Author's response to reviews

Title: Whole exome sequencing in an Indian family links Coats plus syndrome and dextrocardia with a homozygous novel CTC1 and a rare HES7 variation.

Authors:

Manjunath Netravathi (sundernetra@yahoo.co.in)
Renu Kumari (renu.kumari@igib.in)
Saketh Kapoor (kapoor.saketh@gmail.com)
Pushkar Dakle (pushkar.dakle@igib.in)
Manish K Dwivedi (mk.dwivedi@igib.in)
Sumitabho Deb Roy (sumitabhodebroy@gmail.com)
Paritosh Pandey (paritosh2000@gmail.com)
Jitender Saini (jsaini76@gmail.com)
Anil Ramakrishna (anilramakrishna78@yahoo.co.in)
Devaraddi Navallia (devaraddi.navalli@rediffmail.com)
Parthasarathy Satishchandra (drpsatishchandra@yahoo.com)
Pramod Kumar Pal (pal.pramod@rediffmail.com)
Arun Kumar (karun@mrdg.iisc.ernet.in)
Mohammed Faruq (faruq.mohd@igib.in)

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Author's response to reviews: see over
To

The Editor,

BMC Medical Genetics

Subject: revised submission of our manuscript MS: 171256222136259, case report

We are sincerely thankful to the editor for accepting our manuscript. We are submitting our revised manuscript entitled “Whole exome sequencing in an Indian family links Coats plus syndrome and dextrocardia with a homozygous novel CTC1 and a rare HES7 variation.” We have done changes and provide the information as required/suggested by the editor. Below are our responses to editorial comments:

Editor’s comments:

1. Please include details of the ethics committee that approved this work (or include a statement as to why approval was not required). Research involving human subjects, human material, or human data, must have been performed in accordance with the Declaration of Helsinki and must have been approved by an appropriate ethics committee. A statement detailing this, including the name of the ethics committee and the reference number where appropriate, must appear in all manuscripts reporting such research. If a study has been granted an exemption from requiring ethics approval, this should also be detailed in the manuscript (including the name of the ethics committee that granted the exemption). Further information and documentation to support this should be made available to Editors on request. Manuscripts may be rejected if the Editor considers that the research has not been carried out within an appropriate ethical framework. In rare cases, Editors may contact the ethics committee for further information.

Response: As we had mentioned in our ethics statement that “This research followed the tenets of the Declaration of Helsinki and the guidelines of the Indian Council of Medical Research, New Delhi. A written informed consent was obtained from the parents for
This study was a collaborative effort to diagnose and manage a rare case Coats plus syndrome between three institutes, where a tertiary referral centre National Institute of Mental Health and NeuroSciences (NIMHANS, Deemed University)- Bangalore, India had identified the case and subjected the case for genetic diagnosis to both Indian Institute of Science (IISc) and CSIR-Institute of Genetics and Integrative Biology (CSIR-IGIB). In India, generally these tests are not available routinely through commercial ventures and NIMHANS has taken help of IISc and CSIR-IGIB for getting diagnosis of this case for further management of the patient in study (On the basis of patient’s health care needs). In the course of genetic investigations the identified results are sent for publication with prior approval from family relatives (Consent form has been sent along). The study protocol followed Declaration of Helsinki and the guidelines of the Indian Council of Medical Research, New Delhi for conducting diagnostic research.

2. Please rename the Methods heading in the abstract to Case presentation.
   We have corrected it.

Please find our revised manuscript for your further perusal

Looking forward to a favorable response.

Best regards
Faruq

Dr Mohammed Faruq, MBBS, PhD
Scientist
Genomics and Molecular Medicine,
CSIR-Institute of Genomics and Integrative Biology (CSIR-IGIB),
Mall Road, New Delhi, India.
Tel: 91-11-27666156; Fax: +91-11-27667471
Email: faruq.mohd@igib.res.in/faruq.mohd@igib.in