Author's response to reviews

Title: Female genital mutilation of a karyotypic male presenting as a female with delayed puberty

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

Please find enclosed the revised version of our manuscript *Female Genital Mutilation of a karyotypic male presenting as a case of female with delayed puberty.*

After discussion among the authors, we have made the following changes in the manuscript according to the comments from the reviewers:

1- We have changed the title to: Female genital mutilation of a karyotypic male presenting as a female with delayed puberty.

2- Other affiliations have been added to the first and last authors.

3- Dr. M.A. El-Gohari has revised the manuscript and its language. That is why we have have thanked him in the acknowledgement.

**For the changes reviewer 1 has recommended:**

1- What does “aggressive” FGM mean?

The word aggressive has been explained, as we mean (type IV FGM). The term “aggressive” has been removed for clarity.

2- p. 3: Regarding “psychological” complications, does it matter who performs FGM?

In this sentence we have explained the psychological problem that comes from the pain due to the procedure of FGM itself, because it is an invasive and painful surgical procedure that is usually performed without anesthesia, or with local anesthesia, often resulting in serious psychological and medical complications for the young girls.

3- Regarding the patient’s shyness about sexual feelings, is this typical for the cultural background?

Regarding this point we have tried to explain it by pointing out that the patient thinks of her self as a female and was very shy to tell if she was attracted to males or females; which could be due to her cultural background. But still there could be other psychological background issues which are beyond the scope of this study.

4- Can one comment on whether the patient was given any feedback about the medical evaluation?

In this case it was difficult for the patient’s doctor to inform her about the final medical evaluation other than she is an infertile female. This has now been added to the text. The intersex problem in Sudan has not been given much attention before in clinical practice and there are no national guidelines for management and counseling.
5- How would one address issues of fertility in such a patient?

However, here precise diagnosis and her prognosis including her potential response to hormonal therapy and subsequent fertility remained unclear. This has now been added to the text.

6- If this patient has a female gender identity, is her psychological situation any different from normal biological females who experience FGM?

In the present case, the sex of rearing appeared to be correct, as the patient regarded herself as female. But the combination of FGM and the absence of normal female development probably subjected this patient to a twofold psychological trauma. This has now been added to the text.

7- Fig. 4 seems unnecessary.

All the authors agreed with deleting Fig.4.

8- The language has been corrected by one of my professor who is familiar with intersex conditions.

**Reviewer 2 comments:**

1. This case report is of interest to two groups (1) those who are concerned with the practice of female circumcision in phenotypically normal females and (2) those who are concerned with genital feminization in individuals affected by disorders of sex differentiation. The authors suggest that female circumcision is problematic not only due to the harm that it entails on girls and women, but also because it makes it difficult to pick up cases of 46,XY ntersexuality where presumably the external genitalia are ambiguous at birth. My major problem with this is that we do not know if the reported case exhibited ambiguous genitalia, and depending on the etiology of their condition (for example complete AIS, Swyer syndrome) they may very well have had normal appearing female external genitalia at birth. The authors seem to suggest that if the genitalia in such cases were not altered by practices of female circumcision, then an accurate diagnosis might be possible. However, it is generally accepted that the external genital phenotype is not adequate to make such diagnoses as over-masculinied female pseudohermaphrodites can appear no different from under-masculinized male pseudohermaphrodites. Therefore, the claim that female circumcision precluded proper diagnosis of such cases is unjustified.

Regarding this point we have now explained that FGM might have limited the possibility of clinical assessment, although the configuration of the external genitalia is not always essential for a correct diagnosis, as over-masculinized female pseudohermaphrodites can be similar to under-masculinized male pseudohermaphrodites. The male chromosome complement suggests that the patient suffered from a type of male pseudohermaphroditism, such as complete or incomplete androgen insensitivity syndrome. The low LH and FSH values could support this diagnosis, but the absence of breast
development and the complete absence of pubic hair do not support it. The low LH and FSH could also suggest a delayed puberty condition. However it cannot be excluded that ectopic testicular tissue in or close to the labia majora was excised during the FGM process, thus explaining the absence of puberty signs. Moreover, one cannot exclude non-intersex abnormalities of the male genital organs such as grave hypospadias or conditions of hypogonadotrophic hypogonadism such as Kallmann syndrome. Thus FGM made the surgical management difficult.

2. My second point of disagreement is that these authors suggest that a missed diagnosis lead to incorrect sex of rearing. However, it is stated that this case sees herself as female.

We agree with the reviewer comment on the sex of rearing. This has now been changed in the text.

Mona Ellaithi