Reviewer’s report

Title: Exome sequencing reveals a novel TTC19 mutation in an autosomal recessive spinocerebellar ataxia patient

Version: 2 Date: 15 October 2013

Reviewer: Filippo M. Santorelli

Reviewer’s report:

I suggest minor changes:
The authors should indicate that other more common causes of SCAR have been ruled out.
The authors should explicit the other genomic region IBD do not present known genes associated with SCAR.
The authors should indicate the clinical category of the so termed “145 population-matched disease control sample”. One of the 145 cases harboured the Q277* mutation. Did they look for gene deletions/duplications?
The authors should tell us why they did not test TTC19 protein levels at least in skin fibroblasts.
It is wise to ask editorial assistance by an English native speaker colleague.

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.