

Corrigendum to “Ferroportin disease: A systematic meta-analysis of clinical and molecular findings” J. Hepatol. 2010 Nov;53(5):941–949

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In the above named article there were three errors:

1. In the section on “How to differentiate between SLC40A1 mutations and polymorphisms”, a reference was left out of the following paragraph. The paragraph and reference are now listed below.

Ferroportin disease is genetically heterogeneous with 36 different SLC40A1 mutations reported [8–10,12,26–29,38–41,45–68,78] as summarized in Table 3. In addition, nine ferroportin gene polymorphisms were reported, three of which were associated with increased serum ferritin in various populations (Table 4) [29,34]. The allele frequencies of 29 disease-associated ferroportin mutations had been determined in matched populations and was found to be <1:100. It remains

unclear whether L233V and D270P, each of which have been identified in single patients with iron overload, are disease-causing mutations or represent benign sequence variants [43,65].

[78] Wallace DF, Clark RM, Harley HA, Subramaniam VN. Autosomal dominant iron overload due to a novel mutation of ferroportin1 associated with parenchymal iron loading and cirrhosis. J Hepatol 2004;40:710–3.

2. In Tables 3 and 4 there were some errors. The tables are now reproduced correctly on the following pages.

The authors apologise for these errors.

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Table 3. Molecular genetics of *SLC40A1* mutations. The frequency of mutations highlighted with * in the control population can be inferred from studies, in which control populations have been screened for the presence of another mutation, which affect the same residue.

| mutation (protein) | mutation (cDNA) | EXON | SIFT score | PolyPhen | | Number of patients reported | Biochemical phenotype | Frequency in control population | Reference |
|--------------------|-----------------|------|---|---|----------------|-----------------------------|-----------------------|--|--------------------------------------|
| | | | predicted consequence on protein function | predicted consequence on protein function | PolyPhen score | | | | |
| | c.-59-45del | 1 | | | | 1 | | <1:50 | [42] |
| | c.-188A>G | 1 | | | | 1 | | <1:50 | [47] |
| Y64N | c.190T>C | 3 | affected | probably | 2.736 | 8 | non-classical | <1:100 | [26] |
| V72F | c.214G>T | 3 | affected | possibly | 1.706 | 5 | non-classical | <1:50 | [38] |
| A77D | c.230C>A | 3 | tolerated | possibly | 1.992 | 24 | variable | <1:100 | [34, 44, 48, 49, 50, 51, 52] |
| G80V | c.239G>T | 3 | affected | probably | 2.499 | 2 | classical | <1:50 | [62] |
| G80S | c.238G>A | 3 | affected | possibly | 1.824 | 6 | variable | n/a | [48, 52] |
| R88G | c.262A>G | 3 | affected | probably | 2.429 | 1 | | <1:50 | [42] |
| R88T | c.263G>C | 3 | affected | probably | 2.204 | 7 | variable | <1:60 | [40] |
| N144H | c.430A>C | 5 | tolerated | possibly | 1.736 | 20 | variable | <1:200 | [39, 53, 54] |
| N144D | c.430A>G | 5 | tolerated | possibly | 1.676 | 2 | | n/a (<1:100)* | [78] |
| N144T | c.431A>C | 5 | tolerated | possibly | 1.751 | 1 | | n/a (<1:100)* | [55] |
| I152F | c.758A>T | 5 | affected | possibly | 1.799 | 2 | | n/a | [43] |
| D157G | c.470A>G | 5 | affected | probably | 2.189 | 1 | | <1:80 | [56] |
| D157N | c.469G>A | 5 | affected | possibly | 1.739 | 3 | classical | <1:50 | [38] |
| V162del | c.484_486del3 | 5 | | | | 27 | classical (>95%) | <1:100 | [44, 45, 46, 52, 57, 58, 59, 60, 61] |
| N174I | c.521A>T | 6 | affected | probably | 2.738 | 3 | variable | n/a | [48, 52] |
| R178G | c.532C>A | 6 | affected | probably | 2.429 | 3 | variable | n/a | [46] |
| I180T | c.539T>C | 6 | affected | probably | 2.204 | 1 | | <1:60 | [40] |
| D181V | c.846A>T | 6 | affected | probably | 2.639 | 4 | classical | <1:50 | [62] |
| Q182H | c.546G>T | 6 | tolerated | probably | 2.026 | 1 | classical | <1:80 | [56] |
| R178Q | c.533G>A | 6 | tolerated | possibly | 1.824 | 1 | | <1:50 | [42] |
| N185D | c.553A>G | 6 | tolerated | possibly | 1.838 | 15 | variable | <1:50 | [63] |
| L233P | c.698T>C | 6 | affected | probably | 2.243 | 1 | | n/a | [43] |
| G267D | c.1104G>A | 7 | tolerated | possibly | 1.722 | 1 | | <1:50 | [62] |
| D270V | c.808A>T | 7 | tolerated | probably | 2.099 | 1 | | n/a | [65] |
| G323V | c.968G>T | 7 | affected | probably | 2.499 | 1 | | <1:80 | [56] |
| C326S | c.977G>C | 7 | tolerated | possibly | 1.877 | 7 | non-classical | n/a (<1:100)* | [11, 25] |
| C326Y | c.977G>A | 7 | tolerated | probably | 3.237 | 4 | non-classical | <1:300 in Thai <1:100 in Vietnamese | [8, 10] |
| S338R | c.1014T>G | 7 | affected | possibly | 1.602 | 1 | | <1:94 | [9] |
| splice site** | c.1402G>A | 8 | | | | 1 | | n/a | [66] |
| R489S | c.1467A>C | 8 | affected | probably | 2.204 | 2 | variable | n/a (<1:100)* | [67] |
| R489K | c.1466G>A | 8 | affected | possibly | 1.529 | 6 | classical | <1:100 | [68] |
| G490D | c.1468G>A | 8 | affected | probably | 2.049 | 3 | variable | <1:100 | [41] |
| G490S | c.1468G>A | 8 | affected | possibly | 1.754 | 2 | variable | <1:100 | [42] |
| Y501C | c.1502A>G | 88 | tolerated | possibly | 1.830 | 7 | non-classical | <1:200 | [12] |

** predicted G468S

Corrigendum

Table 4. SLC40A1 non-synonymous single nucleotide polymorphisms. SNPs, which have been associated with high serum iron parameters, are highlighted in bold and italics.

| polymorphism (protein) | base substitution (cDNA) | EXON | SIFT score | | PolyPhen | | Frequency in control population | Reference |
|------------------------|--------------------------|----------|---|---------------|---|----------------|--|------------------------|
| | | | predicted consequence on protein function | catfile score | predicted consequence on protein function | PolyPhen score | | |
| Q248H | c.744G>T | 6 | tolerated | 0.10 | benign | 0.123 | Heterozygosity 11.6%** | [27, 28, 29] |
| F324S | c.971T>C | 7 | affected | 0.01 | probably | 2.507 | n/a** | ENSEMBL |
| G339D | c.1016G>A | 7 | tolerated | 0.62 | possibly | 1.648 | 0.5% in African American | [34] |
| L384M | c.1149T>A | 7 | affected | 0.01 | benign | 1.118 | 1.5% Allelic frequency in African American 10% Italian blood donors with high ferritin | [29, 34] |
| L384V | c.1149T>G | 7 | tolerated | 0.08 | benign | 1.343 | 1.5% Allelic frequency in African Americans 5% in Italian blood donors with high ferritin | [29, 34] |
| F405S | c.1214T>C | 7 | tolerated | 0.31 | benign | 0.982 | n/a** | ENSEMBL ³ |
| M432V | c.1294A>G | 7 | tolerated | 0.5 | benign | 0.76 | Heterozygosity 0.4%** | NCBI ² |
| P443L | c.1328C>T | 7 | tolerated | 0.67 | possibly | 1.758 | Heterozygosity 4.4%** | NCBI ² |
| R561G | c.1681A>G | 7 | tolerated | 0.28 | benign | 0.015 | Heterozygosity 2.8%** | NCBI ² [35] |

** predicted G468S

² as reported in the NCBI SNP database

(http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?chooseRs=coding&go=Go&locusId=30061)

³ http://www.ensembl.org/Homo_sapiens/Variation/Summary?v=ENSSNP12180430;vdb=variation