RE: Manuscript for Review - MESA Genetics P&P

MESA Genetics PP <genpp@uw.edu>

Tue 9/21/2021 1:30 PM

To: MESA Genetics PP <genpp@uw.edu>; Stephanie Battle <sbattl10@jhmi.edu>; MESA Genetics PP <genpp@uw.edu>

Cc: Dan Arking <arking@jhmi.edu>

External Email - Use Caution

Dear Stephanie,

Congratulations! The MESA Genetics P&P Committee reviewed and approved – with comments – the TOPMed proposal for G 845, A Bioinformatics Pipeline for Estimating Mitochondria DNA Copy Number and Heteroplasmy Levels from WGS Data. Please see the attached memo and let me know if you have any questions. Good luck with journal submission, and please let me know when the paper is accepted and published.

Thank you, Ellen

From: MESA Genetics PP < genpp@uw.edu> Sent: Tuesday, September 7, 2021 11:11 AM

To: Stephanie Battle <sbattl10@jhmi.edu>; MESA Genetics PP <qenpp@uw.edu>

Cc: Dan Arking <arking@jhmi.edu>

Subject: RE: Manuscript for Review - MESA Genetics P&P

Great! Thank you, Stephanie.

Best. Ellen

From: Stephanie Battle <sbattl10@jhmi.edu> Sent: Tuesday, September 7, 2021 10:34 AM To: MESA Genetics PP <qenpp@uw.edu>

Cc: Dan Arking <arking@jhmi.edu>

Subject: Re: Manuscript for Review - MESA Genetics P&P

Hi Ellen.

I attached a copy of the approved proposal.

Thanks so much, Stephanie

From: MESA Genetics PP < genpp@uw.edu > Sent: Tuesday. September 7, 2021 2:49 AM To: Stephanie Battle <sbattl10@jhmi.edu>

Cc: Dan Arking <arking@jhmi.edu>; MESA Genetics PP <genpp@uw.edu>

Subject: RE: Manuscript for Review - MESA Genetics P&P

External Email - Use Caution

Hi Dr. Battle,

Thank you for submitting the TOPMed pen draft A Bioinformatics Pipeline for Estimating Mitochondria DNA Copy Number and Heteroplasmy Levels from WGS Data. Would you please send me a copy of the TOPMed-approved proposal that accompanies the paper? The MESA Genetics P&P Committee requires this prior to reviewing TOPMed papers.

Thank you very much, Ellen

From: Karen S. Hansen < hansenk3@uw.edu> Sent: Thursday, September 2, 2021 1:42 PM To: Stephanie Battle <sbattl10@jhmi.edu>

Cc: Dan Arking <arking@jhmi.edu>; MESA Genetics PP <genpp@uw.edu>

Subject: RE: Manuscript for Review - MESA Genetics P&P

Hi Dr. Battle,

This appears to be a paper that goes under the MESA Genetics P&P Committee review. I just work with the main MESA P&P submissions.

I've copied Ellen Terry via the MESA Genetics P&P email and I'm sure that she will get back to you when she is able. I believe that Ellen is typically working Monday – Wednesday, so she may not get back to you until next week.

Thank you, Karen

Karen S. Hansen MESA P&P Program Coordinator Web Information Specialist Collaborative Health Studies Coordinating Center University of Washington, Box 354922 6200 NE 74th Street, Building 29, Suite 210 Seattle, WA 98115

Phone: 206-897-1939 Email: hansenk3@uw.edu

From: Stephanie Battle <sbattl10@jhmi.edu> Sent: Thursday, September 2, 2021 12:07 PM To: Karen S. Hansen < hansenk3@uw.edu>

Cc: Dan Arking <arking@jhmi.edu> Subject: Re: Manuscript for Review

My apologies, I provided the wrong manuscript number. The correct number is #3129.

Thank you, Stephanie

From: Stephanie Battle

Sent: Thursday, September 2, 2021 2:52 PM

To: hansenk3@uw.edu <hansenk3@uw.edu>

Cc: Dan Arking <arking@jhmi.edu> Subject: Manuscript for Review

Dear Karen Hansen and the MESA P&P Committee,

Please accept the attached manuscript entitled 'A Bioinformatics Pipeline for Estimating Mitochondria DNA Copy Number and Heteroplasmy Levels from WGS Data' for review by the MESA P&P Committee. All co-authors have reviewed and approved of the manuscript. This work was completed as part of TOPMed manuscript proposal #4041 ("Association of mitochondrial DNA copy number and heteroplasmy with DNA methylation, gene expression, and metabolomics in MESA"). The target journal for publication would be Nucleic Acids Research.

The lay summary is below:

"Mitochondrial diseases are a heterogeneous group of disorders that affect 1 in 5,000 individuals and can be caused by mutations in the nuclear or mitochondrial genome. This paper describes a bioinformatic process that take DNA sequencing data and outputs mitochondrial DNA mutations. Using DNA sequencing data from TOPMed cohorts ARIC and MESA, which includes thousands of samples, we generate mitochondrial genome sequences for each sample and accurately identify mutations for each sample."

If you have any questions, please do not hesitate to contact me.

Stephanie L Battle, PhD

Postdoctoral Fellow | Arking Lab

Department of Genetic Medicine

Johns Hopkins University School of Medicine