SUPPLEMENTARY APPENDIX

Prevalence and management of iron overload in pyruvate kinase deficiency: report from the Pyruvate Kinase Deficiency Natural History Study

Eduard J. van Beers, ¹ Stephanie van Straaten, ¹ D. Holmes Morton, ² Wilma Barcellini, ³ Stefan W. Eber, ⁴ Bertil Glader, ⁵ Hassan M. Yaish, ⁶ Satheesh Chonat, ⁷ Janet L. Kwiatkowski, ⁸ Jennifer A. Rothman, ⁹ Mukta Sharma, ¹⁰ Ellis J. Neufeld, ¹¹ Sujit Sheth, ¹² Jenny M. Despotovic, ¹³ Nina Kollmar, ¹⁴ Dagmar Pospíšilová, ¹⁵ Christine M. Knoll, ¹⁶ Kevin Kuo, ¹⁷ Yves D. Pastore, ¹⁸ Alexis A. Thompson, ¹⁹ Peter E. Newburger, ²⁰ Yaddanapudi Ravindranath, ²¹ Winfred C. Wang, ¹¹ Marcin W. Wlodarski, ²² Heng Wang, ²³ Susanne Holzhauer, ²⁴ Vicky R. Breakey, ²⁵ Madeleine Verhovsek, ²⁵ Joachim Kunz, ²⁶ Melissa A. McNaull, ²⁷ Melissa J. Rose, ²⁸ Heather A. Bradeen, ²⁹ Kathryn Addonizio, ³⁰ Anran Li, ³⁰ Hasan Al-Sayegh, ³⁰ Wendy B. London ³⁰ and Rachael F. Grace ³⁰

'Van Creveldkliniek, University Medical Centre Utrecht, University of Utrecht, the Netherlands; ²Central Pennsylvania Clinic for Special Children & Adults, Belleville, PA, USA; Lancaster Ĝeneral Hospital, Lancaster, PA, USA; ³Fondazione IRCCS Ca' Granda, Ospedale Maggiore Policlinico, Milan, Italy; ⁴Schwerpunktpraxis für Pädiatrische Hämatologie-Onkologie and Children's Hospital, Technical University, Munich, Germany; Lucile Packard Children's Hospital, Stanford University, Palo Alto, CA, USA; Primary Children's Hospital, University of Utah, Salt Lake City, UT, USA; Emory University School of Medicine, Aflac Cancer and Blood Disorders Center, Children's Healthcare of Atlanta, GA, USA; [®]Children's Hospital of Philadelphia and Perelman School of Medicine of the University of Pennsylvania, Philadelphia, PA, USA; Duke University Medical Center, Durham, NC, USA; Children's Mercy Hospital, University of Missouri, Kansas City, MO, USA; "St. Jude Children's Research Hospital, Memphis, TN, USA; 12Weill Cornell Medical College, New York Presbyterian Hospital, NY, USA; 13 Texas Children's Hematology Center, Baylor College of Medicine, Houston, TX, USA; 14 Klinikum Kassel GmbH, Germany; 15 Fakultni nemocnice Olomouc, Czech Republic; ¹⁶Phoenix Children's Hospital, AZ, USA; ¹⁷University of Toronto, University Health Network, ON, Canada; ¹⁸CHU Sainte-Justine, Montreal, QC, Canada; 19Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA; 20University of Massachusetts Medical School, Worcester, MA, USA; 21 Children's Hospital of Michigan, Wayne State University School of Medicine, Detroit, MI, USA; 22 Department of Pediatrics and Adolescent Medicine, Division of Pediatric Hematology and Oncology, Medical Center, Faculty of Medicine, University of Freiburg, Germany; 23DDC Clinic for Special Needs Children, Middlefield, OH, USA; 24 Charite, Berlin, Germany; 25 McMaster University, Hamilton, ON, Canada; 26 Zentrum für Kinder-und Jugendmedizin, University of Heidelberg, Heidelberg, Germany; 27 University of Mississippi Medical Center, Jackson, MS, USA; 28 Nationwide Children's Hospital, The Ohio State University College of Medicine, Columbus, OH, USA; ²⁹The University of Vermont Children's Hospital, Burlington, VT, USA and 30Dana-Farber Boston Children's Cancer and Blood Disorder Center, Boston, MA, USA

Correspondence: rachael.grace@childrens.harvard.edu doi:10.3324/haematol.2018.196295

SUPPLEMENTAL TABLE

Supplemental Table 1: Patient characteristics, comparing those with iron monitoring to those without monitoring, in the 12 months prior to enrollment.

	Ferritin monitoring (n=242)			MRI monitoring for liver iron		
				assessment, in the non-Amish patient		
				cohort (n=189)		
	Obtained in	Not obtained	p-value**	Obtained in	Not obtained	p-value**
	the 12	in the 12		the last 12	in the last 12	
	months prior	months prior		months prior	months prior	
	to enrollment	to enrollment		to enrollment	to enrollment	
	(n=175)*	(n=67)*		(n=18)*	(n=171)*	
Median*** Age at enrollment	21.8	16.3	0.3	21.1	19.1	0.5
at enrollment	(1.3-69.9)	(1.4-60.4)		(2.2-46.0)	(1.3-69.9)	
Transfused in	72/175	9/67	<0.0001	11/18	67/171	0.08
the 12 months	(41%)	(13%)		(61%)	(39%)	
prior to						
enrollment (%)						
Regularly	43/175	1/67	<0.0001	7/18	36/171	0.1
Transfused (%)	(25%)	(1%)		(39%)	(21%)	
Median***	8.8	9.6	0.01	8.6	8.7	0.3
Hemoglobin	(5.2-14.1)	(6.5-13.0)		(6.2-11.1)	(5.2-14.1)	
value (g/dl)	n=174	n=65		(0.2-11.1)	n=168	
	11-17-	11–00			11–100	
Splenectomized	112/175	37/67	0.2	15/18	83/171	0.006
(%)	(64%)	(55%)		(83%)	(49%)	

^{*}Number of patients with known data (n) are presented in the column headers unless otherwise indicated in the table. **Using Fisher's exact test for categorical variables and Wilcoxon ranksum test for continuous variables. ***Median presented with ranges.