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Age at diagnosis associated with mortality in patients with Hereditary Hemochromatosis

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Background

Hereditary Haemochromatosis (HH) a disorder of iron metabolism, is the most common autosomal recessive disorder in Caucasians, with Ireland having the highest prevalence in the world. Despite this, no formal screening exists for the condition. Treatment by therapeutic phlebotomy reduces morbidity and mortality.

Methods

- This study aimed to identify factors associated with mortality in HH patients attending a Hemochromatosis service.

Results

- 1043 patients with HH were identified; 65% were male and 35% were female. Fatigue was the most common presenting complaint (37%), with family history (22%), incidental finding of raised iron studies (16%) and screening programs (13%) being other common causes of referral to the service. Homozygosity for C282Y was the most common HFE genetic mutation in this cohort (68%) with compound HFE heterozygosity accounting for 26%.

- Median Ferritin at diagnosis was 332 ug/L (range 22-4655ug/L). Mean Transferrin saturation at diagnosis: 68% (SD+/-17).

- At the time of assessment, 0.03% (30/1043) of patients had died, with a median follow up time of 10 years (range 0.06-26). Of those that died, 70% were male, 43% were homozygous for C282Y, and 36% were compound heterozygotes. Median ferritin at diagnosis was 265ug/L (dead) vs 264ug/L (alive). Median age at diagnosis was significantly higher in those that died, 63.9yrs vs 49.1yrs for those alive at follow up ($p<0.0001$). No significant difference was observed between groups based on gender, HFE genotype, or serum ferritin at diagnosis.

Discussion

- In a large sample of patients with patients with Haemochromatosis, age at diagnosis appears to be associated with death. Further investigation is warranted to detail the causes of death, and to determine whether HH screening in an at-risk population would reduce morbidity and mortality.