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Author(s): Richard E. Green

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COVER PAGE

2020-DQ-BX-0014

Evaluation of nuclear DNA from rootless hair for forensic purposes

PI: Richard E. Green – Professor of Biomolecular Engineering, University of California, Santa Cruz

ed@soe.ucsc.edu
1156 High Street
Santa Cruz CA 95064
+1 831-502-7394

University of California, Santa Cruz

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\$747,384

SUMMARY OF THE PROJECT

The major goals of this project were to determine the suitability and limitations of DNA from rootless hair for forensic purposes. From a panel of volunteers, we collected saliva and head and pubic hair. We sought to assess the range and characteristics of DNA in hair, develop a procedure for efficient recovery of DNA from hair, and develop bioinformatic procedures for using the DNA information for forensic analyses including finding genetic relatives. We also aimed to develop a low-cost method of performing hybridization capture of DNA library molecules that would aid genotype inference and reduce the overall cost of this approach.

The design of the project involved first enrolling 50 anonymous volunteers to submit a saliva and up to two hair samples. DNA was collected from the saliva for deep sequencing and for microarray genotype analysis. DNA was also collected from each individual hair. We analyzed the characteristics of this DNA including its length distribution, the range of recoverable DNA per hair, and its utility in inferring accurate genotypes. The DNA from the saliva sample was used to assess the accuracy of the genotype inference.

We need not speculate on the applicability of this research. During the course of this project, this technology has been commercialized and is in active use in case work.

PARTICIPANTS AND OTHER COLLABORATING ORGANIZATIONS

There were no funded participants or collaborating organizations.

CHANGES IN APPROACH FROM ORIGINAL DESIGN

There were no notable changes from the original design, i.e., all project goals were met. However, our work on this project stimulated development of a bioinformatic method not described in the original proposal. This approach allows one to compare DNA sequence information from a limited sample, such as a single, rootless hair to a known genotype file. This method has been published and is being actively used in casework.

OUTCOMES

We completed all research activities as described in the proposal. The findings of this research project resulted in the following findings:

Five centimeters of rootless hair yielded in 77 of 80 hairs, enough DNA data from 300 M sequence reads to achieve 1-fold average genome coverage.

1-fold average genome coverage is sufficient DNA information to accurately infer genotypes (>99% allele call accuracy) to perform genetic genealogy analysis. Specifically, the closest genetic relative in the GEDMatch database is identified from hair genotypes as was identified from deep sequencing of saliva DNA.

ARTIFACTS

Publications

1. Sundararaman B, Vershinina A.O, Hershauer S, Kapp J.D, Dunn S, Shapiro B and Green R.E (2023)
A method to generate capture baits for targeted sequencing
Nucleic Acids Research, 2023 Jun; gkad460, <https://doi.org/10.1093/nar/gkad460>.
2. Nguyen R, Kapp JD, Sacco S, Myers SP, Green RE (2023)
A computational approach for positive genetic identification and relatedness detection from low-coverage shotgun sequencing data
Journal of Heredity, 2023. doi:10.1093/jhered/esad041.
3. Kapp JK, Wanket C, Nguyen R, Tzadikaria T, Shapiro B, and Green RE (in prep for *Science*)
Rootless hair as a reliable source of forensic genetic information

Conference presentations

"A computational approach for positive genetic identification and relatedness detection from low-coverage shotgun sequencing data." National Institute of Justice 2023 National Research Conference, May 23 – 25, 2023 Arlington, VA. [Remy Nguyen presenting]

"A computational approach for positive genetic identification and relatedness detection from low-coverage shotgun sequencing data." 2023 NHGRI Research Training and Career Development Annual Meeting, April 2 – 4, 2023, Salt Lake City, UT. [Remy Nguyen presenting]

Probes for Targeted Genotyping by Sequencing of Rootless Hair for Investigative Genetic Genealogy, National Institute of Justice, National Research Conference 2023, May 22 - 25, 2023 in Arlington, VA. [Balaji Sundararman presenting]

CNERs: Circular Nucleic acid Enrichment Reagents for targeted sequencing (Oral presentation). Plant and Animal Genome Conference (PAG 30), Jan 12 - 18, 2023 at the The Town & Country Resort and Conference Center, San Diego, CA. [Balaji Sundararman presenting]

Probes for targeted sequencing to detect and genotype *Toxoplasma gondii* oocysts in environmental samples (Oral presentation). International Congress on Toxoplasmosis and *Toxoplasma gondii* Research, May 22 – May 26, 2022 at The Mission Inn, Riverside, CA. [Balaji Sundararman presenting]

"Rootless hair DNA for forensic purposes" (Invited Oral Presentation). Considerations for court admissibility of SNP-based identity verification meeting Organized by Verogen and FBI. San Diego, CA, May 15, 2023 [Ed Green presenting]

"Evaluation of DNA from rootless hair for forensic identification" (Invited main session presentation) Sept. 22, 2022 at Washington, D.C. ISHI [Ed Green Presenting]

"DNA from rootless hair for forensic purposes" (Invited main session presentation) October 1, 2021 at Orlando, FL ISHI 2021 [Ed Green Presenting]

"DNA-based forensics" (Invited department seminar) March 30, 2023 at Fresno State University [Ed Green Presenting]

"New developments in DNA based forensics" (Invited department seminar) March 2, 2021 at Morehouse College, Atlanta, GA (via zoom) [Ed Green presenting]

Technologies

Methods for genetic identification and relatedness detection

United States Patent Application: US20230105167A1

Inventors: Remy Nguyen and Richard E. Green

IBDgem software implementation of this invention is available via github at this URL:
<https://github.com/Paleogenomics/IBDgem>

Methods of producing target capture nucleic acids

United States Patent Application: US20230348955A1

Inventors: Balaji Sundararaman and Richard E. Green

Datasets

Dissemination activities

2023 (several sessions throughout the year) - Participated as an invited panelist in the ForenSeq Delphi Panel Focus Group organized by Baylor University to develop best practices and legal guidelines for Forensic Genetic Genealogy

Invited speaker at Missing and Unidentified Persons Conference – California Department of Justice, Garden Grove, California. May 4, 2022

Invited external guest speaker for SWGDAM meetings on March 29, 2022 and January 11, 2021.