Genetic disarray follows mutant KLF1-E325K expression in a congenital dyserythropoietic anemia patient

Lilian Varricchio,¹ Antanas Planutis,² Deepa Manwani,³ Julie Jaffray,⁴ W. Beau Mitchell,⁵ Anna Rita Migliaccio¹.6,* and James J. Bieker¹.2,७,8,*

¹Tisch Cancer Institute, Icahn School of Medicine at Mount Sinai, New York, NY, USA; ²Department of Cell, Developmental, and Regenerative Biology, Icahn School of Medicine at Mount Sinai, New York, NY, USA; ³Division of Hematology/Oncology, The Children's Hospital at Montefiore, Albert Einstein College of Medicine, Bronx, NY, USA; ⁴Children's Hospital Los Angeles, University of Southern California Keck School of Medicine, Los Angeles, CA, USA; ⁵Department of Pediatrics, Icahn School of Medicine at Mount Sinai, New York, NY, USA; ⁶Dipartimento di Scienze Biomediche e NeuroMotorie, Alma Mater Studiorum, Università di Bologna, Bologna, Italy; ⁷Black Family Stem Cell Institute, Icahn School of Medicine at Mount Sinai, New York, NY, USA and ⁸Mindich Child Health and Development Institute, Icahn School of Medicine at Mount Sinai, New York, NY, USA

*ARM and JJB contributed equally as senior authors on this work.

©2019 Ferrata Storti Foundation, This is an open-access paper, doi:10.3324/haematol.2018.209858

Received: October 22, 2018. Accepted: March 12, 2019. Pre-published: March 14, 2019.

Correspondence: JAMES J. BIEKER - james.bieker@mssm.edu

Supplemental information for:

Genetic disarray follows mutant KLF1-E325K expression in a congenital dyserythropoietic anemia patient

Lilian Varricchio, Antanas Planutis, Deepa Manwani, Julie Jaffray, W Beau Mitchell, Anna Rita Migliaccio, and James J Bieker

Supplemental Figure 1

Figure S1. Analysis of patient RNA. (A) RT-qPCR analysis of γ - and β-globin RNA expressed in the proliferation/differentiation CDA series (P d11, P d15, D d5) and presented as $\gamma/(\gamma+\beta)$. (B) cDNA sequence chromatograms confirming transcription of both alleles (WT codon GAG (glu: E) and mutant codon AAG (lys: K)) in the CDA cells in the proliferation/differentiation series (P d11, P d15, Diff d5); WT only expresses codon GAG (glu: E).

Supplemental Figure 2

Figure S2. Expression levels of megakaryocyte genes. Relative FPKM expression values of selected megakaryocyte genes ⁶¹ from the proliferation/differentiation series, grouped together and color coded as in Figure 2.

Supplemental Table 1

FPKM values of all six samples (CDA d11, d15, difft d5, and WT d11, d15, difft d5) for all genes.

Supplemental Table 2

Genes exclusively expressed within selected subsets (relevant to Figure 4).

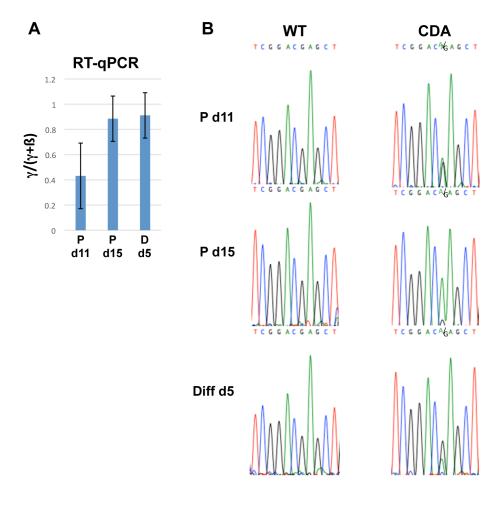


Figure S1. Analysis of patient RNA. (A) RT-qPCR analysis of γ - and ß-globin RNA expressed in the proliferation/differentiation CDA series (P d11, P d15, Diff d5) and presented as $\gamma/(\gamma+\beta)$. (B) cDNA sequence chromatograms confirming transcription of both alleles (WT codon GAG (glu: E) and mutant codon AAG (lys: K)) in the CDA cells in the proliferation/differentiation series (P d11, P d15, Diff d5); WT only expresses codon GAG (glu: E).

Supplemental FIGURE S2



Figure S2. Expression levels of megakaryocyte genes. Relative FPKM expression values of selected megakaryocyte genes (ref 61) from the proliferation/differentiation series, grouped together and color coded as in Figure 2.