

SUPPLEMENTARY MATERIAL

A simple clinical score to promote and enhance ferroportin disease screening

Amandine Landemaine^{*1, 2}, Houda Hamdi-Roze^{*1, 2}, Séverine Cunat³, Véronique Loustaud-Ratti⁴, Xavier Causse⁵, Si Nafa Si Ahmed⁵, Bernard Drenou⁶, Christophe Bureau⁷, Gilles Pelletier⁸, Caroline De Kerguenec⁹, Nathalie Ganne-Carrie¹⁰, Stéphane Durupt¹¹, Fabrice Laine^{1, 2}, Olivier Loréal^{2, 12}, Martine Ropert^{1, 2}, Lenaick Detivaud^{1, 2}, Jeff Morcet¹, Patricia Aguilar-Martinez³, Yves M. Deugnier^{1, 2}, Edouard Bardou-Jacquet^{1, 2, 12}

Affiliations:

¹ Univ Rennes, CHU Rennes, INSERM CIC1414, F-35000, Rennes, France.

² National Reference Center for Hemochromatosis and iron metabolism disorder, CHU Rennes, F-35000, Rennes, France.

³ CHU Montpellier, Montpellier, Competence center for Hemochromatosis and iron metabolism disorder, Reference center on rare red cell disorders, France

⁴ CHU Limoges, U1248, INSERM, F-87000, Limoges, France.

⁵ Department of Hepatology and Gastroenterology, Centre Hospitalier Régional (CHR), Orléans, France.

⁶ CH Emile Muller, F-68100 Mulhouse, France; Institut de Recherche en Hématologie et Transplantation (IRHT), F-68100 Mulhouse, France

⁷ CHU Toulouse, Liver Unit, University Hospital of Toulouse and University Paul Sabatier, Toulouse, France.

⁸ AH-HP, Hôpital Paul Brousse, Centre Hépato-Biliaire, Villejuif, France.

⁹ AP-HP, Hôpital Beaujon, Département d'Hépatologie, Clichy, France.

¹⁰ AP-HP Hopitaux Universitaire Paris Saine Saint-Denis, APHP, Liver Unit, University Paris 13, Sorbonne Paris Cité; INSE RM, S1138 FunGeST F-75006, Paris, France.

¹¹ Department of Internal and Vascular Medicine, Hôpital Lyon Sud, Hospices Civils de Lyon, Pierre-Bénite, France.

¹² INSERM, Univ Rennes, CHU Rennes, UMR1241, Institut NuMeCan, Rennes.

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Supplementary Table 1. Characteristics of the derivation cohort without patients with missing data in the variables used for the score.

Results are shown as Mean (\pm SD) or N (%) as appropriate. HBP, high blood pressure; BMI, body mass index; LIC, liver iron concentration; TSat, transferrin saturation; ALT, alanine aminotransferase; GGT, gamma-glutamyltransferase; CRP, C reactive protein; WHO rec, World Health Organization recommendations regarding alcohol consumption. Comparison between patients with and without *SLC40A1* pathogenic variant. ^a p value of the logistic binary regression.

N=663	Total	No pathogenic	Pathogenic variant	p value ^a
Sex				
	Male	547 (82.5%)	515 (83.7%)	32 (66.7%)
	Female	116 (17.5%)	100 (16.3%)	16 (33.3%)
Alcohol				
	None	354 (53.4%)	329 (53.5%)	25 (52.1%)
	< WHO rec	213 (32.1%)	198 (32.2%)	15 (31.2%)
	> WHO rec	35 (5.3%)	32 (5.2%)	3 (6.3%)
	Missing	61 (9.2%)	56 (9.1%)	5 (10.4%)
Dyslipidemia				
	No	371 (55.9%)	341 (55.5%)	30 (62.5%)
	Yes	253 (38.2%)	237 (38.5%)	16 (33.3%)
	Missing	39 (5.9%)	37 (6.0%)	2 (4.2%)
HBP				
	No	428(64.5%)	389(63.3%)	39(81.2%)
	Yes	228(34.4%)	219(35.6%)	9(18.8%)
	Missing	7(1.1%)	7(1.1%)	0(0%)
Diabetes				
	No	499(75.3%)	456(74.1%)	43(89.6%)
	Yes	61(9.2%)	59(9.6%)	2(4.2%)
	Missing	103(15.5%)	100(16.3%)	3(6.2%)
HBP or Diabetes				
	No	402 (60.6%)	364 (59.2%)	38 (79.2%)
	Yes	261 (39.4%)	251 (40.8%)	10 (20.8%)
Age (y)		57.42 (\pm 12.96)	57.75 (\pm 12.94)	53.13 (\pm 12.59)
BMI (kg/m²)		25.57 (\pm 3.87)	25.57 (\pm 3.83)	25.62 (\pm 4.33)

LIC ($\mu\text{mol/g}$)	169 (± 77)	164 (± 75)	226 (± 84)	p<0.01
TSat (%)	2.23 (± 0.42)	2.23 (± 0.41)	2.23 (± 0.54)	p=0.75
Ferritin level ($\mu\text{g/L}$)	1370.7	1280.8 (± 1078.4)	2522.4 (± 2391.0)	p<0.01
ALT level (IU/L)	38.11 (± 26.77)	37.88 (± 26.84)	41.28 (± 25.97)	p=0.44
GGT level (IU/L)	71.44	71.17 (± 168.85)	75.32 (± 183.24)	p=0.89
Hemoglobin (g/dL)	14.67 (± 1.73)	14.70 (± 1.75)	14.24 (± 1.34)	p=0.12
CRP (mg/L)	3.53 (± 6.03)	3.39 (± 5.98)	53.13 (± 12.59)	p=0.11

Supplementary Table 2. Patients' characteristics of the validation cohort and univariate comparison with the derivation cohort (logistic regression).

Clinical characteristics of patients with and without *SLC40A1* pathogenic variant in the validation cohort, and comparison of the derivation and validation cohort with Chi square. Results are shown as Mean (\pm SD) or N (%) as appropriate. HBP, high blood pressure; LIC, liver iron concentration.

		<i>No pathogenic</i>	<i>Pathogenic</i>	<i>Total Validation</i>	<i>Comparison to</i>
Sex	Male	155 (79.5%)	8 (80.0%)	163 (79.5%)	p=0.82
	Female	40 (20.5%)	2 (20.0%)	42 (20.5%)	
Age at diagnosis		55.1 (\pm 14.2)	55.1 (\pm 15.5)	55.1 (\pm 14.2)	p=0.47
HBP or Diabetes	No	105(67.3%)	8(88.9%)	113(68.5%)	p=0.001
	Yes	51(32.7%)	1(11.1%)	52(31.5%)	
LIC (μmol/g) (N=205)		151 (\pm 83)	248 (\pm 73)	156 (\pm 85)	p=0.10
Ferritin level		1136 (\pm 981)	2178 (\pm 1154)	1188 (\pm 1013)	p=0.09