

Haematologica  
HAEMATOL/2017/180778  
Version 3

Constitutional SAMD9L mutations cause familial myelodysplastic  
syndrome and transient monosomy 7

Victor B. Pastor, Sushree Sahoo, Jessica Boklan, Georg C. Schwabe,  
Ebru Saribeyoglu, Brigitte Strahm, Dirk Lebrecht, Matthias Voss, Yenan  
T. Bryceson, Miriam Erlacher, Gerhard Ehninger, Marena Niewisch,  
Brigitte Schlegelberger, Irith Baumann, John C. Achermann, Akiko  
Shimamura, Jochen Hochrein, Ulf Tedgård, Lars Nilsson, Henrik Hasle,  
Melanie Boerries, Hauke Busch, Charlotte M. Niemeyer, and Marcin W.  
Wlodarski

Disclosures: none

Contributions: MWW, VBP, SS designed the study and wrote the manuscript. VBP, SS, DL, MV, YTB, MB, JH, HB, and MWW performed experiments and analyzed data. CMN, JCA, BS, and ME contributed to study conception and data interpretation. JB, GCS, ES, BS, GE, MN, BSch, IB, AS, UT, LN, HH, CMN were involved in patient care, testing procedures and sample acquisition. All authors contributed to writing and approved the manuscript.