

Dear Editor-in-Chief

Thank you for the constructive comments that help to improve the quality of our paper. Kindly find below the authors' reply to the reviewers' comments.

Reviewer #1:

1. The findings of Chromosomal Microarray Analysis (CMA), karyotyping and whole-exome sequencing should be mentioned in the Results section.

Reply: Based on the given detailed ophthalmological and medical assessment, we applied comprehensive genetic testing by performing karyotyping and chromosomal microarray analysis. Karyotype showed normal male (46, XY) with no evidence of clinically significant numerical or structural chromosome abnormalities. Moreover, we performed whole-exome sequencing (WES) that allowed us to screen the whole exome and to focus in particular on the ESCO2 gene that is known to be linked with RS and other genes related to primary congenital glaucoma such as the CYP1B1 and LTBP2 genes. Despite our detailed review of the WES data, negative results were found with no observed genetic variants or incidental findings.

2. The authors need to give differential diagnosis.

Reply: Thalidomide-induced phocomelia

Holt-Oram syndrome

Thrombocytopenia with absent radius syndrome

Sporadic phocomelia

3. The authors need to mention prenatal conditions, prenatal exposures and risk factors.

Reply: The baby was the second child of a healthy non-consanguineous couple and a product of an uneventful pregnancy with spontaneous vaginal delivery at 40 wk. Birth weight was 3 kg. There was neither a family history of similar conditions nor a prenatal history of exposure to any known teratogenic medication.

Reviewer #2:

Revision and language polishing are done.

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