Haematologica
HAEMATOL/2018/189845
Version 3

The SLC40A1 R178Q mutation is a recurrent cause of hemochromatosis and is associated with a novel pathogenic mechanism

Chandran Ka, Julie Guellec, Xavier Perpermans, Caroline Kannengiesser, Cécile Ged, Wim Wuyts, David Cassiman, Victor de Ledinghen, Bruno Varet, Caroline de Kerguenec, Claire Oudin, Isabelle Gourlaouen, Thibaud Lefebvre, Claude Férec, Isabelle Callebaut, and Gerald Le Gac
Collaborative Groups: French National Network for the Molecular Diagnosis of Inherited Iron Overload Disorders)

Disclosures: None

Contributions: G. Le Gac, X. Pepermans and I. Callebaut designed the study. C. Ka, J. Guellec, I. Gourlaouen, C. Oudin, T. Lefebvre and I. Callebaut conducted experiments. C. Ged, C. Kannengiesser, W. Wuyts, D. Cassiman, V. de Ledinghen, B. Varet and C. de Kerguenec provided data. G. Le Gac, C. Ka, J. Guellec, I. Callebaut and C. Férec analyzed data. G. Le Gac wrote the manuscript. All authors contributed to the editing of the final manuscript.