The Prevalence and Patient Characteristics of Hepatic Porphyria: A National Database Study

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BACKGROUND

- Hepatic porphyria (HP) is a relatively rare clinical entity that could be a hereditary or acquired condition of defective heme synthesis that lead to the accumulation of heme precursors.
- Patients with acute HP exhibit episodic symptoms, including severe abdominal, neurologic, psychiatric, or cardiovascular symptoms.
- Often intensive care is required to treat attacks; hence, early detection and awareness of the prevalence are of dire importance. Our study aimed to assess the prevalence and GI manifestation of HP in the US cohort.

METHODS

- A large multi-center database (Explorys Inc., Cleveland, OH, USA) of aggregated electronic health records of 26 different healthcare systems with 360 hospitals.
- The database covers ~25% of the US population. A cohort of patients with a SNOMED-CT diagnosis of "Hepatic porphyria" between 1999 to 2022 was identified.
- Subsequently, two sub-cohorts of patients were identified: those diagnosed with HP and those who were not.

	Hepatic Porphyria	Without Hepatic Porphyria	OR	CI	p-value
Age					
18-65	810 (55%)	47944950 (68%)	0.5656	0.5106 to 0.6265	< 0.0001
>65	670 (45%)	21252540 (30%)	1.9119	1.7259 to 2.117	< 0.0001
Race					
African-American	80	7024810	0.5153	0.4114 to 0.6455	< 0.0001
White	1260	37846710	4.9224	4.2656 to 5.680	< 0.0001
Gender					
Female	640	38458330	0.6323	0.5705 to 0.7008	< 0.0001
Male	830	31417300	1.5834	1.4289 to 1.7546	< 0.0001
Symptoms					
Severe abdominal pain	570	9662680	3.9356	3.5444 to 4.3700	< 0.0001
Acute polyneuropathy	180	1231180	7.7761	6.6536 to 9.0878	< 0.0001
Seizures	80	773020	5.1451	4.1071 to 6.4453	< 0.0001
Hyponatremia	10	96790	4.9394	2.6519 to 9.1998	< 0.0001
Chronic liver disease	460	904290	34.6458	31.0338 to 38.6781	< 0.0001
Liver transplantation	20	41800	23.0494	14.8244 to 35.8379	< 0.0001
chronic kidney disease	250	2302560	6.0089	5.2449 to 6.8842	< 0.0001
Hypertension	270	3500580	4.2628	3.7360 to 4.8640	< 0.0001
Hepatocellular carcinoma	20	25530	37.7473	24.2758 to 58.6944	< 0.0001

Table 1: A comparison of the baseline characteristics and GI manifestation of patients with and without Hepatic Porphyria. Univariate analysis used to calculate OR.

DISCUSSION

- This is one of the largest population-based studies, which entails comparative data for GI manifestation of HP in the US population.
- HP patients have a greater risk of developing liver disease, including CLD and HCC; hence a special focus should be provided for patients who develop GI manifestation.

RESULTS

- Among the 70,376,230 individuals screened in this database, there were a total of 1,480 individuals diagnosed with HP—a prevalence rate of 0.21 per 100,000 in the US population.
- In contrast to those without, patients who were diagnosed with HP tended to be older >65 (OR 1.91), predominantly of Caucasian (OR 4.92) race and to be males (OR 1.58).
- In terms of neurological manifestations, population with HP was more prone to develop seizures (OR 5.15), hyponatremia (OR 4.94), and acute polyneuropathy (OR 7.78).
- In addition, the HP cohort was more likely to develop hypertension (OR 4.26), chronic liver disease (CLD) (OR 34.65), hepatocellular carcinoma (HCC) (OR 37.75), and chronic kidney disease (CKD) (OR 6.01) (table 1).

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