hsc THE UNIVERSITY *of* NORTH TEXAS
HEALTH SCIENCE CENTER *at* FORT WORTH

CENTER FOR HUMAN IDENTIFICATION

Kinship and Data Analysis Panel
(WTC 9/11/2001)

THE CITY OF NEW YORK • OFFICE OF CHIEF MEDICAL EXAMINER

Event and One of the Responses

- September 11, 2001, ~2800 people were killed in terrorist attacks on the World Trade Center (WTC) in New York City.
    - Initially thought that 5000-10,000 perished
- The number of victims, the condition of their remains, and the duration of the recovery effort made the identification of the victims the most difficult ever undertaken by the forensic community in this country.
- NYSP and OCME requested support from NIJ/DOJ.
- KADAP was formed to provide recommendations to help identify those who perished in the WTC attack.
    - Think tank and beyond
    - Charged with assisting the OCME in the development of procedures, standards of evidence, and processes related to the DNA identification effort

Many Volunteered

- **Joan E. Bailey-Wilson, Ph.D.,** Inherited Disease Research Branch National Human Genome Research Institute National Institutes of Health Baltimore, MD
- **Jack Ballantyne, Ph.D.,** University of Central Florida Department of Chemistry Orlando, FL
- **Howard Baum, Ph.D.,** Office of the Chief Medical Examiner New York, NY
- **Frederick R. Bieber, Ph.D.,** Brigham and Women’s Hospital Harvard Medical School Boston, MA
- **Les Biesecker, M.D.,** National Human Genome Research Institute National Institutes of Health, Bethesda, MD
- **Charles Brenner, Ph.D.,** University of California, Berkeley School of Public Health Oakland, CA
- **Bruce Budowle, Ph.D.,** Federal Bureau of Investigation Laboratory Division Quantico, VA
- **John Butler, Ph.D.,** National Institute of Standards and Technology Gaithersburg, MD
- **George R. Carmody, Ph.D.,** Carleton University Ottawa, Ontario, Canada
- **P. Michael Conneally, Ph.D.,** Indiana University School of Medicine Department of Medical and Molecular Genetics Indianapolis, IN
- **W. Mark Dale, MBA,** Northeast Regional Forensic Institute University at Albany, State University of New York Albany, NY
- **Barry Duceman, Ph.D.,** New York State Police Forensic Investigation Center Albany, NY
- **Arthur J. Eisenberg, Ph.D.,** DNA Identity Laboratory Department of Pathology and Anatomy University of North Texas Health Science Center Fort Worth, TX

Many Volunteered

- **Kenneth K. Kidd, Ph.D.,** Yale University School of Medicine Department of Genetics New Haven, CT
- **Lisa Forman, Ph.D.,** National Institute of Justice, U.S. Department of Justice Washington, D.C.
- **Benoît Leclair, Ph.D.,** Myriad Genetic Laboratories, Inc. Salt Lake City, UT
- **Steve Niezgoda, MBA,** Technical Consultant, National Institute of Justice Niezgoda Consulting Bristow, VA
- **Elizabeth Pugh, Ph.D.,** Johns Hopkins University Center for Inherited Disease Research Baltimore, MD
- **Robert Shaler, Ph.D.,** Office of the Chief Medical Examiner New York, NY
- **Stephen Sherry, Ph.D.,** National Center for Biotechnology Information National Library of Medicine National Institutes of Health Bethesda, MD
- **Amanda Sozer, Ph.D.,** Technical Consultant, National Institute of Justice DNA Technology Consulting Services, LLC Fairfax Station, VA
- **Thomas J. Parsons, Ph.D.,** Department of Defense DNA Registry Armed Forces DNA Identification Laboratory Armed Forces Institute of Pathology Rockville, MD
- **Anne Walsh, Ph.D.,** Biggs Laboratory Wadsworth Center New York State Department of Health Albany, NY
- **Many other scientists and organizations also supported the effort**
- **Multidisciplinary - human geneticists, statisticians, bioethicists, forensic DNA scientists/managers, genetic researchers, information technologists, database managers, program managers,…**

Complex Set of Issues

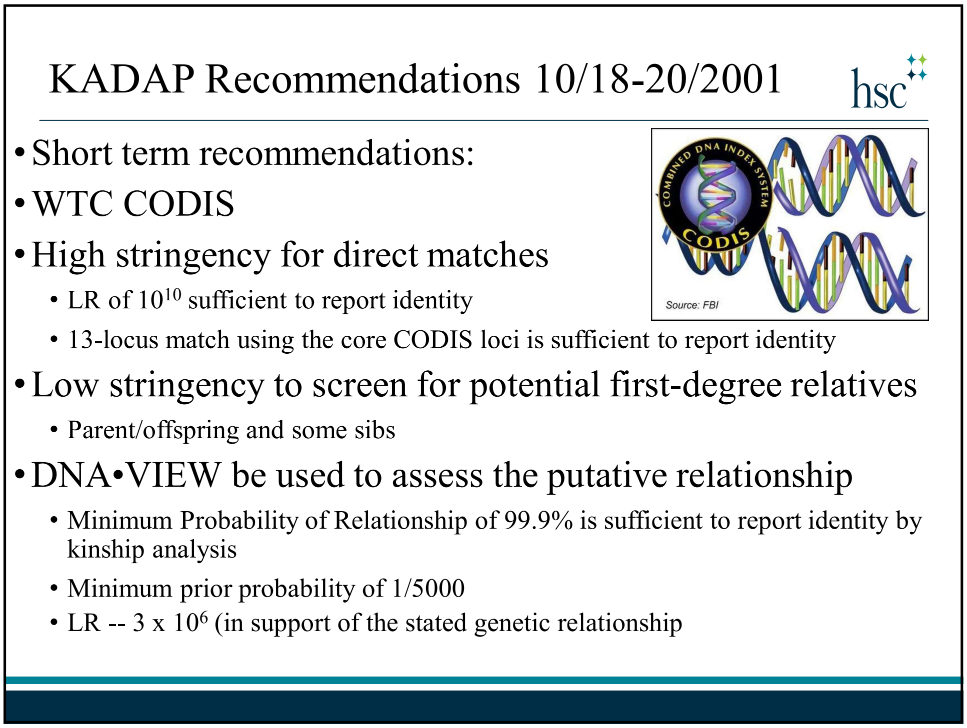
- Sample collection, preservation, shipping, and storage
- Tracking and chain of custody issues
- Clean, secure laboratory facilities
- Quality assurance and quality control practices
- Managing the work
- DNA extraction and typing
- Interpretation of results
- Automation
- New technologies
- Use of IT/software for sample tracking and data management
- Family and public education and communication
- Privacy issues

Similar to Missing Persons Id

- But with surge capacity!
- Methods of human identification
  - Anthropology
  - Odontology
  - Fingerprints
  - Prosthetics
  - Personal items
  - Tattoos
  - etc
- Circumstances indicated that DNA would be the primary tool

Identifications

- OCME – remains; NYSP – reference samples
- Laboratory scientists do not make identifications
- MEs/Coronors/JPs make identifications
  - Statutory responsibility and authority to identify the deceased and issue a death certificate
- Associations were made based on DNA with some threshold criteria
  - Some meta data – i.e., missing person manifest
- Consequences of a misidentification can have emotional and legal ramifications well beyond a specific case
- Huge challenge was that NY decided to type all (possible) bone fragments

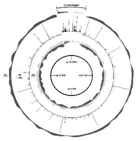


KADAP Recommendations 10/18-20/2001

Source: FBI

- Short term recommendations:
- WTC CODIS
- High stringency for direct matches
  - LR of 10^{10} sufficient to report identity
  - 13-locus match using the core CODIS loci is sufficient to report identity
- Low stringency to screen for potential first-degree relatives
  - Parent/offspring and some sibs
- DNA•VIEW be used to assess the putative relationship
  - Minimum Probability of Relationship of 99.9% is sufficient to report identity by kinship analysis
  - Minimum prior probability of 1/5000
  - LR -- 3×10^6 (in support of the stated genetic relationship

KADAP Recommendations 10/18-20/2001



- mtDNA testing
  - Concerns that focus on mtDNA would dilute the effort to yield sufficient STR loci in what were likely to be difficult samples
  - Might hinder the identification process by adding less powerful methods of identification before all efforts to reveal unique identities had been exhausted.
    - mtDNA data are insufficient to resolve identity, but….
  - Should be a last resort after additional test reanalysis
  - Explored on a case-by-case basis
  - Should be performed on all maternal lineage relative's appropriate samples
  - Should not be performed on personal effect samples until other appropriate approaches have been considered

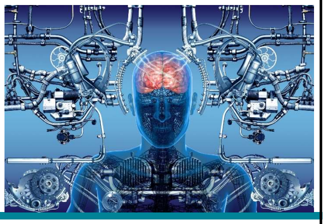
KADAP Recommendations 11/20/2001

- Data transfer between the NYSP and the OCME required new information technology infrastructure and support
  - Critical to the success of the WTC identification project that the OCME and NYSP share rapid access to the same data sets via immediate installation of a T1 line
  - A primary data repository allowing shared access to analysts outside OCME was set up on a secure server at the National Center for Biotechnology Information, in Bethesda, Maryland
- Recognize that requests for prioritization of analyses of particular samples have significant implications for the overall process
  - Impede the overall progress of identification
  - Increase the chances of analytical or interpretive errors
  - Increase costs
- Minimize requests for prioritization

KADAP Recommendations 2/21-22/2002

- Collaboration and information sharing is a critical component to maximum identification throughput
- Information management and software integration
- Additional resources such as hardware, software, expert systems, and personnel.

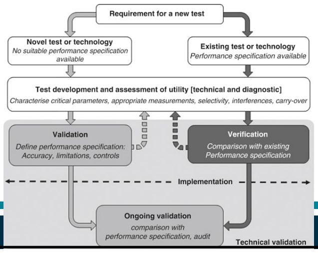
- OCME and NYSP each hire/contract two (2) additional information technology FTEs so that present staff experienced in the current process can be solely dedicated to the WTC effort
- OCME and NYSP each hire/contract five (5) additional forensic analyst FTEs to be solely dedicated to the WTC effort



KADAP Recommendations 2/21-22/2002

Validation and Quality Control:

- Documented validation protocols for software programs and interfaces
- Dedicated personnel and equipment for validation
- Objective unbiased peer review process to implement valid systems
- Appropriate test genetic data should be integrated into the WTC CODIS for efficient validation of all software
- Procedures to confirm matches are appropriate
- The probability of miscalling alleles that would lead to false inclusions is so small
  - not necessary to review EPGs previously reviewed by vendor laboratories for uncomplicated STR cases that meet previous recommendations for LRs

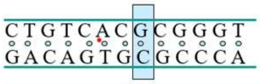


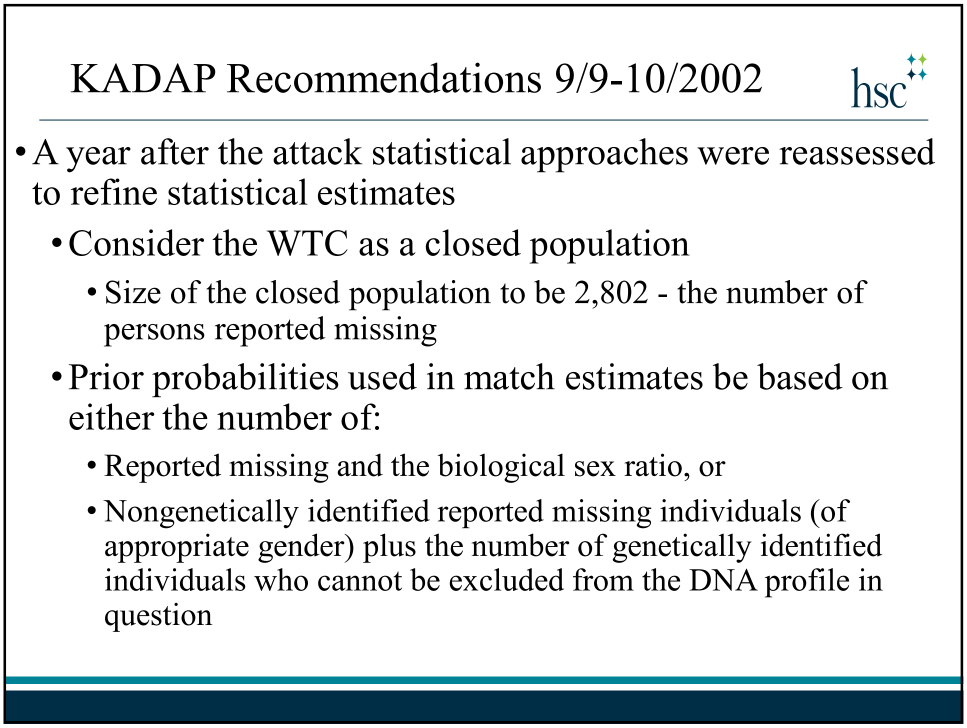
KADAP Recommendations 4/24-25/2002

- A mechanism of data synchronization should be created
  - NCBI [National Center for Biotechnology Information] should host the secure FTP resource
- The Forensic Biology Unit of the OCME needs Internet access with adequate bandwidth and tools for secure access
- Cases involving difficult kinship interpretations, including such things as mutations, should be reviewed by members of the AABB Parentage Testing Community to recommend disposition to OCME
- Kinship used to confirm a personal effect match should be accepted at a Probability of Relationship of 99.9% using a Prior Probability of 0.5.
- Compromised samples can be considered associated with samples that were previously matched through DNA if the LR of shared loci is $>10^8$.
  - Equivalent to one divided by the random match probability of the shared loci between the two profiles.

KADAP Recommendations 7/15-16/2002

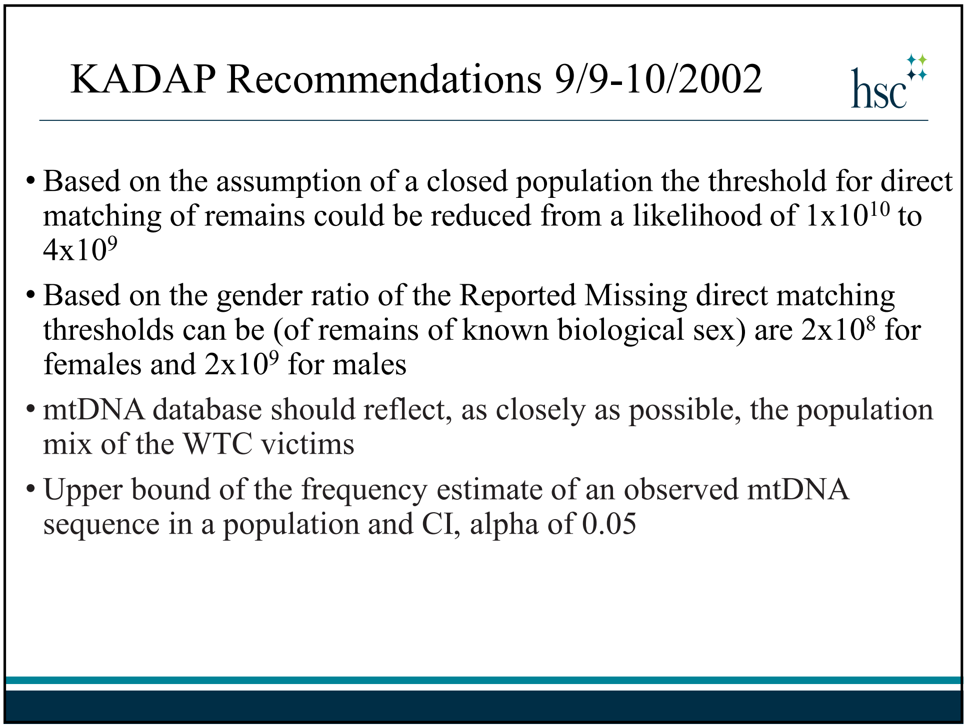
- Proceed with the pilot use of the ORCHID/Genescreen SNP panels on WTC samples
  - 70 SNP loci some of which are closely linked with each other and with the CODIS STR loci
- Linkage (LD) between SNPs can impact kinship estimates
- A KADAP subcommittee on SNPs met on 12 July 2002 in Washington, D.C.
  - Supported the potential pending additional studies
- Sample consumption issues must be appropriately addressed before SNP analysis proceeds
- The KADAP SNP subcommittee pursue further statistical analysis of existing SNP data





KADAP Recommendations 9/9-10/2002

- A year after the attack statistical approaches were reassessed to refine statistical estimates
  - Consider the WTC as a closed population
    - Size of the closed population to be 2,802 - the number of persons reported missing
  - Prior probabilities used in match estimates be based on either the number of:
    - Reported missing and the biological sex ratio, or
    - Nongenetically identified reported missing individuals (of appropriate gender) plus the number of genetically identified individuals who cannot be excluded from the DNA profile in question

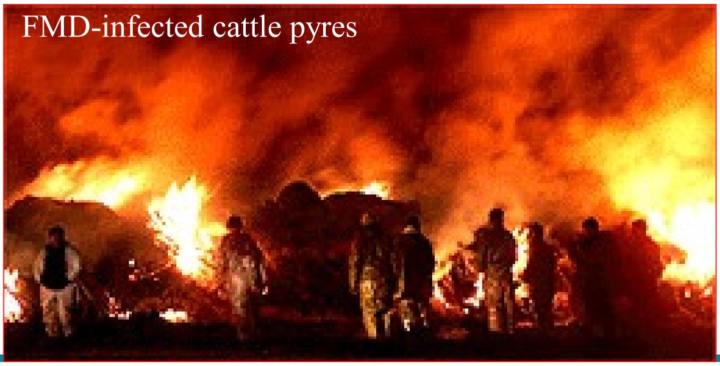


KADAP Recommendations 9/9-10/2002

- Based on the assumption of a closed population the threshold for direct matching of remains could be reduced from a likelihood of $1x10^{10}$ to $4x10^{9}$
- Based on the gender ratio of the Reported Missing direct matching thresholds can be (of remains of known biological sex) are $2x10^{8}$ for females and $2x10^{9}$ for males
- mtDNA database should reflect, as closely as possible, the population mix of the WTC victims
- Upper bound of the frequency estimate of an observed mtDNA sequence in a population and CI, alpha of 0.05

Planning ahead is better than at the moment!

- We may not make things better, but we could make it much worse!
- Better to be scared by the improbable possibility, than to be unprepared for the catastrophic reality’
    - Congressman Christopher Shays (R-CT), Chairman of the Subcommittee on National Security, Veterans Affairs, and International Relations


FMD-infected cattle pyres

Prepare/Plan/Execute

- Before a mass fatality incident occurs, laboratories should develop a plan for all aspects
    - Necessary but unrealistic
- Chain of custody - many moving parts
    - Intake, storage, reference samples, DRS, remains, degradation, analysis, communication to the public, information about processes, collection of FRS, IT, etc
- Create a repository of information, protocols, validation data, etc

Other Challenges

- Determining the minimum fragment size
  - Typically, 1 to 10 centimeters
- Need to not consume entirely
- Minimum sample affects outcome
- Three criteria:
  - Maximizing the probability that all victims are identified;
  - Recognizing the emotional needs of the victims' families and friends; and
  - Providing forensically relevant information
- Closed or Open population

Science 310:1122-1123, 2005

POLICY FORUM

EPIDEMIOLOGY

DNA Identifications After the 9/11 World Trade Center Attack

Leslie G. Biesecker,* Joan E. Bailey-Wilson, Jack Ballantyne, Howard Baum, Frederick R. Bieber, Charles Brenner, Bruce Budowle, John M. Butler, George Carmody, P. Michael Conneally, Barry Duceman, Arthur Eisenberg, Lisa Forman, Kenneth K. Kidd, Benoit Leclair, Steven Niezgoda, Thomas J. Parsons, Elizabeth Pugh, Robert Shaler, Stephen T. Sherry, Amanda Sozer, Anne Walsh

Before the September 11, 2001, World Trade Center (WTC) attack, the use of DNA profiling for victim identifica-

from a few nearly complete bodies to multitudes of tiny fragments of charred bone, often difficult to distinguish from inorganic mate-

- Technical improvements led to better DNA yields generating useful DNA profiles in ~40% of samples (when standard method was not successful).
- Two-stage drilling process to isolate uncontaminated bone powder and modifications to the wash incubations, buffer concentrations, and elution times for DNA extractions.
- ~ 20% of the DNA identifications were made solely from mini-STRs.
- SNP analysis alone identified about 10 individuals
- 10 more identifications made with SNPs supplementing partial STR profiles.
- Additional identifications were made when mtDNA typing results were used to screen for potential matches, followed by DNA re-extraction and mini-STR retyping.
- No DNA-based identifications were accomplished by mtDNA alone.

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- OCME and New York State personnel
- KADAP
- Federal and State Agencies
- Academia
- Vendors
- All others who contributed
- Promega Corporation
- **Most importantly – the Victims!**