Author's response to reviews

Title: Screening of Y chromosome microdeletions in 46,XY partial gonadal dysgenesis and in patients with a 45,X/46,XY karyotype or its variants

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Author's response to reviews: see over
Answers to the Editor's comments:

The authors have satisfactorily addressed the reviewers' comments. The manuscript would benefit from a few minor changes, as suggested below:

Abstract: ??(Yp, centromere and Yq) where analyzed ?? > ??(Yp, centromere and Yq) were analyzed ??

Sure! It was corrected.

Introduction, p. 4, 1st paragraph: ?Whatever the phenotype, this mosaic is associated with short stature, dysmorphisms and cardiovascular and renal anomalies, among other congenital and acquired conditions? What does ?other congenital and acquired conditions? mean?

This part of the phrase is indeed not clear. Our purpose was to state that there are other congenital and also acquired conditions associated to the 45,X/46,XY mosaicism. Maybe it would be better to write "and other congenital and acquired conditions". The change was made in the text.

Introduction, p. 4, 2nd paragraph: ?? evidences of a linkage between Y microdeletions and 45,X/46,XY mosaicism?; rather than linkage this is an association (using genetic terms appropriately)

The Editor is right. This change was also made.

Subjects, p. 5, last paragraph: ??marker chromosome, which origin was elucidated?? > ??marker chromosome, whose origin was elucidated??

The Editor is right. This change was also made.

Methods, p. 5, 2nd paragraph: ?a)? dysgenetics testis; b) two dysgenetics testes? > ?? dysgenetic testis; two dysgenetics testes:

Actually, it must be "a streak gonad and a dysgenetic testis; b) two dysgenetic testes". It was corrected.

Results, p. 7, 3rd paragraph: ??by two individual PCR of each STS? © ??by two individual PCRs of each STS?

OK, it was corrected.

Results, p. 7, 3rd paragraph: ??of the identified deletion for each patient ?? > ??of the identified deletions for each patient ??
Discussion, p. 8, 1st paragraph: ??region, which is rich testis-specific genes ?? > ??region, which is rich in testis-specific genes ??

OK, it was corrected.

Conclusions, p. 11, last paragraph: ?Absence of deletions in individuals with PGD suggests that the Y chromosome is intact, indicating that it is not likely that its instability in the gonads is one of the causes of this condition; further studies that aim to identify the etiology of this condition are needed.? > ?Absence of deletions in individuals with PGD suggests that the Y chromosome is intact, indicating that it is not likely that its instability in the gonads is one of the causes of this condition; further studies that aim to identify the etiology of this condition are needed.?

The Editor's suggestion was accepted.

Conclusions, p. 11-12: ?However, deletions identified in the second group indicate that mosaicism may be associated with Y chromosome abnormalities detectable only at the molecular level, indicating that microdeletions can be found in patients carrying an apparently normal Y chromosome in karyotyping? > ?However, deletions identified in the second group indicate that mosaicism may be associated with Y chromosome abnormalities detectable only at the molecular level?.

The Editor's suggestion was accepted.