Hereditary hemochromatosis and iron-induced diabetes: a missed diagnosis?

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Context: Hereditary hemochromatosis (HH) is a genetic disorder, that can lead to unregulated absorption of iron with resultant iron overload. The accumulation of iron in the entire body will eventually lead to fibrosis, cirrhosis, diabetes,? wich will lead to a higher morbidity and mortality. This can be prevented completely by timely diagnosis and treatment (phlebotomy). In family medicine we are less aware of HH and are unfamiliar with the complications, although the genetic prevalence is about one per 200-250 individuals. Diabetes mellitus is a common disease and we are trained to screen and treat patients in time. But are we considering diabetes as a common complication of HH?

Onderzoeksvraag: In our literature research we want to investigate if phlebotomy has an effect on the glucose metabolism in diabetic HH patients. Can phlebotomy ameliorate the glucose metabolism in these patients? With our project we want to investigate if the screening goals for HH in diabetic patients are reached in family medicine and in an academic hospital. And we want to know if genetic testing is done and if the indications for genetic testing are correct in family medicine and in an academic hospital.

Methode: We conducted a systematic literature research in the database MEDLINE with the search filter OVID from 18/05/2012 till 18/06/2012. As search terms, hemochromatosis, phlebotomy, bloodletting, iron overload, diabetes mellitus, hyperglycemia and hyperinsulinism were used. For our project we wanted to compare the data from three different sources: a general practice (?De Drempel), an epidemiological network of general practices (Intego), and an academic hospital (UH Leuven). A population born between 1941 and 1966 was selected. All patients with diabetes mellitus type two were selected. Their medical file was searched for ferritin (F) and transferrin saturation (TS) values, and if genetic testing for HH was ever done.

Resultaten: In our literature research we found 16 articles dealing with the effect of phlebotomy on the glucose metabolism in diabetic HH patients. Only few articles report some improvement of the glucose metabolism. Less advanced stages of diabetes will show more improvement. In our project both F and TS values were lacking in 7,55%-23,20%-27,11% (De Drempel ? Intego ? UH Leuven). Both parameters were known in 15,09%-25,54%-72,62%. Genetic testing was only done in UH Leuven, in 11,38% of the patients with abnormal results.

Conclusie: Phlebotomy can improve glucose metabolism in less than 50% of the diabetic HH patients. Patients not requiring insulin appear more likely to improve during iron depletion therapy. F and TS are not determined on a regular basis in diabetic patients in both general medicine and a third line hospital. Both parameters are lacking in 7,54 to 27,11% of the patients. In general medicine F is taken regularly but TS is hardly ever determined. In a third line hospital F and TS are usually taken together. Genetic testing is hardly done in both general medicine and third line hospital. The screening goals for HH in diabetic patients are not met.