Abstract

Autosomal dominant polycystic kidney disease is an inherited chronic disease which is characterized by the cysts formations. It´s the most frequent congenital of kidney disease. Incidence occurs at about 1 : 1000. The presence of multiple cysts cause the wastage of functional parenchym, kidneys get bigger and deform. The disease progression is slow and at the beginning it is asymptomatic. The symptoms appear in the course of life by every person affected by the disease. Between the typical symptoms belong the backpain in the lumbal area, frequent urinary infections, haematuria and anterial hypertension. In the course of time the complications increase and the disease gets worse as the result of the functional glomerulus decrease. ADPKD almost everytime flow to chronic renal insufficiency at adulthood.

The disease is mostly diagnosted at the higher age, because of the asymptomatic phase at the beginning. However the accidental sonography can reveal the disease at the childhood, because the cysts already begin to form. Diagnostic is done by imaging metods - mostly sonography and by radiologic methods - computer tomography. These metods serve for the prove of the multiple cysts and increase of the kidneys. Discovery of these symptoms is an indication for the molecular genetic examination which prove the presence of the PKD1 or PKD2 gene mutation. During the PKD2 gene mutation the disease has a milder progession and the kidney failure come at the higher age compared to the patients with the PKD1 gene mutation. With the age increasing begin to show the clinic symptoms.

The bachelor thesis is mostly focused on the laboratory diagnostic and the basic measured laboratory parameters for the examinations of the renal functions by the polycystic kidney disease with the autosomal dominant type. All the laboratory tests and parameters have been done in the Karovy Vary hospital. The nefrologic ambulance in the Karlovy Vary hospital currently has 3 patients with the ADPKD. After the causuistic stating we are going to monitor the disease progression, beginning with the patient´s arrival to the nephrology ambulance and subsequently in 2014-2017. We are going to be focused mostly on the most important biochemic and haematologic parameters that are the sign of the disease progressions. Laboratory examinations are important for the monitoring and evaluation of the patien´s state of health, for the well timed start of the medical therapy as well as for the suitable medication and the regime measures.

Autosomal dominant kidney disease leads by every individual to the chronic kidney failure. The first step of the medical therapy by the patients with the chronic renal insufficiency is the conservative therapy. It lies in the symptoms postponement, complications suppression and in the deceleration of the disease progression. In the terminal phase of the renal failure is the kidney activity replaced by dialysis or transplantation.