Manuscript no. HAEMATOL.2010.028977 entitled “Integration of molecular and clinical data of 40 unrelated VWD families in a Spanish locus-specific mutation database: first release including 58 mutations”

Authors: Irene Corrales, Lorena Ramírez, Júlia Ayats, Carme Altisent, Rafael Parra, Francisco Vidal

Information about the contributions of each person named as having participated in the study

1) Guarantor(s), i.e., person(s) who is (are) responsible for the integrity of the work as a whole:
   • Francisco Vidal, Unitat de Diagnòstic i Teràpia Molecular, Banc de Sang i Teixits, fvidal@bstcat.net
   • Irene Corrales, Unitat de Diagnòstic i Teràpia Molecular, Banc de Sang i Teixits, icorrales@ir.vhebron.net

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2) Authors who participated in the conception of the study: Francisco Vidal, Irene Corrales, Rafael Parra and Carme Altisent.

3) Design & Methods. The following authors were responsible for specific investigations (please detail):
   • Francisco Vidal, was the principal investigator, coordinated the research, created the database and takes primary responsibility for the paper.
   • Irene Corrales, performed the complete sequencing of patients and the STR analysis, designed the family trees and compiled all data available in the database.
   • Lorena Ramírez, participated in the laboratory work for this study.
   • Júlia Ayats, Carme Altisent and Rafael Parra diagnosed and recruited the patients and clinical data.

4) Results. The following authors were responsible for specific portions of the results, including figures and tables (please indicate the person responsible for each figure and each table):
   • Francisco Vidal and Irene Corrales were responsible for all figures and tables.
   • Lorena Ramirez, Júlia Ayats, Carme Altisent and Rafael Parra were responsible for the diagnosis and laboratory test results of the patients (Table 1.)

5) Writing the manuscript. The following authors were responsible for writing the manuscript:
   • Francisco Vidal and Irene Corrales were responsible for writing the manuscript.
   • Lorena Ramirez, Júlia Ayats, Carme Altisent and Rafael Parra were responsible for the revision of the drafting and the approval of the final version of the manuscript.

6) Contributors Listed in Acknowledgments:
   This work was supported in part with a grant (FIS, PI080385) from the Fondo de Investigaciones Sanitarias, Spanish Ministry of Science and Innovation, and by the Fundació Privada Catalana de l’Hemofilia.