Reviewer's report

Title: Allele frequencies of hemojuvelin gene (HJV) I222N and G320V missense mutations in white and African American subjects from the general Alabama population

Version: 1 Date: 26 September 2004

Reviewer: Clara Camaschella

Reviewer's report:

General

The paper by Barton et al reports the study of two mutations of juvenile hemochromatosis (HJV) gene in normal whites and African American controls from Alabama. The problem is clearly illustrated, the methodology adequate and the results well described. The results show that a single white allele out of the 480 examined was positive for HJV I222N and none for the “recurrent” G320V mutation.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

It is unclear whether iron parameters were available or have been measured in the studied population.

The first sentence of the Abstract is obscure. Mutations in the coding region of the HJV gene at the homozygous or compound heterozygous state are associated with juvenile hemochromatosis and not “with primary iron overload in some adults who lack typical hemochromatosis-associated HFE ........”. Obviously patients with juvenile hemochromatosis may present in young adulthood! The sentence should be changed according to the first Background sentence.

References are puzzling! References 1 and 2 are indicated as the papers that correlate HJV mutations and juvenile hemochromatosis, but the largest series of HJV mutations reported in the literature is in the article by Lanzara et al (ref 7), that is quoted only referring to I122N and G320V mutations! This inconsistency should be corrected.

G320V mutation has been screened in a Greek cohort of blood donors. The paper is available in Haematologica on line (Politis et al Haematologica ,2004). This paper should be quoted in the discussion of the frequency of G320V and other HJV mutations.

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions
Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:

None