



# NIJ and CHORI – Collaboration with SoftGenetics® and California Department of Justice

## Customizing NextGENe Software for Forensic Applications

Moving Knowledge from Research to Impact



*“Mitochondrial analysis holds significant potential for the forensic community and represents a great market opportunity for SoftGenetics®. As part of Dr. Calloway’s NIJ funding, we were able to initiate a collaboration to customize the NextGENe® software for forensic use.”*

John Fosnacht, Co-Founder/VP Sales & Marketing  
SoftGenetics®vv

### Synopsis of Problem and Solution

Degraded and mixed DNA samples are often encountered in forensic cases and pose interpretation challenges. Mitochondrial DNA (mtDNA) markers are ideal targets for analyzing these samples, as the high copy number of mtDNA and the large number of nucleotide polymorphisms can allow for discrimination among samples. A software program that could analyze the aspects of mtDNA significant to forensics could enable more accurate and rapid DNA analysis.

Dr. Cassandra Calloway of Children’s Hospital Oakland Research Institute (CHORI) recognized that NextGENe software, from SoftGenetics, offered a user-friendly, versatile program that could handle analysis of data generated by many different Massively Parallel Sequencing (MPS) platforms. Dr. Calloway led the research effort with SoftGenetics® to modify NextGENe®, commercial software tool to improve analysis of mtDNA in a forensic context.

### NIJ-Funded Research

Through a grant (2010-DN-BX-K141) from the National Institute of Justice (NIJ), Dr. Calloway and colleagues at CHORI collaborated with SoftGenetics to improve the company’s commercially available NextGENe® MPS data analysis software for analysis of mtDNA and STR data. As a direct result, the current commercially available version of the NextGENe® software (v2.4.1) contains alignment analysis modules for STR’s, mtDNA amplicon, and the whole mitochondrial genome.

### Benefits

- ▶ Drives progress toward incorporation of mitochondrial sequence analysis into the US criminal justice system.
- ▶ Supports missing person identification, analysis of degraded samples, and forensic profiling.
- ▶ Adapts easily into current workflow in labs with MPS equipment.
- ▶ Enables quick, easy analysis of the data in comprehensive reports.

## The Future

Dr. Calloway will continue to work with SoftGenetics® to further customize the software for forensic reporting with the input of CADOJ-BFS. The goals of the project are to refine forensic reporting tools and features for mtDNA analysis, as well as to establish appropriate filter parameters and thresholds for base calling for STR and whole mitochondrial genome MPS analysis.

*“We are excited to participate in the evaluation and implementation of a mitochondrial DNA analysis tool that can allow Massively Parallel Sequencing to be used in forensic identification.”*

Daniela Cuenca, MS Criminalist  
 California Department of Justice

## More Information

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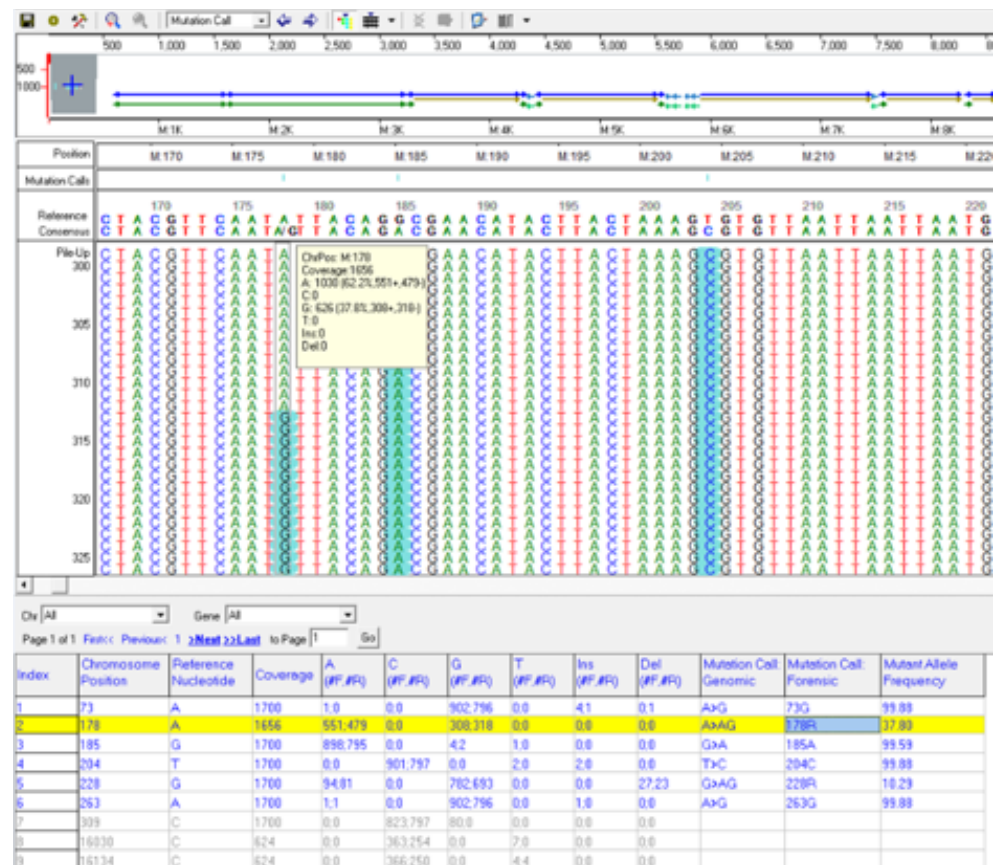
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Through grants from the National Institute of Justice Award 2010-DN-BX-K141 Resolution of DNA Mixtures and Analysis of Degraded DNA Using the 454 DNA Sequencing Technology

## Bringing Research to Practice

The Bureau of Forensic Services in the California Department of Justice (CADOJ-BFS) is evaluating a whole-genome mitochondrial DNA MPS assay to increase the discrimination potential of mitochondrial analysis and to increase casework productivity, particularly for implementation in the CADOJ-BFS Missing Persons DNA Program. An important component of the evaluation and ultimate implementation of such an assay is a robust analysis software tool that can accurately align MPS reads to the circular mitochondrial reference genome, filter and trim low quality sequence reads to improve accuracy, and report DNA sequence information in a format that follows approved forensic nomenclature for the mitochondrial genome. Dr. Calloway's work to empower SoftGenetics NextGENE® accomplishes these requirements with the added benefit of user friendliness.



Alignment window with mutation report. This feature allows the user to either confirm or delete a mutation after determining its authenticity.

*“Software for mixture analysis in forensic applications was not available when we initiated this grant. Our collaboration with SoftGenetics® and California DOJ is enabling its use in the identification of individuals in complex mixtures as well as degraded samples.”*

Cassandra Calloway, PhD, Assistant Scientist  
 Children's Hospital Oakland Research Institute