Reviewer's report

Title: Fanconi anaemia with bilateral diffuse pulmonary arterio venous fistulae: A case report

Version: 2 Date: 31 January 2012

Reviewer: Marc Tischkowitz

Reviewer's report:

Samarakoon et al Fanconi anaemia with bilateral diffuse pulmonary arterio venous fistulae: A case report

This is a concise and well written report detailing a rare pulmonary presentation in FA.

Comments:

Please give the precise details for the chromosome studies that established the diagnosis.

Do the authors have an image of the PAVMs they could include in the article?

Was there any evidence that the case may have had a concurrent diagnosis that would explain the PAVMs e.g. Hereditary Hemorrhagic Telangiectasia

P6 L2 Please write “Complementation Group A” rather than “Type A”

P6 L2 Please correct “atersia” to “atresia”

P6 L4 thumb polydactyly is a relatively common manifestation of FA.

P6 last paragraph. Could the low incidence mean that it is a chance association?

The case in 1973 would not have had diagnostic testing by chromosome breakage so the diagnosed is a clinical one in that case.

P7 Given the extreme rarity of this association with FA, I do not think one can recommend routine screening for PAVMs in FA. Rather it can be suggested if there is evidence of desaturation.

Level of interest: An article whose findings are important to those with closely related research interests

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