Forensic Mathematics and 9/11

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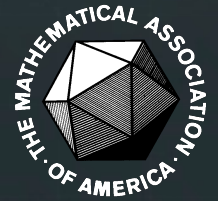
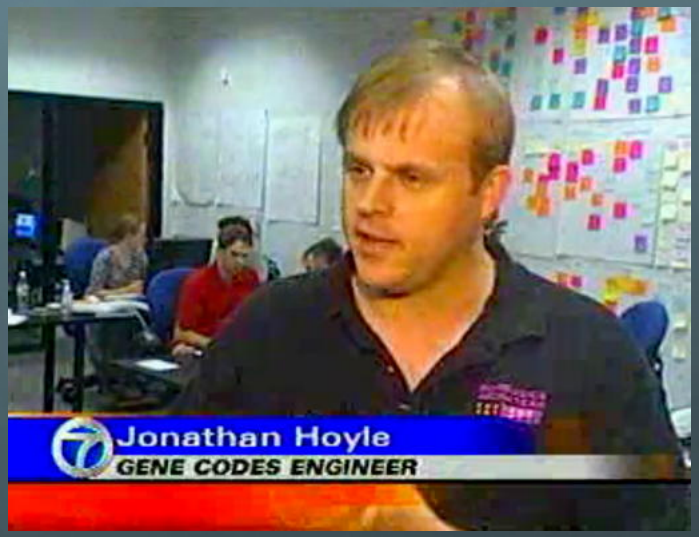
Overview

- Introduction
- World Trade Center Project
  - 9/11 and NYC
  - Direct Matching (STR Analysis)
  - Kinship Analysis
  - Mitochondrial DNA
  - SNP's
- Summary
- Q & A



Introduction

- I am Jonathan Hoyle.
- From 2001-2005, Mathematician and Software Engineer with *Gene Codes Forensics*.
- Updated Presentation



- Both my Undergraduate (University of Delaware) and Graduate (University of Michigan) studies were in Mathematics with a Computer Science minor.
- Involved with ***M-FISys*** (pronounced “emphasis”), the forensic DNA software used to identify the victims of the World Trade Center attacks.

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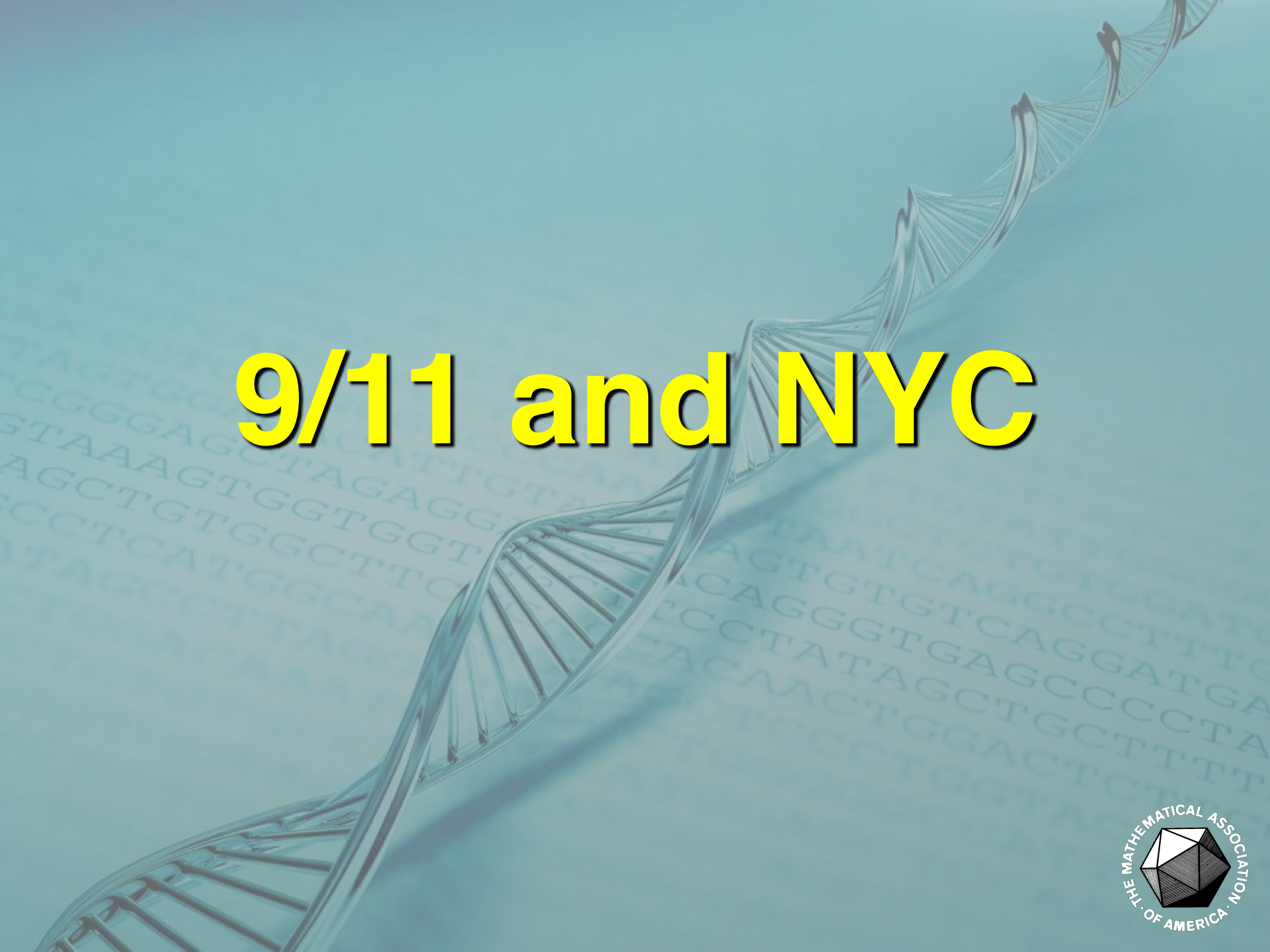
9/11 and NYC



Ground Zero

- Two 110 story towers
- 15 buildings over 16 acres
- Six basement levels and four subway lines
- 24,000 gallons of jet fuel
- Fires burned at 1800˚F for over 3 months
- 2 billion pounds of rubble
- Existing DNA were incapable of handling level of magnitude

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Barclay St.
West Broadway St.
Washington St.
Verizon Building
7 WTC (47 Stories)
Old Post Office Building
Vesey St.
6 WTC (9 Stories)
5 WTC (9 Stories)
American Express Building
Fulton St.
Millenium Hotel
Church St.
West St.
1 WTC (110 Stories)
Austin J. Tobin Plaza
Dey St.
Merrill Lynch Building
3 WTC (22 Stories)
2 WTC (110 Stories)
4 WTC (9 Stories)
Cortlandt St.
1 Liberty Plaza Building
Liberty St.
Washington St.
Greenwich St.
90 West Building
Bankers Trust
Credit: NOAA
September 23, 2001
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The Victims

- Unknown number of casualties early on
- Some family members afraid to come forward
- 20,000 total remains
- Some victims found in up to 200 fragments
- Majority of remains required DNA analysis
- 2,606 total victims

The Recovery

Thousands of rescue workers work around the clock from 9/11/01 through 5/30/02 in the recovery effort.

Forensic DNA Identification Project with NYC Chief Medical Examiner's Office continued for three years.

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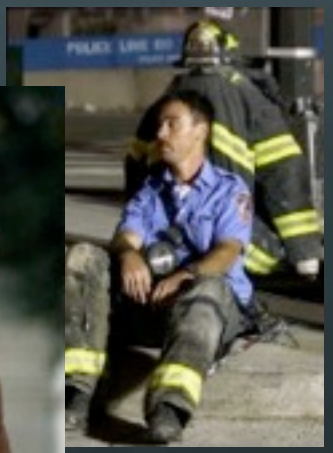

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Staten Island Triage

Trucks ship tons of debris from Ground Zero were sent to the Staten Island Recovery Site

Forensic anthropologists examine the debris to determine if it contains any human remains

Human remains found were sent to the Forensic Investigation Center in Albany, NY

Staten Island Recovery Site

Victim samples are typed using many DNA fingerprinting techniques, such as STR, MitoDNA & SNP to match against a personal effect.

Family members are cheek swabbed for their DNA so that Kinship identification can be made when direct matching is not available

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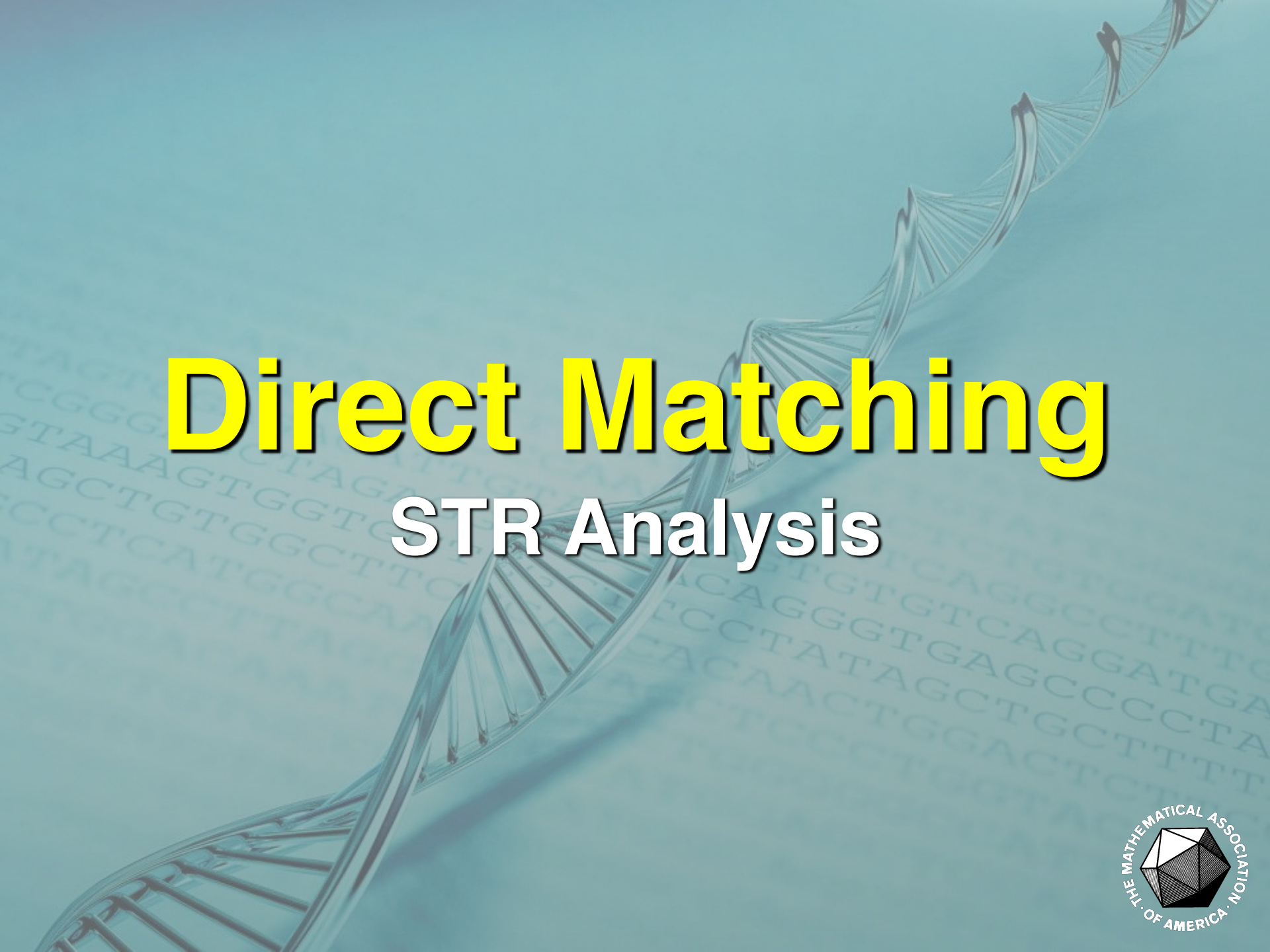
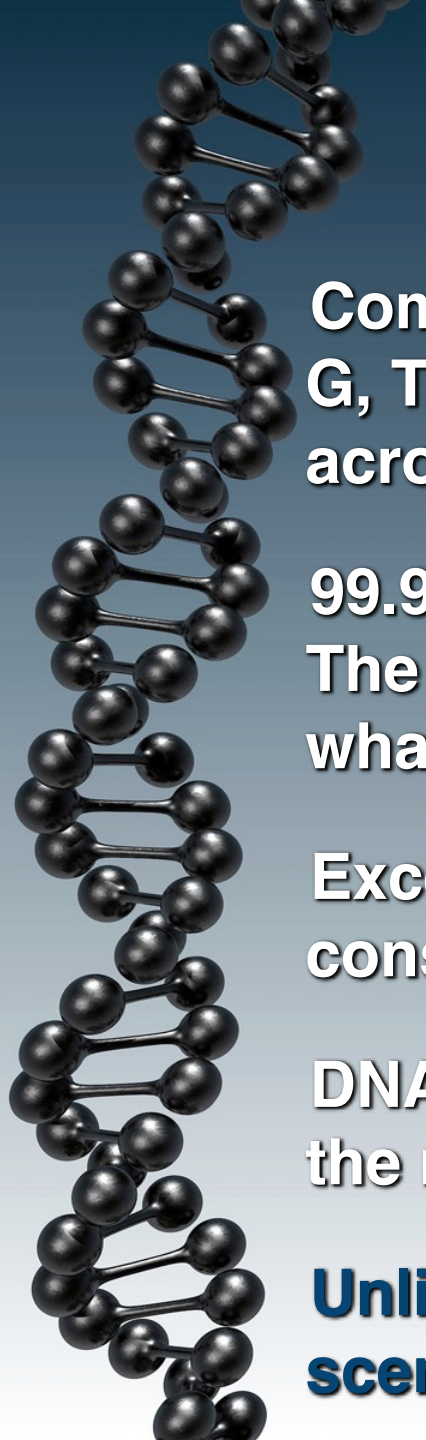
M-FISys Team Meeting

Direct Matching

STR Analysis



DNA



Composed of an alphabet of four chemicals: A, C, G, T, human DNA consists of 3.5 Billion base pairs across 23 chromosomes.

99.9% of your DNA is shared with all of humanity. The remaining 0.1% (3.5 million base pairs) are what distinguishes us.

Except for identical twins, each person's DNA is considered unique.

DNA began to be used for forensic analysis in the mid-1980's.

Unlike plane crashes and most other crime scenes, this is an N-to-N problem.

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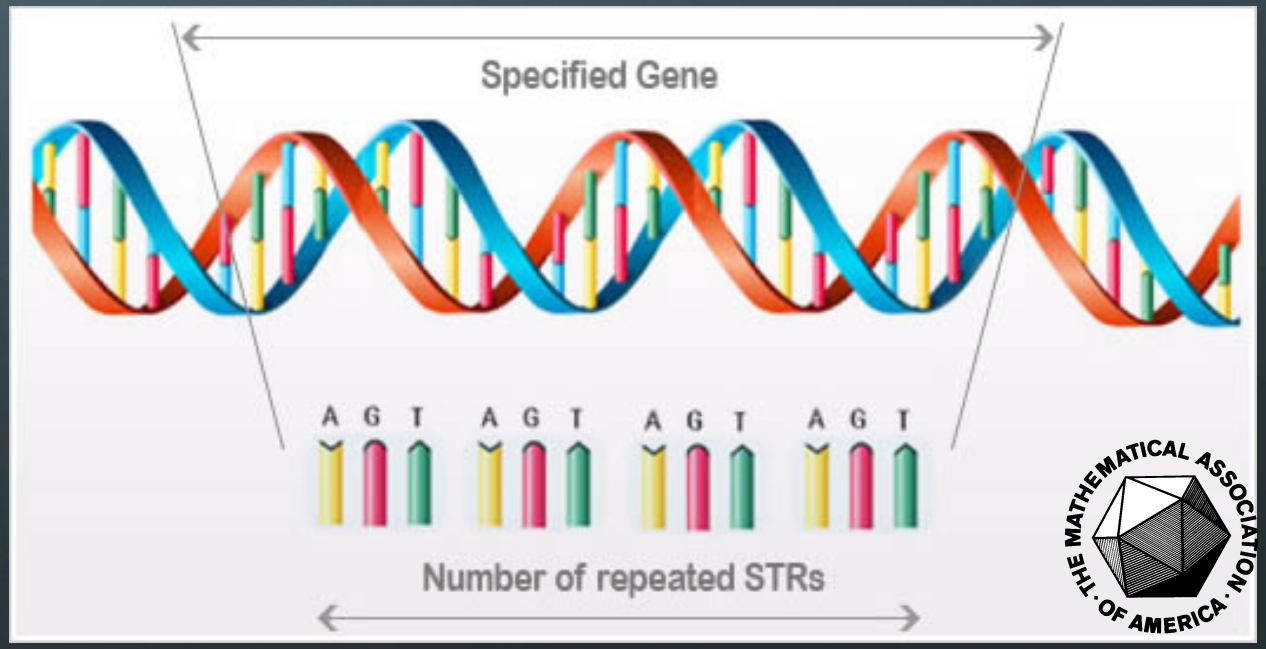
STR: Short Tandem Repeats

- A repeat of a short sequence of bases (usually 4 or 5):

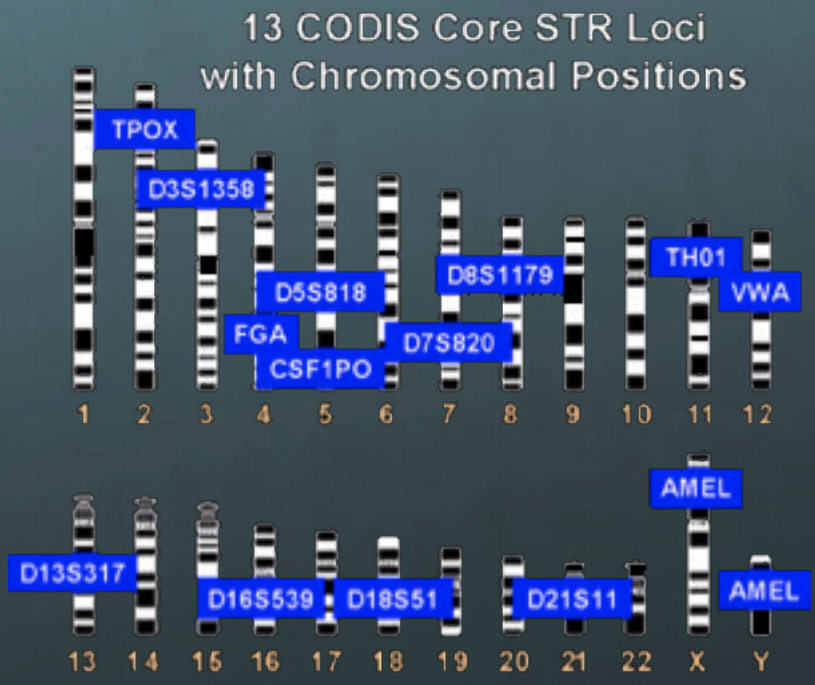
```
...gcctggatagatagatagatagatgttta...
```

- The above is repeated 5 times with a partial 3 bases
- The value for this STR locus is `5.3` (called its *allele*)
- Each locus contains a pair of alleles (inherited one from each parent), eg: `5.3 / 8`



STR Profiles

- In 1997, the FBI standardized on 13 core STR loci for its national database, CoDIS.
- STR analysis is the forensic standard for identification.
- Includes two PowerPlex loci: Penta D and Penta E
- When both allele values are the same, it is called homozygous; otherwise, it is called heterozygous.
- Gender: XX or XY
- These loci are “unlinked” and thus independent



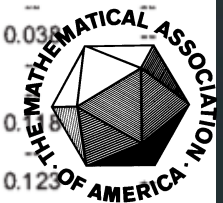
Allele Frequencies

TABLE 1—*U.S. Caucasian allele frequencies for 15 autosomal STR loci (N =302).*

Allele	CSF1PO	FGA	TH01	TPOX	VWA	D3S1358	D5S818	D7S820	D8S1179	D13S317	D16S539	D18S51	D21S11	D2S1338	D19S433
5	--	--	0.002	0.002	--	--	--	--	--	--	--	--	--	--	--
6	--	--	0.232	0.002	--	--	--	--	--	--	--	--	--	--	--
7	--	--	0.190	--	--	--	0.002	0.018	--	--	--	--	--	--	--
8	0.005	--	0.084	0.535	--	--	0.003	0.151	0.012	0.113	0.018	--	--	--	--
8.1	--	--	--	--	--	--	--	0.002	--	--	--	--	--	--	--
9	0.012	--	0.114	0.119	--	--	0.050	0.177	0.003	0.075	0.113	--	--	--	--
9.3	--	--	0.368	--	--	--	--	--	--	--	--	--	--	--	--
10	0.217	--	0.008	0.056	--	--	0.051	0.243	0.101	0.051	0.056	0.008	--	--	0.002
10.3	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--
11	0.301	--	0.002	0.243	--	0.002	0.361	0.207	0.083	0.339	0.321	0.017	--	--	0.005
12	0.361	--	--	0.041	--	--	0.384	0.166	0.185	0.248	0.326	0.127	--	--	0.081
12.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.002
13	0.096	--	--	0.002	0.002	--	0.141	0.035	0.305	0.124	0.146	0.132	--	--	0.253
13.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.007
14	0.008	--	--	--	0.094	0.103	0.007	0.002	0.166	0.048	0.020	0.137	--	--	0.369
14.2	--	--	--	--	--	--	--	--	--	--	--	0.002	--	--	0.018
15	--	--	--	--	0.111	0.262	0.002	--	0.114	0.002	--	0.159	--	0.002	0.152
15.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.035
16	--	--	--	--	0.200	0.253	--	--	0.031	--	--	0.139	--	0.033	0.050
16.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.015
17	--	--	--	--	0.281	0.215	--	--	--	--	--	0.126	--	0.182	0.008
17.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	0.002
18	--	0.026	--	--	0.200	0.152	--	--	--	--	--	0.076	--	0.079	--
18.2	--	--	--	--	--	--	--	--	--	--	--		--	--	0.002
19	--	0.053	--	--	0.104	0.012	--	--	--	--	--	0.038	--	0.114	--
19.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--
20	--	0.127	--	--	0.005	0.002	--	--	--	--	--	0.022	--	0.146	--
21	--	0.185	--	--	0.002	--	--	--	--	--	--	0.008	--	0.041	--
21.2	--	0.005	--	--	--	--	--	--	--	--	--	--	--	--	--
22	--	0.219	--	--	--	--	--	--	--	--	--	0.008	--	0.038	--
22.2	--	0.012	--	--	--	--	--	--	--	--	--	--	--	--	--
22.3	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--
23	--	0.134	--	--	--	--	--	--	--	--	--	--	--	0.[illegible]	--
23.2	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--
24	--	0.136	--	--	--	--	--	--	--	--	--	--	--	0.123	--

J Forensic Sci, July 2003, Vol. 48, No. 4

http://www.cstl.nist.gov/strbase/pub_pres/Butler2003a.pdf



Allele Frequencies

- **According to the Hardy-Weinberg Principle:**
  - p^2 for homozygous alleles, p = frequency of allele
  - $2pq$ for heterozygous alleles, p,q = frequency of alleles
- **This assumes an sufficiently large population.**
- **Since the population is relatively small, we must introduce the inbreeding coefficient θ:**
  - $p^2 + p(1-p)\theta$ for homozygous alleles
  - $2pq(1-\theta)$ for heterozygous alleles
- **Because θ is very small (0.03), we round on the side of being conservative:**
  - $p^2 + p(1-p)\theta$ for homozygous alleles
  - $2pq$ for heterozygous alleles

Profile Frequency

Locus	Victim	Sample	Equation	Prob	Likelihood
Gender	XY	XY	1/2	0.5000	2.00
D3S1358	14/16	14/16	2pq	0.0650	15.38
vWA	15/16	-			1.00
FGA	20/24	20/24	2pq	0.0401	24.95
D8S1179	12	12	$p^2+p(1-p)\theta$	0.0224	44.68
D21S11	28/31.2	28/31.2	2pq	0.0330	30.31
D18S51	14/17	-			1.00
D5S818	8/11	8/11	2pq	0.0106	94.47
D13S317	8	8	$p^2+p(1-p)\theta$	0.0108	92.62
D7S820	10/13	10/13	2pq	0.0172	58.13
D16S539	9	9	$p^2+p(1-p)\theta$	0.0117	85.12
TH01	6/9	-			1.00
TPOX	8/10	-			1.00
CSF1PO	10/12	10/12	2pq	0.1650	6.06
Penta D	9	-			1.00
Penta E	8/12	-			1.00

2.7E+14

Allelic Dropout

Locus	Victim	Sample	Equation	Prob	Likelihood
Gender	XY	XY	1/2	0.5000	2.00
D3S1358	14/16	14/16	2pq	0.0650	15.38
vWA	15/16	-			1.00
FGA	20/24	20/24	2pq	0.0401	24.95
D8S1179	12	12	$p^2+p(1-p)\theta$	0.0224	44.68
D21S11	28/31.2	28/31.2	2pq	0.0330	30.31
D18S51	14/17	-			1.00
D5S818	8/11	8	2p	0.3205	3.12
D13S317	8	8	$p^2+p(1-p)\theta$	0.0108	92.62
D7S820	10/13	10/13	2pq	0.0172	58.13
D16S539	9	9	$p^2+p(1-p)\theta$	0.0117	85.12
TH01	6/9	-			1.00
TPOX	8/10	-			1.00
CSF1PO	10/12	10/12	2pq	0.1650	6.06
Penta D	9	-			1.00
Penta E	8/12	-			1.00

9.0E+12

Likelihood Threshold

- How good is *good enough*?
- OCME wanted a minimum likelihood threshold set such that a chance of *any* mismatch would be less than one in a million.
- What does this mean mathematically?
- Choose n such that identifications are satisfied when the likelihood value of a sample is $\geq 10^n$.
- The probability of a fortuitous match of such a sample is thus $\mathrm{p} = 10^{-n}$. No mismatch $\mathrm{q} = 1 - 10^{-n}$.
- Unknown population size, but early estimates assumed a population as high as 5000.

Likelihood Threshold

- The probability of no mismatches is thus: q^{5000}
- The probability of any mismatch in the population:

$$1 - q^{5000} = 1 - (1 - 10^{-n})^{5000}$$

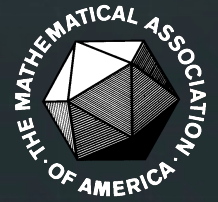
- For this to be a *"less than one in a million chance"* occurrence yields the equation:

$$1 - (1 - 10^{-n})^{5000} < 0.000001$$

- Solving for n we get:

$$n > log_{10}(1 - \sqrt[5000]{\quad} 0.999999) = 9.6989\ldots$$

- Thus we choose $n = 10.$



DNA Matching

- 12,000 personal effects were collected from families.
- A sample can be identified to a personal effect if:
  - ✓ Has at least 7 common alleles
  - ✓ No more than one mismatch due to allelic dropout
  - ✓ Likelihood value $\geq 10^{10}$
- ~30% of the victim samples had complete profiles.
- ~20% had partial profiles with likelihoods $\geq 10^{10}$.
- ~20% had partial profiles with likelihoods $< 10^{10}$.
- ~30% of the STR profiles had no data at all.
- STR analysis alone would not be sufficient.

Kinship Analysis

Kinship Analysis

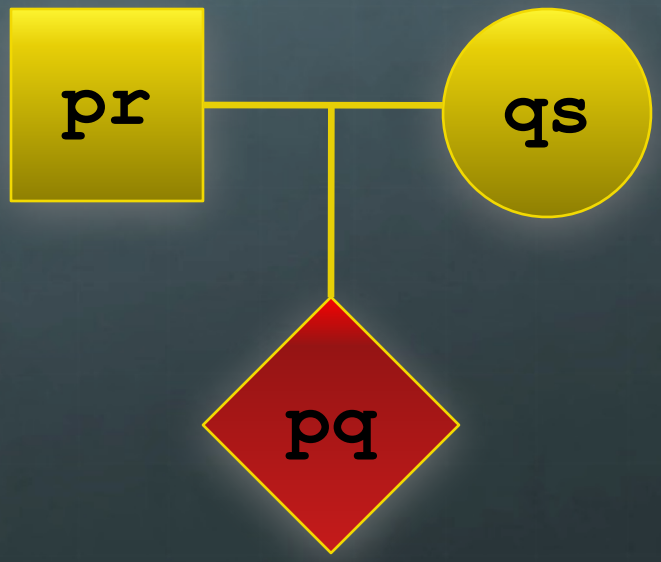
- Many personal effects lacked sufficient DNA.
- Others were contaminated by external DNA.
- Cheek swabs from family members were taken at Pier 94, so that a pedigree tree could be generated.
- A product of common loci can be used to produce kinship likelihood ratios (identifications $\geq 10^6$)
- A likelihood ratio is the ratio of the probability that the sample is a member of the given pedigree (H_1) over the probability that it is unrelated (H_0).

Kinship Example #1

Let **p**, **q**, **r**, **s** represent alleles and let p, q, r, s represent the probabilities of these alleles. (Let $p = 0.005, q = 0.02$)

A victim sample with allele **pq** and two Pedigree #1 containing two parents: father **pr** and mother **qs**.

$$LR = P(H_1) \div P(H_0) = P(\mathbf{pq} \mid \mathbf{pr} + \mathbf{qs}) \div P(\mathbf{pq} \mid \text{unrelated})$$



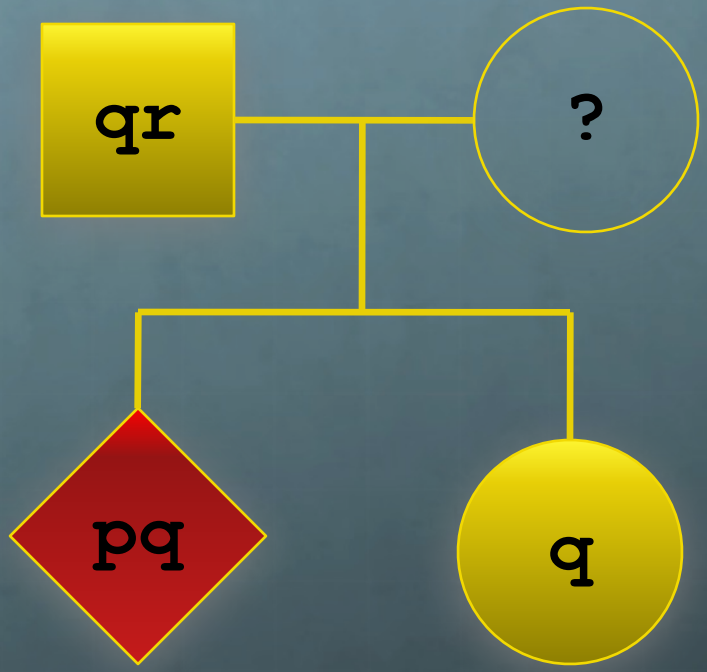
$$P(H_1) = \tfrac{1}{2} \times \tfrac{1}{2} \times 2pr \times 2qs = pqrs$$

$$P(H_0) = 2pq2pr2qs = 8p^2q^2rs$$

$$LR = pqrs \div 8p^2q^2rs = 1/8pq$$

$$= \mathbf{1250}$$

Kinship Example #2



The same victim sample with Pedigree #2 containing father **qr** and sister **q**.

For the **pq** victim sample to fit, the mother must be **pq** for H_1.

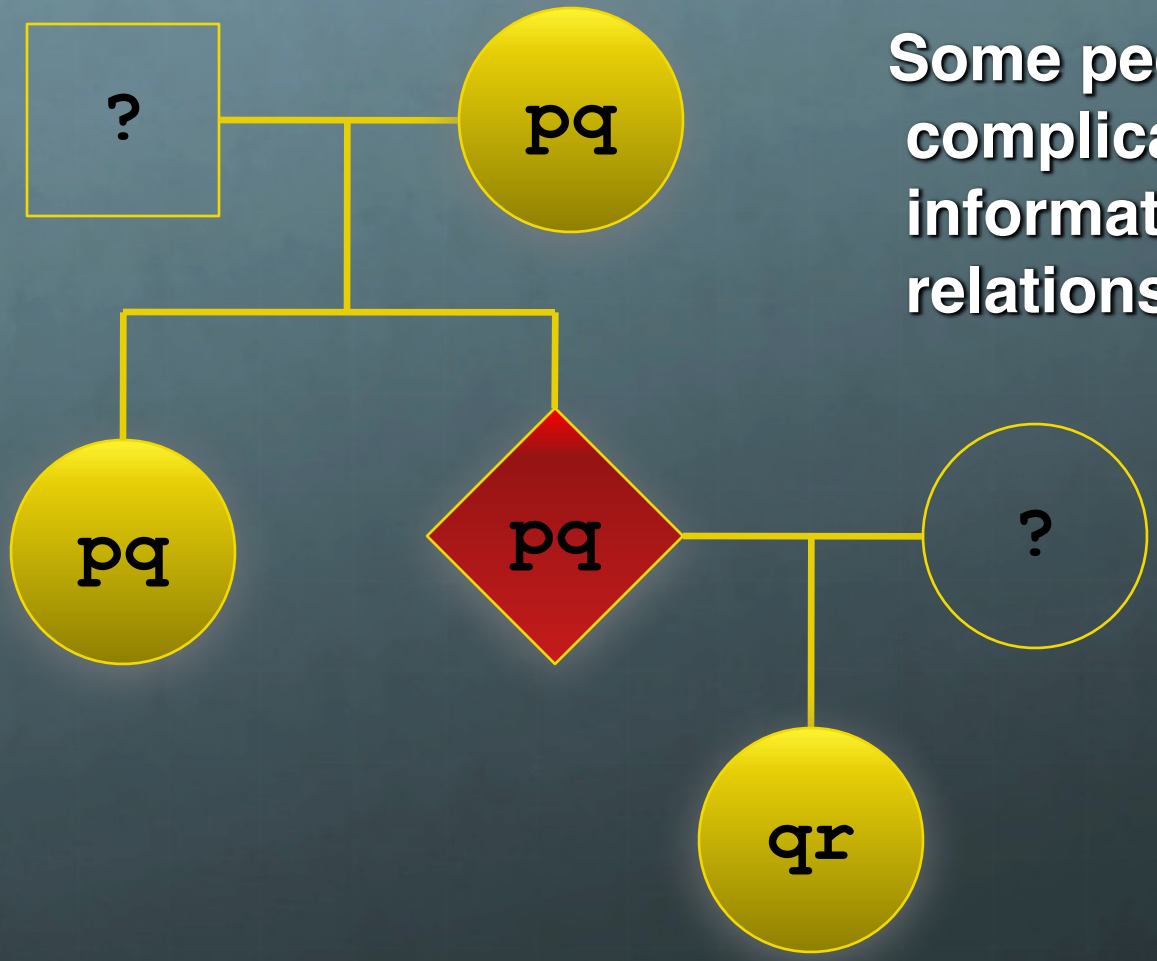
$$P(H_1) = \tfrac{1}{4} \times \tfrac{1}{4} \times 2pq2qr = \tfrac{1}{4}\, pq^2r$$

In H_0, mother may be **q** or **qX**, thus $P(H_0) = P(H_q) + P(H_{qx})$

$$P(H_q) = 2pq^4r \quad P(H_{qx}) = 2pq^3(1\text{-}q)r \rightarrow P(H_0) = 2pq^3r$$

$$LR = P(H_1) \div P(H_0) = 1/8q = \mathbf{6.25}$$

Kinship Example #3



Some pedigrees can be complicated, with partial information and extended relationships.

Relations may involve half siblings, cousins and any number of combinations.

$LR = (1+p+q) / 8pq =$ **1281.25**

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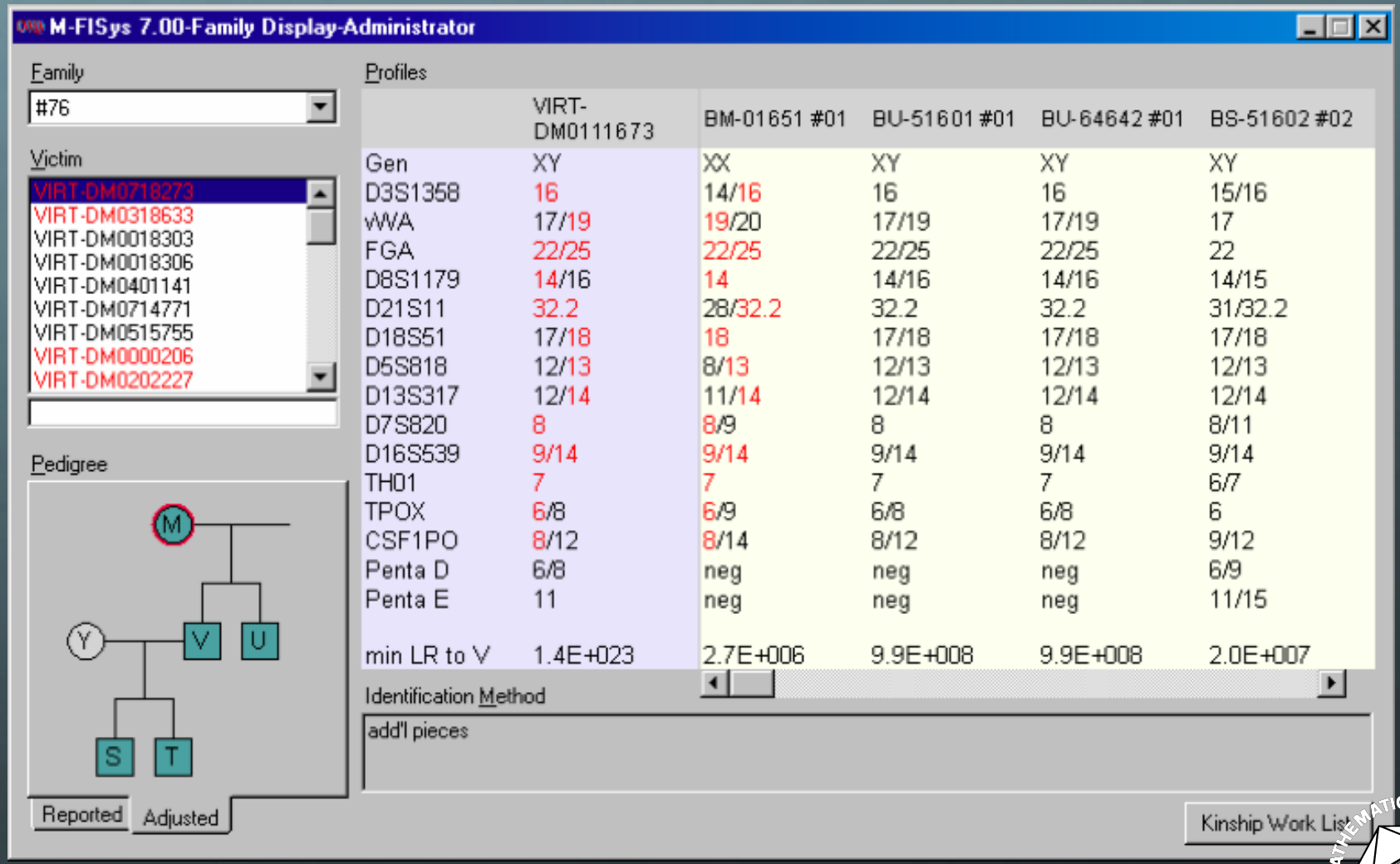
Kinship Equations

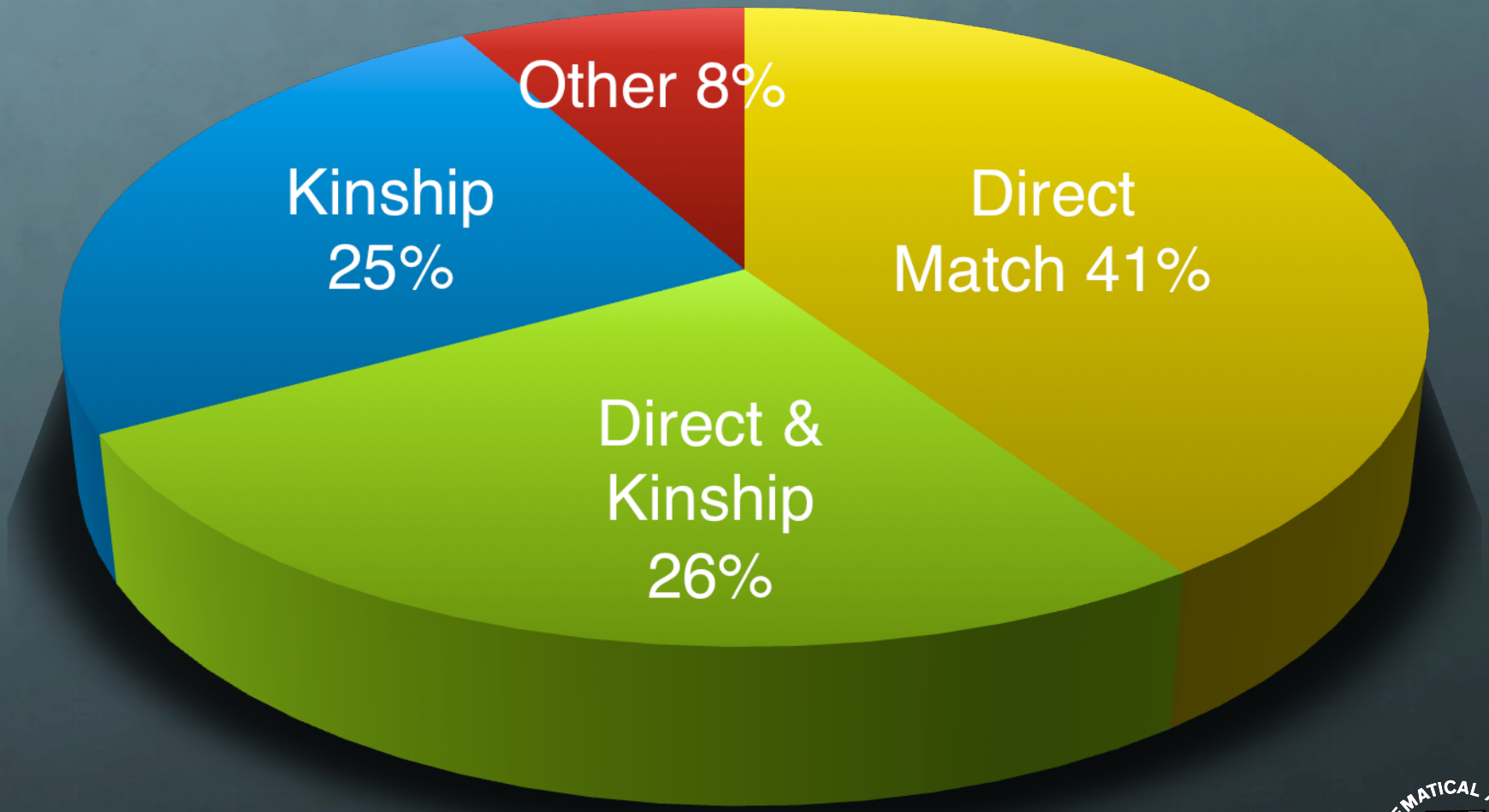
	VIRT-DM8180705	☑ BM-50527 #...	☑ BU-50527 #03	☑ BD-05721 #02	VIRT-DM8180705
Gen	XY	XX	XX	XX	-
D3S1358	15/16	15	14/15	15/16	1/4p
vWA	15/18	14/15	14/18	16/18	(1+q)/8pq
FGA	23/24	24	23/24	21/24	(1+p)/4pq
D8S1179	10/13	10/13	10/13	13/14	(p+q+pp+2pq+qq)/(8ppq+8pqq)
D21S11	30/31	30/33.2	30/31	27/31	(1+q)/8pq
D18S51	12/17	12/13	12/16	12/14	(1+q)/8pq
D5S818	10	10/12	10/12	9/10	(1+p+q)/(4pp+4pq)
D13S317	8/11	8/11	11/13	11	(p+q)/8pq
D7S820	10/12	11/12	11/12	10/11	1/8q
D16S539	10/12	12	12	12	1/4q
TH01	8/9.3	7/9.3	7/10	8/9.3	1/8p
TPOX	8/9	8/9	8/9	8/11	(p+q+pp+2pq+qq)/(8ppq+8pqq)
CSF1P0	9	9/11	9/11	9/13	(1+p+q)/(4pp+4pq)
Penta D	-	-	-	-	-
Penta E	-	-	-	-	-
					-
Likelihood	1.01e+18	1.54e+17	4.33e+16	2.01e+17	99.990883%
Kinship LR	99.990883%	2.59e+5	4.59e+5	5.73e+5	

M-FISys Kinship Form†



†presented in *Bioinformatics for 9/11*, Dr. Simon Mercer, Bio IT World, 2004.

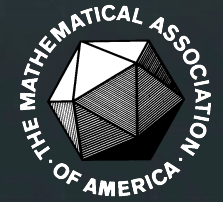
Match Methods on Remains



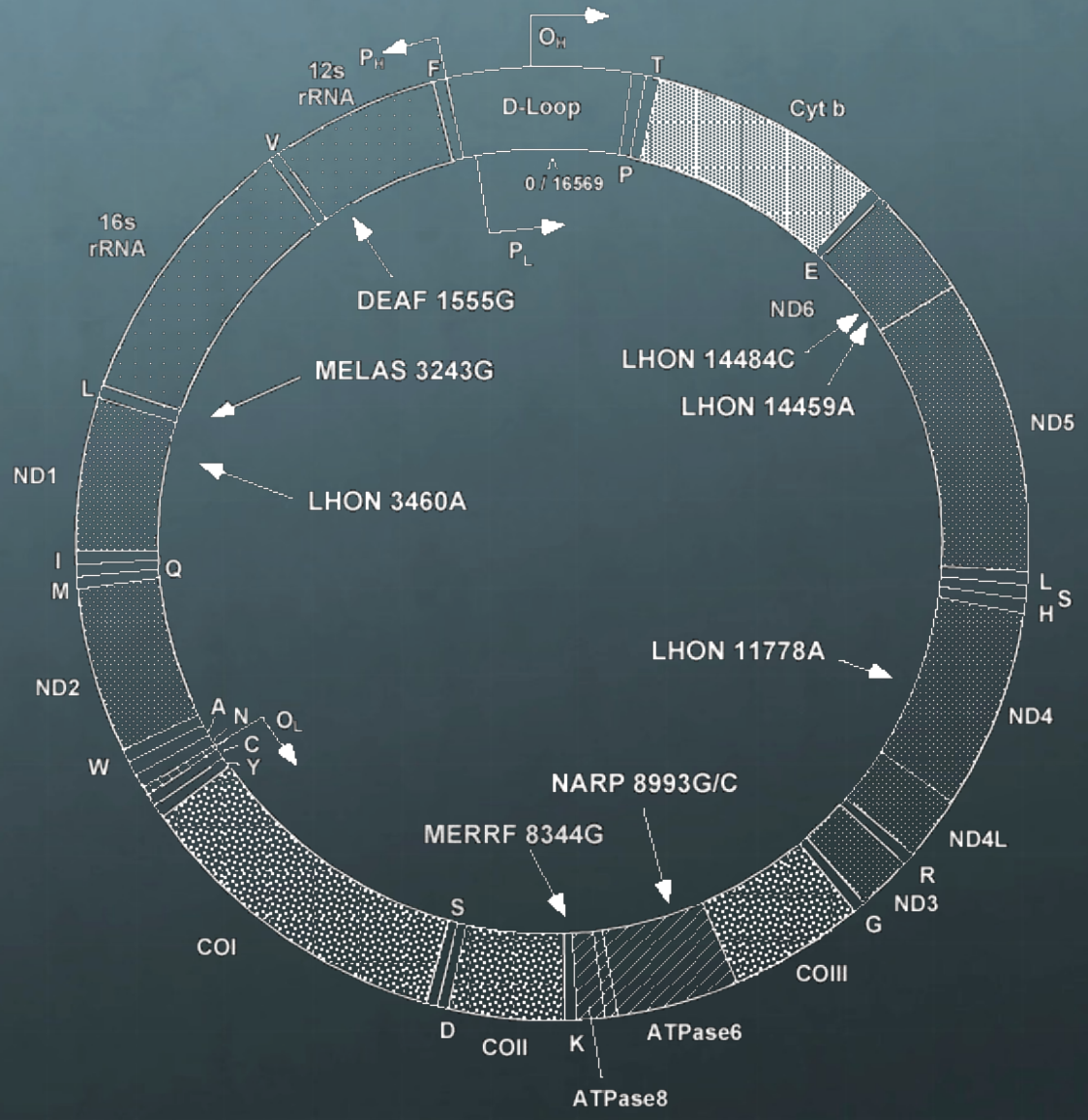
Mitochondrial DNA

Mitochondrial DNA

- Some victim samples were so degraded that STR analysis could not yield an identification.
- Mitochondrial DNA (mtDNA) is heartier material, surviving under extreme conditions.
- mtDNA is a 16,569-based circular genome.
- Being circular (unlike the double helix of nuclear DNA), it is more stable and less prone to damage.
- Although each cell contains only two copies of nuclear DNA, it has thousands of copies of mtDNA.
- mtDNA has been retrieved from ancient bones, including woolly mammoths and Neanderthals



mtDNA Map


O_H
P_H
12s rRNA
F
T
D-Loop
Cyt b
V
0 / 16569
P
16s rRNA
P_L
DEAF 1555G
E
ND6
LHON 14484C
MELAS 3243G
LHON 14459A
L
ND5
ND1
LHON 3460A
I
Q
M
L
S
H
LHON 11778A
ND2
ND4
A
N
O_L
C
Y
W
NARP 8993G/C
MERRF 8344G
ND4L
R
ND3
S
G
COI
COIII
D
COII
K
ATPase6
ATPase8

mtDNA Typing

- Mito-typing involves direct sequencing of two highly variable regions of mtDNA (HV1, HV2).
- Differences from *the Anderson Sequence* (an internationally accepted standard) are tracked.

mtDNA profile	
16093:	C
16224:	D
16311:	C
195:	C
263:	G
315.1:	C

- mtDNA is not unique, it is maternally inherited.
- Thus matching can be done against a personal effect or from maternal relatives (eg: mother, full sibling, maternal half-sibs, not father or paternal half-sibs).
- 75% of the victims had maternal relatives providing sample mtDNA for potential matches.

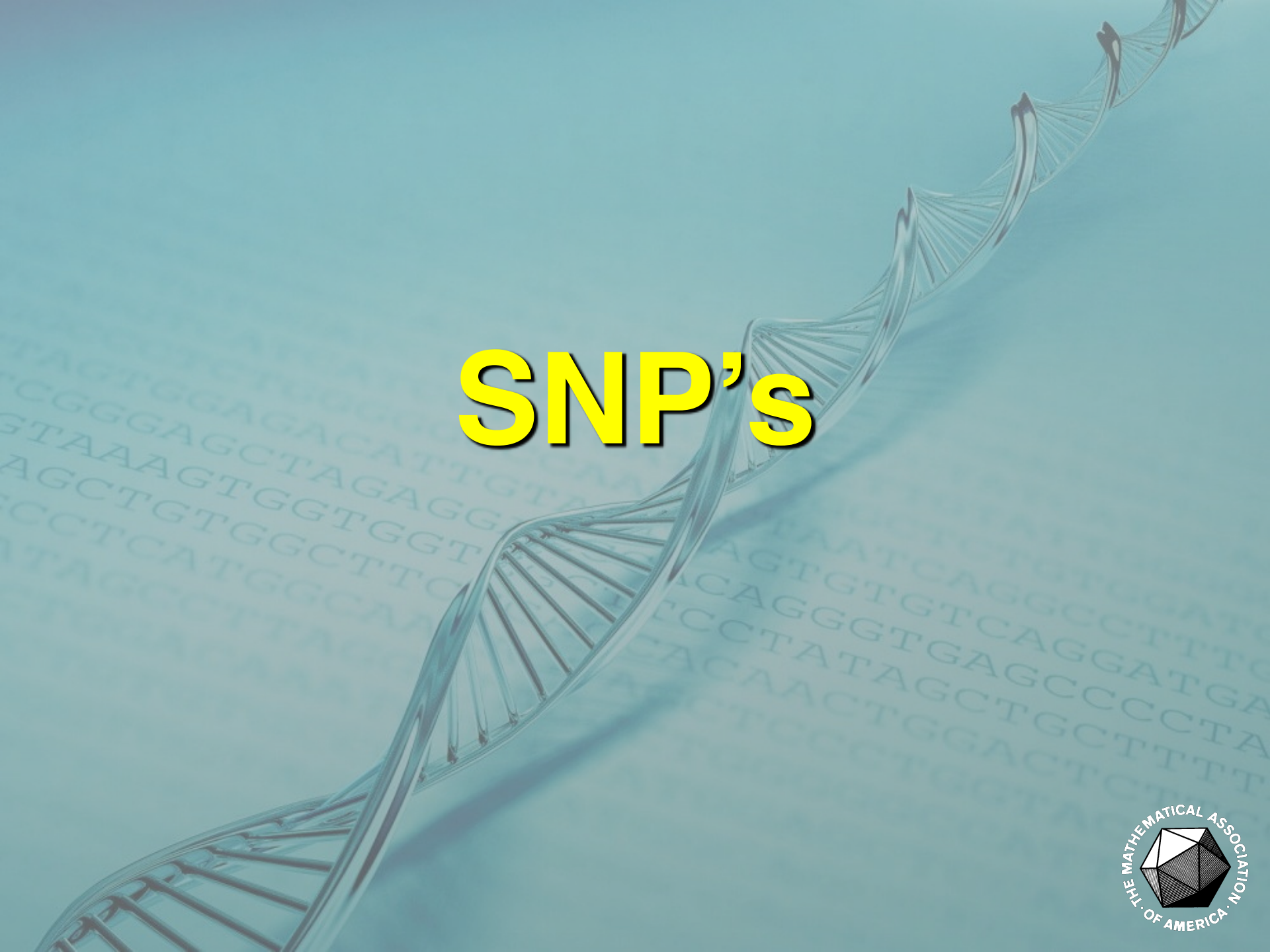
mtDNA Likelihood

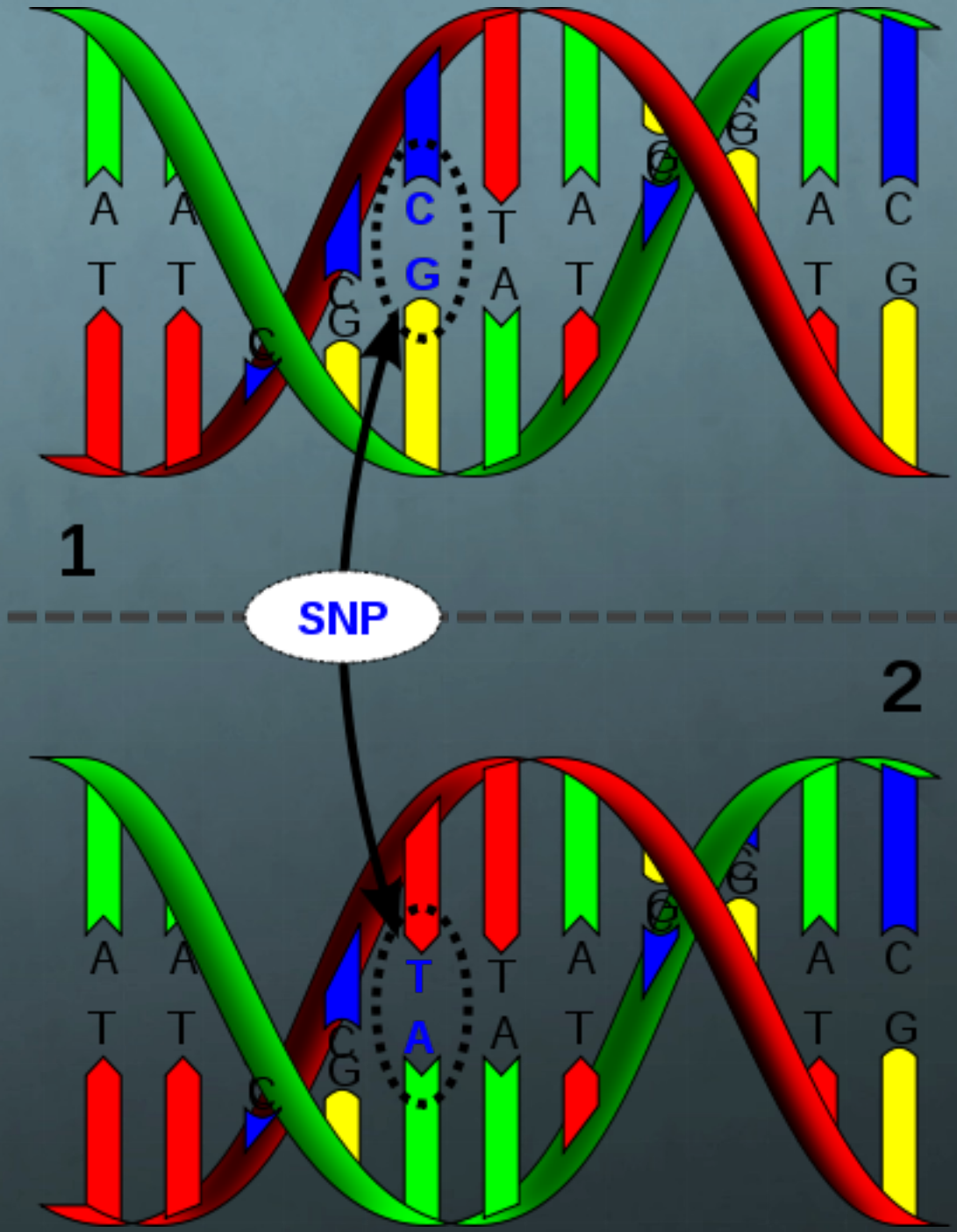
- Likelihood for a given mitotype is determined by the number of hits x in the FBI's CoDIS^{mt} database, of size n *(~5000)*. Thus we have probability $p = x/n$.
- For a Binomial distribution, we have the equations: $\mu = p$ (mean) and $\sigma = \sqrt{p(1-p)}$ (standard deviation).
- The 95% confidence interval is defined by the formula:

$$[\ \mu - 1.96\sigma/\sqrt{n},\ \mu + 1.96\sigma/\sqrt{n}\ ]$$

- Which reduces to an upper bound of $x/n + 2\sqrt{x(n-x)/n}$.
- If no database entries, we use: $1 - \alpha^{1/n}$ with $\alpha = 0.05$
- mtDNA is independent of STR, so can be multiplied.

SNP's





Single Nucleotide Polymorphisms, representing single base differences from the genome.

Useful for badly degraded samples.

Mutation rate is 100,000 times lower than STR's.

Occur on both nuclear and mitochondrial DNA

SNP's occur on average every 100 – 300 base pairs.

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SNP's

	Victim	PE	BF #01	BM #01	BU #02
Amel	CC	CC	-	TT	TT
65882	TC	TC	-	TC	TC
68532	-	TC	-	TC	CC
234217	CC	CC	-	TC	CC
231480	TT	TT	-	TT	TT
62059	-	-	-	TT	TT
56608	-	TC	-	TC	TC
61955	-	TT	-	TC	TC
220875	-	TT	-	TT	TT
58388	-	TT	-	TT	TT
63799	CC	CC	-	CC	TC
219561	TT	TT	-	TT	TT
60188	-	CC	-	CC	CC
182622	-	TC	-	TT	TT
85187	-	TC	-	TC	TC
212605	CC	CC	-	CC	CC
58091	-	TT	-	TT	TT
66026	-	TT	-	TC	TC
63836	-	CC	-	CC	CC
214373	TC	TC	-	TC	TT
238155	TT	TT	-	TT	TT

Two out of three SNP's involve replacing a C with a T.

Of these, there is a panel of 70 chosen by Orchid BioSciences in for each C and T are equally likely.

Many more SNP's are needed to reach STR likelihood levels.

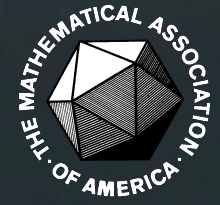
Can be used with Kinship Analysis

SNP Likelihood

- The Center for Genome Information concluded that although these 70 SNP's lack theoretical independence, allelic dependence was low enough for use in forensic identification.
- Conservative likelihoods can be calculated even without the assumption of equi-probability. Heterozygous SNP's have a minimum likelihood of 2:

$$f = 2pq = 2p(1-p) \leq 0.5 \ \forall p \in [0,1]; \ \therefore L = 1/f \geq 2$$

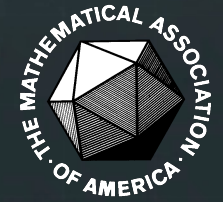
- Thus the minimum likelihood of a SNP profile containing n heterozygous alleles is 2^n.
- Average profile has ~35 heterozygous alleles, giving a minimum likelihood of $2^{35} \approx 10^{10}$.



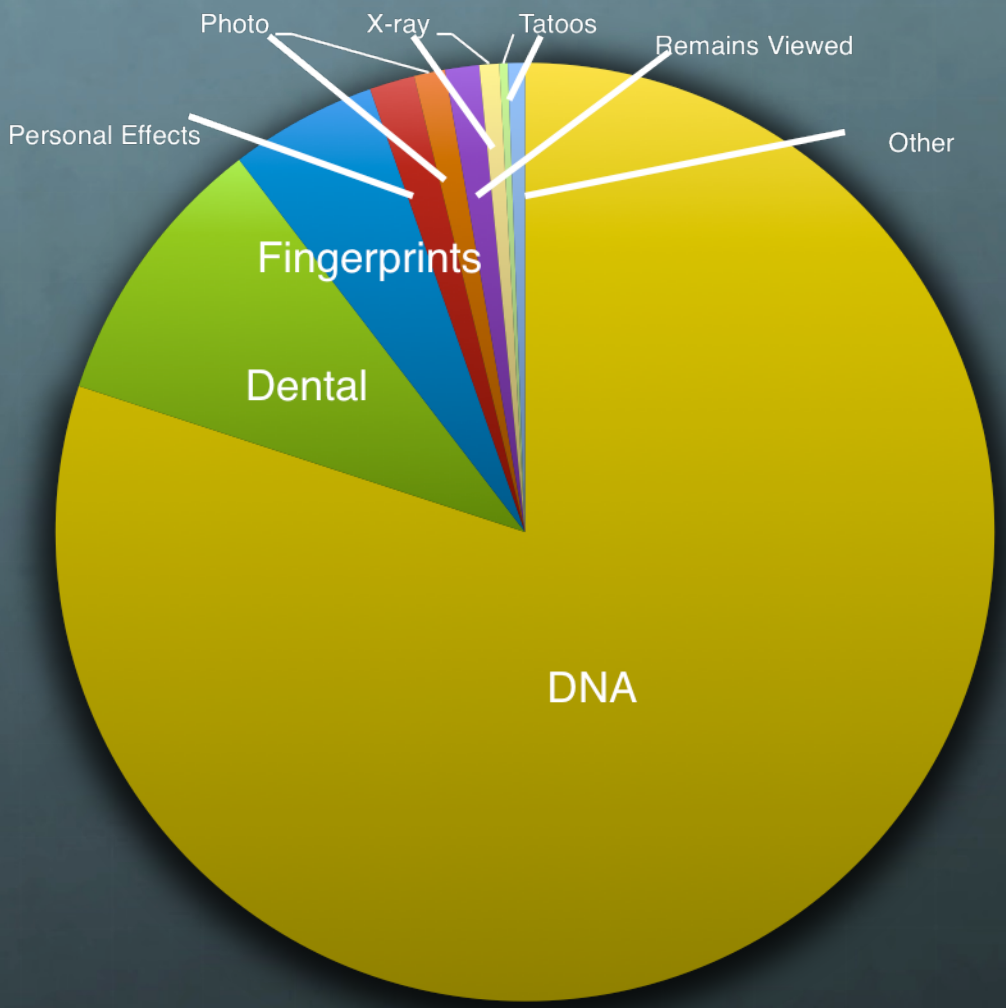
Summary

Statistics

- 2,606 victims (not including 10 hijackers)
- 21,905 total remains recovered
- 52,528 STR profiles
- 31,155 mtDNA profiles
- 16,938 SNP profiles
- Victims identified: 1,640 (59%)
- Hijackers identified: 3 (out of 10)
- Remains identified: 14,320 (65%)



Identification Modalities



Of all the victims identified by a single modality, DNA represented 81% of the identifications

Of identifications made with multiple modalities, 87% included DNA.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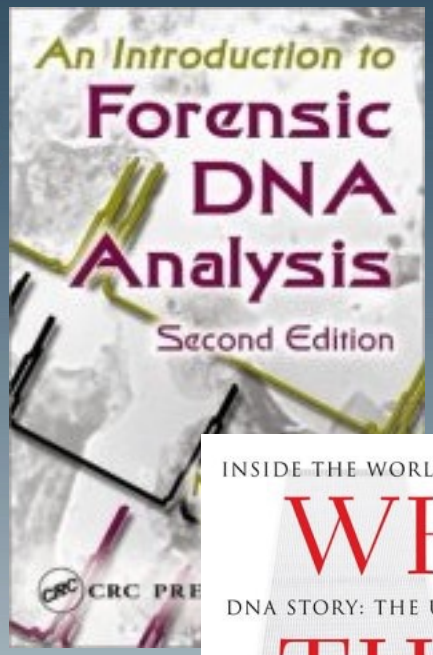
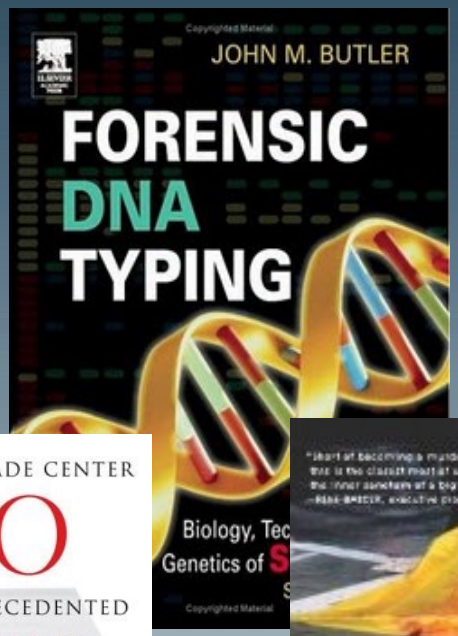
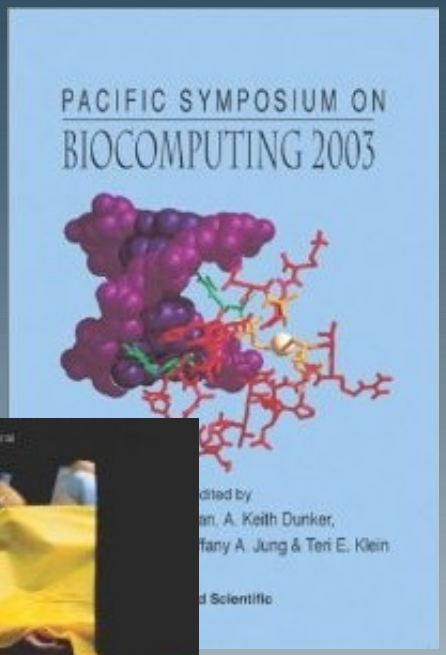
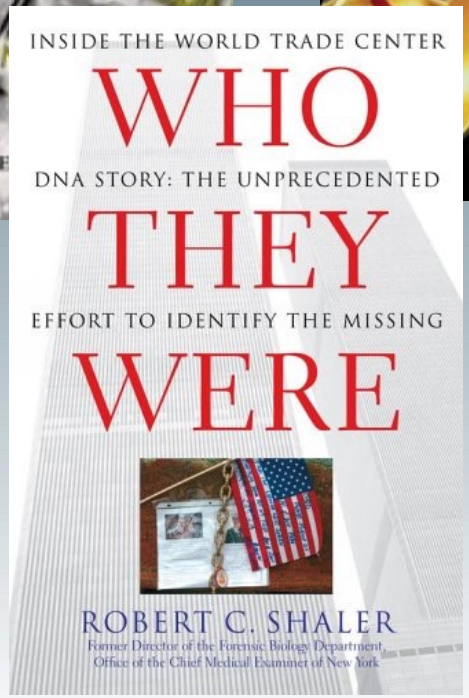
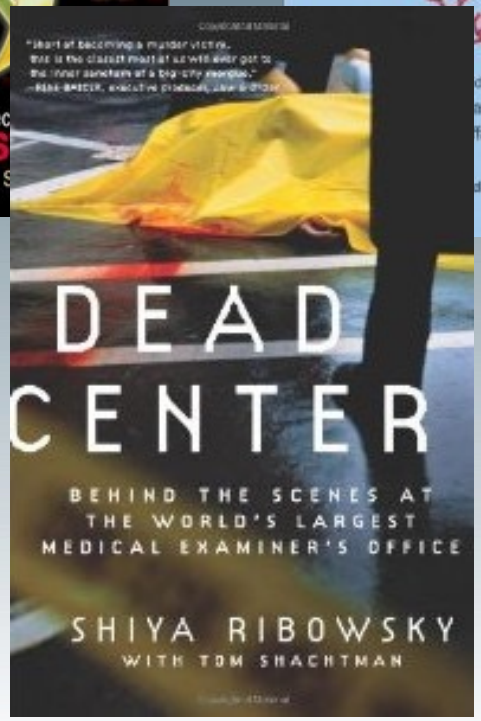
Further Reading











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More Information

Presentation related to a paper published at the Pacific Symposium on Bioomputing, 2003.

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An update to the Forensic Mathematics presentation given at St. Bonaventure University in October 2011.

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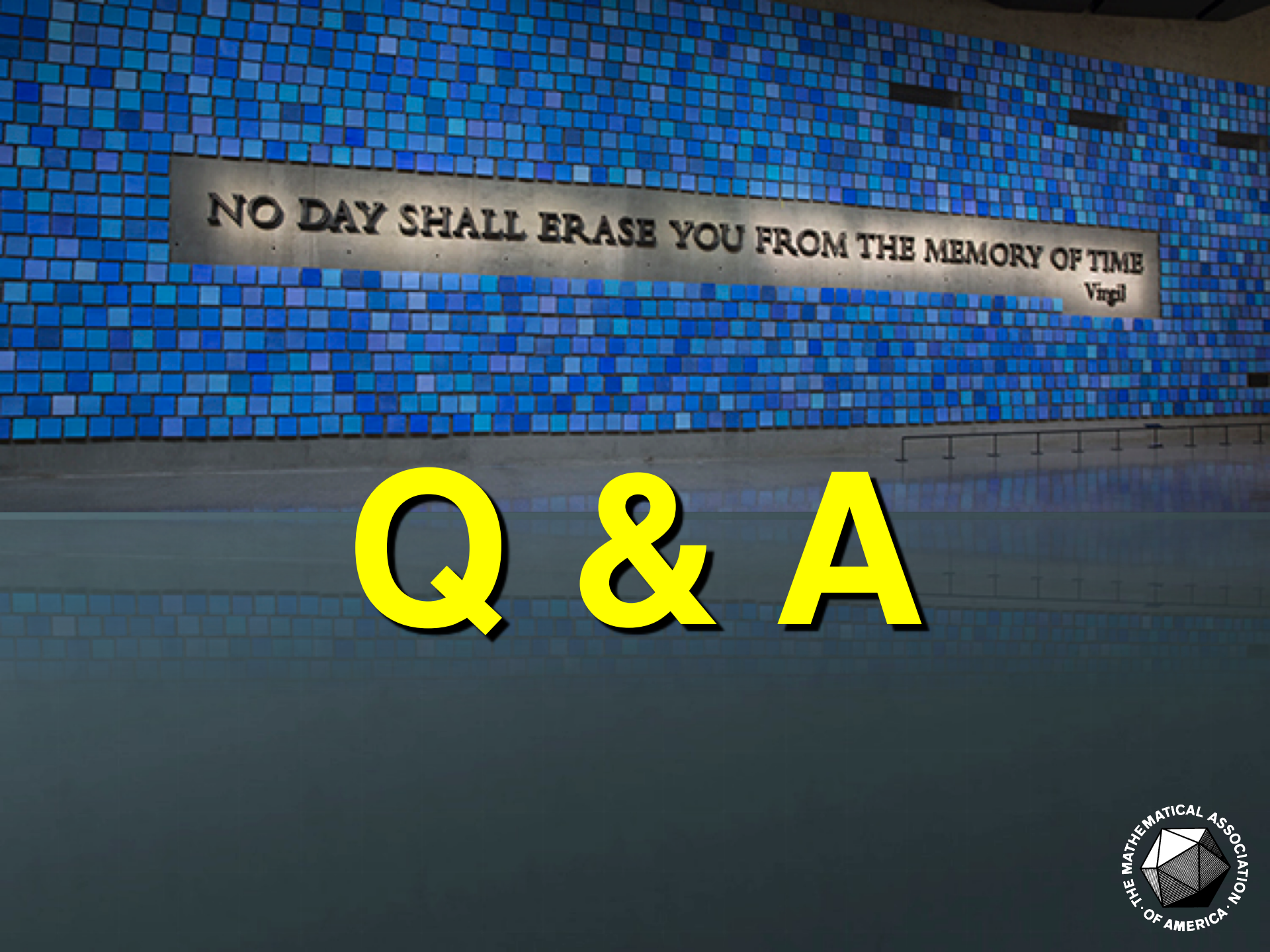

Q & A

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