Clinical aspects of acute intermittent porphyria in northern Sweden: A population-based study.

https://arctichealth.org/en/permalink/ahliterature89204

Author: BylesjoIngemar
WikbergAgneta
AnderssonChrister

Author Affiliation: Department of Family Medicine, University of Umeå, Umeå, Sweden.

Source: Scand J Clin Lab Invest. 2009 Apr 28;:1-7

Date: Apr-28-2009

Language: English

Publication Type: Article

Abstract: The objective of this study was to update the clinical issues of acute intermittent porphyria (AIP), as they have not been in focus for years, and to be aware of potentially associated disorders and social consequences. A total of 356 gene carriers of AIP from northern Sweden participated in this retrospective population-based study. Eight mutations were found with a predominance of W198X (89%). Clinical manifestations of AIP (manifest AIP) were identified in 42%, 65% were women. Women were more severely stricken by AIP attacks concerning number and duration, hospital admission and early onset. Men reporting most attacks were > 40 years of age. In addition to traditional symptoms during attacks, fatigue was commonly described. Chronic AIP symptoms and disability pension due to AIP were reported in about 20% of subjects. Precipitating factors were reported with evident sex differences. Half of the gene carriers who were on medications used drugs considered not safe (in 1999), mainly antihypertensive drugs. Smoking was associated with high AIP attack frequency. Elevated levels of ALT, bile acids, creatinine, U-ALA and U-PBG and decreased levels of creatinine clearance were associated with manifest AIP. The same was true for hypertension and myalgia in the legs. Hepatoma was strikingly overrepresented. The high prevalence of manifest AIP in this study could be explained by a mutation-dependent penetrance. Our results emphasize the importance of early diagnosis, counselling and treatment of attacks, screening and treatment of associated disorders.

PubMed ID: 19401933 View in PubMed