Assessing the health status of ≥16-year-old patients with early-diagnosed PKU from the retrospective analysis of the full French insurance claims cohort 2006-2018

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Background

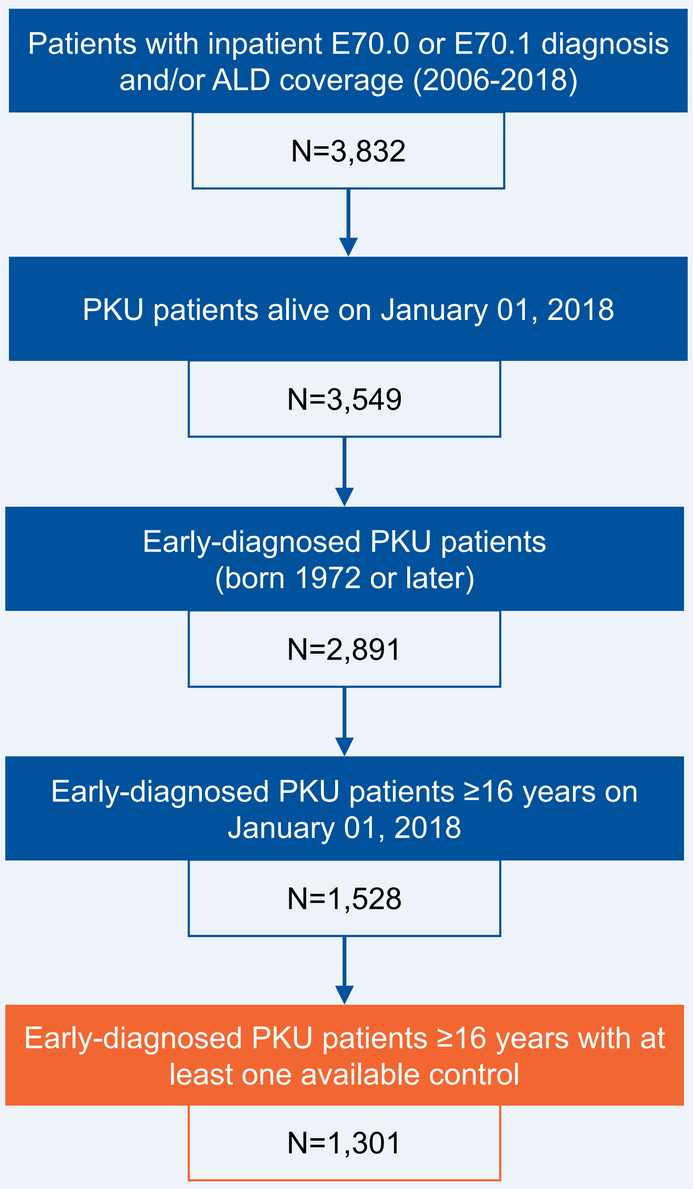
- Phenylketonuria (PKU) is an inherited deficiency in the enzyme phenylalanine hydroxylase
- Untreated PKU results in deficient growth, microcephaly, seizures, and intellectual impairment
- Management of PKU should therefore start soon after diagnosis to prevent irreversible damage. For this reason, newborn screening for PKU was implemented in 1972 in France
- While PKU effects on childhood development are well covered in the literature, consequences on adulthood require further analysis
- **Thus, this study aimed at evaluating the health status and healthcare consumptions of ≥16-year-old patients with early-diagnosed PKU in France**

Methods

- This retrospective observational study used health insurance claims data from the French database SNDS, including data from over 66 million French inhabitants
- PKU patients were identified between 2006-2018 by ICD-10 diagnosis codes E70.0/ E70.1 documented as chronic condition (affection de longue durée – ALD) or in the inpatient setting
- PKU patients born after implementation of the newborn screening in France in 1972, before 2002, and alive on January 01, 2018, were included and matched to controls without PKU regarding age, gender, and region
- Comorbidities of interest were assessed using the algorithms collected in 2015 by Quantin and the French Sickness Fund[1] for selected diseases
- Medications were assessed by using 1-digit Anatomical-Therapeutic-Chemical (ATC) classification codes. Sapropterin was identified via ATC code A16AX07 and dietary amino acid supplements were identified by the PRS_NAT_REF code 3517
- Outcomes were analyzed for the year 2018

Results – Population and Demographics

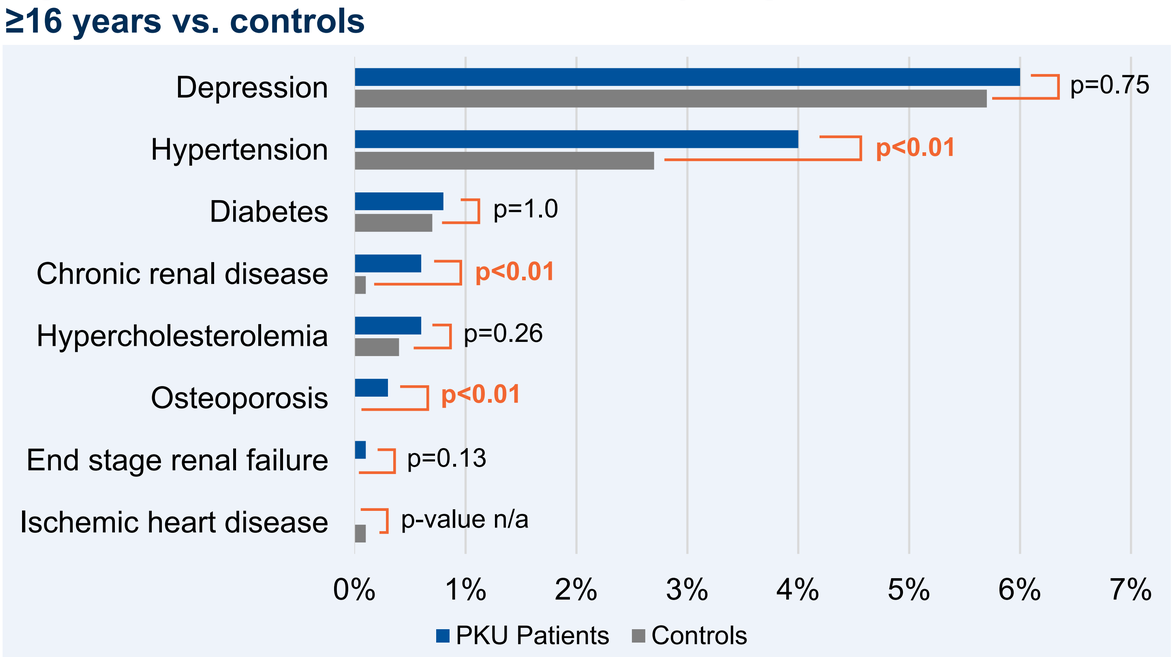
Figure 1. Flowchart



- Of 3,832 patients with a PKU diagnosis between 2006-2018 in France, 1,528 patients were still alive in the beginning of 2018, categorized as early-diagnosed and at least 16 years old on January 01, 2018
- Among those, 1,301 early-diagnosed PKU patients had at least one healthcare consumption in 2018 and could be matched to 6,457 controls without PKU **(Figure 1)**
- The mean age of the PKU population and the controls was 28.4 (±8.4) years. The age ranged from 16 to 46 years
- More than half of both matched populations (59.6%) was female

Results – Comorbidities of Interest

Figure 2. Comorbidities of interest in early-diagnosed PKU patients ≥16 years vs. controls

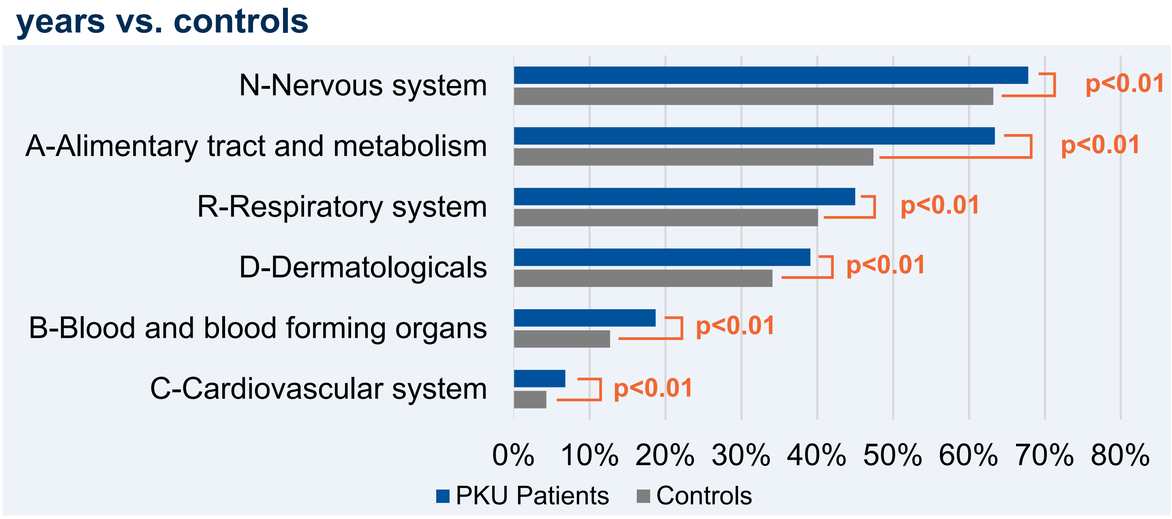


- The study results indicated a significantly higher prevalence of hypertension (4.0% vs 2.7%), chronic renal disease (0.6% vs 0.1%), and osteoporosis (0.3% vs 0.0%) in PKU patients than in controls (**Figure 2**)
- The mean Charlson Comorbidity Index was comparable for both cohorts (0.0±0.3 in PKU cohort vs 0.0±0.4 in controls; p=0.47)

Results – Pharmaceutical Treatment

- 7% of the early-diagnosed PKU patients ≥16 years received sapropterin and 40.4% received dietary amino acid supplements
- Significantly more PKU patients than controls received medications for e.g., nervous system (67.8% vs 63.2%; p<0.01), respiratory system (45.0% vs 40.1%; p<0.01), and cardiovascular system (6.8% vs 4.3%; p<0.01) **(Figure 3)**

Figure 3. Selected ATC classes in early-diagnosed PKU patients ≥16 years vs. controls



Discussion

- This study utilized the French database SNDS including data from over 66 million French inhabitants, leading to the largest PKU population analyzed
- The results indicate that even early-diagnosed PKU seems to have an overall clinical impact on adult health status, with a significantly higher risk of comorbid hypertension, chronic renal disease, and osteoporosis compared to non-PKU controls, along with increased healthcare consumptions in terms of pharmaceutical treatment
- However, less than expected early-diagnosed PKU patients were treated with sapropterin and dietary amino acid supplements
- Claims data analyses are subject to inherent limitations, as claims data are primarily collected for reimbursement purposes. This means they do not cover clinical parameters, self-treatment or over-the-counter medications. Additionally, socio-economic information is not available

Conflict of Interest

The study, data analysis, writing, editing, and poster production was funded by BioMarin Europe Limited (BioMarin). JBA, FM, and SC received expert honoraria from BioMarin. JT, CJ, and KMS are full-time employees of Xcenda GmbH acting as contractor for BioMarin for the execution of this study. SB is a full-time employee of CEMKA acting as a contractor of Xcenda GmbH for the execution of this study.

Associated Poster

Assessing the health status patients with late-diagnosed PKU from the retrospective analysis of the full French insurance claims cohort 2006-2018.

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