Reviewer’s report

Title: Familial hypercholesterolemia mutations in Petrozavodsk: no similarity to St. Petersburg mutation spectrum

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Reviewer: George Dedoussis

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In this article Komarova et al report the detection of new LDLR mutations in a population of Petrozavodsk. Besides the novelty of these mutations and the evident added value to the DNA diagnosis of the disease in this population, the authors report that these variants were not detected in their neighboring Russian populations.

The article is well written and the results are clear.

Minor comments.

1. The document needs editing by an English native speaker
2. It is not clear what was the discovery rate, in how many patients a mutation was discovered.
3. In the discussion the text claiming that the p.G20R is not a pathogenic variant is confusing. An heterozygous profile in FH is a disease profile. Please clarify.

Level of interest: An article of importance in its field

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