Author's response to reviews

Title: Testicular histological and immunohistochemical aspects in a post-pubertal patient with 5 alpha-Reductase type 2 Deficiency. Case report and review of the literature in a perspective of evaluation of potential fertility of these patients.

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Version: 2
Date: 10 January 2014

Author's response to reviews: see over
Editors
BMC Endocrine Disorders

January 10th, 2014

Dear Editors:

Please find enclosed the revised version of the manuscript entitled “Testicular histological and immunohistochemical aspects in a post-pubertal patient with 5α-Reductase type 2 Deficiency. Case report and review of the literature in a perspective of evaluation of potential fertility of these patients.” by Lavinia Vija, Sophie Ferlicot, Diana Paun, Hélène Bry-Gauillard, Gabriela Berdan, Issam Abd-Alsamad, Marc Lombès and Jacques Young, that we are submitting for consideration to BMC Endocrine Disorders.

As requested, we added the e-mail addresses of all authors on the title page as well as their names, institutions, countries and full postal address of the submitting author. We also correctly entitled the “Conclusions” and the “List of abbreviations” sections and uploaded the figures as a single figure file and not as an additional file.

To our knowledge, we present here the first testicular histological and immunohistochemical description in a postpubertal patient, with a genetic
confirmation of the 5α-R2 deficiency. Our genetic confirmation is important, insofar as the simple clinical and hormonal analysis of DSD and particularly of 5α-Reductase type 2 Deficiency cases can sometimes lead to wrong diagnoses. This point was raised by a number of recent publications on genetically analyzed DSD patients.

Finally, in this manuscript we also make a detailed review of the literature and we will discuss on the whole our and literature's histological findings, in the more general context of evaluating the fertility potential of these patients if they were raised as males. The literature on these patients, essentially pediatric, does not address these issues that are crucial for these patients when they become adults. We believe that this case and review that accompanies it is important not only for pediatricians but also for urologists and andrologists who will have to take care of these patients. We think therefore that, on the whole, our paper makes a significant contribution to medical knowledge and has educational value, highlighting the need for a change in clinical practice or diagnostic/prognostic approaches of this condition, as required by BMC Endocrine Disorders editorial policy.

As potential reviewers we suggest: Rodolfo Rey, Ilpo Huhtaniemi, Jorma Toppari.

We wish not to have the manuscript reviewed by Charles Sultan or Patrick Fénichel for reason of conflict of interest.
We hope you will find our work to be of interest, and would be pleased to provide you with any further information you may require.

Sincerely yours,

Professor Jacques Young, MD, PhD

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