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Hereditary haemorrhagic telangiectasia and genetic thrombophilia

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Govani and Shovlin¹ recently described the clinical and diagnostic implications of hereditary haemorrhagic telangiectasia (HHT). The authors¹ do not seem to be paying attention to the possible presence of genetic thrombophilic risk in HHT patients in relation to pharmacological treatment. Recently, we published² a case of HHT and genetic thrombophilia with pharmacological complications attributed to the coexistence of both genetic conditions.

In the review paper, the authors¹ also highlight the risks of life-threatening maternal complications in pregnant HHT women. It is well known that up to 65% of vascular gestational abnormalities can be accounted for by genetic thrombophilias.³ Therefore, it is possible that women with HHT may have recurrent pregnancy loss or other pregnancy complications because of to thrombophilic gene mutations. In the presence of thrombophilic risk, therapy with acetylsalicylic acid and low molecular heparin is recommended, but this may increase the risk of haemorrhages in the presence of HHT. To our knowledge, there are no data on the incidence of pregnancy complications, such as fetal loss or venous thrombosis,

in HHT patients. As reported by Undas *et al.*,⁴ in the absence of life-threatening haemorrhages and detectable vascular malformations, oral anticoagulation could be considered with strictly haematological and clinical follow-up. An associated thrombotic tendency may confer a survival advantage for HHT patients by decreasing the severity of their bleeding problems.⁵

We suggest that for the clinical management of these patients, genetic tests and counselling for inherited thrombophilia may be useful to prevent vascular and pregnancy complications and that more appropriate pharmacological treatment in consideration of the possible presence of both genetic conditions is used.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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Reply to Bianca *et al*

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We thank Bianca *et al*¹ for their interesting comments and hereditary haemorrhagic telangiectasia (HHT) case reports. Within our structured 2009 *EJHG* review,² we did not have space to discuss in detail all of the implications of the cited studies, including, of relevance here, our large thrombosis³ and pregnancy⁴ HHT data series (*EJHG* references 9 and 48).

Concern regarding thrombophilic risk in HHT patients in relation to pharmacological treatment was one of the main conclusions of our cited (*EJHG* reference 9) series of 309 HHT-affected individuals.³