Question for written answer E-012656/2011
to the Commission
Rule 117
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Subject: Early diagnosis and cost-effective management of hereditary haemochromatosis

Hereditary haemochromatosis (HH) is a genetic condition marked by iron overload in the blood. While the prevalence of this genetic defect (homozygosity of the C282Y mutation in the HFE gene) varies widely among Member States, it is estimated that 2 million inhabitants in the EU are potentially affected by the disease. Scientists and patients' groups have already worked on raising awareness of hereditary haemochromatosis which, if not diagnosed early, can result in severe damage to vital organs (cirrhosis and carcinoma of the liver, diabetes, polyarthritis and hormonal disorders) inducing not only poor quality of life but also disabilities and even death.

At present, simple and cost-effective ways already exist to diagnose and treat hereditary haemochromatosis effectively. A suspected diagnosis can be given by means of very simple and low-cost blood tests (transferrin saturation and ferritin) and can easily be confirmed by a sensitive and specific genetic test (search for C282Y mutation). If the test is positive it is extended to the patient's relatives. The current treatment consists of blood-letting or phlebotomy. Phlebotomies are periodically repeated in order to eliminate iron overload.

Given the cross-border dimension of hereditary haemochromatosis as well as the number of European patients potentially affected, and while recognising the primary responsibility of Member States for the organisation and delivery of health services and medical care, could the Commission answer the following questions:

1. Is the Commission considering initiatives or programmes which could enhance awareness of hereditary haemochromatosis, notably targeting risk-group populations and health professionals (particularly general practitioners)?

2. Would the Commission consider the development of specific guidelines to enhance early detection and diagnosis of hereditary haemochromatosis, thus allowing cost-effective management of the disease for healthcare systems in Europe by reducing the number of late-diagnosed patients?

3. Does the Commission intend to continue and step up its support for research into hereditary or genetic diseases?