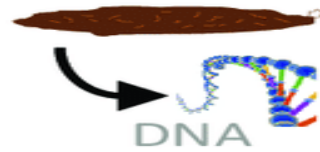
DNA BARCODING



How does DNA Barcoding work ?



Faecal sample



A. DNA BARCODING

PCR

CATGCTTTTGTAAATGATTTCTTTATAGTCATACCTATTATGAT

Myotis daubentonii

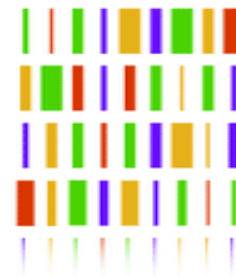


B. METABARCODING

Insect trap
bulk
sample



PCR



Microtendipes pedellus

Oecetis ochracea

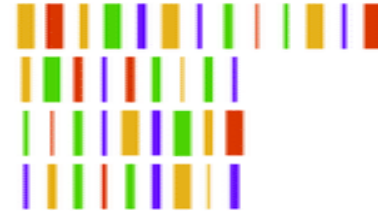
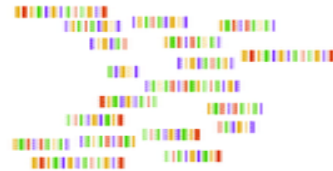
Metellina segmentata

Tipula recondita

...

C. METAGENOMICS

PCR



DNA from:

Bat

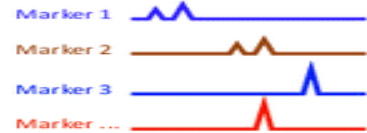
Insects

Bacteria

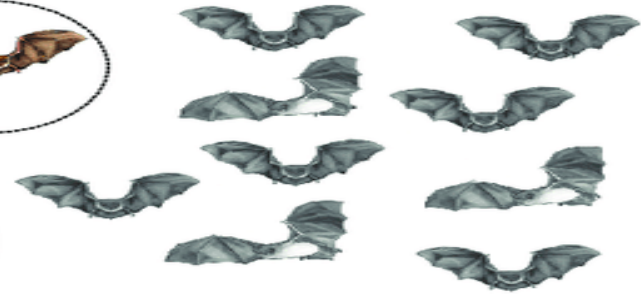
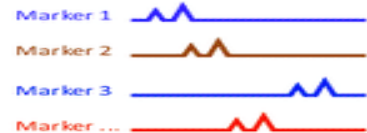
Viruses

D. MICROSATELLITE GENOTYPING

Sample A



Sample B





Thank

you

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