

# Hereditary Hyperferritinemia Cataract Syndrome: Ferritin L Gene and Physiopathology behind the Disease – Report of New Cases

Table S1. Reported mutations in HHCS disease.

N	HGVS Nomenclature	Mutation Type	Mutation Position	First Publication	Old Nomenclature
1	c.-216C>A	IRE regulatory	Promoter <i>FTL</i>	[1]	NA
2	c.-193C>G + c.-160A>G	IRE regulatory	lower stem + hexanucleotide loop	[2]	+7C>G & +40A>G
3	c.-190C>T	IRE regulatory	lower stem	[3]	+10C>U
4	c.-186C>G	IRE regulatory	lower stem	[4]	+14C>G
5	c.-184C>T	IRE regulatory	lower stem	[3]	+16C>U
6	c.-182C>T + c.-178T>G	IRE regulatory	lower stem	[5]	Paiva-2 + 18C>U & 22U>G
7	c.-176T>C	IRE regulatory	lower stem	[6]	+24U>C
8	c.-171C>G	IRE regulatory	lower stem	[7]	Torino +29C>G
9	c.-168G>A	IRE regulatory	lower stem	[5]	Pavia-1 +32G>A
10	c.-168G>C	IRE regulatory	lower stem	[8]	Baltimore-1 +32G>C
11	c.-168G>T	IRE regulatory	lower stem	[9]	Paris-2 or Milano-1 +32G>U
12	c.-167C>A	IRE regulatory	C bulge	[10]	Paris +33C>A
13	c.-167C>T	IRE regulatory	C bulge	[11]	Madrid or Philadelphia +33C>U
14	c.-166T>C	IRE regulatory	upper stem	[12]	Paris +34U>C
15	c.-164C>A	IRE regulatory	upper stem	[13]	London-2 +36C>A
16	c.-164C>G	IRE regulatory	upper stem	[3]	Milano +36C>G
17	c.-164C>T	IRE regulatory	upper stem	[14]	Badalona +36C>U
18	c.-163A>C	IRE regulatory	upper stem	[15]	Pavia +37A>C
19	c.-163A>G	IRE regulatory	upper stem	[3]	Milano +37A>G
20	c.-163A>T	IRE regulatory	upper stem	[16]	Zaragoza +37A>U
21	c.-161C>A	IRE regulatory	hexanucleotide loop	[17]	Geelong +39C>A
22	c.-161C>G	IRE regulatory	hexanucleotide loop	[18]	Paris +39C>G
23	c.-161C>T	IRE regulatory	hexanucleotide loop	[13]	London-1 +39C>U
24	c.-160A>G	IRE regulatory	hexanucleotide loop	[19]	Paris-1 or Montpellier-1 +40A>G
25	c.-160A>G + c.-159G>C	IRE regulatory	hexanucleotide loop	[4]	Paris-1 or Montpellier-1 +40A>G & Verona-1 +41G>C
26	c.-159G>C	IRE regulatory	hexanucleotide loop	[20]	Verona-1 +41G>C
27	c.-157G>A	IRE regulatory	hexanucleotide loop	[21]	Salt Lake City +43G>A
28	c.-154T>G	IRE regulatory	upper stem	[22]	+46U>G
29	c.-153G>A	IRE regulatory	upper stem	[12]	Paris +47G>A
30	c.-151A>C	IRE regulatory	lower stem	[2]	+49A>C
31	c.-151A>G	IRE regulatory	lower stem	[23]	Ghent +49A > G
32	c.-150C>A	IRE regulatory	lower stem	[24]	+50C>A
33	c.-149G>C	IRE regulatory	lower stem	[25]	Torino +51G>C
34	c.-148G>C	IRE regulatory	lower stem	[14]	Heidelberg +52G>C
35	c.-144A>T	IRE regulatory	lower stem	[15]	Paris +56A>U
36	c.-110C>T	IRE regulatory	5' UTR	[3]	+90C>U
37	c.-220_-196del25	IRE regulatory	new transcription starting site (resulting IRE lacks nt 1-24)	[26]	NA
38	c.-190-162del29	IRE regulatory	eliminating IRE	[27]	Verona-2 +10_38del29
39	c.-182_-174 delCGGGTCTGT insAGGGGCCCGG \$	IRE regulatory	eliminating part of lower stem	[28]	+18_+26 delCGGGTCTGT insAGGGGCCCGG
40	c.-178_-173del6	IRE regulatory	eliminating part of lower stem	[29]	+22_27del6
41	c.-168_-165delGCTT	IRE regulatory	eliminating C bulge	[30]	+32_35delGCTT
42	c.-164_158del7	IRE regulatory	eliminating part of hexanucleotide loop	[31]	Esplugues +36_42d
43	c.-161delC	IRE regulatory	eliminating IRE	[32]	+39delC
44	c.-162_-161delCA	IRE regulatory	eliminating part of hexanucleotide loop	[33]	+38_39del AC
45	c.-158_-143del16	IRE regulatory	eliminating part of hexanucleotide loop	[33]	+42_57del16

46	c.-153_-152 delGGinsCT	IRE regulatory	eliminating part of upper stem	[34]	+47_48delGGinsCT
47	c.-44delT	IRE regulatory	eliminating IRE	[3]	+176delT

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