SUMMARY

BACKGROUND: Hereditary hemochromatosis (HH) is one of the most common hereditary diseases of the Euro-American population. The disease is caused by increased absorption of iron from the intestine, regardless of its current stores in the body. Excess iron is stored in tissues and organs and causes their significant damage. HH treatment consists of regular blood withdrawal, activation of erythropoesis causes the utilization of excess iron and normalization of its reserves. The standard method of blood withdrawal is venipuncture, an alternative method with the possibility of collecting larger red cell volume in one procedure is erythrocytapheresis. The aim of the research was to create a group of patients with newly diagnosed HH, apply erythrocytapheresis, verify their effectiveness, optimize and standardize treatment to reduce ferritin levels in the induction phase of treatment below 50 μg/l and subsequently keep ferritin levels below 100 μg / l in the maintenance phase of treatment.

METHODS: The group of patient was created in cooperation with hepatology departments of the Hradec Králové region, by testing related patients with HH and by screening blood donors. For erythrocytapheresis, a Haemonetics MCS+ separator (Haemonetics Corp., Braintree, MA, USA) was used. The SW of MCS+ can modify red cell collection to each patient. Erythrocytaphereses in the induction phase were performed with a planned interval of 10–14 days between two procedures in order to collect 25–35 % of the patient's total erythrocyte volume (TEV) in each of procedures. In the maintenance phase, erythrocytapheresis was performed individually depending on the rate of increase of the ferritin level.

RESULTS: A total of 47 patients (37 men and 10 women) were treated with erythrocytapheresis, the average age at the start of treatment was 52 years. 33 patients carried the homozygous C282Y mutation, 4 patients were H63D homozygous, 3× C282Y/H63D heterozygous. The average ferritin level before treatment was 1429 μg/l, 51 % of patients had ferritin level above 1000 μg/l. A total of 1086 erythrocytapheresis, 501 in the induction phase and 585 in the maintenance phase were evaluated. In the induction phase, an average of 532 ml of red cells were collected in one apheresis (550 ml in men and 464 ml in women), which represents a collection of 26.5 % TEV in men and 31.6 % TEV in women. Treatment in the induction phase was completed by 42 patients, in 5 patients the treatment was not completed (medical reasons, moving, death). In patients with completed treatment, the average ferritin level dropped to 26 μg/l, and in all of them a decrease in ferritin levels below 50 μg/l was achieved. Adverse reactions were observed in 10.4 % of induction and in 6,8 % of maintenance procedures: mild vasovagal reactions or mild manifestations citrate toxicity. Only 2 reactions were classified as moderate (1× hypotension and 1× citrate toxicity). A total of 33 patients underwent maintenance treatment, and 1 – 3 erythrocytaphereses were performed annually.

CONCLUSION: Erythrocyteapheresis is an effective, safe and well-tolerated treatment procedure for patients with HH. The effectiveness, safety and tolerability of this method is proven by the analysis of the test group. This analysis is one of the largest published files on the treatment of HH. The aims of the research were fully met, other findings were processed outside the original intention: the importance of monitoring ferritin as a simple prevention parameter of significant reduction of iron stores in first-time donors before entering the donation of blood and blood components.