Reviewer’s report

Title: Achondroplasia with 47, XXY karyotype: a case report of the neonatal diagnosis of an extremely unusual association

Version: 1 Date: 4 April 2012

Reviewer: Oliver Quarrell

Reviewer’s report:

Thank you for the invitation to review this case report. I would like to make the following comments:

Minor essential revisions

In the discussion, I am concerned about the use of the word "mandatory". It is very helpful for a gynaecologist, paediatrician and geneticist to work together but the word "mandatory" is, in my opinion, too strong.

I am concerned about the use of the word "mandatory" in relation to mutation testing. Given that there are only 5 recorded cases of the combination of Klinefelter and achondroplasia, I think the clinicians at the local hospital can be forgiven for accepting the chromosome report. Identifying a specific skeletal dysplasia during pregnancy is difficult, testing for achondroplasia is often a diagnosis of exclusion.

I think the term rhizomelic dwarfism at the end of page 3 should be replaced with rhizomelic shortening. We are trying to move away from terms like dwarfism.

On page 4 the upper limb bones are written in the singular form but if they are bilateral then it would be better to use the plural humeri radii and ulnae or ulnas.

Discreionary revisions.

I think the clauses in the sentence would be better the other way around:"Our case was initially diagnosed prenatally in a different institution of non-mosaic Klinefelter syndrome, based on the karyotype analysis ordered because of abnormal sonographic findings." Based on the karyotype analysis ordered because of abnormal sonographic findings, our case ..... 

I think this sentence could be adjusted "Similarly in our cases the FGFR3 mutation dominated the clinical picture and the Klinefelter syndrome was not able to counter the achondroplasia’s effects on height."

Similarly, in our case, height was dominated by the effect of the FGFR3 mutation rather than by the XXY aneuploidy.
Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests