Table 3.1 Types of biological material that can be recovered from a crime scene. The DNA profiles generated from crime scene material are compared against reference profiles that are provided by suspects (or to a collection of reference samples held on a DNA database), and in some cases, the victims.

Scenes of Crime	Reference Samples
Blood	Blood
Semen	Buccal swabs
Hair	Pulled hairs (containing roots)
Epithelial cells – shed skin cells:	
Saliva	
Dandruff	
Clothing	
Cigarette butts	
Drinking vessels/food	
Urine	
Vomit	
Faeces	
Touch DNA	

Table 5.1 The PCR reaction can theoretically multiply DNA over 1 billion fold after 32 cycles – in reality it is not 100% efficient but is still extremely powerful.

Cycle	Number of PCR products	
1	0	
2	0	
3	2	
4	4	
5	8	
6	16	
7	32	
8	64	
9	128	
10	256	
20	262,144	
28	67,108,864	Standard cycle number using Applied Biosystems SGM Plus and Identifiler Kits
30	268,435,456	
32	1,073,741,824	Standard cycle number using Promega PowerPlex 16 Kit
34	4,294,967,296	Maximum number of cycles normally used in forensic analysis

Table 6.1 The development of STR systems. Two STR systems, the quadruplex (QUAD) and SGM were developed by the Forensic Science Service in the UK. The AmpFℓSTR® SGM Plus® became commercially available in 1998 and has been adopted by a large number of laboratories for routine forensic casework. The AmpFℓSTR® Identifiler® and PowerPlex® 16 both analyse 15 STR including the 13 loci CODIS loci that are required to be analysed for forensic casework in the USA. The two kits are used widely worldwide, particularly for kinship testing; other kits with additional loci are also available (see Chapter 10)

QUAD	SGM	SGM Plus®	Identifiler®	PowerPlex® 16
vWA	Amelogenin	Amelogenin	Amelogenin	Amelogenin
TH01	vWA	D3S1358	D3S1358	D3S1358
F13A1	D8S1179	vWA	vWA	vWA
FES	D21S11	D16S539	D16S539	D16S539
	D18S51	D8S1179	D8S1179	D8S1179
	TH01	D21S11	D21S11	D21S11
	FGA	D18S51	D18S51	D18S51
		TH01	TH01	TH01
		FGA	FGA	FGA
			D13S317	D13S317
			CSF1PO	CSF1PO
			D7S820	D7S820
			TPOX	TPOX
			D5S818	D5S818
		D2S1338	D2S1338	Penta D
		D19S433	D19S433	Penta E

Table 6.2 Profiles have been generated from the same DNA sample using three commercial kits, the AmpFℓSTR® SGM Plus®, AmpFℓSTR® Identifiler® and the PowerPlex® 16. The alleles that are detected in the loci that are common between the kits are all identical.

	Profile					
Locus	SGM Plus®		Identifiler®		PowerPlex® 16	
Amelogenin	X	Y	X	Y	X	Y
D3S1358	15	17	15	17	15	17
vWA	14	17	14	17	14	17
D16S539	11	13	11	13	11	13
D8S1179	11	13	11	13	11	13
D21S11	30	31.2	30	31.2	30	31.2
D18S51	14	15	14	15	14	15
TH01	9	9.3	9	9.3	9	9.3
FGA	21	21	21	21	21	21
D13S317			10	14	10	14
CSF1PO			9	12	9	12
D7S820			8	10	8	10
TPOX			11	11	11	11
D5S818			11	13	11	13
D2S1338	24	25	24	25		
D19S433	14	15.2	14	15.2		
Penta D					12	13
Penta E					12	14

Table 8.1 The profile frequency is estimated using the principles of the Hardy Weinberg Law and an allele frequency database that was constructed using 400 alleles. Because the loci do not display any linkage and appear to be inherited independently of each other, the product rule can be used, multiplying each genotype frequency together to calculate the overall profile frequency.

Locus	Allele	Allele frequency	HWE	Genotype frequency
D3S1358	15 17	0.2825 0.2225	*2pq*	0.1257
vWA	14 17	0.0850 0.2500	*2pq*	0.0425
D16S539	11 13	0.2975 0.1750	*2pq*	0.1041
D2S1338	24 25	0.1000 0.1200	*2pq*	0.0240
D8S1179	11 13	0.0625 0.3475	*2pq*	0.0434
D21S11	30 31.2	0.2625 0.1050	*2pq*	0.0551
D18S51	14 15	0.1675 0.1425	*2pq*	0.0477
D19S433	14 15.2	0.3275 0.0250	*2pq*	0.0164
TH01	9 9.3	0.1375 0.3500	*2pq*	0.0963
FGA	21 21	0.1775 0.1775	p^2	0.0315
		Profile frequency		**$7.579\ 10^{-14}$**

Table 8.2 The profile frequency has been recalculated from Table 8.1 using the Balding correction for sampling bias. The impact of this correction factor is greatest on the rare alleles.

Locus	Alleles	Allele frequency	Allele Count	Corrected allele frequency	HWE	Genotype proportion
D3S1358	15	0.2825	113	115/404 = 0.2847	*2pq*	0.1282
	17	0.2225	89	91/404 = 0.2252		
vWA	14	0.0850	34	36/404 = 0.0891	*2pq*	0.0450
	17	0.2500	100	102/404 = 0.2525		
D16S539	11	0.2975	119	121/404 = 0.2995	*2pq*	0.1068
	13	0.1750	70	72/404 = 0.1782		
D2S1338	24	0.1000	40	42/404 = 0.1040	*2pq*	0.0257
	25	0.1200	48	50/404 = 0.1238		
D8S1179	11	0.0625	25	27/404 = 0.0668	*2pq*	0.0466
	13	0.3475	139	141/404 = 0.3490		
D21S11	30	0.2625	105	107/404 = 0.2649	*2pq*	0.0577
	31.2	0.1050	42	44/404 = 0.1089		
D18S51	14	0.1675	67	69/404 = 0.1708	*2pq*	0.0499
	15	0.1425	57	59/404 = 0.1460		
D19S433	14	0.3275	131	133/404 = 0.3292	*2pq*	0.0196
	15.2	0.0250	10	12/404 = 0.0297		
TH01	9	0.1375	55	57/404 = 0.1411	*2pq*	0.0992
	9.3	0.3500	140	142/404 = 0.3515		
FGA	21	0.1775	71	75/404 = 0.1856	p^2	0.0345
	21	0.1775	71	75/404 = 0.1856		
				Profile frequency		**$1.4225\ 10^{-13}$**

Table 8.3 The effect of different correction methods on the profile frequency calculated in Table 8.1. With this profile, applying a minimum allele frequency of 0.0125 would have no impact because the rarest allele frequency is 0.025.

Calculation method	Profile Frequency	Fold reduction relative to uncorrected frequency
Uncorrected	$7.58\ 10^{-14}$	
Size Bias	$1.42\ 10^{-13}$	2 (1.88)
Subpopulation: $\theta = 0.01$	$2.44\ 10^{-13}$	3 (3.29)
Subpopulation: $\theta = 0.03$	$1.62\ 10^{-12}$	21
Profile ceiling – 1 in 1 billion	$1.00\ 10^{-9}$	13,195

Table 10.1 Core STR loci as defined by different agencies, which facilitate the sharing of DNA profiles across international borders. As databases increase in size there is a requirement for more loci to be added in order to avoid adventitious (false) matches.

INTERPOL	ESS	ESS - extended	CODIS
D3S1358	D3S1358	D3S1358	D3S1358
TH01	TH01	TH01	TH01
D21S11	D21S11	D21S11	D21S11
D18S51	D18S51	D18S51	D18S51
vWA	vWA	vWA	vWA
D8S1179	D8S1179	D8S1179	D8S1179
FGA	FGA	FGA	FGA
		D1S1656	
		D2S441	
		D10S1248	
		D12S391	
		D22S1045	
			TPOX
			CSF1PO
			D5S818
			D7S820
			D13S317
			D19S433

Table 11.1 The numerator and denominator that should be used when calculating a paternity index are determined by the genotypes of the child (G_C), mother (G_M), and tested man (G_{TM}). The alleles are represented by A, B, C and D where $A \neq B \neq C \neq D$. Based on Lucy (2006) [15] p174 and Evett and Weir (1998) [16] p168.

G_C	G_M	G_{TM}	Numerator	Denominator	PI
$A\,A$	$A\,A$	$A\,A$	1	p_A	$\frac{1}{p_A}$
		$A\,B$	½	p_A	$\frac{1}{2p_A}$
		$B\,C$	0	p_A	0
	$A\,B$	$A\,A$	½	$\frac{p_A}{2}$	$\frac{1}{p_A}$
		$A\,B$	¼	$\frac{p_A}{2}$	$\frac{1}{2p_A}$
		$A\,C$	¼	$\frac{p_A}{2}$	$\frac{1}{2p_A}$
		$B\,C$	0	$\frac{p_A}{2}$	0
$A\,B$	$A\,A$	$B\,B$	1	P_B	$\frac{1}{p_B}$
		$A\,B$	½	P_B	$\frac{1}{2p_B}$
		$B\,C$	½	P_B	$\frac{1}{2p_B}$
		$C\,D$	0	P_B	0
	$A\,B$	$A\,A$	½	$\frac{P_A + P_B}{2}$	$1/p_A + p_B$
		$A\,B$	½	$\frac{P_A + P_B}{2}$	$\frac{1}{p_A + p_B}$
		$A\,C$	¼	$\frac{P_A + P_B}{2}$	$\frac{1}{2(p_A + p_B)}$
		$B\,C$	¼	$\frac{P_A + P_B}{2}$	$\frac{1}{2(p_A + p_B)}$
		$C\,D$	0	$\frac{P_A + P_B}{2}$	0
	$A\,C$	$B\,B$	½	$\frac{P_B}{2}$	$\frac{1}{p_B}$
		$A\,B$	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$
		$B\,C$	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$
		$B\,D$	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$
		$C\,D$	0	$\frac{P_B}{2}$	0

Table 11.2 The result of a paternity test using the Powerplex® 16 STR Kit (Promega). The alleles that the child could have inherited from the mother are underlined and the paternal alleles are shown in bold. The $^{A,B,C \text{ and } D}$ symbols correspond to symbols in Table 11.2. The allele frequencies were taken from Marino et al. (2006) [17].

Locus	Child (G_C)	Mother (G_M)	Tested Man (G_{TM})	Num	Denom	PI	$P_{i/j}$	PI
D3S1358	**15**^{A} – **15**^{A}	14^B – 15^A	**15**^{A} – 19^C	¼	$\frac{P_A}{2}$	$\frac{1}{2p_A}$	0.3239	1.54
VWA	**17**^{B} – 18^A	16^C – 18^A	**17**^{B} – 18^A	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$	0.2715	1.84
D16S359	11^A – **12**^{B}	11^A – 13^C	**12**^{B} – 13^C	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$	0.2773	1.80
D8S1179	10^A – 13^B	10^A – 13^B	**10**^{A} – **10**^{A}	½	$\frac{P_A + P_B}{2}$	$\frac{1}{p_A + p_B}$	0.0630 0.3033	2.73
D21S11	30^A – **32.2**^{B}	30^A – 31^C	27^D – **32.2**^{B}	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$	0.1245	4.02
D18S51	13^A – **14**^{B}	13^A – 14^B	12^C – **14**^{B}	¼	$\frac{P_A + P_B}{2}$	$\frac{1}{2(p_A + p_B)}$	0.1326 0.2063	1.48
TH01	9^A – **9.3**^{B}	9^A – 9.3^B	6^C – **9.3**^{B}	¼	$\frac{P_A + P_B}{2}$	$\frac{1}{2(p_A + p_B)}$	0.1407 0.2624	1.24
FGA	18^A – **23**^{B}	18^A – 25^C	**23**^{B} – **23**^{B}	½	$\frac{P_B}{2}$	$\frac{1}{p_B}$	0.1440	6.94
D13S317	8^A – **13**^{B}	8^A – 11^C	11^C – **13**^{B}	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$	0.1444	3.46
CSF1PO	11^A – **11**^{A}	11^A – 13^B	**11**^{A} – 13^B	¼	$\frac{P_A}{2}$	$\frac{1}{2p_A}$	0.2916	1.71
D7S820	9^A – **9**^{A}	9^A – 10^B	**9**^{A} – 11^C	¼	$\frac{P_A}{2}$	$\frac{1}{2p_A}$	0.0998	5.01
TPOX	**8**^{B} – 10^A	10^A – 11^C	**8**^{B} – **8**^{B}	½	$\frac{P_B}{2}$	$\frac{1}{p_B}$	0.5243	1.91
D5S818	**11**^{A} – **12**^{B}	11^A – 12^B	**11**^{A} – **12**^{B}	½	$\frac{P_A + P_B}{2}$	$\frac{1}{p_A + p_B}$	0.3618 0.2992	1.51
Penta D	**13**^{B} – 15^A	12^C – 15^A	12^D – **13**^{B}	¼	$\frac{P_B}{2}$	$\frac{1}{2p_B}$	0.1726	2.90
Penta E	10^A – **18**^{B}	10^A – 10^A	16^C – **18**^{B}	½	P_B	$\frac{1}{2p_B}$	0.0304	16.4
						Combined PI		**2,920,823**

Table 11.3 Formulae used to calculate paternity indices in cases where no mother is available for testing. The alleles are represented by *A*, *B* and *C* where $A \neq B \neq C$ [21].

G_C	G_{TM}	PI
A A	*A A*	$\frac{1}{p_A}$
	A B	$\frac{1}{2p_A}$
A B	*A A*	$\frac{1}{2p_A}$
	B B	$\frac{1}{2p_B}$
	A B	$\frac{p_A + p_B}{4p_A\, p_B}$
	A C	$\frac{1}{4p_A}$

Table 11.4 Without the mother's genotype from the paternity case shown in Table 11.2 we can recalculated the paternity index for what is now a motherless case. The results, while still providing very strong support for paternity, are approximately 28-fold weaker than when the mother was available for testing.

Locus	Child (G_C)	Tested Man (G_{TM})	PI	$P_{A/B}$	PI
D3S1358	**15**^{A} – **15**^{A}	**15**^{A} – 19^B	$\frac{1}{2p_A}$	0.3239	1.54
VWA	**17**^{A} – **18**$^{\boldsymbol{B}}$	**17**^{A} – **18**$^{\boldsymbol{B}}$	$\frac{p_A + p_B}{4p_A\,p_B}$	0.2715 0.1816	2.30
D16S359	11^B – **12**^{A}	**12**^{A} – 13^C	$\frac{1}{4p_A}$	0.2773	0.90
D8S1179	**10**$^{\boldsymbol{A}}$ – 13^B	**10**^{A} – **10**^{A}	$\frac{1}{2p_A}$	0.0630	7.94
D21S11	30^B – **32.2**^{A}	27^C – **32.2**^{A}	$\frac{1}{4p_A}$	0.1245	2.01
D18S51	13^B – **14**^{A}	12^C – **14**^{A}	$\frac{1}{4p_A}$	0.2063	1.21
TH01	9^B – **9.3**^{A}	6^C – **9.3**^{A}	$\frac{1}{4p_A}$	0.2624	0.95
FGA	18^B – **23**^{A}	**23**^{A} – **23**^{A}	$\frac{1}{2p_A}$	0.1440	3.47
D13S317	8^B – **13**^{A}	11^C – **13**^{A}	$\frac{1}{4p_A}$	0.1444	1.73
CSF1PO	**11**$^{\boldsymbol{A}}$ – **11**^{A}	**11**^{A} – 13^B	$\frac{1}{2p_A}$	0.2916	1.71
D7S820	**9**$^{\boldsymbol{A}}$ – **9**$^{\boldsymbol{A}}$	**9**^{A} – 11^B	$\frac{1}{2p_A}$	0.0998	5.01
TPOX	**8**^{A} – 10^B	**8**$^{\boldsymbol{A}}$ – **8**$^{\boldsymbol{A}}$	$\frac{1}{2p_A}$	0.5243	1.91
D5S818	**11**^{A} – **12**^{B}	**11**^{A} – **12**^{B}	$\frac{p_A + p_B}{4p_A\,p_B}$	0.3618 0.2992	1.53
Penta D	**13**^{A} – 15^B	12^C – **13**^{A}	$\frac{1}{4p_A}$	0.1726	1.45
Penta E	10^B – **18**^{A}	16^C – **18**^{A}	$\frac{1}{4p_A}$	0.0304	8.22
			Combined PI		**104,759**

Table 11.5 The impact of prior probabilities on the probability of paternity is shown with two paternity indexes: one with a value of 1,000 and the other taken from the above example, with a value of 2,920,823.

	Paternity Index	
Prior Odds	1,000	2,920,823
0.0001	0.090917356	0.996588329
0.0010	0.500250125	0.99965809
0.0100	0.909918107	0.999966107
0.1000	0.991080278	0.999996919
0.5000	0.999000999	0.999999658
0.7500	0.999666778	0.999999886
0.9000	0.999888901	0.999999962

Table 12.1 A comparison of the properties of SNPs and STRs.

	STR	**SNP**
Frequency of occurrence	Once every 15 Kb	Once every 500 bp
Typical rate of Mutation	10^{-3}	2-3 x 10^{-8}
Typical number of alleles	Between 5 and 20	2
Potential to multiplex	Currently a maximum of 16 STR loci examined at one time	Difficult to amplify more than 50 SNPs in one reaction
Number of loci required to have a P_M of 1 in 1 billion	10	~ 60
Method of Detection	Capillary Gel Electrophoresis (CGE)	CGE, Microarrays, Mass Spectroscopy
Automation Potential	Medium	High
Artefacts	Amplification of STRs can produce artefacts such as stutter and split peaks.	No stutter artefacts associated with the amplification of the SNPs
Amount of DNA required	~0.5 ng to 1 ng	~100 pg
Size of amplicon	Amplicon sizes typically between 100 bp and 400 bp	Amplicon sizes can be less than 100 bp
Mixtures	Interpretation of mixtures of STR loci is possible	Mixtures of SNP loci can be highly problematic to interpret
Predicting geographical origin	Limited ethnic identification from STR loci	Some SNPs can be associated with particular ethnic groups
Phenotypic information	No possibility to infer phenotype	Possible to predict some hair colour, eye colour, skin colour.

Table 13.1 An example where a mtDNA profile has been generated from the HV-I of five bones that were found in close proximity. The mtDNA profiles of three women who were maternal relatives of three missing individuals are also shown. Maternal reference 1 clearly matches the bone, while maternal reference 2 and 3 can be excluded as potential maternal relatives as they have different mtDNA types. In this particular case the mtDNA profiling helped to establish the identification of the human remains, and that the bones all came from the same person [39].

Sample	HV-I Sequence			
Right Femur	16,189C	16,223T	16,271C	16,278T
Left Femur	16,189C	16,223T	16,271C	16,278T
Right Pelvis	16,189C	16,223T	16,271C	16,278T
Left Ulna	16,189C	16,223T	16,271C	16,278T
Left Tibia	16,189C	16,223T	16,271C	16,278T
Maternal Reference 1	16,189C	16,223T	16,271C	16,278T
Maternal Reference 2	Same as Cambridge Reference Sequence			
Maternal Reference 3	16,278T	16,293G	16,311C	

Table 13.2 The Y chromosome STR loci that are commonly used in forensic analysis.

Minimal Haplotype	Extended Haplotype	PowerPlex® Y	AmpFℓSTR® Yfiler®
DYS19	DYS19	DYS19	DYS19
DYS385 a/b	DYS385 a/b	DYS385 a/b	DYS385 a/b
DYS389 I	DYS389 I	DYS389 I	DYS389 I
DYS389 II	DYS389 II	DYS389 II	DYS389 II
DYS390	DYS390	DYS390	DYS390
DYS391	DYS391	DYS391	DYS391
DYS392	DYS392	DYS392	DYS392
DYS393	DYS393	DYS393	DYS393
	DYS438	DYS437	DYS437
	DYS439	DYS438	DYS438
		DYS439	DYS439
			DYS448
			DYS456
			DYS458
			DYS635
			GATA H4