SUPPLEMENTARY MATERIALS

L-Ferritin, one gene - five diseases; from hereditary hyperferritinaemia to hypoferritinaemia. Report of new cases.

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Supplementary Materials Table S1. Summary of *FTL* identified mutations.

Table summarizing all mutations in *FTL* described up to now in the literature that may cause one of the five diseases. The table shows for each mutation, the conventional nomenclature according to HGVS (corresponding to [NCBI:NM_000146.3] reference sequence), the traditional nomenclature, the position in the IRE structure, and the corresponding published report. NA (not available).

N	Enfermedad	HGVS nomenclature	Mutation type	Mutation position	First Publication	Old Nomenclature
1	**HHCS**	c.-216C>A	IRE regulatory	Promoter *FTL*	[1]	NA
2	HHCS	c.-193C>G + c.-160A>G	IRE regulatory	lower stem + hexanucleotide loop	[2]	+7C>G & +40A>G
3	HHCS	c.-190C>T	IRE regulatory	lower stem	[3]	+10C>U
4	HHCS	c.-186C>G	IRE regulatory	lower stem	[4]	+14C>G
5	HHCS	c.-184C>T	IRE regulatory	lower stem	[3]	+16C>U
6	HHCS	c.-182C>T + c.-178T>G	IRE regulatory	lower stem	[5]	Paiva-2 + 18C>U & 22U>G
7	HHCS	c.-176T>C	IRE regulatory	lower stem	[6]	+24U>C
8	HHCS	c.-171C>G	IRE regulatory	lower stem	[7]	Torino +29C>G
9	HHCS	c.-168G>A	IRE regulatory	lower stem	[5]	Pavia-1 +32G>A
10	HHCS	c.-168G>C	IRE regulatory	lower stem	[8]	Baltimore-1 +32G>C
11	HHCS	c.-168G>T	IRE regulatory	lower stem	[9]	Paris-2 or Milano-1 +32G>U
12	HHCS	c.-167C>A	IRE regulatory	C bulge	[10]	Paris +33C>A
13	HHCS	c.-167C>T	IRE regulatory	C bulge	[11]	Madrid or Philadelphia +33C>U
14	HHCS	c.-166T>C	IRE regulatory	upper stem	[12]	Paris +34U>C
15	HHCS	c.-164C>A	IRE regulatory	upper stem	[13]	London-2 +36C>A
16	HHCS	c.-164C>G	IRE regulatory	upper stem	[3]	Milano +36C>G
17	HHCS	c.-164C>T	IRE regulatory	upper stem	[14]	Badalona +36C>U
18	HHCS	c.-163A>C	IRE regulatory	upper stem	[15]	Pavia +37A>C
19	HHCS	c.-163A>G	IRE regulatory	upper stem	[3]	Milano +37A>G
20	HHCS	c.-163A>T	IRE regulatory	upper stem	[16]	Zaragoza +37A>U
21	HHCS	c.-161C>A	IRE regulatory	hexanucleotide loop	[17]	Geelong +39C>A

22	HHCS	c.-161C>G	IRE regulatory	hexanucleotide loop	[18]	Paris +39C>G
23	HHCS	c.-161C>T	IRE regulatory	hexanucleotide loop	[13]	London-1 +39C>U
24	HHCS	c.-160A>G	IRE regulatory	hexanucleotide loop	[19]	Paris-1 or Montpellier-1 +40A>G
25	HHCS	c.-160A>G + c.-159G>C	IRE regulatory	hexanucleotide loop	[4]	Paris-1 or Montpellier-1 +40A>G & Verona-1 +41G>C
26	HHCS	c.-159G>C	IRE regulatory	hexanucleotide loop	[20]	Verona-1 +41G>C
27	HHCS	c.-157G>A	IRE regulatory	hexanucleotide loop	[21]	Salt Lake City +43G>A
28	HHCS	c.-154T>G	IRE regulatory	upper stem	[22]	+46U>G
29	HHCS	c.-153G>A	IRE regulatory	upper stem	[12]	Paris +47G>A
30	HHCS	c.-151A>C	IRE regulatory	lower stem	[2]	+49A>C
31	HHCS	c.-151A>G	IRE regulatory	lower stem	[23]	Ghent +49A > G
32	HHCS	c.-150C>A	IRE regulatory	lower stem	[24]	+50C>A
33	HHCS	c.-149G>C	IRE regulatory	lower stem	[25]	Torino +51G>C
34	HHCS	c.-148G>C	IRE regulatory	lower stem	[14]	Heidelberg +52G>C
35	HHCS	c.-144A>T	IRE regulatory	lower stem	[15]	Paris +56A>U
36	HHCS	c.-110C>T	IRE regulatory	5′ UTR	[3]	+90C>U
37	HHCS	c.-220_-196del25	IRE regulatory	new transcription starting site (resulting IRE lacks nt 1-24)	[26]	NA
38	HHCS	c.-190-162del29	IRE regulatory	eliminating IRE	[27]	Verona-2 +10_38del29
39	HHCS	c.-182_-174delCGGGTCTGTinsAGGGGCCGG $	IRE regulatory	eliminating part of lower stem	[28]	+18_+26 delCGGGTCTGTinsAGGGGCCGG
40	HHCS	c.-178_-173del6	IRE regulatory	eliminating part of lower stem	[29]	+22_27del6
41	HHCS	c.-168_-165delGCTT	IRE regulatory	eliminating C bulge	[30]	+32_35delGCTT
42	HHCS	c.-164_158del7	IRE regulatory	eliminating part of	**THIS**	Esplugues +36_42del7

				hexanucleotide loop	**STUDY**	
43	HHCS	c.-161delC	IRE regulatory	eliminating IRE	[31]	+39delC
44	HHCS	c.-162_-161delCA	IRE regulatory	eliminating part of hexanucleotide loop	[32]	+38_39del AC
45	HHCS	c.-158_-143del16	IRE regulatory	eliminating part of hexanucleotide loop	[32]	+42_57del16
46	HHCS	c.-153_-152delGGinsCT	IRE regulatory	eliminating part of upper stem	[33]	+47_48delGGinsCT
47	HHCS	c.-44delT	IRE regulatory	eliminating IRE	[3]	
1	**Neuroferritinopathy**	c.[474G>A]; p.(Ala96Thr)	Missense	exon3	[34]	
2	Neuroferritinopathy	c.641_642 4bp_dup	Frameshift	exon 4	[35]	
3	Neuroferritinopathy	c.646InsC	Frameshift	exon 4	[36]	
4	Neuroferritinopathy	c.458dupA	Frameshift	exon 4	[37]	
5	Neuroferritinopathy	c.460InsA	Frameshift	exon 4	[38]	
6	Neuroferritinopathy	c.468_483 dup16nt	Frameshift	exon 4	[39]	
7	Neuroferritinopathy	c.469_484 dup16nt	Frameshift	exon 4	[40]	
8	Neuroferritinopathy	c.498InsTC	Frameshift	exon 4	[41]	
9	Neuroferritinopathy	c.468dupT	Frameshift	exon 4	[42]	
10	Neuroferritinopathy	c.467_470dupGTGG	Frameshift	exon 4	[43]	
1	**Beningn Hyperferritinaemia**	c.[77A<T]; p.(Gln26Leu)	Missense	exon 1	[44]	
2	Beningn Hyperferritinaemia	c.[80C>T]; p.(Ala27Val)	Missense	exon 1	[44]	
3	Beningn Hyperferritinaemia	c.[89C>T]; p.Thr30Ile	Missense	exon 1	[45]	
1	**L-ferritin deficiency Dominant**	c[1A>G]; p.(M1V)	Missense	start codon	[46]	
2	L-ferritin deficiency Dominant	c.375+2T>A	Splicing	intronic	**THIS STUDY**	
1	**L-ferritin deficiency**	c.[310G>T];p. (E104X]	Nonsense	exon3	[47]	

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6	NOTES:
7	[$]Previously reported by HGMD as c.-182_-176delCGGGTCTinsAGGGGCC, correct: c.-182_-174delCGGGTCTGTinsAGGGGCCGG

Supplementary Materials Table S2. DNA sequence for *FTL* RNA fold predictions

WT-IRE	5′-GCAGTTCGGCGGTCCCGCGGGTCTGTCTCTTGCTTCAACAGTGTTTGGACGGA ACAGATCCGGGGACTCTCTTCC-3′
Mut-IRE	5′-GCAGTTCGGCGGTCCCGCGGGTCTGTCTCTTGCTTGTTTGGACGGAACAGATC CGGGGACTCTCTTCC-3′

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