

Supplementary Materials

Table S1. Performance of the TSAT/hepcidin ratio in distinguishing IRIDA patients from IDA controls in absence of inflammation and without recent iron therapy.

	IRIDA patients (n = 20) versus IDA controls (n = 39)						
TSAT/hepcidin ratio	AUC	95% CI of AUC	Cut-off value	Sensitivity	95% CI of Sensitivity	Specificity	95% CI of Specificity
<i>A. Hepcidin <0.5 nM = 0.25 nM</i>							
Total IRIDA group versus IDA controls	1.000	1.000-1.000	<5.6%/nM	100%	84-100%	100%	91-100%
Biallelic IRIDA patients versus IDA controls	1.000	1.000-1.000	<4.3%/nM	100%	74-100%	100%	91-100%
Monoallelic IRIDA patients versus IDA controls	1.000	1.000-1.000	<5.6%/nM	100%	70-100%	100%	91-100%
<i>B. Hepcidin <0.5 nM = 0.49 nM</i>							
Total IRIDA group versus IDA controls	1.000	1.000-1.000	<3.9%/nM	100%	84-100%	100%	91-100%
Biallelic IRIDA patients versus IDA controls	1.000	1.000-1.000	<2.6%/nM	100%	74-100%	100%	91-100%
Monoallelic IRIDA patients versus IDA controls	1.000	1.000-1.000	<3.9%/nM	100%	70-100%	100%	91-100%
<i>C. Exclusion of IDA control with a monoallelic pathogenic TMPRSS6 variant without IRIDA phenotype</i>	1.000	1.000-1.000	<5.9%/nM	100%	84-100%	100%	91-100%

Hepcidin levels below the lower limit of detection (<0.5 nM) were imputed with a value of 0.25 nM (A) or with a value of 0.49 nM (B). 95% CI of AUC calculated with SPSS version 22.0 and 95% CI of sensitivity and specificity calculated with GraphPad Prism version 8.4.2. AUC, area under the curve; CI, confidence interval.