Hereditary Haemochromatosis (HH) type 1, HFE gene mutations frequency in a region of southern Europe

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BACKGOUND-AIM

HH type I is one of the most common autosomal recessive diseases in the caucasian population. It is due to mutations in the HFE gene of the short arm of chromosome 6. This disease is characterized by a disorder in iron metabolism, producing an increase in intestinal absorption of iron and the consequent storage in multiple tissues causing damage.

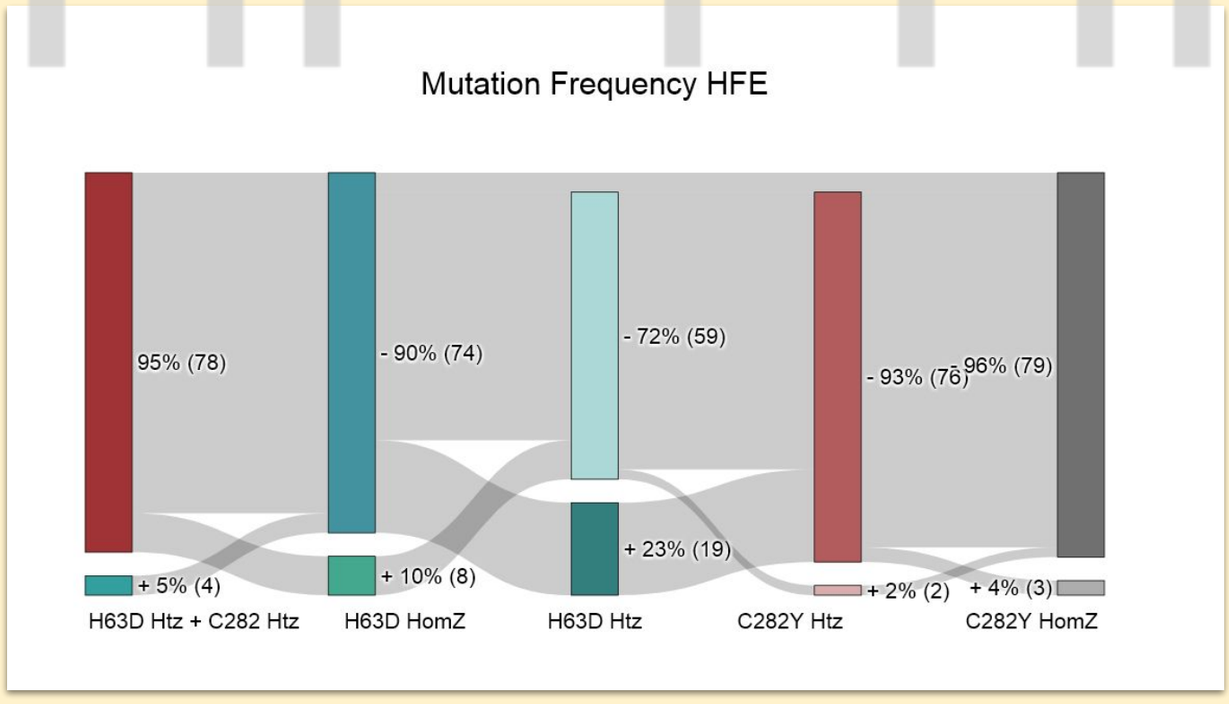
The aim of this study is to establish the frequency (%) of mutations for HH type I in our population in order to establish the genetic algorithm that fits this region of Baix Empordà, Catalunya.

METHODS

Eighty-two patients were screened for HFE gene mutations C282Y, H63D and S65C using molecular genetics assays (real time PCR) during 2021 and 2022. The inclusion criteria consisted of patients with situations of Ferritin> 300 ng/mL and Transferrin Saturation Index > 45%. The data in this prospective study was analyzed by SPSS from IBM.

RESULTS

The frequency (%) obtained for mutations in HFE consists of 2 heterozygotes for C282Y (2.43%), 3 homozygous for C282Y (3.65%), 19 heterozygotes for H63D (23.17%) and 8 homozygotes for H63D (9.75%). We also found a compound heterozygous genotype (C282Y/H63D) in 4 individuals (4.87%). We found no patients for S65C mutations (0.0%).



CONCLUSION

The distribution of HFE gene mutations found in our group matches the trends observed in other European countries: high frequency for the H63D mutation, followed by C282Y mutations and low or no frequency for the S65C. Despite the migratory exchange in our area of the Baix Empordà our distribution of mutations is in line with the total of the Spanish State. These results allow us to maintain our molecular diagnosis algorithm giving priority to the study of mutations in H63D followed by C282Y.