Author’s response to reviews

Title: New Genetic Variant in the SERPINC1 gene: Hereditary Antithrombin Deficiency Case Report, Familial Thrombosis, and Considerations on Genetic Counseling.

Authors:

Margarita Polyak (margaritapolyak@gmail.com)

Elena Zaklyazminskaya (helenezak@gmail.com)

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In response to the review of the manuscript "New Genetic Variant in the SERPINC1: Hereditary Antithrombin Deficiency, Familial Thrombosis and Considerations on Genetic Counseling" (MGTC-D-19-00607R1).

We would like to thank the reviewers for the time taken to read, comment, and make suggestions!

In order to response to Dr. Xenofontos’ remarks, we have made the following changes to our manuscript. We have modified the “Materials and Methods” section and stated the colorimetric method used for AT quantification (Line 84), oligoprimer design (Line 87), the sequencing methodology (Line 88) and data analysis software (Line 88). We find though that detailed description of our patient’s phenotype is somewhat excessive in this case because no phenotypic features had been described for hereditary antithrombin deficiency. However, following Dr. Xenofontos’ comments, we have stated that no findings specific for syndromic or metabolic diseases were made in our patient (Lines 82-83). We also stated that repetitive measurements in our patient were performed in order to confirm the low levels of antithrombin (Line 101).

We provided a figure showing a sequence electropherogram in order to show a heterozygous state of the genetic variant c.662G>C in our patient (Figure 1B).

We have also accepted all grammar remarks.

We would like to thank Dr. Akdemir for recommended article of multiple thrombophilic gene mutations. We do fully agree that thrombophilia investigations are highly important in order to prevent pregnancy complications. However, our experience in observing patients with antithrombin deficiency is based on the only case in question. As much as we would like to comment on VTE risk and adverse perinatal outcomes we believe that it is beyond the scope of the clinical case under discussion, especially considering the fact that we do not have a history of monitoring the pregnancy of our patient so far.

We highly appreciate all the comments and suggestions.
Sincerely yours,

Margarita Polyak, MD

Laboratory of Medical Genetics
Petrovsky Russian Scientific Surgery Centre
Moscow, Russia, 119991
+74952485495
margaritapolyak@gmail.com