Reviewer's report

Title: Ovarian dysfunction and FMR1 alleles in a large Italian family with POF and FRAXA disorders: Case report

Version: 2 Date: 24 January 2007

Reviewer: Daniela Toniolo

Reviewer's report:

General

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

The revised manuscript still has the defect of presenting a family where the POF patients may derive from completely independent causes without a clear statement about this likely possibility: no further evidence is provided of any relationships between the two disorders (POF due to FMR1 premutations and POF without) by the study of additional cases and a better description of the family. To make completely clear that there is no evidence also part of the abstract and of the background sections should be modified

1. In the conclusion the sentence “Our case study represents a helpful observation to improve the clinical counseling of familial POF disorders and to avoid incorrect classification of ovarian dysfunctions” is misleading. Should be changed to “Our case study represents a helpful observation and will provide familial cases with heterogeneous etiology that could be further studied when candidate genes in addition to the FMR1 premutation will be available.

2. “The aim of this study is to offer new information regarding the complexity of familial POF conditions by presenting a large pedigree in which two different POF disorders coexist, one FMR1-related and one not FMR1-related”. Also this sentence should be changed as there is not really any new information, but just the presentation of a large pedigree: “The aim of this study is to present a large pedigree in which two different POF disorders coexist, one FMR1-related and one not FMR1-related”.

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

Discretionary Revisions (which the author can choose to ignore)

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

Level of interest: An article of limited interest

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