

Subject: RE: Revised manuscript on height and BMI heritability estimation from WGS data
Date: Tuesday, 12 February 2019 at 6:30:27 am Australian Eastern Standard Time
From: Mitchell, Braxton
To: Peter Visscher
CC: Jian Yang, Pierrick Wainschtein
Category: Green
Attachments: 20190211_TOPMed_WGS_Revision-bdm.docx

Peter et al.,

This is a nice piece of work with a very clear result.
Attached are a few very lite edits, including to my affiliation, and addition of Amish-specific acknowledgments. Good to go on the Amish front.

Best wishes, Brackie

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Monday, February 11, 2019 8:10 AM
To: jaind@uw.edu; folso001@umn.edu; adrienne@bu.edu; aladdinhs@yahoo.com; bmck@uw.edu; moore.b.shoemaker@vanderbilt.edu; Mitchell, Braxton <bmitchel@som.umaryland.edu>; psaty@uw.edu; clk@fredhutch.org; dan.roden@Vanderbilt.Edu; darbar@uic.edu; donna.arnett@uky.edu; regane@njc.org; Eric.Boerwinkle@uth.tmc.edu; jrotter@labiomed.org; mallison@ucsd.edu; mmcdonald@uab.edu; chungm@ccf.org; nlsmith@uw.edu; ellinor@mgh.harvard.edu; vasan@bu.edu; rmathias@jhmi.edu; ssr4n@virginia.edu; heckbert@uw.edu; sredline1@rics.bwh.harvard.edu; xguo@labiomed.org; jchen@labiomed.org; CALBERT@mgh.harvard.edu; ryan.hernandez@ucsf.edu; stephen_mcgarvey@brown.edu; kari_north@unc.edu; LESLIE.LANGE@UCDENVER.EDU; bsweir@uw.edu; cclaurie@uw.edu
Cc: Jian Yang <jian.yang@uq.edu.au>; Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>
Subject: Revised manuscript on height and BMI heritability estimation from WGS data
Importance: High

CAUTION: This message originated from a non UMB, UMSOM, FPI, or UMMS email system. Whether the sender is known or not known, hover over any links before clicking and use caution opening attachments.

Dear Colleagues,

You are receiving this email because you are a named author on the attached TOPMed manuscript on height and BMI. It is a revised version of a ms sent round in December last year.

Many of you responded with helpful comments (Thank You!) and with the names of additional authors. We really hope that we have the authorship correct but please let us know if otherwise. We have tried to deal with all comments as best as we could, and have performed additional checks to test the robustness of the inference.

Please could we ask you to:

- 1) Check your affiliations (we've tried our best but are likely to have made mistakes)
- 2) Add more cohort-level information if/when relevant. We have created a new section with cohort-specific acknowledgements.
- 3) Let us know if we need additional permission(s) before submission.
- 4) Let us know if you have any other comments or suggestions.

We'd love to submit the (final) ms for publication by February 23rd if possible.

Best wishes,

Peter, on behalf of the UQ and UW teams

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

Subject: RE: Consent for submission of paper containing ARIC TOPMed data
Date: Saturday, 23 February 2019 at 4:57:18 am Australian Eastern Standard Time
From: Boerwinkle, Eric
To: Peter Visscher
CC: Pierrick Wainschtein, aricjhu@jhu.edu
Category: Green

Peter:

Sorry for the late reply. Took me too long to engage.

1. I approve of submission. Very nice work.
2. I will make comments and send to you, as a coauthor. You can incorporate (or not) during the revision phase.

I hope you are doing well.

Eric.

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Thursday, February 21, 2019 9:19 AM
To: Boerwinkle, Eric <Eric.Boerwinkle@uth.tmc.edu>
Cc: Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>; aricjhu@jhu.edu
Subject: Consent for submission of paper containing ARIC TOPMed data
Importance: High

Dear Eric,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript (on heritability estimation from WGS data) for publication. Are you in a position to give this consent (in an email reply) for the ARIC cohort or should we go through a more formal process? All currently listed authors have approved the ms for submission.

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com



**Research
with Heart.**

Memorandum

To: Peter Visscher
From: Richey Sharrett, for the ARIC Publications Committee
CC: ARIC Publications Committee; Hunter Holbrook
Subject: Review of ARIC MS # TOPMed03
Title: Recovery of trait heritability from whole genome sequence data
Date: Friday, February 22, 2019

On behalf of the ARIC Steering Committee, I am happy to inform you that the above-referenced paper has been approved.

ARIC Publication Policies Require:

1. To comply with NIH Public Access Policy, all ARIC manuscripts are required to be submitted to PubMed Central upon acceptance for publication. If your paper is accepted for publication in a journal that deposits all final published articles in PubMed Central, no author involvement is required. If the journal does not automatically deposit all final manuscripts into PubMed Central, you (or a co-author) **MUST** deposit the final peer-reviewed manuscript in PubMed Central via the NIH Manuscript Submission System (NIHMS): <http://www.nihms.nih.gov/>. More information regarding NIH Public Access Policy can be found here: <http://publicaccess.nih.gov/>
2. Enclosed are the instructions to authors, per our policy, as well as a sentence acknowledging ARIC staff, which should be added to the paper.
3. Please remember to add reference to “ARIC,” in either the title of the paper or the abstract, so that the study can be identified by reference searches. In addition, when the paper has been published, please email the citation and pdf to both Hunter Holbrook (aricpub@unc.edu) and Kendra Hay (aricjhu@jhu.edu).
4. Please follow procedures local to your center for archiving a copy of the programs which created this paper. At Johns Hopkins provide a copy to Natalie Daya (ndaya1@jhu.edu).

Congratulations and good luck with this interesting paper’s submission.

Subject: RE: TOPMED paper
Date: Wednesday, 27 February 2019 at 4:37:26 pm Australian Eastern Standard Time
From: Chung, M.D., Mina
To: Peter Visscher
CC: Pierrick Wainschtein
Category: Green

I thought I had already. Please proceed to submit on behalf of the CCAF cohort.
Best regards,
Mina

Mina K. Chung, M.D.
Cardiac Pacing and Electrophysiology
Department of Cardiovascular Medicine, Heart and Vascular Institute
Department of Cardiovascular & Metabolic Sciences, Lerner Research Institute
Cleveland Clinic
Cleveland Clinic Lerner College of Medicine of Case Western Reserve University
9500 Euclid Avenue, Desk J2-2
Cleveland, OH 44195
office: 216-444-2290
FAX: 216-636-6951
email: chungm@ccf.org
CCF pager: 24843

From: Peter Visscher [mailto:peter.visscher@uq.edu.au]
Sent: Wednesday, February 27, 2019 1:36 AM
To: Chung, M.D., Mina <CHUNGGM@ccf.org>
Cc: Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>
Subject: [EXT] FW: TOPMED paper
Importance: High

Dear Mina,

It would be wonderful if you could give us explicit consent to submit the paper on behalf of the CCAF cohort.

For information, I attached the manuscript.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsngenomics.com

From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>
Date: Friday, 22 February 2019 at 5:53 AM
To: Peter Visscher <peter.visscher@uq.edu.au>, Mina Chung <>, Patrick Ellinor <ellinor@mgh.harvard.edu>
Cc: "Roden, Dan" <dan.roden@vumc.org>

Subject: RE: TOPMED paper

Hi Peter, I can give you permission for VAFAR and VU_AF, but CCAF and MGH_AF are Mina Chung and Patrick Ellinor's cohorts, respectively. I have cc'd them.

Thanks,

-Ben

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Thursday, February 21, 2019 10:24 AM
To: Shoemaker, Moore Benjamin <moore.b.shoemaker@vumc.org>
Cc: Roden, Dan <dan.roden@vumc.org>
Subject: Re: TOPMED paper

Dear Ben,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the AFGen cohorts (CCAF, MGH_AF, VAFAR, VU_AF) or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>
Date: Thursday, 27 December 2018 at 22:37 PM
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: "Roden, Dan" <dan.roden@vumc.org>
Subject: TOPMED paper

Hi Peter,

Thanks for including us on the paper. Dawood Darbar is another author that should be included. Our author list and affiliations are below

M. Benjamin Shoemaker, MD, MSCI (please include my first name's initial), Department of Medicine, Vanderbilt University Medical Center, Nashville, TN

Dan Roden, MD. Departments of Medicine, Pharmacology and Bioinformatics. Vanderbilt University Medical Center, Nashville, TN

Dawood Darbar, MD. Department of Medicine. University of Illinois-Chicago. Chicago, IL

Best,

-Ben

M. Benjamin Shoemaker, MD, MSCI
Assistant Professor, Division of Cardiovascular Medicine
Vanderbilt Center for Arrhythmia Research and Therapeutics (VanCART)
Vanderbilt University Medical Center

Please consider the environment before printing this e-mail

Cleveland Clinic is currently ranked as the No. 2 hospital in the country by U.S. News & World Report (2017-2018). Visit us online at <http://www.clevelandclinic.org> for a complete listing of our services, staff and locations. Confidentiality Note: This message is intended for use only by the individual or entity to which it is addressed and may contain information that is privileged, confidential, and exempt from disclosure under applicable law. If the reader of this message is not the intended recipient or the employee or agent responsible for delivering the message to the intended recipient, you are hereby notified that any dissemination, distribution or copying of this communication is strictly prohibited. If you have received this communication in error, please contact the sender immediately and destroy the material in its entirety, whether electronic or hard copy. Thank you.

Subject: Fwd: Draft paper TOPMed Anthropometry & Adiposity Working Group
Date: Wednesday, 16 January 2019 at 4:44:44 pm Australian Eastern Standard Time
From: Peter Visscher
To: Pierrick Wainschtein
Attachments: image001.jpg, image002.jpg

Peter
Sent from my Phone. Apologies for typos etc.

Begin forwarded message:

From: Susan R Heckbert <heckbert@uw.edu>
Date: 16 January 2019 at 08:56:07 GMT+5:30
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: Bruce M Psaty <psaty@uw.edu>, Barbara McKnight <bmck@uw.edu>, "Nicholas L. Smith" <nlsmith@uw.edu>
Subject: Re: Draft paper TOPMed Anthropometry & Adiposity Working Group

Hello Peter,

This note is to let you know that this manuscript has been approved by the CHS Publications and Steering Committees, with the requirement that the CHS Acknowledgment must be added to the Acknowledgments section (which was incomplete in the version I submitted). The CHS Acknowledgments are:

Cardiovascular Health Study: This research was supported by contracts HHSN268201200036C, HHSN268200800007C, HHSN268201800001C, N01HC55222, N01HC85079, N01HC85080, N01HC85081, N01HC85082, N01HC85083, N01HC85086, and grants U01HL080295, U01HL130114, and R01HL059367 from the National Heart, Lung, and Blood Institute (NHLBI), with additional contribution from the National Institute of Neurological Disorders and Stroke (NINDS). Additional support was provided by R01AG023629 from the National Institute on Aging (NIA). A full list of principal CHS investigators and institutions can be found at CHS-NHLBI.org. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

Best wishes for success with this manuscript,
Susan

Susan R. Heckbert, MD PhD
Professor, Department of Epidemiology
University of Washington
Cardiovascular Health Research Unit
1730 Minor Ave, Suite 1360
Seattle, WA 98101
206.221.7775 | heckbert@uw.edu
<http://epi.washington.edu>

 DEPARTMENT OF EPIDEMIOLOGY
UNIVERSITY of WASHINGTON
School of Public Health

From: Susan R Heckbert <heckbert@uw.edu>
Date: Thursday, December 20, 2018 at 8:35 PM
To: Peter Visscher <peter.visscher@uq.edu.au>
Subject: Re: Draft paper TOPMed Anthropometry & Adiposity Working Group

Peter,
Thank you for circulating this interesting and well-written manuscript.

Please add my middle initial, "R."
My affiliation:
Cardiovascular Health Research Unit and Department of Epidemiology, University of Washington. Seattle, WA, USA 98195

I have submitted this manuscript to the CHS Publications Committee for review and will let you know when it is approved.

Best regards,
Susan

Susan R. Heckbert, MD PhD
Professor, Department of Epidemiology
University of Washington
Cardiovascular Health Research Unit
1730 Minor Ave, Suite 1360
Seattle, WA 98101
206.221.7775 | heckbert@uw.edu
<http://epi.washington.edu>



From: Peter Visscher <peter.visscher@uq.edu.au>
Date: Monday, December 17, 2018 at 9:23 PM
To: "moore.b.shoemaker@vanderbilt.edu" <moore.b.shoemaker@vanderbilt.edu>, "bmitchel@som.umaryland.edu" <bmitchel@som.umaryland.edu>, Bruce M Psaty <psaty@uw.edu>, "clk@fredhutch.org" <clk@fredhutch.org>, "dan.roden@Vanderbilt.Edu" <dan.roden@Vanderbilt.Edu>, "donna.arnett@uky.edu" <donna.arnett@uky.edu>, "ed.silverman@channing.harvard.edu" <ed.silverman@channing.harvard.edu>, "Eric.Boerwinkle@uth.tmc.edu" <Eric.Boerwinkle@uth.tmc.edu>, "ichen@labiomed.org" <ichen@labiomed.org>, "crapoj@NJHealth.org" <crapoj@NJHealth.org>, "chungm@ccf.org" <chungm@ccf.org>, "Nicholas L. Smith" <nlsmith@uw.edu>, "rmathias@jhmi.edu" <rmathias@jhmi.edu>, Susan R Heckbert <heckbert@uw.edu>, "sredline1@rics.bwh.harvard.edu" <sredline1@rics.bwh.harvard.edu>, "Ramachandran, Vasan" <vasan@bu.edu>, "CALBERT@mgh.harvard.edu" <CALBERT@mgh.harvard.edu>, "folso001@umn.edu" <folso001@umn.edu>
Cc: Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>, Jian Yang <jian.yang@uq.edu.au>, "Cathy C. Laurie" <cclaurie@uw.edu>, "Lange, Leslie" <LESLIE.LANGE@UCDENVER.EDU>
Subject: Draft paper TOPMed Anthropometry & Adiposity Working Group

Dear all,

You are receiving this email because you are a named author on the attached DRAFT research paper on the inference of genetic variance of height and BMI from (TOPMed) WGS data . This draft has recently been sent to members who are on the Working Group list-server, but not all currently named authors are on that list.

We realise that the timing is awkward and that we need to collect additional cohort-specific information to add to the paper, but we wanted to share the (exciting) science with you now. I showed and discussed the main results at a TOPMed Working Group call in July 2018 and present some of the findings at ASHG in San Diego this year.

We would appreciate feedback on a number of fronts:

- science
- authorship (have we left anybody out who should be named?)
- cohort-specific consent for submission if required
- authors-specific affiliations

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

Subject: FW: Revised manuscript on height and BMI heritability estimation from WGS data
Date: Wednesday, 13 February 2019 at 1:02:42 am Australian Eastern Standard Time
From: Peter Visscher
To: Pierrick Wainschtein
Category: Green
Attachments: image001.jpg

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cns.genomics.com

From: Susan R Heckbert <heckbert@uw.edu>
Date: Tuesday, 12 February 2019 at 7:46 AM
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: Bruce M Psaty <psaty@uw.edu>, "Nicholas L. Smith" <nlsmith@uw.edu>, Barbara McKnight <bmck@uw.edu>
Subject: Re: Revised manuscript on height and BMI heritability estimation from WGS data

Hello Peter,

Thank you for the update on this ms, which as you know has been approved by the CHS Steering Committee.

On affiliations, please note that this is my affiliation (which is a subset of footnote #7):
Cardiovascular Health Research Unit and Department of Epidemiology, University of Washington, Seattle, WA, USA;

The CHS Acknowledgments are correct.
Best of luck with this submission!
Susan

Susan R. Heckbert, MD PhD
Professor, Department of Epidemiology
University of Washington
Cardiovascular Health Research Unit
1730 Minor Ave, Suite 1360
Seattle, WA 98101
206.221.7775 | heckbert@uw.edu
<http://epi.washington.edu>

 **DEPARTMENT OF EPIDEMIOLOGY**
UNIVERSITY of WASHINGTON
School of Public Health

From: Peter Visscher <peter.visscher@uq.edu.au>

Date: Monday, February 11, 2019 at 5:10 AM

To: Deepti Jain <jaind@uw.edu>, "folso001@umn.edu" <folso001@umn.edu>, "adrienne@bu.edu" <adrienne@bu.edu>, "aladdinhs@yahoo.com" <aladdinhs@yahoo.com>, Barbara McKnight <bmck@uw.edu>, "moore.b.shoemaker@vanderbilt.edu" <moore.b.shoemaker@vanderbilt.edu>, "bmitchel@som.umaryland.edu" <bmitchel@som.umaryland.edu>, Bruce M Psaty <psaty@uw.edu>, "clk@fredhutch.org" <clk@fredhutch.org>, "dan.roden@Vanderbilt.Edu" <dan.roden@Vanderbilt.Edu>, "darbar@uic.edu" <darbar@uic.edu>, "donna.arnett@uky.edu" <donna.arnett@uky.edu>, "regane@njc.org" <regane@njc.org>, "Eric.Boerwinkle@uth.tmc.edu" <Eric.Boerwinkle@uth.tmc.edu>, "jrotter@labiomed.org" <jrotter@labiomed.org>, Matthew Allison <mallison@ucsd.edu>, "mmcdonald@uab.edu" <mmcdonald@uab.edu>, "chungm@ccf.org" <chungm@ccf.org>, "Nicholas L. Smith" <nlsmith@uw.edu>, "ellinor@mgh.harvard.edu" <ellinor@mgh.harvard.edu>, "Ramachandran, Vasan" <vasan@bu.edu>, "rmathias@jhmi.edu" <rmathias@jhmi.edu>, "ssr4n@virginia.edu" <ssr4n@virginia.edu>, Susan R Heckbert <heckbert@uw.edu>, "sredline1@rics.bwh.harvard.edu" <sredline1@rics.bwh.harvard.edu>, "xguo@labiomed.org" <xguo@labiomed.org>, "jchen@labiomed.org" <jchen@labiomed.org>, "CALBERT@mgh.harvard.edu" <CALBERT@mgh.harvard.edu>, "ryan.hernandez@ucsf.edu" <ryan.hernandez@ucsf.edu>, "stephen_mcgarvey@brown.edu" <stephen_mcgarvey@brown.edu>, "kari_north@unc.edu" <kari_north@unc.edu>, "LESLIE.LANGE@UCDENVER.EDU" <LESLIE.LANGE@UCDENVER.EDU>, Bruce S Weir <bsweir@uw.edu>, "Cathy C. Laurie" <cclaurie@uw.edu>
Cc: Jian Yang <jian.yang@uq.edu.au>, Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>
Subject: Revised manuscript on height and BMI heritability estimation from WGS data

Dear Colleagues,

You are receiving this email because you are a named author on the attached TOPMed manuscript on height and BMI. It is a revised version of a ms sent round in December last year.

Many of you responded with helpful comments (Thank You!) and with the names of additional authors. We really hope that we have the authorship correct but please let us know if otherwise. We have tried to deal with all comments as best as we could, and have performed additional checks to test the robustness of the inference.

Please could we ask you to:

- 1) Check your affiliations (we've tried our best but are likely to have made mistakes)
- 2) Add more cohort-level information if/when relevant. We have created a new section with cohort-specific acknowledgements.
- 3) Let us know if we need additional permission(s) before submission.
- 4) Let us know if you have any other comments or suggestions.

We'd love to submit the (final) ms for publication by February 23rd if possible.

Best wishes,

Peter, on behalf of the UQ and UW teams

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

Subject: Re: Draft paper TOPMed Anthropometry & Adiposity Working Group
Date: Friday, 22 February 2019 at 1:45:20 am Australian Eastern Standard Time
From: Ed Silverman
To: Peter Visscher
CC: James Crapo, Pierrick Wainschtein
Category: Green

Dear Peter,
On behalf of COPDGene, I support submission of this manuscript.
Best wishes,
Ed

On Thu, Feb 21, 2019 at 10:24 AM Peter Visscher <peter.visscher@uq.edu.au> wrote:

Dear Ed,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the COPDGene cohort or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience,
University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w:
cnsgenomics.com

From: Ed Silverman <reeks@channing.harvard.edu>
Date: Thursday, 20 December 2018 at 12:14 PM
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: James Crapo <james.crapo@gmail.com>
Subject: Re: Draft paper TOPMed Anthropometry & Adiposity Working Group

Dear Peter,

Thanks very much for sending along this fascinating and important paper.

Although James Crapo and I are pleased that the COPDGene study was able to contribute to your efforts, we do not believe that our contributions to the article warrant named authorship.

We hope that the COPDGene members of the Anthropometry Working Group (Elizabeth Regan and Merry-Lynn McDonald)

will be named authors on this paper.

Best wishes,

Ed Silverman

On Tue, Dec 18, 2018 at 12:22 AM Peter Visscher <peter.visscher@uq.edu.au> wrote:

Dear all,

You are receiving this email because you are a named author on the attached DRAFT research paper on the inference of genetic variance of height and BMI from (TOPMed) WGS data . This draft has recently been sent to members who are on the Working Group list-server, but not all currently named authors are on that list.

We realise that the timing is awkward and that we need to collect additional cohort-specific information to add to the paper, but we wanted to share the (exciting) science with you now. I showed and discussed the main results at a TOPMed Working Group call in July 2018 and present some of the findings at ASHG in San Diego this year.

We would appreciate feedback on a number of fronts:

- science
- authorship (have we left anybody out who should be named?)
- cohort-specific consent for submission if required

-- authors-specific affiliations

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience,
University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w:
cnsgenomics.com

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The information in this e-mail is intended only for the person to whom it is addressed. If you believe this e-mail was sent to you in error and the e-mail contains patient information, please contact the Partners Compliance HelpLine at <http://www.partners.org/complianceline> . If the e-mail was sent to you in error but does not contain patient information, please contact the sender and properly dispose of the e-mail.

Subject: RE: Consent for submission of TOPMed paper containing FHS data
Date: Friday, 22 February 2019 at 2:15:47 am Australian Eastern Standard Time
From: Ramachandran, Vasana
To: Ann Walsh, Peter Visscher, Mutalik, Karen M
CC: Pierrick Wainschtein, Cupples, L Adrienne
Category: Green

Thanks, Ann and Peter.

At FHS, Adrienne and I have been okaying Ms and this one is good to go (ready for submission) from our standpoint.

Hope this is helpful.

Vasana

From: Walsh, Ann (NIH/NHLBI) [E] <walshac@nhlbi.nih.gov>
Sent: Thursday, February 21, 2019 10:41 AM
To: Peter Visscher <peter.visscher@uq.edu.au>; Mutalik, Karen M <kmutalik@bu.edu>
Cc: Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>; Ramachandran, Vasana <vasana@bu.edu>
Subject: RE: Consent for submission of TOPMed paper containing FHS data

Dear Peter,

I defer to Vasana and Karen.

Are you aware of the TOPMed publication policy?

<https://www.nhlbiwgs.org/topmed-publications-policy>

Thank you for your consideration,
Ann Walsh

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Thursday, February 21, 2019 10:31 AM
To: Walsh, Ann (NIH/NHLBI) [E] <walshac@nhlbi.nih.gov>; kmutalik@bu.edu
Cc: Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>; vasana@bu.edu
Subject: Consent for submission of TOPMed paper containing FHS data
Importance: High

Dear Ann and Karen and Vasana,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the FHS cohort or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular
Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346
6348 | w: cnsgenomics.com

Subject: Revised manuscript on height and BMI heritability estimation from WGS data
Date: Tuesday, 26 February 2019 at 10:00:20 pm Australian Eastern Standard Time
From: RASIKA Mathias
To: Pierrick Wainschtein, Lisa Yanek
CC: Peter Visscher
Category: Green

Pierrick, yes you have authorization from me on behalf of GeneSTAR for the submission.

Rasika Mathias, ScD.

Associate Professor of Medicine,
Johns Hopkins University.
Director of Genomics, GeneSTAR Research program, General Internal Medicine.
Director, Data Analysis Unit, Allergy and Clinical Immunology.
5501 Hopkins Bayview Circle, 3A.62A
Baltimore, MD 21224
Tel: 410-550-2487

Subject: Re: Revised manuscript on height and BMI heritability estimation from WGS data
Date: Friday, 22 February 2019 at 2:34:28 am Australian Eastern Standard Time
From: Arnett, Donna
To: Peter Visscher
CC: Pierrick Wainschtein, Claas, Steve
Category: Green

Yes I consent for GOLDN

Sent from my iPhone

On Feb 21, 2019, at 10:39 AM, Peter Visscher <peter.visscher@uq.edu.au> wrote:

Dear Donna,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the GOLDN cohort or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "Arnett, Donna" <donna.arnett@uky.edu>

Date: Monday, 18 February 2019 at 7:27 AM

To: Peter Visscher <peter.visscher@uq.edu.au>

Subject: Re: Revised manuscript on height and BMI heritability estimation from WGS data

Peter,

I have read thru the manuscript and I commend you on the analysis. I think it adds to the literature regarding missing heritability. I don't have anything to add – I was happy to see that the addition of rare variants improved the estimates. My affiliation is correct.

Best regards,

Donna

From: Peter Visscher <peter.visscher@uq.edu.au>

Date: Monday, February 11, 2019 at 8:12 AM

To: "jaind@uw.edu" <jaind@uw.edu>, Aaron Folsom <folso001@umn.edu>, Adrienne Cupples <adrienne@bu.edu>, "aladdinhs@yahoo.com" <aladdinhs@yahoo.com>, "bmck@uw.edu" <bmck@uw.edu>, "moore.b.shoemaker@vanderbilt.edu" <moore.b.shoemaker@vanderbilt.edu>, "bmitchel@som.umaryland.edu" <bmitchel@som.umaryland.edu>, "psaty@uw.edu" <psaty@uw.edu>, "clk@fredhutch.org" <clk@fredhutch.org>, Dan Roden <dan.roden@Vanderbilt.Edu>, "darbar@uic.edu" <darbar@uic.edu>, "Arnett, Donna" <donna.arnett@uky.edu>, "regane@njc.org" <regane@njc.org>, Eric Boerwinkle <Eric.Boerwinkle@uth.tmc.edu>, Jerry Rotter <jrotter@labiomed.org>, Matthew Allison <mallison@ucsd.edu>, "mmcdonald@uab.edu" <mmcdonald@uab.edu>, "chungm@ccf.org" <chungm@ccf.org>, "nlsmith@uw.edu" <nlsmith@uw.edu>, "ellinor@mgh.harvard.edu" <ellinor@mgh.harvard.edu>, Vasan Ramachandran <vasan@bu.edu>, "rmathias@jhmi.edu" <rmathias@jhmi.edu>, Steve Rich <ssr4n@virginia.edu>, "heckbert@uw.edu" <heckbert@uw.edu>, "sredline1@rics.bwh.harvard.edu" <sredline1@rics.bwh.harvard.edu>, "xguo@labiomed.org" <xguo@labiomed.org>, "jchen@labiomed.org" <jchen@labiomed.org>, "CALBERT@mgh.harvard.edu" <CALBERT@mgh.harvard.edu>, "ryan.hernandez@ucsf.edu" <ryan.hernandez@ucsf.edu>, "stephen_mcgarvey@brown.edu" <stephen_mcgarvey@brown.edu>, Kari North <kari_north@unc.edu>, "LESLIE.LANGE@UCDENVER.EDU" <LESLIE.LANGE@UCDENVER.EDU>, Bruce Weir <bsweir@uw.edu>, "cclaurie@uw.edu" <cclaurie@uw.edu>

Cc: Jian Yang <jian.yang@uq.edu.au>, Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>

Subject: Revised manuscript on height and BMI heritability estimation from WGS data

Dear Colleagues,

You are receiving this email because you are a named author on the attached TOPMed manuscript on height and BMI. It is a revised version of a ms sent round in December last year.

Many of you responded with helpful comments (Thank You!) and with the names of additional authors. We really hope that we have the authorship correct but please let us know if otherwise. We have tried to deal with all comments as best as we could, and have performed additional checks to test the robustness of the inference.

Please could we ask you to:

- 1) Check your affiliations (we've tried our best but are likely to have made mistakes)
- 2) Add more cohort-level information if/when relevant. We have created a new section with cohort-specific acknowledgements.
- 3) Let us know if we need additional permission(s) before submission.
- 4) Let us know if you have any other comments or suggestions.

We'd love to submit the (final) ms for publication by February 23rd if possible.

Best wishes,

Peter, on behalf of the UQ and UW teams

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

<20190221_TOPMed_WGS.docx>

Subject: RE: Revised manuscript on height and BMI heritability estimation from WGS data
Date: Friday, 22 February 2019 at 3:57:08 pm Australian Eastern Standard Time
From: Nicholas L. Smith
To: Peter Visscher
CC: Jian Yang, Pierrick Wainschtein, Susan R Heckbert
Category: Green

Peter:

Consent given. Good luck with the submission.

Nick

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Thursday, February 21, 2019 08:21
To: Nicholas L. Smith <nsmith@uw.edu>
Cc: Jian Yang <jian.yang@uq.edu.au>; Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>; Susan R Heckbert <heckbert@uw.edu>
Subject: Re: Revised manuscript on height and BMI heritability estimation from WGS data

Dear Nick,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for HVH or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cns.genomics.com

From: "Nicholas L. Smith" <nsmith@uw.edu>
Date: Tuesday, 12 February 2019 at 8:43 AM
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: Jian Yang <jian.yang@uq.edu.au>, Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>, Susan R Heckbert <heckbert@uw.edu>
Subject: RE: Revised manuscript on height and BMI heritability estimation from WGS data

[Peter and colleagues:](#)

I really enjoyed reading the final version of the manuscript. I have 3 affiliations and if you could add the 3rd one, I would be appreciative. The 3rd is as follows: Seattle Epidemiologic Research and Information Center, Department of Veterans Affairs Office of Research and Development, Seattle WA USA.

Thanks and good luck with the submission.

Nick

Nicholas L. Smith, PhD
Professor, Department of Epidemiology, University of Washington
Director, Seattle Epidemiologic Research and Information Center, VA Cooperative Studies Program

From: Peter Visscher [<mailto:peter.visscher@uq.edu.au>]

Sent: Monday, February 11, 2019 5:10 AM

To: Deepti Jain <jaind@uw.edu>; folso001@umn.edu; adrienne@bu.edu; aladdinhs@yahoo.com; Barbara McKnight <bmck@uw.edu>; moore.b.shoemaker@vanderbilt.edu; bmitchel@som.umaryland.edu; Bruce M Psaty <psaty@uw.edu>; clk@fredhutch.org; dan.roden@Vanderbilt.Edu; darbar@uic.edu; donna.arnett@uky.edu; regane@njc.org; Eric.Boerwinkle@uth.tmc.edu; jrotter@labiomed.org; mallison@ucsd.edu; mmcdonald@uab.edu; chungm@ccf.org; Nicholas L. Smith <nlsmith@uw.edu>; ellinor@mgh.harvard.edu; vasan@bu.edu; rmathias@jhmi.edu; ssr4n@virginia.edu; Susan R Heckbert <heckbert@uw.edu>; sredline1@rics.bwh.harvard.edu; xguo@labiomed.org; jchen@labiomed.org; CALBERT@mgh.harvard.edu; ryan.hernandez@ucsf.edu; stephen_mcgarvey@brown.edu; kari_north@unc.edu; LESLIE.LANGE@UCDENVER.EDU; Bruce S Weir <bsweir@uw.edu>; Cathy C. Laurie <cclaurie@uw.edu>

Cc: Jian Yang <jian.yang@uq.edu.au>; Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>

Subject: Revised manuscript on height and BMI heritability estimation from WGS data

Importance: High

Dear Colleagues,

You are receiving this email because you are a named author on the attached TOPMed manuscript on height and BMI. It is a revised version of a ms sent round in December last year.

Many of you responded with helpful comments (Thank You!) and with the names of additional authors. We really hope that we have the authorship correct but please let us know if otherwise. We have tried to deal with all comments as best as we could, and have performed additional checks to test the robustness of the inference.

Please could we ask you to:

- 1) Check your affiliations (we've tried our best but are likely to have made mistakes)
- 2) Add more cohort-level information if/when relevant. We have created a new section with cohort-specific acknowledgements.

- 3) Let us know if we need additional permission(s) before submission.
- 4) Let us know if you have any other comments or suggestions.

We'd love to submit the (final) ms for publication by February 23rd if possible.

Best wishes,

Peter, on behalf of the UQ and UW teams

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

Subject: Re: TOPMed proposal involving the Mayo(VTE) study
Date: Wednesday, 27 February 2019 at 8:24:13 am Australian Eastern Standard Time
From: Peter Visscher
To: de Andrade, Mariza, Ph.D.
CC: Pierrick Wainschtein
Category: Green

Wonderful – thank you so much.

We presume that your affiliation is correct.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "de Andrade, Mariza, Ph.D." <mandrade@mayo.edu>
Date: Wednesday, 27 February 2019 at 8:20 AM
To: Peter Visscher <peter.visscher@uq.edu.au>
Subject: RE: TOPMed proposal involving the Mayo(VTE) study

I approve the use of the TOPMed VTE data!

Mariza de Andrade, Ph.D. | Consultant | Division of Biomedical Statistics and Informatics | Professor of Biostatistics | College Of Medicine | 507-284-1032 | Secretary: 507-284-5567 | Fax: 507-284-9542 | mandrade@mayo.edu
Mayo Clinic | 200 First Street SW | Rochester, MN 55905 | mayoclinic.org

From: Peter Visscher [<mailto:peter.visscher@uq.edu.au>]
Sent: Thursday, February 21, 2019 10:23 AM
To: de Andrade, Mariza, Ph.D.
Cc: Pierrick Wainschtein
Subject: [EXTERNAL] Re: TOPMed proposal involving the Mayo(VTE) study
Importance: High

Dear Mariza,

Due to conflicting information on the dbGaP and TopMed sites we have failed to include you on previous emails with draft versions of our manuscript (sent round in December 2018 and January 2019). I am really sorry about that and apologise. Attached is the latest (near-final) version.

We have added you as an author because of the inclusion of Mayo(VTE) data in the study. Please could you let us know if that is ok (& check your affiliation) and also if we need to include more authors from the Mayo on the paper?

In addition, it was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the Mayo(VTE) cohort or should we go through a more formal process?

Apologies once more about the oversight.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "de Andrade, Mariza, Ph.D." <mandrade@mayo.edu>
Date: Sunday, 17 September 2017 at 22:52 PM
To: Peter Visscher <peter.visscher@uq.edu.au>
Cc: Bruce Weir <bsweir@uw.edu>, Cathy Laurie <cclaurie@u.washington.edu>, Jian Yang <jian.yang@imb.uq.edu.au>
Subject: Re: TOPMed proposal involving the Mayo(VTE) study

Dear Dr. Visscher,

I approve the use of the Mayo VTE data for your proposal,

Sincerely,

Mariza de Andrade

Mariza de Andrade, Ph.D. | Consultant | Division of Biomedical Statistics and Informatics | Professor of Biostatistics | College Of Medicine | 507-284-1032 | Secretary: 507-284-5567 | Fax: 507-284-9542 | mandrade@mayo.edu
Mayo Clinic | 200 First Street SW | Rochester, MN 55905 | mayoclinic.org

From: Peter Visscher <peter.visscher@uq.edu.au>
Date: Sunday, September 17, 2017 at 2:09 AM
To: Mariza de Andrade <mandrade@mayo.edu>
Cc: "bsweir@uw.edu" <bsweir@uw.edu>, Cathy Laurie <cclaurie@u.washington.edu>, Jian Yang <jian.yang@imb.uq.edu.au>

Subject: TOPMed proposal involving the Mayo(VTE) study

Dear Dr de Andrade,

Re: TOPMed proposal on the genetic architecture of height and BMI

I am contacting you as the named PI of the Mayo(VTE) study to request your collaboration involving the use of your study's data in a proposed research project from the TOPMed Anthropometrics Working Group (attached).

This proposal is from Drs Bruce Weir and Dr Cathy Laurie from the University of Washington and Dr Jian Yang and myself from the University of Queensland (Australia).

In addition to seeking your collaboration we would of course offer authorship to you and other relevant contributors to your study on any papers arising from the proposed research.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

Subject: Re: Corrections and Additions - RE: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity
Date: Friday, 22 February 2019 at 2:30:14 am Australian Eastern Standard Time
From: Rotter, Jerome
To: Peter Visscher, Thorington, Daune
CC: Pierrick Wainschtein, Stephen Rich, Lange, Leslie, Guo, Xiuqing
Category: Green

peter:

one:

either i or steve rich can give the consent (i am mesa wgs pi and he is co-pi).
with this email, i am giving my consent as mesa wgs pi.

two:

in addition, the manuscript requires approval from the mesa genetics p and p committee.
it is my understanding that leslie lange, as mesa's lead representative to the topmed anthropology group (joined by xiuqing guo and ida chen), has submitted or is in the process of submitting to the mesa genetics p and p.

leslie:

please confirm.

once again, thank you for including mesa in this interesting work.

From: Peter Visscher <peter.visscher@uq.edu.au>

Sent: Thursday, February 21, 2019 8:20 AM

To: Thorington, Daune

Cc: Rotter, Jerome; Pierrick Wainschtein

Subject: Re: Corrections and Additions - RE: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity

[External Email]

Dear Daune,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you or Jerome in a position to give this consent (in an email reply) or should we go through a more formal process?

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cns.genomics.com

From: Peter Visscher <peter.visscher@uq.edu.au>
Date: Wednesday, 13 February 2019 at 1:17 AM
To: "Thorington, Daune" <daunet@labiomed.org>, "Cathy C. Laurie" <cclaurie@uw.edu>
Cc: "Rotter, Jerome" <jrotter@labiomed.org>, Pierrick Wainschtein <p.wainschtein@imb.uq.edu.au>
Subject: Re: Corrections and Additions - RE: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity

Dear Daune,

Thank you for your quick response. We will make the changes!

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "Thorington, Daune" <daunet@labiomed.org>
Date: Tuesday, 12 February 2019 at 5:57 AM
To: "Cathy C. Laurie" <cclaurie@uw.edu>, Peter Visscher <peter.visscher@uq.edu.au>
Cc: "Rotter, Jerome" <jrotter@labiomed.org>
Subject: Corrections and Additions - RE: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity

Good Morning,

In reviewing the affiliations and acknowledgment sections of the paper, please make the following corrections/additions:

The affiliation for Drs. Rotter, Guo, Chen (#15) should read:

The Institute for Translational Genomics and Population Sciences, Department of Pediatrics, Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center, Torrance, CA, USA

Cohort Specific Acknowledgements should include:

MESA and the MESA SHARe project are conducted and supported by the National Heart, Lung, and Blood Institute (NHLBI) in collaboration with MESA investigators. Support for MESA is provided by contracts HHSN268201500003I, N01-HC-95159, N01-HC-95160, N01-HC-95161, N01-HC-95162, N01-HC-95163, N01-HC-95164, N01-HC-95165, N01-HC-95166, N01-HC-95167, N01-HC-95168, N01-HC-95169, UL1-TR-000040, UL1-TR-001079, UL1-TR-001420. The provision of genotyping data was supported in part by the National Center for Advancing Translational Sciences, CTSI grant UL1TR001881, and the National Institute of Diabetes and Digestive and Kidney Disease Diabetes Research Center (DRC) grant DK063491 to the Southern California Diabetes Endocrinology Research Center.

Thank you!
Daune

Daune B. Thorington
Administrative Manager to Jerome I. Rotter, M.D.
Institute for Translational Genomics and Population Sciences
LABioMed at Harbor-UCLA Medical Center
1124 W. Carson Street
Torrance, CA 90502

T. (310) 974-9339
F. (310) 533-8519
daunet@labiomed.org

Begin forwarded message:

From: "Cathy C. Laurie" <cclaurie@uw.edu>
To: "North, Kari E" <kari_north@unc.edu>, "Lange, Leslie" <LESLIE.LANGE@UCDENVER.EDU>, "topmed_anthropometry_wg@uw.edu" <topmed_anthropometry_wg@uw.edu>
Cc: "Anne Justice" <aejustice1@geisinger.edu>
Subject: Re: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity

[External Email]

Hi All,

Peter Visscher and I would appreciate your consideration on the call tomorrow of the attached manuscript for submission to the TOPMed publications system.

Best wishes,

Cathy

From: Topmed_anthropometry_wg <topmed_anthropometry_wg-bounces@mailman12.u.washington.edu> **On Behalf Of** North, Kari E
Sent: Sunday, February 10, 2019 5:33 PM
To: Lange, Leslie <LESLIE.LANGE@UCDENVER.EDU>; topmed_anthropometry_wg@uw.edu
Cc: Anne Justice <aejustice1@geisinger.edu>
Subject: [Topmed_anthropometry_wg] TOPMed Anthropometry and Adiposity

Dear colleagues,

This is a reminder for our TOPMed Anthropometry and Adiposity call scheduled for 1pm ET / Noon CT / 11am MT / 10am PT this Tuesday- TODAY.

For the call, please use the usual phone number and access code:

Toll-free US and Canada: [1-866-477-1355](tel:1-866-477-1355)

Conference code: 271-923-2731

Please bring any agenda items that you'd like discussed.
We will discuss the working papers, attached.
Anne will send around a link to take attendance.

Notes

1. Group updates
 1. Leslie and Steve have stepped down as leaders. Kari, Ching-Ti, and Anne will step in to co-lead (contact info below)
 2. Kari North – University of North Carolina (kari_north@unc.edu)
 3. Ching-Ti Liu – Boston University (ctliu@bu.edu)
 4. Anne Justice – Geisinger Health (anne.justice@unc.edu or aejustice1@geisinger.edu)
2. Progress on the waist traits (Elise Lim and Ching-Ti).
 1. See attachment (Topmed-freeze5b_wc_whr.pptx)
 2. In addition to WC, WCadjBMI, and WHRadjBMI, we will perform analyses for WHR (unadjusted for BMI) to address the potential for collider bias in significant associations.
 3. Currently using genome-wide significance threshold for rare variant (gene-based) test. We will relax the threshold to account for the number of genes tested, but this will not affect the results.
 4. The GC lambdas for gene-based test are not reliable, as the lambda test was created to assess inflation on single variants tests, but the distribution of test statistics for SKAT are more complex, and therefore are not expected to follow the same distribution. The QQ plots may still be examined to detect obvious inflation, but we can ignore the lambda values.
 5. Consider doing ancestry-specific analyses to ensure that one ancestry is not weighting the results.
 6. (Slide 23) when conditioning on the top variant for each chromosome, all other significant results seem to go away. Elise will look further into this to figure out why the conditional analysis has such a large influence (especially for chr X).
 7. We will look into different masks for filtering the variants in the SKAT test (i.e. consider removing synonymous variants, prediction algorithms [polyphen]).
 8. Consider sex-specific analysis.
 9. Jen Brody is working on masks for non-coding regions.
3. Analysis Commons
 1. If you need access, please contact Jen Brody
4. **Next Meeting on Tuesday, February 12th at 1:00 PM, EST**
 1. We will review active paper proposals

Thanks!
Kari

recipient, please notify us by return email, and delete this message from your computer at once.

Subject: Re: TOPMED paper
Date: Thursday, 28 February 2019 at 12:47:53 am Australian Eastern Standard Time
From: Ellinor, Patrick Thomas, M.D., Ph.D.
To: Peter Visscher
CC: Pierrick Wainschtein
Priority: High
Category: Green

Peter,

Of course, happy to support the inclusion of the MGH AF Study data and the manuscript. I have updated the acknowledgements and my disclosures.

It looks like a subset of the AF studies was included, but not all, I was curious, was there some reason why you were unable to include them? (Some of the studies are Partners, WGHS). This isn't a big deal, but it has led to some confusion in our group so I am just trying to see how we can be helpful in the future.

In the meantime, good luck w the submission!

Best,
Patrick

On Feb 27, 2019, at 1:56 AM, Peter Visscher <peter.visscher@uq.edu.au> wrote:

External Email - Use Caution

Dear Patrick,

I would be wonderful if you could give us explicit consent to submit the manuscript on behalf of the MGH_AF cohort.

For your information, I attach the manuscript.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>
Date: Friday, 22 February 2019 at 5:53 AM
To: Peter Visscher <peter.visscher@uq.edu.au>, Mina Chung <chungm@ccf.org>, Patrick Ellinor < >

Cc: "Roden, Dan" <dan.roden@vumc.org>

Subject: RE: TOPMED paper

Hi Peter, I can give you permission for VAFAR and VU_AF, but CCAF and MGH_AF are Mina Chung and Patrick Ellinor's cohorts, respectively. I have cc'd them.

Thanks,

-Ben

From: Peter Visscher <peter.visscher@uq.edu.au>

Sent: Thursday, February 21, 2019 10:24 AM

To: Shoemaker, Moore Benjamin <moore.b.shoemaker@vumc.org>

Cc: Roden, Dan <dan.roden@vumc.org>

Subject: Re: TOPMED paper

Dear Ben,

It was brought to my attention that we need explicit consent from each cohort to submit the manuscript for publication. Are you in a position to give this consent (in an email reply) for the AFGen cohorts (CCAF, MGH_AF, VAFAR, VU_AF) or should we go through a more formal process?

I attach the latest (~final) version.

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cns.genomics.com

From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>

Date: Thursday, 27 December 2018 at 22:37 PM

To: Peter Visscher <peter.visscher@uq.edu.au>

Cc: "Roden, Dan" <dan.roden@vumc.org>

Subject: TOPMED paper

Hi Peter,

Thanks for including us on the paper. Dawood Darbar is another author that should be included. Our author list and affiliations are below

M. Benjamin Shoemaker, MD, MSCI (please include my first name's initial),
Department of Medicine, Vanderbilt University Medical Center, Nashville, TN

Dan Roden, MD. Departments of Medicine, Pharmacology and Bioinformatics.
Vanderbilt University Medical Center, Nashville, TN

Dawood Darbar, MD. Department of Medicine. University of Illinois-Chicago.
Chicago, IL

Best,

-Ben

M. Benjamin Shoemaker, MD, MSCI
Assistant Professor, Division of Cardiovascular Medicine
Vanderbilt Center for Arrhythmia Research and Therapeutics (VanCART)
Vanderbilt University Medical Center
<20190227_TOPMed_WGS.docx>

The information in this e-mail is intended only for the person to whom it is addressed. If you believe this e-mail was sent to you in error and the e-mail contains patient information, please contact the Partners Compliance Helpline at <http://www.partners.org/complianceline> . If the e-mail was sent to you in error but does not contain patient information, please contact the sender and properly dispose of the e-mail.

Subject: FW: TOPMED paper
Date: Friday, 22 February 2019 at 2:51:47 pm Australian Eastern Standard Time
From: Peter Visscher
To: Pierrick Wainschtein
Category: Green

FYI

Best wishes,

Peter

Peter M. Visscher | Program in Complex Trait Genomics | Institute for Molecular Bioscience, University of Queensland | St Lucia, Brisbane, Australia, 4072 | t: +61 7 3346 6348 | w: cnsgenomics.com

From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>
Date: Friday, 22 February 2019 at 5:53 AM
To: Peter Visscher <peter.visscher@uq.edu.au>, Mina Chung <chungm@ccf.org>, Patrick Ellinor <ellinor@mgh.harvard.edu>
Cc: "Roden, Dan" <dan.roden@vumc.org>
Subject: RE: TOPMED paper

Hi Peter, I can give you permission for VAFAR and VU_AF, but CCAF and MGH_AF are Mina Chung and Patrick Ellinor's cohorts, respectively. I have cc'd them.

Thanks,

-Ben

From: Peter Visscher <peter.visscher@uq.edu.au>
Sent: Thursday, February 21, 2019 10:24 AM
To: Shoemaker, Moore Benjamin <moore.b.shoemaker@vumc.org>
Cc: Roden, Dan <dan.roden@vumc.org>
Subject: Re: TOPMED paper

Dear Ben,

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I attach the latest (~final) version.

Best wishes,

Peter

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From: "Shoemaker, Moore Benjamin" <moore.b.shoemaker@vumc.org>

Date: Thursday, 27 December 2018 at 22:37 PM

To: Peter Visscher <peter.visscher@uq.edu.au>

Cc: "Roden, Dan" <dan.roden@vumc.org>

Subject: TOPMED paper

Hi Peter,

Thanks for including us on the paper. Dawood Darbar is another author that should be included. Our author list and affiliations are below

M. Benjamin Shoemaker, MD, MSCI (please include my first name's initial), Department of Medicine, Vanderbilt University Medical Center, Nashville, TN

Dan Roden, MD. Departments of Medicine, Pharmacology and Bioinformatics. Vanderbilt University Medical Center, Nashville, TN

Dawood Darbar, MD. Department of Medicine. University of Illinois-Chicago. Chicago, IL

Best,

-Ben

M. Benjamin Shoemaker, MD, MSCI
Assistant Professor, Division of Cardiovascular Medicine
Vanderbilt Center for Arrhythmia Research and Therapeutics (VanCART)
Vanderbilt University Medical Center



MEMORANDUM

Date: March 7, 2019

To: Pierrick Wainschtein
Peter Visscher

From: Lindsey Bull
P&P Committee Coordinator

Subject: Manuscript 3856 – Recovery of trait heritability from whole genome sequence data

Congratulations! The Publications and Presentations (P&P) Committee has approved this paper with recommended changes. Please see the review at the end of this memo for details.

You may now submit your manuscript for publication. It is important to keep WHI updated of your manuscript's status on an ongoing basis. Please notify P&P when your manuscript is submitted to a journal and accepted for publication by e-mailing (p&p@whi.org). As they become available, please send (1) a copy of the manuscript as accepted by the journal, (2) the published manuscript in PDF form, and (3) the NIH Manuscript Submission number/PubMed Central number. Authors are required to submit their published manuscript to PubMed Central; see <http://publicaccess.nih.gov> if you are unfamiliar with the NIH Public Access Policy.

Please be aware that lead authors are responsible for identifying and reporting any conflicts of interest among co-authors and noting these in the final manuscript, per journal requirements. Please ensure that all authors complete and report their conflicts on this paper. In addition, please be sure to keep all co-authors informed of your manuscript's progress. If a journal requires revisions, please solicit and incorporate co-author input and inform the P&P Chairs of any major disagreements among the writing group.

Please be reminded that, in accordance with the data use agreement you signed to obtain WHI data for this paper, the data may only be used for this approved analysis and may not be shared with anyone without authorization from WHI.

WHI Policy requires papers to be published with an appendix acknowledging WHI Investigators; please use the “short” list of WHI investigators that is available online at <https://www.whi.org/researchers/Documents%20%20Write%20a%20Paper/WHI%20Investigator%20Short%20List.pdf>. All WHI publications must also include an acknowledgement that approximates the following: “The WHI program is funded by the National Heart, Lung, and Blood Institute, National Institutes of Health, U.S. Department of Health and Human Services through contracts HHSN268201600018C, HHSN268201600001C, HHSN268201600002C, HHSN268201600003C, and HHSN268201600004C.” We also request that you consult www.whi.org for exact wording if you include a description of the WHI trials in your manuscript.

Review 1:

Comments:

This is a well-crafted paper and potentially adds to the knowledge of explaining heritability. The only suggestion is to add the acknowledgements for the Women's Health Initiative cohort in that respective section.