Author's response to reviews

Title: A Novel RUNX2 Missense Mutation Predicted to Disrupt DNA Binding Causes Cleidocranial Dysplasia in a Large Chinese Family with Hyperplastic Nails

Authors:

Shaohua Tang (tsh006@163.com)
Qiyu Xu (0105062@sohu.com)
Xueqin Xu (xxq@yilian.com.cn)
Jicheng Du (wzDu.Jicheng@163.com)
Xuemei Yang (yxmcqzd@126.com)
Yusheng Jiang (yusheng0104@hotmail.com)
Xiaoqing Wang (wxqwz@126.com)
Nancy Speck (Nancy.Speck@Dartmouth.edu)
Taosheng Huang (huangts@uci.edu)

Version: 2 Date: 14 September 2007

Author's response to reviews: see over
Sept 14, 2007

Editor-in-Chief
BMC Medical Genetics

Dear Sir/Madam

I just submitted a manuscript entitled “A Novel RUNX2 Missense Mutation Predicted to Disrupt DNA Binding Causes Cleidocranial Dysplasia in a Large Chinese Family with Hyperplastic Nails” We report a large Chinese family with CCD with a new phenotype, hyperplastic nail. The clinical features show a significant intrafamilial variation. The family carries a c.T407C (p.L136P) mutation. Based on molecular modeling, this novel missense mutation in the RUNT domain may disrupt RUNX2-DNA interaction.

If the manuscript is accepted, I will appreciate your consideration to use the molecular model (Figure 4B) as the cover of the journal.

The author(s) declare that they have no competing interests.

Yours,

Taosheng Huang, M.D., Ph.D.

Department of Pediatrics & Developmental Biology, Pathology
Director, MitoMed Molecular Diagnostic Lab
Robert R. Sprague Hall, Room 314 Zot 3950
College of Medicine
University California
Irvine, CA 92697
Huangts@uci.edu
949-824-9346 (o)
949-824-9466 (L) Fax 824-9776