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Invitation from Shree Ram Singh to review a manuscript for BMC Genetics - GTIC-D-15-00167

1 message

BMC Genetics Editorial Office <em@editorialmanager.com> Fri, Oct 16, 2015 at 8:54 AM
Reply-To: BMC Genetics Editorial Office <bmcgenet@biomedcentral.com>
To: Shicheng Guo <scguo@ucsd.edu>

GTIC-D-15-00167
The CYLD p.R758X worldwide recurrent nonsense mutation detected in patients with multiple familial trichoepithelioma type 1, Brooke-Spiegler syndrome and familial cylindromatosis represents a mutational hotspot in the gene
Nikoletta Nagy; Katalin Farkas; Barbara Kocsis Deák; Laura Cubells Sánchez; Ana Mercedes Victoria Martínez; Juan José Vilata Corell; Alfredo Montoro Botella; Goitzane Marcaida Benito; Raquel Rodríguez López; Tomas Vanecek; Dmitry V. Kazakov; Joan N. R. Kromosoeto; Ans M.W. van den Ouweland; János Varga; Márta Széll
BMC Genetics

Dear Dr. Guo,

I would like to invite you to review the manuscript above which has been submitted to BMC Genetics. Further details including the full abstract can be found at the end of this email.

If you are able to review this submission please click on this link:

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In order to keep delays to a minimum, please accept or decline this invitation online within the next few days. If you are unable to review the manuscript, we would be most grateful if you could suggest alternative reviewers.

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Thank you for your time, and I look forward to hearing from you.

Best wishes,

Shree Ram Singh
BMC Genetics
http://www.biomedcentral.com/bmcgenet

GTIC-D-15-00167
Research article
The CYLD p.R758X worldwide recurrent nonsense mutation detected in patients with multiple familial trichoepithelioma type 1, Brooke-Spiegler syndrome and familial cylindromatosis represents a mutational hotspot in the gene
BMC Genetics

Abstract: Background Multiple familial trichoepithelioma type 1 (MFT1; MIM 601606), a rare monogenic skin disease with autosomal dominant inheritance, is characterized by the development of multiple skin-colored papules on the central area of the face, frequently occurring in the nasolabial area. The disease is associated with various mutations in the cylindromatosis (CYLD) gene that are also responsible for familial cylindromatosis (FC) and Brooke-Spiegler syndrome (BSS).
Methods Recently we have identified a Spanish MFT1 pedigree with two affected family members (father and daughter). Direct sequencing of the CYLD gene revealed a worldwide recurrent heterozygous nonsense mutation (c.2272C/T, p.R758X) in the patients.
Results This mutation has already been detected in patients with all three clinical variants - BSS, FC and MFT1 - of the CYLD-mutation spectrum. Haplotype analysis was performed for the Spanish patients with MFT1, Dutch patients with FC and an Austrian patient with BSS, all of whom carry the same heterozygous nonsense p.R758X CYLD mutation.
Conclusions Our results indicate that this position is a mutational hotspot on the gene and that patients carrying the mutation exhibit high phenotypic diversity.