International Alliance of Hemochromatosis Associations Symposium

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The Italian point of view on hemochromatosis

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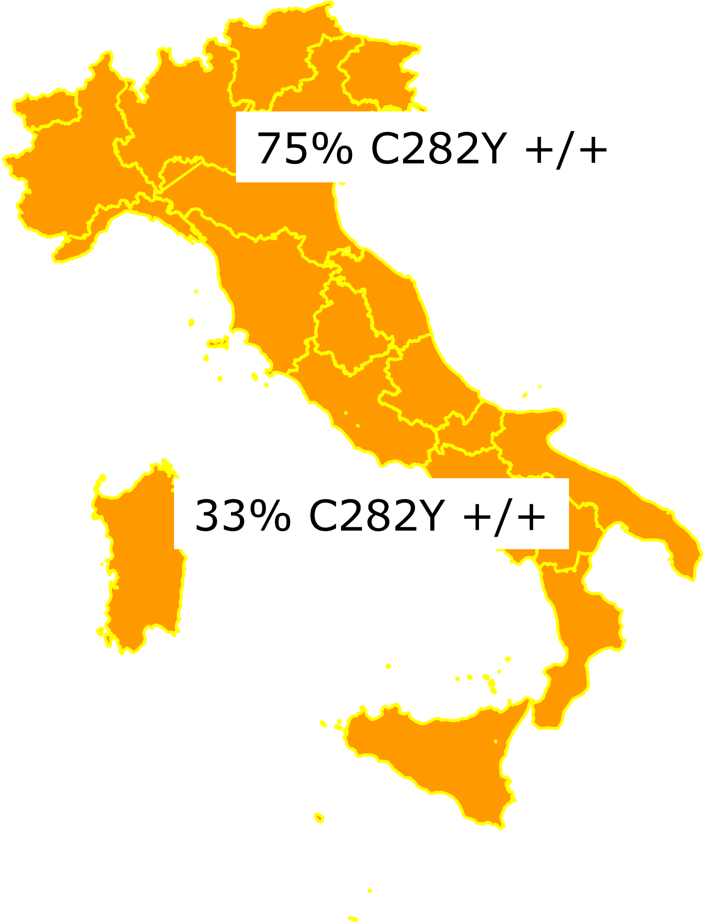
on behalf of the Italian Association for Hemochromatosis and Iron Overload Disorders –ONLUS – Monza

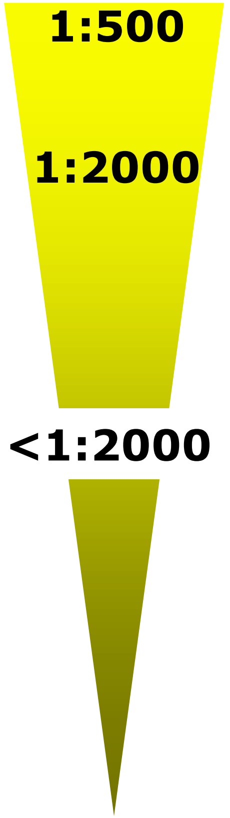
HH is not homogeneously distributed in Italy: the North to South gradient

This heterogeneity influences several issues: screening options, quality of care, information.

Frequency of C282Y +/+ in patients with HH phenotype



C282Y +/+ prevalence



HH in Italy is in the national register of rare diseases

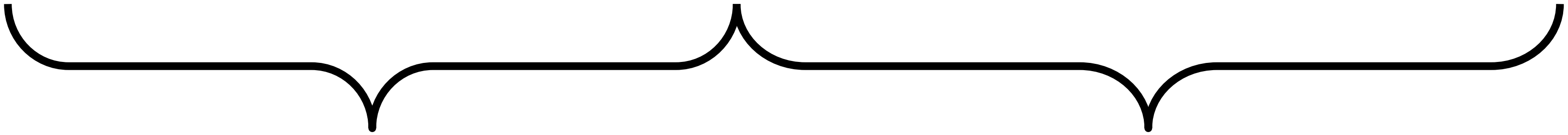
- Since 2001 patients affected by HH are covered by the National Health Service.
- The National Health Service supports HH patients for diagnostic tests, follow-up and therapies related to the disease, and for family screening. To get this each patient should be registered as affected, but this is not always done by referral centers, geneticists or experts.
- Private health insurance is confined to a small part of population and there is no relevant social or bank discrimination.

HH in Italy is in the national register of rare diseases

- According to the decree of the Ministry of Health in 2001, every region had to identify referral Centers for the disease. However, criteria to define these Centers were not clearly defined. This is more relevant in Centre and South of Italy where the disease is rare.
- Since 2000, the Association produced recommendation for diagnosis and therapy of HH available for physicians on the website *www.emocromatosi.it*.
- A very recent project from the Ministry of Health was aimed to identify groups of experts within each region to produce shared practice guidelines for some diseases including HH.

Correct use of genetic testing: analysis of 1133 molecular HFE test

Wildtype	**H63D +/-**	**C282Y +/-**	**H63D +/+**	**C282Y/H63D**	**C282Y +/+**
N=452	**N=257**	**N=77**	**N=88**	**N=99**	**N=160**
(39.9%)	**(22.7%)**	**(6.8%)**	**(7.8%)**	**(8.7%)**	**(14.1%)**

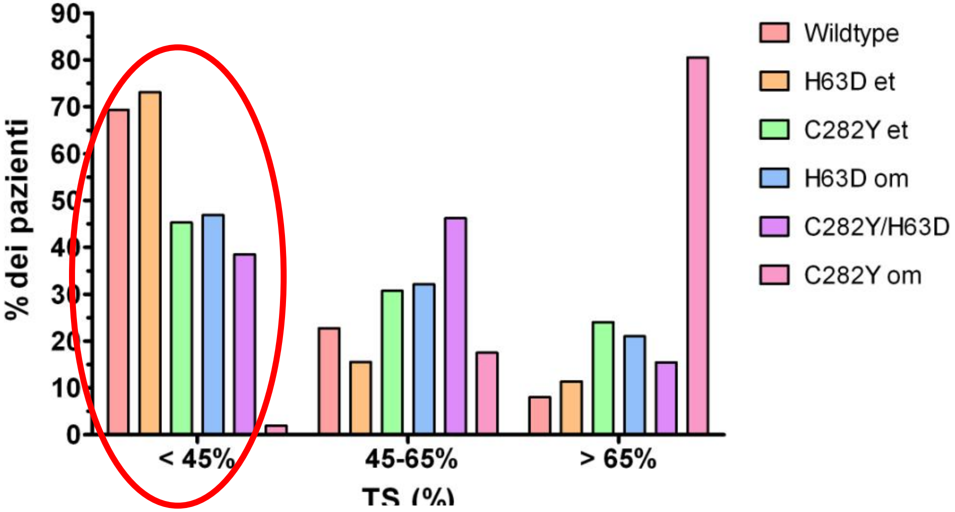


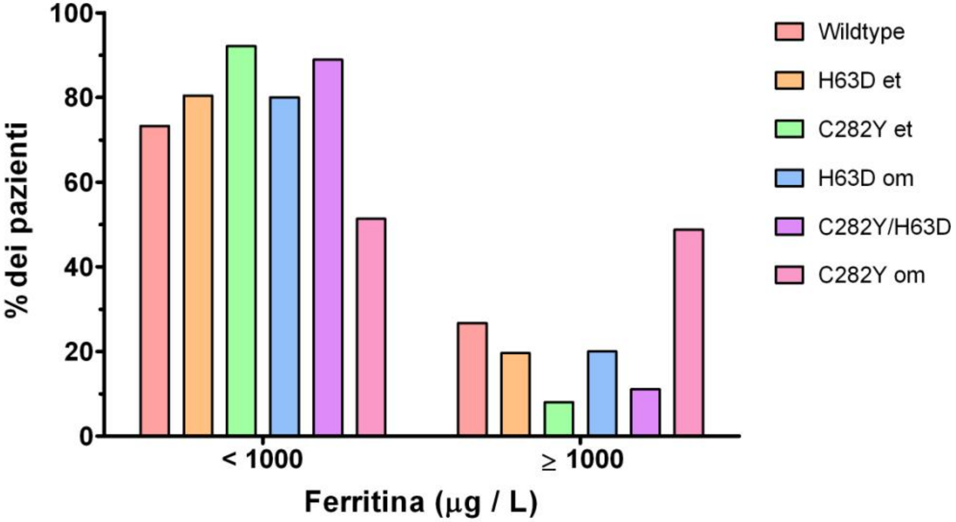
N=786
(69.4%)

N=347
(30.6%)

Genetic testing for HH is offered by many laboratories and counseling is not required. Results indicate that criteria defined by International and National Societies based on correct definition of the clinical phenotype of HH are often overlooked.

Adequateness of genetic testing: analysis of 1133 molecular HFE test





Genetic testing was offered to subjects who have not the biochemical features of HH: high transferrin saturation and serum ferritin.

Many of them have hyper-ferritinemia alone and most have dysmetabolic alterations, NAFLD or other associated chronic liver diseases.

Venesections: procedure, cost and use of blood

- Venesections are performed within referral Centers or, more often, Blood Banks.
- Referral Centers able to take global care of HH patient including therapy are few and mainly located in the North.
- Cost are covered by the National Health Service when patients are registered as affected.
- Use of blood for transfusions is not routinely accepted, but there is not an absolute prejudice. There is no national rule. HH patients can be refused as donors, accepted only in the maintenance therapy or when ferritin decreases below 600 ng/ml.

Venesections: The DoEmo (donor with hemochromatosis) project in Monza

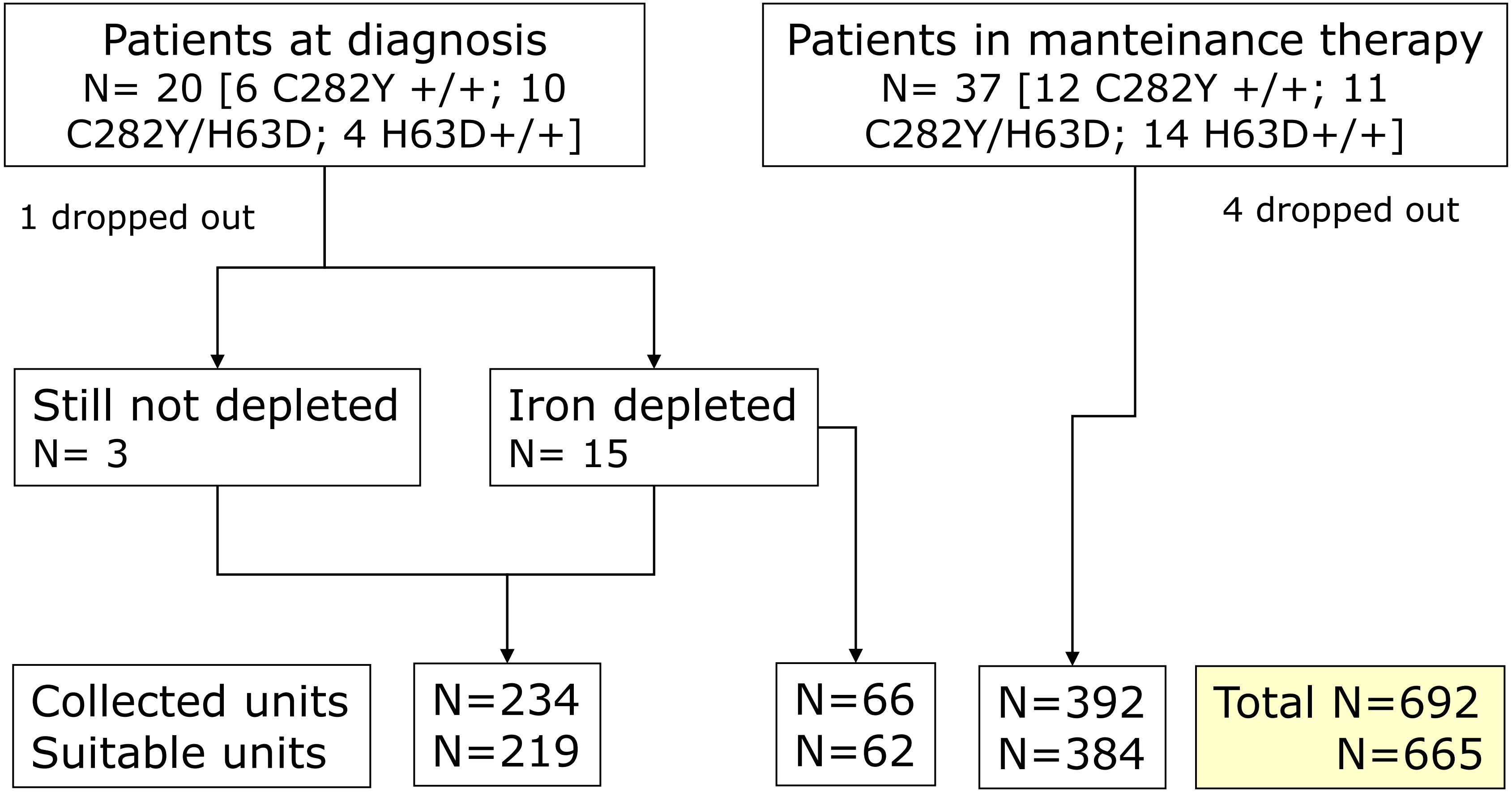
- Since the end of 2005 a project was approved whose aim was to offer to individuals with HH identified in the Clinical Unit the possibility to become blood donors.
- Inclusion criteria:

  -Subjects with normal hepatic tests, high TS %, serum ferritin below 1000 ng/ml, no clinical complications, consistent HFE genotype [C282Y +/+, C282Y/H63D, H63D +/+].

  -First diagnosis or maintenance therapy.
- Exclusion criteria:

  - hepatic fibrosis, coexistence of high alcohol intake or chronic hepatic diseases.

Venesections: The DoEmo (DOnor with hEMOchromatosis) project in Monza



Patients at diagnosis
N= 20 [6 C282Y +/+; 10 C282Y/H63D; 4 H63D+/+]

Patients in manteinance therapy
N= 37 [12 C282Y +/+; 11 C282Y/H63D; 14 H63D+/+]

1 dropped out

4 dropped out

Still not depleted
N= 3

Iron depleted
N= 15

Collected units
Suitable units

N=234
N=219

N=66
N=62

N=392
N=384

Total N=692
N=665

December 2009

Future efforts and aims

- To define the map of referral centers in Centre and South of Italy which can offer adequate counseling for the disease.
- To stimulate Scientific Societies to expand the knowledge of guidelines or recommendations for a correct diagnostic approach to the disease: not underestimate but also not overestimate HH.
- To define the map of centers which can offer upper diagnostic levels: rare HFE mutations, rare HH forms, non invasive quantification of hepatic iron, alternative therapies.
- To stimulate the Italian Society of Transfusional Medicine to establish national rules for the use of blood of HH subjects.

Italian Association for Hemochromatosis and Iron Overload Disorders

- Born in 1996 in Monza

Aims

- Inform physicians (general practitioners and specialists), patients and relatives on hemochromatosis.
- Support scientific research in the field of hemochromatosis and iron overload disorders.
- Support information to general population through the website [www.emocromatosi.it], social networks and a four-monthly journal.
- A hemochromatosis week is scheduled this year according to the initiative of the European association.

The Italian point of view on hemochromatosis


The global care of the patients

quality

Social issues

Medical issues

Information