Author's response to reviews

Title: Evaluation of microdeletions and microduplications associated with cognitive impairment in a large cohort of subjects with autism spectrum disorders identifies duplications at 15q11-q13, 22q11 and Xp11/TM4SF2

Authors:

Guiqing Cai (guiqing.cai@mssm.edu)
Lisa Edelmann (lisa.edelmann@mssm.edu)
Juliet E Goldsmith (jaykat28@aol.com)
Ninette Cohen (ninette.cohen@mssm.edu)
Alisa Nakamine (alisa.nakamine@mssm.edu)
Jennifer G Reichert (jennifer.reichert@mssm.edu)
Ellen J Hoffman (ellen.hoffman@mssm.edu)
Danielle M Zurawiechi (danielle.zurawiecki@mssm.edu)
Jeremy M Silverman (jeremy.silverman@mssm.edu)
Catalina Betancur (Catalina.Betancur@creteil.inserm.fr)
Joseph D Buxbaum (joseph.buxbaum@mssm.edu)

Version: 2 Date: 5 March 2008

Author's response to reviews: see over
March 5th, 2008

To the Editors, BMC-Medical-Genomics:

Enclosed please find a manuscript for consideration as an article, entitled “Evaluation of microdeletions and microduplications associated with cognitive impairment in a large cohort of subjects with autism spectrum disorders identifies duplications at 15q11-q13, 22q11 and Xp11/TM4SF2.”

Autism spectrum disorders (ASDs) are pervasive developmental disorders with strong genetic basis. Known chromosomal microdeletions and microduplications associated with mental retardation have been reported to present with autistic features in some cases, but whether such chromosomal abnormalities are found in non-syndromal cases, ascertained for ASDs, has not been fully explored. In the current report, we studied the majority of common microdeletions/microduplications associated with cognitive impairment (16 recurrent autosomal deletions/duplications regions, 15 loci on the X chromosome and 2 loci on the Y chromosome), in a large cohort of 279 unrelated affected ASD patients. Two small, atypical de novo duplications in 15q11-q13 and a partial duplication of the TM4SF2 gene on Xp11.4 were identified and are here for the first time reported as potential causes of ASD. We also identified duplications in 22q11 as likely contributing to ASD. Based on the scope of the study and the novel findings in this cohort, we think our manuscript would be suitable for publication in BMC Medical Genomics.

All authors on this manuscript agree to the BMC Medical Genomics editorial policies. Furthermore, there are no competing interests identified.

We would suggest the following individuals as potential reviewers as they are highly qualified to review this work:
Astrid M. Vicente
Instituto Nacional de Saúde Dr. Ricardo Jorge, Av. Padre Cruz, 1649-016 Lisboa, Portugal, and Instituto Gulbenkian de Ciência, Ap. 14, 2781-901, Oeiras, Portugal
email: avicente@igc.gulbenkian.pt

Edwin H. Cook Jr
Institute of Juvenile Research, Department of Psychiatry, University of Illinois at Chicago, Chicago, IL 60612, USA
email: ecook@psych.uic.edu

Michael L. Cuccaro
Miami Institute for Human Genomics, Miller School of Medicine, University of Miami, Miami, FL 33101, USA
email: mcuccaro@med.miami.edu

Gerard D. Schellenberg
GRECC S-182B, Veterans Affairs Puget Sound Health Care System, 1660 S. Columbian Way, Seattle, WA 98108, USA
email: zachdad@u.washington.edu

James S. Sutcliffe
Department of Molecular Physiology and Biophysics, Center for Molecular Neuroscience, 702 Light Hall, Vanderbilt University, Nashville, TN 37232-0615, USA
email: james.s.sutcliffe@vanderbilt.edu

As the manuscript was transferred from BMC Genomics to BMC Medical Genomics, some changes have been made: Methods section has been moved to the place after Background; Two typographical errors have been corrected (one author name “Ellen J. Hofman” in the title page should be “Ellen J. Hoffman”, and “5’ UTR” in the last paragraph of Discussion should be “3’ UTR”).
We would be very grateful if you would consider our manuscript for publication in BMC Medical Genomics.

Sincerely yours,

Guiqing Cai and Joseph D. Buxbaum