Manuscript no. HAEMATOL/2012/070508 entitled "Polycythemia due to Croatian homozygous VHL (571C>G:H191D) mutation has a different phenotype than Chuvash polycythemia (VHL598C>T:R200W)"

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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:

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The guarantors of this manuscript confirm that all persons designated as authors qualify for authorship, and that each author has participated sufficiently in the work to take public responsibility for appropriate portions of the content.

2) Authors who participated in the conception of the study: Nikica Ljubas Tomasic, Ernest Bilic, and Josef T. Prchal

3) Design & Methods. The following authors were responsible for specific investigations:

• Nikica Ljubas Tomasic was responsible for identifying the propositus, collecting and consenting all studied individuals
• Lucie Piterkova was responsible for designing analysis of the VHL H191D mutation, in vitro assay of the sensitivity of erythroid progenitors to EPO, and real-time PCR assay
• Chad Huff was responsible for designing an analysis of recent shared ancestry
• Ernest Bilic was responsible for identifying the propositus and contributed to study design
• Donghoon Yoon was responsible for designing analysis of in vitro expanded erythroid progenitors in liquid cultures
• Galina Y. Miasnikova, Adelina I. Sergueeva, and Sergei Nekhai were responsible for conducting the study for comparison with Chuvash polycythemia
•Victor Gordeuk was responsible for comparison with Chuvash polycythemia and designed comparison of clinical variables and biological markers among VHL genotypes
• Josef T. Prchal was responsible for final approval of study design

4) Results. The following authors were responsible for specific portions of the results, including figures and tables:

• Nikica Ljubas Tomasic was responsible for the molecular defect identification and analysis of the VHL H191D mutation (Figure 1A and 1B) and comparison of clinical variables between individuals with VHL H191D or VHL R200W mutations (Table 1), as well as responsible for comparison and data analyses of serum EPO levels in VHL H191D Croatian patients (Figure 5)
• Lucie Piterkova was responsible for the molecular defect identification and analysis, in vitro assay of the sensitivity of erythroid progenitors to EPO and real-time PCR assay (Figure 3A and 3B and Figure 4)
• Chad Huff was responsible for analysis of recent shared ancestry (Figure 2)

• Donghoon Yoon was responsible for in vitro expansion of human erythroid progenitors in liquid culture (Figure 3C)
• Galina Y. Miasnikova, Adelina I. Sergueeva, Sergei Nekhai, Victor Gordeuk and Nikica Ljubas Tomasic were responsible for comparison of serum EPO levels in VHL H191D Croatian patients to Chuvash polycythemia.(Figure 5)
• Xiaomei Niu was responsible for biochemical studies and interpretation of data

5) Writing the manuscript. The following authors were responsible for writing the manuscript:
• Nikica Ljubas Tomasic was responsible for Introduction section, "Patient characteristics" and "Analysis of the VHL H191D mutation" in Results section
• Lucie Piterkova was responsible for Introduction section, Design and Methods section, "Analysis of the VHL H191D mutation". Response of erythroid progenitors to EPO", "Effect of the VHL H191D mutation on expression of HIF-1 and/or HIF-2 target genes" in Results section
• Chad Huff was responsible for "Analysis of recent shared ancestry" in Design and Methods and Results section
• Victor Gordeuk was responsible for "Comparison of clinical variables and biological markers among VHL genotypes" in Results section and for Discussion and critically revised the manuscript
• Josef T. Prchal was responsible for Discussion section and critically revised the manuscript

6) Contributors Listed in Acknowledgments: Not applicable