

Relatedness and Kinship Statistics

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Relatedness

“Relatedness is a consequence of sharing identical alleles. Alleles that have descended from a single ancestral allele are said to be Identical by Descent (IBD).”*
Alleles that are IBD will have the same base sequence.

However, alleles with the same base sequence are not necessarily IBD, they can be Identical By State and not originate from the same ancestral allele. This may be true in an individual that is homozygous at a given locus.

* B.S. Weir, 1996. Genetic Data Analysis II. (p 204).

The genetic profile of a suspect matches the genetic profile of an evidentiary sample, therefore, the suspect cannot be excluded as a potential contributor of the biological evidentiary sample.

Popstats can then be used to calculate the frequency with which a person taken at random from a population of potential perpetrators has the profile in question?



Single Sample Target Pro...

Reference:

Evidence

Locus	Allele 1	Allele 2
D3S1358	17	17
VWA	14	16
FGA	20	23
D8S1179	13	15
D21S11	29	31.2
D18S51	12	18
D5S818	11	12
D13S317	8	12
D7S820	8	11
CSF1PO	12	13
TPOX	8	9
TH01	7	9.3
D16S539	12	13

Reference = Evidence

Single Sample Target Pro...

Reference:

Suspect

Locus	Allele 1	Allele 2
D3S1358	17	17
VWA	14	16
FGA	20	23
D8S1179	13	15
D21S11	29	31.2
D18S51	12	18
D5S818	11	12
D13S317	8	12
D7S820	8	11
CSF1PO	12	13
TPOX	8	9
TH01	7	9.3
D16S539	12	13

Reference = Suspect



Summary of Probability Statistics

Locus	CAU	BLK	SEH	SWH
D3S1358	4.6529E-02	4.1600E-02	2.7701E-02	1.7185E-02
VWA	4.1106E-02	3.5938E-02	3.6987E-02	4.4303E-02
FGA	4.6005E-02	1.8050E-02	3.5152E-02	2.0049E-02
D8S1179	7.4442E-02	9.5057E-02	8.5725E-02	7.5293E-02
D21S11	3.6039E-02	2.8637E-02	4.0358E-02	3.5239E-02
D18S51	2.3427E-02	1.5228E-02	1.4971E-02	1.0950E-02
D5S818	2.9041E-01	1.8569E-01	2.4943E-01	2.4480E-01
D13S317	6.1431E-02	3.5080E-02	5.2533E-02	2.8834E-02
D7S820	6.5690E-02	7.7793E-02	6.4360E-02	5.6800E-02
CSF1PO	4.6424E-02	3.2880E-02	4.9027E-02	5.0685E-02
TPOX	1.3412E-01	1.3395E-01	8.4350E-02	3.7185E-02
TH01	1.0530E-01	9.2329E-02	1.1869E-01	1.6298E-01
D16S539	1.1082E-01	6.1615E-02	8.2615E-02	5.9165E-02
Total	4.709E-16	1.420E-17	6.379E-17	3.231E-18



Inverse Summary of Probability Statistics



Locus	CAU	BLK	SEH	SWH
D3S1358	21	24	36	58
VWA	24	28	27	23
FGA	22	55	28	50
D8S1179	13	11	12	13
D21S11	28	35	25	28
D18S51	43	66	67	91
D5S818	3	5	4	4
D13S317	16	29	19	35
D7S820	15	13	16	18
CSF1PO	22	30	20	20
TPOX	7	7	12	27
TH01	9	11	8	6
D16S539	9	16	12	17

	CAU	BLK	SEH	SWH
Total	2,124,000,000,000,000	70,420,000,000,000,000	15,680,000,000,000,000	309,500,000,000,000,000

Relatedness Statistics

Although, unrelated individuals have a very low probability of sharing the same genetic profile with a suspect, the probability increases for relatives.

Relatedness Statistics

What is the probability of a person with a given degree of relatedness having the identical genotype as the known genotype of our suspect?

Relatedness Statistics

Calculations for related individuals
are **conditional probabilities**.

They answer the question for a
specific genotype

In general, the closest relationship that has
an impact on calculations is
full-sibs

Coefficient of Kinship (F)

definition:

- randomly select an allele from each of two individuals
- the probability that the two alleles are identical by descent (IBD) is called F , the coefficient of kinship
- the expected proportion of alleles that are IBD between two people is $2F$



Single Sample Target Pro...



LDAS

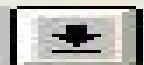


LDIS



Reference:

Suspect



	Locus	Allele 1	Allele 2
X	D3S1358	17	17
X	VWA	14	16
X	FGA	20	23
X	D8S1179	13	15
X	D21S11	29	31.2
X	D18S51	12	18
X	D5S818	11	12
X	D13S317	8	12
X	D7S820	8	11
X	CSF1PO	12	13
X	TPOX	8	9
X	TH01	7	9.3
X	D16S539	12	13

Reference = Suspect



Summary of Probability Statistics

Locus	CAU	BLK	SEH	SWH
D3S1358	4.6529E-02	4.1600E-02	2.7701E-02	1.7185E-02
VWA	4.1106E-02	3.5938E-02	3.6987E-02	4.4303E-02
FGA	4.6005E-02	1.8050E-02	3.5152E-02	2.0049E-02
D8S1179	7.4442E-02	9.5057E-02	8.5725E-02	7.5293E-02
D21S11	3.6039E-02	2.8637E-02	4.0358E-02	3.5239E-02
D18S51	2.3427E-02	1.5228E-02	1.4971E-02	1.0950E-02
D5S818	2.9041E-01	1.8569E-01	2.4943E-01	2.4480E-01
D13S317	6.1431E-02	3.5080E-02	5.2533E-02	2.8834E-02
D7S820	6.5690E-02	7.7793E-02	6.4360E-02	5.6800E-02
CSF1PO	4.6424E-02	3.2880E-02	4.9027E-02	5.0685E-02
TPOX	1.3412E-01	1.3395E-01	8.4350E-02	3.7185E-02
TH01	1.0530E-01	9.2329E-02	1.1869E-01	1.6298E-01
D16S539	1.1082E-01	6.1615E-02	8.2615E-02	5.9165E-02
Total	4.709E-16	1.420E-17	6.379E-17	3.231E-18

Popstats 5.3 - [Relatedness Statistics]

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CAU

BLK

SEH

SWH

Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	4.6529E-02	2.1180E-01	3.6711E-01	1.2833E-01	8.6594E-02
VWA	4.1106E-02	1.5175E-01	3.3615E-01	9.6428E-02	6.8767E-02
FGA	4.6005E-02	1.5180E-01	3.3740E-01	9.8902E-02	7.2453E-02
D8S1179	7.4442E-02	2.2450E-01	3.8086E-01	1.4947E-01	1.1196E-01
D21S11	3.6039E-02	1.4030E-01	3.2916E-01	8.8169E-02	6.2104E-02
D18S51	2.3427E-02	1.0970E-01	3.1071E-01	6.6564E-02	4.4996E-02
D5S818	2.9041E-01	3.8210E-01	5.1365E-01	3.3626E-01	3.1333E-01
D13S317	6.1431E-02	2.0410E-01	3.6741E-01	1.3277E-01	9.7098E-02
D7S820	6.5690E-02	1.8230E-01	3.5757E-01	1.2400E-01	9.4843E-02
CSF1PO	4.6424E-02	1.9825E-01	3.6073E-01	1.2234E-01	8.4381E-02
TPOX	1.3412E-01	3.3375E-01	4.5040E-01	2.3393E-01	1.8402E-01
TH01	1.0530E-01	2.3890E-01	3.9578E-01	1.7210E-01	1.3870E-01
D16S539	1.1082E-01	2.5125E-01	4.0333E-01	1.8103E-01	1.4593E-01
Total	4.709E-16	9.519E-10	2.838E-06	5.299E-12	1.224E-13

Popstats 5.3 - [Relatedness Statistics]

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CAU	BLK	SEH	SWH		
Locus	Unrelated	Parent/Offspring	Full Sibling	Half Sibling/Uncle/Aunt/Nephew/Niece	First Cousin
D3S1358	21	5	3	8	12
VWA	24	7	3	10	15
FGA	22	7	3	10	14
D8S1179	13	4	3	7	9
D21S11	28	7	3	11	16
D18S51	43	9	3	15	22
D5S818	3	3	2	3	3
D13S317	16	5	3	8	10
D7S820	15	5	3	8	11
CSF1PO	22	5	3	8	12
TPOX	7	3	2	4	5
TH01	9	4	3	6	7
D16S539	9	4	2	6	7
Total	2,124,000,000,000,000	1,051,000,000	352,400	188,700,000,000	8,170,000,000,000

Popstats 5.3 - [Relatedness Statistics]

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CAU		BLK	SEH	SWh	
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	4.1600E-02	2.0000E-01	3.6000E-01	1.2000E-01	8.0000E-02
VWA	3.5938E-02	1.6805E-01	3.4301E-01	1.0199E-01	6.8966E-02
FGA	1.8050E-02	9.8600E-02	3.0381E-01	5.8325E-02	3.8188E-02
D8S1179	9.5057E-02	2.1805E-01	3.8279E-01	1.5655E-01	1.2581E-01
D21S11	2.8637E-02	1.3265E-01	3.2348E-01	8.0643E-02	5.4640E-02
D18S51	1.5228E-02	9.4450E-02	3.0103E-01	5.4839E-02	3.5033E-02
D5S818	1.8569E-01	3.0835E-01	4.5060E-01	2.4702E-01	2.1636E-01
D13S317	3.5080E-02	2.5975E-01	3.8865E-01	1.4742E-01	9.1248E-02
D7S820	7.7793E-02	1.9880E-01	3.6885E-01	1.3830E-01	1.0804E-01
CSF1PO	3.2880E-02	1.7740E-01	3.4692E-01	1.0514E-01	6.9010E-02
TPOX	1.3395E-01	2.7510E-01	4.2104E-01	2.0453E-01	1.6924E-01
TH01	9.2329E-02	2.7265E-01	4.0941E-01	1.8249E-01	1.3741E-01
D16S539	6.1615E-02	1.7585E-01	3.5333E-01	1.1873E-01	9.0174E-02
Total	1.420E-17	3.373E-10	1.909E-06	1.160E-12	1.566E-14

Popstats 5.3 - [Relatedness Statistics]

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CAU	BLK	SEH	SwH		
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	24	5	3	8	13
VWA	28	6	3	10	15
FGA	55	10	3	17	26
D8S1179	11	5	3	6	8
D21S11	35	8	3	12	18
D18S51	66	11	3	18	29
D5S818	5	3	2	4	5
D13S317	29	4	3	7	11
D7S820	13	5	3	7	9
CSF1PO	30	6	3	10	14
TPOX	7	4	2	5	6
TH01	11	4	2	5	7
D16S539	16	6	3	8	11
Total	70,420,000,000,000,000	2,965,000,000	523,800	862,100,000,000	63,860,000,000,000

Popstats 5.3 - [Relatedness Statistics]

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	CAU	BLK	SEH	SWH	
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	2.7701E-02	1.6230E-01	3.3774E-01	9.4321E-02	6.0331E-02
VWA	3.6987E-02	1.6880E-01	3.4365E-01	1.0289E-01	6.9940E-02
FGA	3.5152E-02	1.3350E-01	3.2554E-01	8.4326E-02	5.9739E-02
D8S1179	8.5725E-02	2.3820E-01	3.9053E-01	1.6196E-01	1.2384E-01
D21S11	4.0358E-02	1.6230E-01	3.4124E-01	1.0133E-01	7.0844E-02
D18S51	1.4971E-02	9.5550E-02	3.0152E-01	5.5260E-02	3.5116E-02
D5S818	2.4943E-01	3.5525E-01	4.8998E-01	3.0234E-01	2.7589E-01
D13S317	5.2533E-02	1.7190E-01	3.4908E-01	1.1222E-01	8.2374E-02
D7S820	6.4360E-02	1.8440E-01	3.5829E-01	1.2438E-01	9.4370E-02
CSF1PO	4.9027E-02	2.1255E-01	3.6853E-01	1.3079E-01	8.9908E-02
TPOX	8.4350E-02	2.9480E-01	4.1849E-01	1.8957E-01	1.3696E-01
TH01	1.1869E-01	2.4375E-01	4.0155E-01	1.8122E-01	1.4995E-01
D16S539	8.2615E-02	2.0835E-01	3.7483E-01	1.4548E-01	1.1405E-01
Total	6.379E-17	4.841E-10	2.160E-06	2.047E-12	3.507E-14

Popstats 5.3 - [Relatedness Statistics]

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CAU		BLK		SEH		SwH	
Locus	Unrelated	Parent/Offspring	Full Sibling	Half Sibling/Uncle/Aunt/Nephew/Niece	First Cousin		
D3S1358	36	6	3	11	17		
VWA	27	6	3	10	14		
FGA	28	7	3	12	17		
D8S1179	12	4	3	6	8		
D21S11	25	6	3	10	14		
D18S51	67	10	3	18	28		
D5S818	4	3	2	3	4		
D13S317	19	6	3	9	12		
D7S820	16	5	3	8	11		
CSF1PO	20	5	3	8	11		
TPOX	12	3	2	5	7		
TH01	8	4	2	6	7		
D16S539	12	5	3	7	9		
Total	15,680,000,000,000,000	2,066,000,000	463,000	488,500,000,000	28,510,000,000,000		

Popstats 5.3 - [Relatedness Statistics]

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	CAU	BLK	SEH	SWH	
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	1.7185E-02	1.2680E-01	3.1742E-01	7.1439E-02	4.3759E-02
VWA	4.4303E-02	2.1060E-01	3.6638E-01	1.2745E-01	8.5877E-02
FGA	2.0049E-02	1.0590E-01	3.0796E-01	6.2975E-02	4.1512E-02
D8S1179	7.5293E-02	2.2045E-01	3.7905E-01	1.4787E-01	1.1158E-01
D21S11	3.5239E-02	1.4530E-01	3.3146E-01	9.0269E-02	6.2754E-02
D18S51	1.0950E-02	7.8800E-02	2.9214E-01	4.4875E-02	2.7913E-02
D5S818	2.4480E-01	3.5590E-01	4.8915E-01	3.0035E-01	2.7258E-01
D13S317	2.8834E-02	1.4165E-01	3.2803E-01	8.5242E-02	5.7038E-02
D7S820	5.6800E-02	1.9380E-01	3.6110E-01	1.2530E-01	9.1050E-02
CSF1PO	5.0685E-02	2.2845E-01	3.7690E-01	1.3957E-01	9.5126E-02
TPOX	3.7185E-02	2.9425E-01	4.0642E-01	1.6572E-01	1.0145E-01
TH01	1.6298E-01	2.8945E-01	4.3547E-01	2.2622E-01	1.9460E-01
D16S539	5.9165E-02	1.9475E-01	3.6217E-01	1.2696E-01	9.3062E-02
Total	3.231E-18	2.643E-10	1.840E-06	7.319E-13	7.543E-15

Popstats 5.3 - [Relatedness Statistics]

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CAU		BLK		SEH		SWH	
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin		
D3S1358	58	8	3	14	23		
VWA	23	5	3	8	12		
FGA	50	9	3	16	24		
D8S1179	13	5	3	7	9		
D21S11	28	7	3	11	16		
D18S51	91	13	3	22	36		
D5S818	4	3	2	3	4		
D13S317	35	7	3	12	18		
D7S820	18	5	3	8	11		
CSF1PO	20	4	3	7	11		
TPOX	27	3	2	6	10		
TH01	6	3	2	4	5		
D16S539	17	5	3	8	11		
Total	309,500,000,000,000	3,784,000,000	543,500	1,366,000,000,000	132,600,000,000,000		

Relatedness Statistics

Brothers

Prob (Full-Sibs have same given genotype)

$A_i A_i$ (homozygous locus): $(1 + p_i)^2 / 4$

$A_i A_j$ (heterozygous locus): $(1 + p_i + p_j + 2 p_i p_j) / 4$

	ALLELE	ALLELE	ALLELE	ALLELE	FORMULA	FULL SIBLING
LOCUS	i	j	FREQ	FREQ	FULL SIBLING	
D3S1358	17		0.2118		$(1 + p_i)^2 / 4$	0.36711
VWA	14	16	0.1020	0.2015	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.33615
FGA	20	23	0.1454	0.1582	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.33740
D8S1179	13	15	0.3393	0.1097	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.38086
D21S11	29	31.2	0.1811	0.0995	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.32916
D18S51	12	18	0.1276	0.0918	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.31071
D5S818	11	12	0.4103	0.3538	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.51361
D13S317	8	12	0.0995	0.3087	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.36741
D7S820	8	11	0.1626	0.2020	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.35757
CSF1PO	12	13	0.3251	0.0714	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.36073
TPOX	8	9	0.5443	0.1232	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.45040
TH01	7	9.3	0.1724	0.3054	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.39578
D16S539	12	13	0.3391	0.1634	$(1 + p_i + p_j + 2p_i p_j) / 4$	0.40333
frequency (f)						2.838E-06
1 / frequency (1/f)						352,334

Probability that a Brother would have the same profile is 1 in 352,334

Relatedness Statistics

Brothers

Prob (Full-Sibs have same given genotype)

Probability is approximately 1/4 per locus, Therefore, for “L” loci,

The Probability that two full-sibs would have the same given genotype is approximately

$$(1/4)^L$$

Relatedness Statistics Brothers (Full-Sibs)

<u>L</u>	<u>$(1/4)^L$</u>
4	1 / 256
5	1 / 1024
:	:
9	1 / 262,144
:	:
13	1 / 67,108,864

Relatedness Statistics

Father and Son

Prob (Parent-Child have same given genotype)

$$A_i A_i \text{ (homozygote)}: p_i$$

$$A_i A_j \text{ (heterozygote)}: (p_i + p_j) / 2$$

	ALLELE	ALLELE	ALLEL E FREQ	ALLEL E FREQ	FORMULA PARENT CHILD	PARENT & CHILD
LOCUS	i	j	p _i	p _j		
D3S1358	17		0.2118		p _i	0.21180
VWA	14	16	0.1020	0.2015	(p _i + p _j) / 2	0.15175
FGA	20	23	0.1454	0.1582	(p _i + p _j) / 2	0.15180
D8S1179	13	15	0.3393	0.1097	(p _i + p _j) / 2	0.22450
D21S11	29	31.2	0.1811	0.0995	(p _i + p _j) / 2	0.14030
D18S51	12	18	0.1276	0.0918	(p _i + p _j) / 2	0.10970
D5S818	11	12	0.4103	0.3538	(p _i + p _j) / 2	0.38205
D13S317	8	12	0.0995	0.3087	(p _i + p _j) / 2	0.20410
D7S820	8	11	0.1626	0.2020	(p _i + p _j) / 2	0.18230
CSF1PO	12	13	0.3251	0.0714	(p _i + p _j) / 2	0.19825
TPOX	8	9	0.5443	0.1232	(p _i + p _j) / 2	0.33375
TH01	7	9.3	0.1724	0.3054	(p _i + p _j) / 2	0.23890
D16S539	12	13	0.3391	0.1634	(p _i + p _j) / 2	0.25125
frequency (f)						9.517E-10
1 / frequency (1/f)						1,050,718,457

Probability that a Father or Son would have the same profile is 1 in 1,050,718,457

Relatedness Statistics

Half-Brothers

Prob (Half-Sibs have same given genotype)

$$A_i A_i \text{ (homozygous locus): } p_i (1 + p_i) / 2$$

$$A_i A_j \text{ (heterozygous locus): } (p_i + p_j + 4 p_i p_j) / 4$$

	ALLELE	ALLELE	ALLELE FREQ	ALLELE FREQ	FORMULA HALF SIBLING	HALF SIBLING
LOCUS	i	j	p_i	p_j		
D3S1358	17		0.2118		$p_i (1 + p_i) / 2$	0.12833
VWA	14	16	0.1020	0.2015	$(p_i + p_j + 4p_i p_j) / 4$	0.09643
FGA	20	23	0.1454	0.1582	$(p_i + p_j + 4p_i p_j) / 4$	0.09890
D8S1179	13	15	0.3393	0.1097	$(p_i + p_j + 4p_i p_j) / 4$	0.14947
D21S11	29	31.2	0.1811	0.0995	$(p_i + p_j + 4p_i p_j) / 4$	0.08817
D18S51	12	18	0.1276	0.0918	$(p_i + p_j + 4p_i p_j) / 4$	0.06656
D5S818	11	12	0.4103	0.3538	$(p_i + p_j + 4p_i p_j) / 4$	0.33619
D13S317	8	12	0.0995	0.3087	$(p_i + p_j + 4p_i p_j) / 4$	0.13277
D7S820	8	11	0.1626	0.2020	$(p_i + p_j + 4p_i p_j) / 4$	0.12400
CSF1PO	12	13	0.3251	0.0714	$(p_i + p_j + 4p_i p_j) / 4$	0.12234
TPOX	8	9	0.5443	0.1232	$(p_i + p_j + 4p_i p_j) / 4$	0.23393
TH01	7	9.3	0.1724	0.3054	$(p_i + p_j + 4p_i p_j) / 4$	0.17210
D16S539	12	13	0.3391	0.1634	$(p_i + p_j + 4p_i p_j) / 4$	0.18103
frequency (f)						5.298E-12
1 / frequency (1/f)						188,748,573,845

Probability that a Half Brother would have the same profile is 1 in 188,748,573,845

Relatedness Statistics

Uncle-Nephew

Prob (Uncle and Nephew have same given genotype)

$$A_i A_i \text{ (homozygous locus): } p_i (1 + p_i) / 2$$

$$A_i A_j \text{ (heterozygous locus): } (p_i + p_j + 4 p_i p_j) / 4$$

			ALLEL E	ALLEL E	FORMULA	UNCLE & NEPHEW
LOCUS	i	j	FREQ p_i	FREQ p_j	UNCLE NEPHEW	
D3S1358	17		0.2118		$p_i (1 + p_i) / 2$	0.12833
VWA	14	16	0.1020	0.2015	$(p_i + p_j + 4p_i p_j) / 4$	0.09643
FGA	20	23	0.1454	0.1582	$(p_i + p_j + 4p_i p_j) / 4$	0.09890
D8S1179	13	15	0.3393	0.1097	$(p_i + p_j + 4p_i p_j) / 4$	0.14947
D21S11	29	31.2	0.1811	0.0995	$(p_i + p_j + 4p_i p_j) / 4$	0.08817
D18S51	12	18	0.1276	0.0918	$(p_i + p_j + 4p_i p_j) / 4$	0.06656
D5S818	11	12	0.4103	0.3538	$(p_i + p_j + 4p_i p_j) / 4$	0.33619
D13S317	8	12	0.0995	0.3087	$(p_i + p_j + 4p_i p_j) / 4$	0.13277
D7S820	8	11	0.1626	0.2020	$(p_i + p_j + 4p_i p_j) / 4$	0.12400
CSF1PO	12	13	0.3251	0.0714	$(p_i + p_j + 4p_i p_j) / 4$	0.12234
TPOX	8	9	0.5443	0.1232	$(p_i + p_j + 4p_i p_j) / 4$	0.23393
TH01	7	9.3	0.1724	0.3054	$(p_i + p_j + 4p_i p_j) / 4$	0.17210
D16S539	12	13	0.3391	0.1634	$(p_i + p_j + 4p_i p_j) / 4$	0.18103
frequency (f)						5.298E-12
1 / frequency (1/f)						188,748,573,845

Probability that a Uncle or a Nephew would have the same profile is 1 in 188,748,573,845

Relatedness Statistics

First Cousins

Prob (First Cousins have same given genotype)

$$A_i A_i \text{ (homozygous locus): } p_i (1 + 3p_i) / 4$$

$$A_i A_j \text{ (heterozygous locus): } (p_i + p_j + 12 p_i p_j) / 8$$

	ALLELE	ALLELE	ALLEL E FREQ	ALLEL E FREQ	FORMULA	FIRST COUSIN
LOCUS	i	j	p _i	p _j		
D3S1358	17		0.2118		$p_i (1 + 3p_j) / 4$	0.08659
VWA	14	16	0.1020	0.2015	$(p_i + p_j + 12p_ip_j) / 8$	0.06877
FGA	20	23	0.1454	0.1582	$(p_i + p_j + 12p_ip_j) / 8$	0.07245
D8S1179	13	15	0.3393	0.1097	$(p_i + p_j + 12p_ip_j) / 8$	0.11196
D21S11	29	31.2	0.1811	0.0995	$(p_i + p_j + 12p_ip_j) / 8$	0.06210
D18S51	12	18	0.1276	0.0918	$(p_i + p_j + 12p_ip_j) / 8$	0.04500
D5S818	11	12	0.4103	0.3538	$(p_i + p_j + 12p_ip_j) / 8$	0.31326
D13S317	8	12	0.0995	0.3087	$(p_i + p_j + 12p_ip_j) / 8$	0.09710
D7S820	8	11	0.1626	0.2020	$(p_i + p_j + 12p_ip_j) / 8$	0.09484
CSF1PO	12	13	0.3251	0.0714	$(p_i + p_j + 12p_ip_j) / 8$	0.08438
TPOX	8	9	0.5443	0.1232	$(p_i + p_j + 12p_ip_j) / 8$	0.18402
TH01	7	9.3	0.1724	0.3054	$(p_i + p_j + 12p_ip_j) / 8$	0.13870
D16S539	12	13	0.3391	0.1634	$(p_i + p_j + 12p_ip_j) / 8$	0.14593
frequency (f)						1.224E-13
1 / frequency (1/f)						8,171,074,738,912

Probability that a First Cousin would have the same profile is 1 in 8,171,074,738,912

Sexual Assault Case Processed by the Arizona Department of Public Safety, Southern Regional Crime Laboratory in Tucson for the Farmington, New Mexico Police Department

Sexual Assault Case in which the suspects DNA profile matched the DNA profile from the male fraction of the vaginal swab collected from victim

Popstats 5.3

File Edit Profile Case Type Configuration Window Help



Single Sample Target Pro...

Summary of Probability Statistics



LDAS



LDIS



Reference:

Suspect Farmington, NM Rape Case

Locus	Allele 1	Allele 2
D3S1358	15	16
VwA	17	19
FGA	21	27
D8S1179	13	14
D21S11	30	31
D18S51	16	18
D5S818	11	
D13S317	11	12
D7S820	10	12
CSF1PO	9	10
TPOX	8	11
TH01	7	
D16S539	9	13

Reference = Suspect Farmington, NM Rape Case

Locus	Apache	Minnesota	Navajo
D3S1358	2.5916E-01	2.7926E-01	2.2338E-01
VwA	3.9386E-02	5.1540E-02	5.0966E-02
FGA	8.6841E-03	1.1859E-02	2.1050E-02
D8S1179	2.0713E-01	2.3386E-01	1.7463E-01
D21S11	7.4602E-02	5.7296E-02	7.3979E-02
D18S51	3.4844E-02	1.3689E-02	1.8441E-02
D5S818	3.5049E-01	1.9820E-01	3.7487E-01
D13S317	1.1604E-01	1.2297E-01	9.9909E-02
D7S820	1.1685E-01	1.1826E-01	9.4054E-02
CSF1PO	2.9473E-02	6.5583E-02	4.0468E-02
TPOX	2.0366E-01	3.0159E-01	2.6666E-01
TH01	2.2537E-01	2.4171E-01	4.1000E-01
D16S539	3.7885E-02		5.9583E-02
Total	1.162E-14	4.314E-13	5.302E-14



Single Sample Target Pro...



LDAS



LDIS



Reference:

Suspect Farmington, NM Rape Case

Locus	Allele 1	Allele 2
D3S1358	15	16
VWA	17	19
FGA	21	27
D8S1179	13	14
D21S11	30	31
D18S51	16	18
D5S818	11	
D13S317	11	12
D7S820	10	12
CSF1PO	9	10
TPOX	8	11
TH01	7	
D16S539	9	13

Reference = Suspect Farmington, NM Rape Case

Inverse Summary of Probability Statistics

Locus	Apache	Minnesota	Navajo
D3S1358	4	4	4
VWA	25	19	20
FGA	115	84	48
D8S1179	5	4	6
D21S11	13	17	14
D18S51	29	73	54
D5S818	3	5	3
D13S317	9	8	10
D7S820	9	8	11
CSF1PO	34	15	25
TPOX	5	3	4
TH01	4	4	2
D16S539	26		17

	Apache	Minnesota	Navajo
Total	86,060,000,000,000	2,318,000,000,000	18,860,000,000,000

The approximate frequency of this profile in
the Native American (Navajo) Population

is

1 in 18.86 trillion (1.886×10^{13})

The probability of finding an unrelated, random individual from the Native American (Navajo) population that matches the male fraction of the vaginal swab is

1 in 18.86 trillion (1.886×10^{13})

THE BROTHER DID IT?

Defense argued that the suspects brother committed the crime.

Defense called Dr. Dan Krane from Wright State University in Ohio to testify to probability of a brother having the same profile.



Relatedness Statistics



f

1/1

	Apache	Minnesota	Navajo		
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin
D3S1358	2.2338E-01	4.3820E-01	5.1987E-01	3.2064E-01	2.6186E-01
VWA	5.0966E-02	1.8410E-01	3.5066E-01	1.0928E-01	7.1864E-02
FGA	2.1050E-02	8.6550E-02	2.9623E-01	4.9178E-02	3.0492E-02
D8S1179	1.7463E-01	2.9595E-01	4.3900E-01	2.3003E-01	1.9708E-01
D21S11	7.3979E-02	2.7840E-01	4.0157E-01	1.6394E-01	1.0672E-01
D18S51	1.8441E-02	1.0135E-01	3.0241E-01	5.4149E-02	3.0548E-02
D5S818	3.7487E-01	5.8240E-01	6.2600E-01	4.6079E-01	3.9999E-01
D13S317	9.9909E-02	2.1015E-01	3.7708E-01	1.4909E-01	1.1855E-01
D7S820	9.4054E-02	2.1295E-01	3.7670E-01	1.4692E-01	1.1390E-01
CSF1PO	4.0468E-02	1.5525E-01	3.3408E-01	9.0544E-02	5.8192E-02
TPOX	2.6666E-01	3.6265E-01	4.9699E-01	3.1265E-01	2.8765E-01
TH01	4.1000E-01	6.1260E-01	6.5012E-01	4.9394E-01	4.3461E-01
D16S539	5.9583E-02	1.5540E-01	3.3970E-01	1.0170E-01	7.4850E-02
Total	5.302E-14	8.145E-09	9.387E-06	5.051E-11	1.084E-12



Relatedness Statistics



	Apache	Minnesota	Navajo			
Locus	Unrelated	Parent/ Offspring	Full Sibling	Half Sibling/ Uncle/Aunt/ Nephew/Niece	First Cousin	
D3S1358	4	2	2	3	4	
VWA	20	5	3	9	14	
FGA	48	12	3	20	33	
D8S1179	6	3	2	4	5	
D21S11	14	4	2	6	9	
D18S51	54	10	3	18	33	
D5S818	3	2	2	2	3	
D13S317	10	5	3	7	8	
D7S820	11	5	3	7	9	
CSF1PO	25	6	3	11	17	
TP0X	4	3	2	3	3	
TH01	2	2	2	2	2	
D16S539	17	6	3	10	13	
Total	18,860,000,000,000	122,800,000	106,500	19,800,000,000	922,500,000,000	

THE BROTHER DID IT?

The probability of finding a brother of the suspect that matches the male fraction of the vaginal swab is

1 in 106,500

If the suspect had 106,500 brothers,
1 would be expected to have the same
profile as the suspect

THE BROTHER DID IT?

However, it is **1.77×10^8** times more likely to see this profile in an untested brother as opposed to an untested, randomly chosen individual from the Native American (Navajo) population

This is an answer to a different question other than the match probability for the evidentiary sample

THE BROTHER DID IT?

The problem was that the suspect's brother was deceased and not available for testing

Even though the chance of the untested brother having the same profile was very low;

The trial ended with a hung jury

Relatedness Statistics

We can calculate a Likelihood Ratio (for any given profile) in which the probability that the suspect is the contributor of the evidence is compared with the probability that a specified relative is the contributor of the evidence

In the following example the profile at a given locus is either homozygous or heterozygous and the allele frequency is 0.1

Formulas: B.S. Weir, 1996. Genetic Data Analysis II. Table 5.7 (p 221).

Relationship	Homozygous Locus	Heterozygous Locus
Unrelated	100.00	50.00
Full-Sibs	3.31	3.28
Parent-Child	10.00	10.00
Half-Sibs	18.18	16.67
Uncle-Nephew	18.18	16.67
First Cousins	30.76	25.00

Kinship Determination From Genotype Data On Two Individuals

Currently Popstats does not have the capability of calculating the most likely kinship relationship between two individuals whose genotypes are known.

Kinship Determination From Genotype Data On Two Individuals

There are a number of programs that are available to provide statistical support for the kinship relationship between two individuals with observed genotypes.

Currently the FBI has released a contract to develop the appropriate software which would be included in future versions of Popstats.

Kinship Determination From Genotype Data On Two Individuals

The programs that are available to provide statistical support for the kinship relationship between two individuals with observed genotypes essentially make use of the same algorithm.

Let $G_x(l)$ and $G_y(l)$ be the genotypes of two individuals X and Y, the algorithm finds the best support for the kinship relationship between individuals X and Y based upon genotypes $G_x(l)$ and $G_y(l)$ for L loci ($l = 1, 2, \dots, L$)

Kinship Determination From Genotype Data On Two Individuals

The algorithm requires two sets of information.

First, the kinship of two individuals dictate with what probabilities the genotypes of two individuals will have two, one or zero alleles IBD. These three quantities are denoted by:

ϕ_2 , ϕ_1 , and ϕ_0 , ($0 \leq \phi_i \leq 1$, $\phi_2 + \phi_1 + \phi_0 = 1$),

IBD status 2, 1, and 0 are called as events I, T, and O.

Kinship Determination From Genotype Data On Two Individuals

The algorithm requires two sets of information.

Second, the probabilities of $G_y(l)$ given $G_x(l)$ at the l -th locus, under the scenarios of I, T, and O (i.e., under the scenario that $G_x(l)$ and $G_y(l)$ have 2, 1, or 0 alleles IBD)

The three conditional probabilities are:

$P_2(xy) = \text{probability of } G_y(l) \text{ given } G_x(l) \text{ with 2 of their alleles IBD}$

$P_1(xy) = \text{probability of } G_y(l) \text{ given } G_x(l) \text{ with 1 of their alleles IBD}$

$P_0(xy) = \text{probability of } G_y(l) \text{ given } G_x(l) \text{ with 0 of their alleles IBD}$

Kinship Determination From Genotype Data On Two Individuals

The three conditional probabilities are:

$P_2(xy)$ = probability of $G_y(l)$ given $G_x(l)$ with 2 of their alleles IBD

$P_1(xy)$ = probability of $G_y(l)$ given $G_x(l)$ with 1 of their alleles IBD

$P_0(xy)$ = probability of $G_y(l)$ given $G_x(l)$ with 0 of their alleles IBD

The likelihood of observing $G_y(l)$ given $G_x(l)$ for a given kinship (i.e., for a given set of ϕ_2 , ϕ_1 , and ϕ_0) is computed by

$$[P_2(xy) \times \phi_2] + [P_1(xy) \times \phi_1] + [P_0(xy) \times \phi_0]$$

Kinship Determination From Genotype Data On Two Individuals

$$[P_2(xy) \times \phi_2] + [P_1(xy) \times \phi_1] + [P_0(xy) \times \phi_0]$$

The likelihoods that are computed for each locus can be multiplied over all L loci to compute the combined likelihood under a specified kinship relationship. The kinship that provides the maximum likelihood is the best-supported kinship relationship for the observed genotype profiles of individuals X and Y.

At a Single Locus, Two Individuals Can Have the Following Genotypes

<u>#1</u>	<u>#2</u>	
AB	AB	2 alleles shared
AA	AA	
AA	AB	1 allele shared
AB	AC	
AB	CD	0 alleles shared
AA	BB	
AA	BC	

Identity By Descent (IBD) Coefficients:

	ϕ_2	ϕ_1	ϕ_0
• Full sibs	1/4	1/2	1/4
• Parent-child	0	1	0
• Half-sibs	0	1/2	1/2
• Cousins	0	1/4	3/4
• Unrelated	0	0	1

How Are The Identity By Descent (IBD) Coefficients Determined:

	ϕ_2	ϕ_1	ϕ_0
Full sibs	1/4	1/2	1/4

Lets say we have two parents AB and CD, their offspring are either AC, AD, BC, or BD

	AC	AD	BC	BD
AC	2	1	1	0
AD	1	2	0	1
BC	1	0	2	1
BD	0	1	1	2

2 alleles IBD = 4/16 or $\frac{1}{4}$

1 alleles IBD = 8/16 or $\frac{1}{2}$

0 alleles IBD = 4/16 or $\frac{1}{4}$

How Are The Identity By Descent (IBD) Coefficients Determined:

	ϕ_2	ϕ_1	ϕ_0
Parent - Child	0	1	0

Lets say we have two parents AB and CD,
their offspring are either AC, AD, BC, or BD

	AB	CD
AC	1	1
AD	1	1
BC	1	1
BD	1	1

2 alleles IBD = 0/8 or 0

1 alleles IBD = 8/8 or 1

0 alleles IBD = 0/8 or 0

How Are The Identity By Descent (IBD) Coefficients Determined:

	ϕ_2	ϕ_1	ϕ_0
Half-sibs	0	1/2	1/2

Lets say we have three parents AB (m), CD (af1), and EF (af2)
their offspring are either AC, AD, BC, BD or AE, AF, BE, BF

	AE	AF	BE	BF
AC	1	1	0	0
AD	1	1	0	0
BC	0	0	1	1
BD	0	0	1	1

2 alleles IBD = 0/16 or 0

1 alleles IBD = 8/16 or 1/2

0 alleles IBD = 8/16 or 1/2

The Identity By Descent (IBD) Coefficients

	ϕ_2	ϕ_1	ϕ_0
Half-Sibs	0	1/2	1/2
Uncle/Aunt/Nephew/Niece	0	1/2	1/2
Grandparent-Grandchild	0	1/2	1/2

IBD Coefficients are the same for these kinship relationships

Kinship Formula:

C.C. Li and L.Sachs, 1954.
Method of ITO stochastic matrices

Transition matrix for 2 alleles IBD

$$I = \begin{array}{c|ccc} & A_1A_1 & A_1A_2 & A_2A_2 \\ \hline A_1A_1 & 1 & 0 & 0 \\ A_1A_2 & 0 & 1 & 0 \\ A_2A_2 & 0 & 0 & 1 \end{array}$$

Kinship Formula

Transition matrix for 1 allele IBD

$$T = \begin{array}{c|ccc} & A_1A_1 & A_1A_2 & A_2A_2 \\ \hline A_1A_1 & p_1 & p_2 & 0 \\ A_1A_2 & 0.5p_1 & 0.5(p_1 + p_2) & 0.5p_2 \\ A_2A_2 & 0 & p_1 & p_2 \end{array}$$

Kinship Formula:

Transition matrix for 0 alleles IBD

$$O = \begin{array}{c|ccc} & A_1A_1 & A_1A_2 & A_2A_2 \\ \hline A_1A_1 & p_1^2 & 2p_1p_2 & p_2^2 \\ A_1A_2 & p_1^2 & 2p_1p_2 & p_2^2 \\ A_2A_2 & p_1^2 & 2p_1p_2 & p_2^2 \end{array}$$

Kinship Formulas:

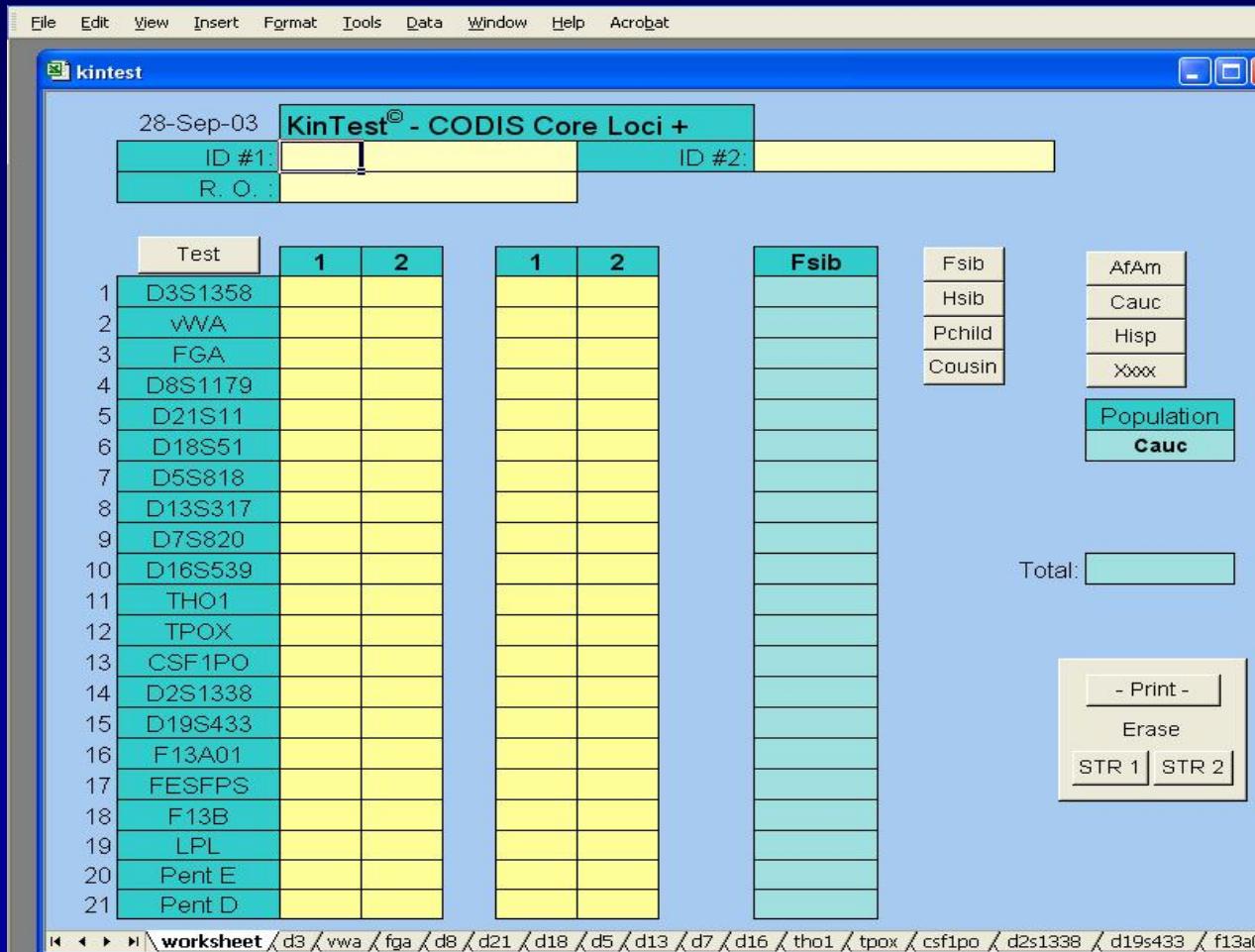
$$[P_2(xy) \times \phi_2] + [P_1(xy) \times \phi_1] + [P_0(xy) \times \phi_0]$$

<u>#1</u>	<u>#2</u>	<u>frequency</u>
AB	AB	$\phi_2 + 0.5\phi_1(p_A + p_B) + 2\phi_0p_Ap_B$
AA	AA	$\phi_2 + \phi_1p_A + \phi_0p_A^2$
AA	AB	$\phi_1p_B + 2\phi_0p_Ap_B$
AB	AC	$0.5\phi_1p_C + 2\phi_0p_Ap_C$
AB	CD	$2\phi_0p_Cp_D$
AA	BB	$\phi_0p_B^2$
AA	BC	$2\phi_0p_Bp_C$

Likelihood Ratios for Full- & Half-Sibs

		full-sib	:	half-sib	:	unrelated
AB	AB	$(1+p+q+2pq)$:	$(p+q+4pq)$:	$8pq$
AA	AA	$(1+p)^2$:	$2p(1+p)$:	$(2p)^2$
AA	AB	$(1+p)$:	$(1+2p)$:	$4p$
AB	AC	$(1+2p)$:	$(1+4p)$:	$8p$
AB	CD	1	:	2	:	4
AA	BB	1	:	2	:	4
AA	BC	1	:	2	:	4

KinTest Program Created by George Carmody, Carleton University, Canada



27-Sep-03

KinTest® - CODIS Core Loci +

ID #1: P-61692

ID #2:

R. O.:

	Test	1	2
1	D3S1358	17	18
2	vWA	15	20
3	FGA	24	25
4	D8S1179	12	15
5	D21S11	27	30
6	D18S51	16	17
7	D5S818	8	13
8	D13S317	11	12
9	D7S820	10	11
10	D16S539	11	12
11	TH01	8	8
12	TPOX	9	10
13	CSF1PO	9	10
14	D2S1338	19	23
15	D19S433	14.2	16
16	F13A01	3.2	15
17	FESFPS	11	12
18	F13B	6	10
19	LPL	11	12
20	Pent E		
21	Pent D		

1	2
16	16
16	20
22	25
12	15
29	30
17	17
8	13
11	12
10	12
11	13
8	8
9	9
10	10
19	19
14	14.2
3.2	15
11	12
9	10
11	12

Fsib
0.25
10.05
2.06
10.09
0.79
1.86
77.57
2.32
0.68
0.71
20.07
2.28
1.24
1.98
7.85
124.46
2.36
0.56
3.45

Fsib
AfAm
Hsib
Pchild
Cousin

AfAm
Cauc
Hisp
Xxxx

Population
Cauc

Total: 3.3E+09

- Print -

Erase

STR 1 STR 2

27-Sep-03

KinTest[®] - CODIS Core Loci +

ID #1: P-61692

ID #2:

R. O.:

	Test	1	2
1	D3S1358	17	18
2	vWA	15	20
3	FGA	24	25
4	D8S1179	12	15
5	D21S11	27	30
6	D18S51	16	17
7	D5S818	8	13
8	D13S317	11	12
9	D7S820	10	11
10	D16S539	11	12
11	TH01	8	8
12	TPOX	9	10
13	CSF1PO	9	10
14	D2S1338	19	23
15	D19S433	14.2	16
16	F13A01	3.2	15
17	FESFPS	11	12
18	F13B	6	10
19	LPL	11	12
20	Pent E		
21	Pent D		

	1	2
	16	16
	16	20
	22	25
	12	15
	29	30
	17	17
	8	13
	11	12
	10	12
	11	13
	8	8
	9	9
	10	10
	19	19
	14	14.2
	3.2	15
	11	12
	9	10
	11	12

	Hsib
	0.50
	10.30
	2.31
	2.50
	1.04
	2.11
	11.11
	1.30
	0.93
	0.96
	4.48
	2.53
	1.49
	2.23
	8.10
	11.59
	1.34
	0.81
	1.55

Fsib
Hsib
Pchild
Cousin

AfAm
Cauc
Hisp
Xxxx

Population
Cauc

Total: 5.0E+06

- Print -

Erase

STR 1 STR 2

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Pent E		
Pent D		

1	2
16	16
16	20
22	25
12	15
29	30
17	17
8	13
11	12
10	12
11	13
8	8
9	9
10	10
19	19
14	14.2
3.2	15
11	12
9	10
11	12

Pchild
0.001
19.60
3.63
4.00
1.08
3.21
21.21
1.59
0.86
0.92
7.96
4.06
1.97
3.45
15.20
22.17
1.68
0.62
2.10

Fsib
Hsib
Pchild
Cousin

AfAm
Cauc
Hisp
Xxxx

Population
Cauc

Total: **4.3E+09**

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STR 1 | STR 2

$$\text{Residual CPI} = 4.3\text{E}+9 \times 0.001 = 4.3\text{E}+6$$

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	1	2
1	16	16
2	16	20
3	22	25
4	12	15
5	29	30
6	17	17
7	8	13
8	11	12
9	10	12
10	11	13
11	8	8
12	9	9
13	10	10
14	19	19
15	14	14.2
16	3.2	15
17	11	12
18	9	10
19	11	12

Cousin
0.75
5.65
1.66
1.75
1.02
1.55
6.05
1.15
0.97
0.98
2.74
1.77
1.24
1.61
4.55
6.29
1.17
0.91
1.28

Fsib
Hsib
Pchild
Cousin

AfAm
Cauc
HisP
Xxxx

Population
Cauc

Total: 4.8E+04

- Print -

Erase

STR 1 STR 2

Kinship Determination For Genotype Data On Two Individuals in Case P-61692

Full – Sibs	3.3E+9
Half – Sibs	5.0E+6
Parent – Child	4.3E+6
First Cousins	4.8E+4

The likelihood ratio comparing Full-Sib to Half-Sib is

$$3.3\text{E}+9 / 5.0\text{E}+6 = 660$$

It is **660 times more likely** given the genotype of individual one, and the genotype of individual two, that the two individuals are Full-Sibs as opposed to Half-Sibs

Kinship Analysis P-61692 at the D5S818 Locus

Individual 1	Individual 2	Allele Freq
8	8	0.0128
13	13	0.1462
	ϕ_2	ϕ_1
Full - Sib	0.25	0.5
Half - Sib	0	0.5
Parent - Child	0	1
First Cousins	0	0.25
Unrelated	1	0
		ϕ_0

$$\phi_2 + 0.5\phi_1(p_A + p_B) + 2\phi_0 p_A p_B$$

Kinship Analysis P-61692 at the D5S818 Locus

$$\phi_2 + 0.5\phi_1(p_A + p_B) + 2\phi_0 p_A p_B$$

Full – Sibs = $0.25 + (0.5 * 0.5(0.0128 + 0.1462)) + (2 * 0.25 * 0.0128 * 0.1462)$

Half – Sibs = $(0.5 * 0.5(0.0128 + 0.1462)) + (2 * 0.5 * 0.0128 * 0.1462)$

Parent – Child = $(0.5 * (0.0128 + 0.1462))$

First Cousins = $(0.5 * 0.25(0.0128 + 0.1462)) + (2 * 0.75 * 0.0128 * 0.1462)$

Unrelated = $2 * 0.0128 * 0.1462$

Kinship Analysis P-61692 at the D5S818 Locus

Full – Sibs	0.291
Half – Sibs	0.042
Parent – Child	0.080
First Cousins	0.023
Unrelated	0.004

Divide each of the kinship likelihood by the likelihood for Unrelated

Kinship Analysis P-61692 at the D5S818 Locus

Full – Sibs	77.67
Half – Sibs	11.12
Parent – Child	21.24
First Cousins	6.06
Unrelated	1

Thank you!

