Reviewer's report

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

Version: 1 Date: 17 November 2006

Reviewer: Daniela Toniolo

Reviewer's report:

General
The manuscript by Miano et al. reports a three generations family segregating Premature Ovarian failure (POF) and the FMR1 premutation. Some of the women presenting POF had the FMR1 premutation. Others did not. As suggested in the discussion, the family could be a further example that different factors may cause POF, as previously suggested from different family and cytogenetic studies. However while this is not a novel finding, there is no proof in the manuscript that the two groups of patients may have some genetic factors in common that may be responsible for the disorder.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)
Only part of the family was studied and only the age of onset of menopause is reported. Are the frequent spontaneous abortions related to the POF condition? What is the age of the women in generation II and III: do they have regular cycles? What about III-1 and III-2? It is likely that a more detailed analysis of the family and of the affected women will make the family more informative and more useful to future studies.

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

Declaration of competing interests:
I declare that I have no competing interests