Reviewer's report

Title: A novel GLI3 Mutation Affecting the Zinc Finger Domain Leads to Isolated Polydactyly

Version: 1
Date: 25 May 2014
Reviewer: Sajid Malik

Reviewer's report:

Major changes required:

1. Mutation should be mentioned in the Abstract.
2. GLI3 has been associated with spectrum of phenotypes. From the given information the phenotypic picture is not clear. Hence, it would be worthwhile if detailed data on preaxial polydactyly, postaxial polydactyly and syndactyly is presented. It is therefore, recommended that the clinical spectrum in all the available affected subjects should be presented in a table in the following format:

<table>
<thead>
<tr>
<th>Subject Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Preaxial polydactyly</td>
</tr>
<tr>
<td>------------------------</td>
</tr>
<tr>
<td>X Bilateral in hands; left hallux duplicated Type A in right hand, type B in left hand</td>
</tr>
<tr>
<td>Y Right thumb bifid; bilateral in feet Type B in both hands</td>
</tr>
<tr>
<td>Z -do- -do- -do-</td>
</tr>
</tbody>
</table>

   Total occurrences in 12 limbs (3 subjects) 6 right hands; 5 left hands; 8 right foot; 3 left foot 6 right hands; 5 left hands; 8 right foot; 3 left foot 6 right hands; 5 left hands; 8 right foot; 3 left foot

3. Title suggestion: Since there is concomitant presence of preaxial polydactyly, postaxial polydactyly and syndactyly, the appropriate title should be “A novel GLI3 mutation affecting the zinc finger domain leads to preaxial-postaxial polydactyly-syndactyly complex.”

4. In this continuum, it would be appropriate not to use the phrase “isolated polydactyly” in the Abstract and other texts of manuscript.

5. Because of the considerable overlap with the preaxial and postaxial polydactyly with Crossed polydactyly (MIM 174700).

Minor changes/modifications required:

1. The most recent references listed in the paper are for year 2010. Please update recent literature on polydactyly.
2. The subjects physically examined and underwent molecular study should be mentioned on the pedigree with appropriate symbols.


4. Page 5, line 108. ‘Non affected’ should be ‘unaffected’.

**Level of interest:** An article of importance in its field

**Quality of written English:** Acceptable

**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.