Author's response to reviews

Title: Exome sequencing identifies a novel mutation in PIK3R1 as the cause of SHORT syndrome

Authors:

Clea Barcena (cbf@degradome.uniovi.es)
Victor Quesada (quesadavictor@uniovi.es)
Annachiara De Sandre-Giovannoli (annachiara.DeSandre@mail.ap-hm.fr)
Diana A. Puente (diana@degradome.uniovi.es)
Joaquin Fernandez-Toral (jftjfb@hotmail.com)
Sabine Sigaudy (Sabine.Sigaudy@mail.ap-hm.fr)
Anwar Baban (anwar.baban@opbg.net)
Nicola Levy (Nicolas.Levy@medicine.univ-mrs.fr)
Gloria Velasco (gvc@uniovi.es)
Carlos Lopez-Otin (clo@uniovi.es)

Version: 3
Date: 22 April 2014

Author's response to reviews: see over
Dear Editor:

We are very thankful for the positive response to the revision of our manuscript entitled “Exome sequencing identifies a novel mutation in PIK3R1 as the cause of SHORT syndrome” (MS ID: 1901502901112736) by Barcena et al.

We have included in the manuscript a statement clarifying that patient 1 and the father of patient 2 gave us an informed consent for the publication of their participation in this study as well as the accompanying images.

Also, we agree to change the article type from Research Article to Case Report, as it will probably be more accurate considering that only two patients were analyzed in this study.

To close, we would like to thank again the positive reception of our revised manuscript.

Yours sincerely,

Professor Carlos López-Otín

Departamento de Bioquímica y Biología Molecular
Facultad de Medicina, Universidad de Oviedo
33006-Oviedo, Spain
Tel. +34 985104201 Fax. +34 985103564
E-mail: clo@uniovi.es
Point by point reply to the Editorial’s requests

1. Please clarify whether patients (or the parents of patients under the age of 16) gave their informed consent for the publication of the pictures in Figure 1. If they did then please include a statement to this effect in the manuscript. If not, then please remove these pictures from the manuscript.

Our response: Both families (patient 1 and father of patient 2) gave us a signed informed consent for the publication of any image of themselves accompanying the manuscript.

2. We note that only 2 patients were analyzed in this study, and so we would like to change the article type from a 'research article' to a 'case report'. We apologize for not noticing this sooner. Please confirm whether you are happy for us to change the article type.

Our response: We confirm that we agree to change the article type.