

Haematologica
HAEMATOL/2019/216267
Version 4

Thrombotic risk in congenital erythrocytosis due to up-regulated hypoxia sensing is not associated with elevated hematocrit

Victor R. Gordeuk, Galina Y. Miasnikova, Adelina I. Sergueeva, Felipe R. Lorenzo, Xu Zhang, Jihyun Song, David W. Stockton, and Josef T. Prchal

Disclosures: The authors declare no conflict of interests

Contributions: GYM and AIS enrolled the CE patients in a registry and followed them for thrombotic complications. DWS, FRL, and JTP collected blood and determined hematological phenotype of EPAS1 mutated family and used genomic DNA for genotyping and performed genetic linkage studies. FRL found the EPAS1 mutation and with JS genotyped available affected and non-affected relatives JS prepared three separate iPSC cell lines from peripheral blood of two affected siblings and their non-affected mother. JTP directed the study of the molecular basis of this family and obtained detailed clinical information of most of the affected subjects and followed and reviewed their clinical phenotype over last two decades. VRG and XZ performed the updated statistical analysis of CE patients and controls and also on the family with the EPAS1 c.1603A>G variant. VRG and JTP drafted the paper.