

TABLE 1 Diagnosis of HHCS in infants (< 1 year old at diagnosis) reported in the literature

Age of reported diagnosis of HHCS	Ferritin levels (µg/L)	Cataract	<i>FTL</i> mutation, previous nomenclature relative to transcription initiation site (TIS) Trivial name, rs	<i>FTL</i> mutation, HGVS nomenclature NC_000019.9(NM_000146.3)	Country of origin	Other affected family members	References
9 months	1500	no	+29 (C>G) Torino	c.-171C>G	Nigeria	mother	This paper
3 months	1020 to 2350	no	+33 (C>T)	c.-167C>T	Spain	6 family members	⁷ Balas 1999
9 weeks	2000	yes	+32 (G>C) Baltimore rs398124635	c.-168G>C	Australia	4 family members	⁸ Craig 2003
6 months	1401	no	+33 (C>T) Madrid	c.-167C>T	Germany	mother	⁹ Nonnenmacher 2011
2 months	4098	no	+37 (A>C)	c.-163A>C	India	mother	¹⁰ Lodh 2012