ABSTRACT

Porphyria is a group of inherited metabolic disorders due to enzymatic defect of the heme biosynthesis resulting in the overproduction of the heme precursors' porphyrins in different body organs. The enzymes of the heme biosynthesis are encoded by corresponding genes in which any defect in any of these genes lead to a specific type of porphyria. Numerous mutations were detected in these genes leading to impairment in the enzyme function and therefore developing of the clinical manifestations of porphyria. The aim of the present work was to investigate the *UROD* gene in patients with porphyria cutanea tarda (PCT) and hepatoerythropoietic protoporphyria (HEP) as well as the *FECH* gene in patients with erythropoietic protoporphyria (EPP) on a molecular level. We identified numerous mutations in the *FECH* and the *UROD* genes in three different populations, Czech, Slovak, and Egyptian. We described the novel mutations in the *UROD* gene in HEP Arabic patients from Egypt as well in the *FECH* gene in patients with EPP of Czech and Slovak origin. We expressed mutatted UROD protein in prokaryotic system and found 19 % of the wild-type enzymatic activity. Moreover, the current study presents for the first time the frequency of the low expression allele IVS3-48c in the *FECH* gene in healthy controls from the Czech population. We analyzed 624 alleles in unrelated individuals from the general Czech population and it was found out to be 5,5 % among the Czech population, similar as in Caucasians from West Europe.

Keywords: Porphyria cutanea tarda, Hepatoerythropoietic porphyria, Eryhtropoietic protoporphyria, Uroporphyrinogen decarboxylase, Ferrochelatase.