Author's response to reviews

Title: Type II diabetes and impaired glucose tolerance due to severe hyperinsulinism in patients with 1p36 deletion syndrome and a Prader-Willi-like phenotype

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Author's response to reviews:

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Title: Type II diabetes and impaired glucose tolerance due to severe hyperinsulinism in patients with 1p36 deletion syndrome and Prader-Willi like phenotype  
Reviewer: Fiorella Gurrieri  
Reviewer's report: The report is of interest because the knowledge of increased diabetes and obesity susceptibility in those patients is important for clinical management.

Minor essential revisions: the manuscript needs robust editing and, possibly, should be shortened as it is too long for a case report.

We thank the Reviewer for the suggestion. The article was reviewer by a native English speaker (American Journal Experts), as for the Editor suggestions.

A methylation test for Prader Willy syndrome may be useful to exclude additional genetic causes for this phenotype.

We thank the Reviewer for the suggestion. We have obviously carried out tests for Prader-Willi syndrome diagnosis. We omitted these data. We have added these tests in the text.

In the methods (page 5 and 7) it seems that array-CGH was done with two different approaches, but it is probably not the case, so please check.

We obviously thank the Reviewer for the suggestion. The two patients carried out some years ago array with another method. We have preferred to carry again array CGH in our hospital laboratory (method with higher resolution). We have deleted the phrase of the first array.

In the first case we used a platform with a resolution of about 100 kb instead in
the second one we used a platform of about 40 kb. The method is the same but they came in different time and the laboratory changed platform.

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

We thank the Reviewer for the suggestion. The article was reviewer by a native English speaker (American Journal Experts), as for the Editor suggestions.

Reviewer's report: Reviewer:Fernando Scaglia

Major compulsory revisions: the authors present two patients with 1p36 deletions and an accompanying phenotype suggestive of Prader-Willi syndrome.

1. Even if the possibility of microdeletions associated with Prader Willi syndrome has been ruled by array CGH on both patients, Prader Willi syndrome can be caused by uniparental disomy, molecular defects affecting the imprinting center, or truncating mutations in the paternal allele of MAGEL2. The authors need to rule out other possibilities by performing methylation studies for Prader Will in both patients.

We thank the Reviewer for the suggestion. We have obviously carried out tests for Padre Willi syndrome diagnosis. We are sorry for the mistake. We have added these tests in the text.

2. The authors give the impression that the array CGH done on both patients are different. They need to clarify this issue in the methods section. They also need to state whether the array is comprehensive and whether it also includes a SNP array that would be useful to exclude cases of Prader Willi due to uniparental disomy.

We obviously thank the Reviewer for the suggestion. The two patients carried out some years ago array with another method. We have preferred to carry again array CGH in our hospital laboratory (method with higher resolution). We have deleted the phrase of the first array.

In the first case we used a platform with a resolution of about 100 kb instead in the second one we used a platform of about 40 kb. The method is the same but they came in different time and the laboratory changed platform.

3. The authors need to improve the English in their report.

We thank the Reviewer for the suggestion. The article was reviewer by a native English speaker (American Journal Experts), as for the Editor suggestions.