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Thank you for submitting an abstract to the **ASHG 2019 Annual Meeting** in Houston, Texas, October 15-19! **This is confirmation that your abstract has been successfully submitted. You will receive a separate emailed confirmation/receipt.**

Your Abstract Control Number: 1920698
Your Abstract Title: Association of Ancient Adaptive Mitochondrial DNA Haplogroups with Type 1 Diabetes

Please see below for full details of your submission. You may retrieve your **submitted abstract** at any time.

If Needed: Revising or Withdrawing Your Abstract

You can go to **ASHG Portal** to revise or withdraw your abstract. All revisions and withdrawals must be completed by June 10, 2019 at 11:59 pm U.S. Eastern Time.

Please note: if you need to change the first author of your abstract, you must first withdraw your abstract and then submit a new abstract with the new name. Abstract submissions are due June 6, 2019 at 11:59 pm U.S. Eastern Time.

Your Next Steps: Registration, Travel, and Hotel

Registration: Please **register** as soon as possible and take advantage of discounted rates. Do not wait until you receive your program confirmation before registering. Submission of an abstract does NOT automatically register you for the meeting.

International attendees: We urge those who require visas to register now and begin the visa application process, which can take up to 6 months. **Further details on this process** are available online.

- Download your **customized letter of invitation** to ASHG 2019 Annual Meeting, which may be needed for your visa application.

Hotel reservations: Please make your **hotel reservations** as soon as possible to ensure you receive the hotel of your choice. If you need to change your arrival/departure dates, you will be able to do so.

Abstract Control Number: 1920698

Abstract was **submitted** on 2019-06-04 12:43:14 (U.S. east coast time)

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Presenting Author Status (when attend the meeting in October): Undergraduate Student

Trainee Research Award Application: None

Presentation Preference: Plenary/Platform talk or Poster

Abstract Topic Choice:
Main Topic Choice: 2. Complex Traits and Polygenic Disorders
Subtopic: D. Diabetes, Obesity, and Metabolic Syndromes

Keywords: 45 123

Track (that my abstract best fits in): Basic

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Title:
Association of Ancient Adaptive Mitochondrial DNA Haplogroups with Type 1 Diabetes

Abstract:

The prevalence of type 1 diabetes (T1D) varies significantly across human populations, with the highest prevalence seen in Caucasian populations. Even among European populations, there are conspicuous differences with the highest prevalence in Finnish population(Patterson et al. 2012). To date, more than 70 T1D loci have been identified, however, the population difference of T1D susceptibility is still largely unknown. It has been well recognized that different ancient adaptive mitochondrial DNA (mtDNA) haplogroups have significantly different functional performance, thus have undergone extensive selection in the evolution of human populations(Tranah et al. 2011). In our study, we examined the association of mtDNA haplogroups with T1D in our T1D GWAS datasets. Population substructures of the sample were analyzed by principal component analysis (PCA) using PLINK1.9, and genetic outliers were removed. Altogether, 979 cases and 2009 controls of European ancestry were used for the association test. We determined the mtDNA haplogroups by the phylogenetic tree based software HaploGrep 2(Weissensteiner et al. 2016), based on 163 mtDNA single nucleotide variants (SNV) genotyped by the Illumina HumanHap550 arrays. We tested 23 mtDNA haplogroups (A, B, C, D, F, G, H, I, J, K, L, M, N, P, Q, R, T, U, V, W, X, Z, and Undetermined) for genetic association by logistic regression adjusted for the first two principal components of the PCA for population stratification, with statistical significance Bonferroni correction level at $\alpha=0.05/23=2.17x10^{-3}$. One haplogroup was significant, i.e. Haplogroup V, OR (95%CI)= 3.200 (1.680, 6.097), $P=^{4}.05x10^{-4}$. Another haplogroup demonstrated nominal significance without Bonferroni correction, i.e. Haplogroup K OR (95%CI)= 0.698(0.528, 0.923), P=0.012. Haplogroup V is most commonly seen in northern Scandinavian, which explains its association with the risk of T1D, but not simply marking Scandinavians (before correction OR (95%CI)=3.627(1.912, 6.879), $P=2.55\ x10^{-5}$). In addition, we identified possible gender effect of Haplogroup K on the age-of-onset of T1D with nominal significance (P=0.048), i.e. later age-of-onset in males [mean of males vs females difference (95%CI)=3.45 (0.033, 6.86)], which as well as the decreased risk of T1D might be explained by its function as an “uncoupling genome” and less oxidative damage, but the sexual dimorphism(Ventura-Clapier et al. 2017) warrants further investigations.

Publication Status:

The work outlined in this abstract Has not been published elsewhere.
The work outlined in this abstract Has not been accepted for future publication.

Relationship(s) to Disclose: No
I intend to discuss unlabeled/off-label use of FDA-approved product(s): No
I intend to discuss investigational product(s) (not FDA-approved): No

If you have included a table in your abstract, it may not appear to be accurately formatted on this confirmation (due to possible wrapping). It is important that you email a copy of any table, formatted as you wish it to appear, to **ashgmeetings@ashg.org** by June 10. Be sure to include your name as first author, the title of your abstract, the abstract control number, and your email address.

What Happens Now: Abstract Review and Programming

After the submission deadline, all abstracts will be reviewed by the 2019 Program Committee and independent reviewers. Program assignment for platform and poster presentations will be available in mid-August. The Annual Meeting website will include searchable scientific abstracts along with an individualized meeting itinerary planner.

- **Abstract FAQs**
- **How Abstracts are Reviewed, Programmed, and Published**

Later: Submit Your Work to *AJHG*

Please consider submitting your work to ASHG's journal, ***The American Journal of Human Genetics***. *AJHG* welcomes submissions of articles and reports on timely subjects concerning all aspects of the genetics of humans. *AJHG* provides rapid publication in a Society Journal with a long-standing reputation for high quality. Publication by ASHG members is free of charge, and trainee member first authors are eligible for the annual **Cotterman Awards**.

Additional Information

- **ASHG 2019 Annual Meeting Website**
- **ASHG Web Portal — renew your membership, change your address, view previous meeting webcasts, and more!**
- Questions or comments? Email **ashgmeetings@ashg.org**.