EFAPH

European Federation of Associations of Patients with Haemochromatosis
Fédération Européenne des Associations de Malades de l'Hémochromatose

10 important points worth knowing about Haemochromatosis !

1. **Geographical spread** of the genetic disease throughout Europe by the migration of the celts and the propagation of the vikings.

2. **Prevalence** of the genetic defect (**homozygosity** of the C282Y mutation in the HFE-Gene) located on chromosome 6: about **1:300** in Europe (rising to 1:80 in Ireland).
   Prevalence of healthy carriers of the **heterozygote** mutation: about **1:8** in Europe.
   Penetrance is unknown, about 30% of homozygotes.

3. **Early symptoms** unspecific: chronic fatigue, joint pain.

4. **Without treatment: potentially lethal !**
   The gradual build-up of toxic iron stores in the organs causes slow progression towards **organ damage** and in the late stages: cirrhosis and carcinoma of the liver, cardiac failure, diabetes, polyarthritis and hormonal disorders.

5. **Diagnosis: simple and non-invasive !**
   Blood testing: Transferrin saturation and ferritin
   Blood or saliva testing: **Genetic test** (search for the C282Y mutation)

6. **Therapy: simple and effective !**
   Phlebotomy-initially frequent in order to reduce iron stores to normal level, then regularly lifelong at a rate to maintain normal level and prevent further excess.

7. **Prevention is possible and crucial !**
   In given individuals: Early blood test for **transferrin saturation and ferritin**.
   In families: Genetic testing as **family screening**.

8. **Early detection means:**
   - No more disease symptoms.
   - No more reduction in life-expectancy or quality of life.
   - No more early death by hepatic cancer or heart failure.
   - Reduced costs for the health care systems.

9. **Problems of Haemochromatosis patients:**
   - Underestimation of frequency of the disease by the medical profession.
   - Misinterpretation of the non-specific symptoms - delay in making correct diagnosis.
   - Lack of standardization of venesection therapy.
   - Lack of correct management of family screening.
   - Great variability in Europe of costs and reimbursement for genetic testing and venesection therapy
   - Exclusion of certain services of insurance companies

10. **Aims of EFAPH (www.efaph.eu):**
    - To raise awareness in the medical community and public
    - To promote and share best practices for the same chance of treatment and survival for all patients
    - To obtain funding for research concerning haemochromatosis.

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