Author’s response to reviews

Title: Bilateral radial agenesis with absent thumbs, complex heart defect, short stature, and facial dysmorphism in a patient with pure distal microduplication of 5q35.2-5q35.3

Authors:

Aleksander Jamsheer (jamsheer@wp.pl)
Anna Sowinska (sowka7@wp.pl)
Malgorzata Jamsheer-Bratkowska (mjamsheer@pzh.gov.pl)
Tomasz Trzeciak (trzeciak@orsk.ump.edu.pl)
Anna Latos-Bielenska (anna.latosbielenska@gmail.com)

Version: 2 Date: 25 May 2012

Author’s response to reviews: see over
FROM:
Aleksander Jamsheer, MD, PhD
Poznan University of Medical Sciences
ul. Grunwaldzka 55, Paw. 15
60-352 Poznan, Poland
Tel: 0048 61 8547345
Fax: 0048 61 8547348
jamsheer@wp.pl

RE: “Bilateral radial agenesis with absent thumbs, complex heart defect, short stature, and facial dysmorphism in a patient with pure distal microduplication of 5q35.2-5q35.3.”

May 25th, 2012

Dear Editor,

We would like to resubmit our manuscript entitled “Bilateral radial agenesis with absent thumbs, complex heart defect, short stature, and facial dysmorphism in a patient with pure distal microduplication of 5q35.2-5q35.3” for your consideration to publish in BMC Medical Genetics. This original report has not been submitted to any other journals, and the results have not been released in an abstract or conference presentation form.

The following points have been addressed and the manuscript is corrected according to your suggestions.

1) Please can you email us a copy of the consent given by the parents - enclosed

2) Please can you rephrase the consent statement to indicate that the parents, and not the patient, gave consent to take part and confirm that they gave permission for publication of the images - corrected accordingly

3) Please can you indicate whether appropriate ethical approval and informed consent was obtained from the patients' parents and the 8 unrelated probands to participate, and include details in the manuscript - corrected accordingly

We would be grateful for your further consideration.

With Kind Regards,

Aleksander Jamsheer, MD, PhD