

Alan Kwong <amkwong@umich.edu>

Re: Fw: TOPMed manuscript M6573: Method paper for Robust HWE

Mak, Angel < ChiYan.Mak@ucsf.edu>

Fri, May 22, 2020 at 4:59 PM

To: Alan Kwong <amkwong@umich.edu>

Cc: "Burchard, Esteban" <Esteban.Burchard@ucsf.edu>, Hyun Min Kang <hmkang@umich.edu>, "Salazar, Sandra" <Sandra.Salazar@ucsf.edu>

Hi Alan,

Thanks for making the edits.

One last thing, please include the funding sources listed in the link below for Dr Burchard: https://docs.google.com/document/d/1llaxHCnH3uu3uVodf52JE4pXRMYms bCXZHD39fs0pyo/edit?usp=sharing

We sign off for GALA II and SAGE.

Good luck with your submission, Angel

From: Alan Kwong <amkwong@umich.edu>

Sent: Friday, May 22, 2020 10:38 AM To: Mak, Angel < ChiYan. Mak@ucsf.edu>

Cc: Burchard, Esteban < Esteban. Burchard@ucsf.edu>; Hyun Min Kang < hmkang@umich.edu>; Salazar, Sandra

<Sandra.Salazar@ucsf.edu>

Subject: Re: Fw: TOPMed manuscript M6573: Method paper for Robust HWE

Hi Angel,

Thanks for the update, I missed the initial email so my apologies for the late reply. The missing SAGE acknowledgement was an error on my part, so I apologize for that also. I added an acknowledgement for the SAGE study on page 23 (see attached), which we obtained from the TOPMed main paper. Please let me know if this looks correct:

NHLBI TOPMed: Study of African Americans, Asthma, Genes and Environment (SAGE)

The Study of African Americans, Asthma, Genes and Environments (SAGE) was supported by the National Heart, Lung, and Blood Institute of the National Institute of Health (NIH) grants R01HL117004 and X01HL134589; study enrollment supported by the Sandler Family Foundation, the American Asthma Foundation, the RWJF Amos Medical Faculty Development Program, Harry Wm. and Diana V. Hind Distinguished Professor in Pharmaceutical Sciences II.

The SAGE study collaborators include Harold J. Farber, Texas Children's Hospital; Emerita Brigino-Buenaventura, Kaiser Permanente; Michael A. LeNoir, Bay Area Pediatrics; Kelley Meade, UCSF Benioff Children's Hospital, Oakland; Luisa N. Borrell, City University of New York; Adam Davis, UCSF Benioff Children's Hospital, Oakland and Fred Lurmann, Sonoma Technologies, Inc.

The authors acknowledge the families and patients for their participation and thank the numerous health care providers and community clinics for their support and participation in SAGE. In particular, the authors thank study coordinator Sandra Salazar; the recruiters who obtained the data: Lisa Caine, Elizabeth Castellanos, Brenda Lopez, MD, Shahdad Saeedi; and the lab researcher Celeste Eng who processed the biospecimens.

On Fri, May 22, 2020 at 1:12 PM Mak, Angel < ChiYan. Mak@ucsf.edu > wrote:

Hi Alan,

Please see the request below. Feel free to let me know if you have any questions.

Best. Angel

From: Mak, Angel < ChiYan. Mak@ucsf.edu>

Sent: Friday, May 15, 2020 7:49 PM

To: Hyun Min Kang <hmkang@umich.edu>

Subject: Re: TOPMed manuscript M6573: Method paper for Robust HWE

Hi Hyun Min,

Thanks for including our team in your manuscript. Could you please make sure the study acknowledgements of the SAGE study is included in the supplementary file? I may be wrong but I couldn't find it.

Study of African Americans, Asthma, Genes and Environment (SAGE) Study phs000921

Thanks, Angel

From: Hyun Min Kang hmkang@umich.edu>

Sent: Thursday, May 7, 2020 8:45 AM

To: Tom Blackwell <tblackw@umich.edu>; Jonathon LeFaive <lefaivej@umich.edu>; Laura Scott < goncalo@umich.edu>; Albert Vernon Smith <albertvs@umich.edu>; Goncalo Abecasis < goncalo@umich.edu>; Michael Boehnke

Soehnke@umich.edu>; Mariza de Andrade <mandrade@mayo.edu>; Kathleen C. Barnes <Kathleen.Barnes@ucdenver.edu>; John Blangero <john.blangero@utrgv.edu>; Eric Boerwinkle <Eric.Boerwinkle@uth.tmc.edu>; Burchard, Esteban <Esteban.Burchard@ucsf.edu>; Daniel Chasman <DCHASMAN@research.bwh.harvard.edu>; Han Chen <Han.Chen.2@uth.tmc.edu>; L. Adrienne Cupples <adrienne@bu.edu>; Yan Gao <ygao@umc.edu>; Guo, Xiuging <xquo@lundquist.org>; Marguerite Ryan Irvin <irvinr@uab.edu>; Tanika N. Kelly <tkelly@tulane.edu>; Wonji Kim <rewki@channing.harvard.edu>; Charles Kooperberg <clk@fredhutch.org>; Mak, Angel <ChiYan.Mak@ucsf.edu>; Manichaikul, Ani W. (am3xa) <am3xa@virginia.edu>; Rasika Mathias <rmathias@jhmi.edu>; May Montasser <mmontass@som.umaryland.edu>; Courtney Montgomery <Courtney-Montgomery@omrf.org>; Solomon Musani <smusani@umc.edu>; Nicholette D. Palmer <nallred@wakehealth.edu>; Gina M. Peloso <qpeloso@bu.edu>; Dandi Qiao <redag@channing.harvard.edu>; Alexander Reiner <apreiner@uw.edu>; Dan Roden <dan.roden@vumc.org>; Shoemaker, Moore Benjamin <moore.b.shoemaker@vumc.org>; Jennifer Smith <smjenn@umich.edu>; Nicholas L. Smith <nlsmith@u.washington.edu>; Jessica Lasky Su <rejas@channing.harvard.edu>; Hemant Tiwari htiwari@uab.edu>; Scott T. Weiss <scott.weiss@channing.harvard.edu>; Patrick Ellinor <ellinor@mgh.harvard.edu>; Steven Lubitz <slubitz@mgh.harvard.edu>; Daniel Weeks <weeks@pitt.edu>; Brian Cade <bcde@bwh.harvard.edu>; John Barnard < barnarj@ccf.org>

Cc: Alan Kwong <amkwong@umich.edu>

Subject: Re: TOPMed manuscript M6573: Method paper for Robust HWE

Dear Co-authors,

Thanks for your feedback on the TOPMed manuscript M6573, entitled 'Robust, flexible, and scalable tests for Hardy-Weinberg Equilibrium across diverse ancestries'. We tried our best to address the comments and to add important details required for P&P committees for individual working groups. We plan to submit the manuscript to DCC as soon as possible, so we will very much appreciate it if you send any additional feedback that can be addressed within the next 48 business hours.

Here are some highlights of the changes made on the manuscript:

- * The length of manuscript has been reduced substantially but clarified more details when needed. Some errors were also corrected.
- * We now have a complete list of co-authors in the paper. Please let me know if there are still errors or missing names.
- * We added acknowledgements of individual studies, similar to the TOPMed flagship paper as a Supplementary Material. We understand that this may be required for P&P committee review, so please let us know if there are any important details missing.

And many thanks for providing detailed comments and requests on the manuscript, and apologies for my silence. However, I believe that we addressed most of the comments, I will respond individually how the comments are incorporated.

Thanks, Hyun.

Hvun Min Kang, Ph.D. Associate Professor of Biostatistics University of Michigan, Ann Arbor Email: hmkang@umich.edu

On Tue, Apr 14, 2020 at 1:29 PM Hyun Min Kang hmkang@umich.edu wrote:

Dear Co-authors.

Attached please find a draft of the TOPMed manuscript M6573, entitled 'Robust, flexible, and scalable tests for Hardy-Weinberg Equilibrium across diverse ancestries'. The manuscript is separated into three pieces, (1) main text with references, (2) figures and tables, and (3) supplementary material for your convenience.

We will appreciate it very much if you could review these documents at your convenience and give any comments or suggested revisions (preferably with track changes). In addition, please check the spelling of your name, your institutional affiliation(s). If you want to add study-specific acknowledgements, please let us know. We would like to submit this to the TOPMed DCC in two weeks, April 28th, 2020.

We are considering submission of this manuscript to Genetics upon TOPMed approval, but please feel free to give us any suggestions regarding recommended journals.

We included co-authors whom we received responses from our earlier emails in alphabetical order in the middle before the TOPMed banner. We included the study names for those studies we need nomination from co-authors. For the studies whose co-authors are not yet nominated, please let us know the names as soon as possible. Alan Kwong (cc'ed) will follow up with PIs of individual studies regarding this.

We wanted to include as many authors as we can, but given the nature that this is a method paper, we were concerned that the number of authors that the journal expects is somewhat limited. As a result, we set up the following policy in the number of co-authors representing the contributing study in the following ways:

- (1) We grouped each study by PIs, and assigned 1-2 co-authors for each group (unless the studies need to nominate different co-authors from each study).
- (2) If the study contains fewer than 2,000 contributing samples in total, we requested to nominate one co-author. If more than 2,000, we requested nomination of two co-authors. We understand this strategy is not ideal for everyone, but we had to make a choice as we still expect to have 40 or more authors in this manuscript.

We look forward to receiving feedback and submitting this paper soon. If you have any other questions or comments, please feel free to let me and Alan know.

Sincerely, Hyun.

Hyun Min Kang, Ph.D.

Associate Professor of Biostatistics University of Michigan, Ann Arbor

Email: hmkang@umich.edu