ABSTRACT

Trombophilia means an increased disposition to creation of trombs. Health complication incurred as a consequence of hypercoagulation can be very serious. When a trombophilic mutation is found at a patient, it brings necassity of thrombosis – control in risk situations (e.g. pregnancy, operation) for the rest of the patient´s life.

There were filed 300 people (206 women and 94 men) with trombophilic mutations into my study of clinical signification of trombophilic mutations. These people were examinated in years 2008 – 2010.

Most of positive medical findings – 266 people, were recorded in the area of MTHFR (C677T i A1298C) mutations. There were less findings in the field of FV Leiden and FII prothrombin mutations. Multipath trombophilic mutations were found at 99 patients.

I accordance with foreign literature, our results advert to clinical consequences of trombophilic mutations like: repeated spontanious aborts, cerebrovascular akcident (CA), ischaemic heart disease (IHD), thrombosis, flebothrombosis, pulmonary embolism, varicose veins, aseptic necrosis of hip bone, arterial sclerosis and aortic stenosis. Mutations MTHFR C677T and MTHFR A1298C we found mainly at patients with CA, IM and IHD. Leiden mutation was most often found at patients with thrombosis, flebothrombosis and pulmonary embolism.

We verified familiar presence of trombophilic mutations. All the patients inherited trombophilic mutations from one or both parents. We have not recorded any new mutation. FV Leiden and FII prothrombin mutations are inherited in autosomal dominant way. Mutations MTHFR C677T and MTHFR A1298C are inherited in autosomal incomplete dominant way.

There are 51 female patients who went through spontaneous abort in my file of female patients with mutation MTHFR A1298C in homozygous phase tend to have spontaneous abort – 64% of these female patients (n = 14) had it – which is a very high percentage. Female patients with mutation MTHFR C677T in homozygous phase had spontaneous abort in 47,6 % (n = 21). We also recorded fissure disease occurrence at these patients.

The main asset of this diagnostic is a possibility to choose an individual approach to treatment of patients and through it a possibility to increase effectivity of the treatment. At the same time, these findings can contribute to envelopment of new therapeutical methods, higher effectivity of genetical couselling and pre-konceptual treatment.