PTH-129

DO H63D HOMOZYGOTE PATIENTS HAVE CLINICALLY SIGNIFICANT IRON OVERLOAD?

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Introduction Homozygosity H63D is present in 2% of the normal Caucasian population and is thought to carry a small risk of iron overload. The authors aim to characterise patients referred with evidence of iron overload found to be H63D homozygous.

Table 1 PTH-129 Blood tests median (range)

Ferritin	TS	ALT	Bili	GGT	ALP	Alb	INR	Chol	Tri
572 (55–1460)	53 (42–76)	54 (22–106)	11 (8–54)	150 (52–462)	83 (59–143)	40 (30–44)	1 (1–1.2)	5.3 (4–7.4)	1.65 (0.8–2.8)

Methods Retrospective analysis of all H63D homozygotes with abnormal iron indexes identified between 2005 and 2009. Approximately 650 patients were referred with abnormal iron indexes in this time period. 16 patients were identified who fitted these criteria, medical database and notes were reviewed to ascertain these individuals clinical outcome.

Results 69% of patients were male, median age 49.5 (22–75) and 88% were from a White British ethnic group.

38% of H63D patients had iron overload on liver biopsy and were all male. Biopsy demonstrated hepatocellular patten iron overload grades I (3), II (2), III (0), IV (1). 5 showed steatohepatitis, 5 fibrosis none cirrhosis. Only one patient had isolated primary iron overload. Secondary co-factors were identified in 88% patients with abnormal iron storage (NASH, ALD, Hep C). All venesected patients (4) had a marked reduction in ferritin. One patient died due to an unrelated problem.

Conclusion The incidence of H63D homozygote on its own leading to iron overload is relatively low but may still be encountered during clinical practice. Although majority of individuals had co-factors that may increase iron indexes these had hepatocellular iron deposition. These deposits may be directly related to H63D gene making it clinically significant.

Competing interests None.