Health Economic Impact of Phenylketonuria – A Retrospective Claims Database Study in France

Paul Okhuoya[1], Julia Altevers[2], Jean-Baptiste Arnoux[3], Stéphane Bouée[4], Sybil Charrière[5], Christian Jacob[2], Ashok Jha[1], Francois Maillot[6], Kim Maren Schneider[2]

[1]1BioMarin Europe Ltd., London, UK; [2]Xcenda GmbH, Hannover, Germany; [3]Centre de Référence des Maladies Héréditaires du Métabolisme, Hôpital Necker-Enfants Malades, Paris, France; [4]CEMKA, Bourg-La-Reine, France; [5]Fédération d'endocrinologie, diabétologie, maladies métaboliques et nutrition, Hôpital Louis Pradel, Hospices Civils de Lyon, Université Claude Bernard Lyon 1, INSERM U1060 CarMen, Lyon, France; [6]Service de médecine interne, CHRU et université de Tours, Tours, France

Background

- Phenylketonuria (PKU) is an inborn error of metabolism that results in elevated levels of the essential amino acid phenylalanine and reduced levels of tyrosine.
- If untreated, PKU results in irreversible damage such as neurological impairment or mental retardation.
- Understanding the impact of PKU on patients and healthcare systems needs to consider exhaustive healthcare consumptions of PKU patients.
- **This study aimed at evaluating the health economic impact of PKU in France.**

Methods

Data Source

- This retrospective observational study used health insurance claims data from the French database SNDS, which covers around 99% of the French population.

Study Population

- PKU patients were identified between 2006 and 2018 by the following ICD-10 diagnosis codes documented as chronic condition (affection de longue durée – ALD) or in the inpatient setting:
  - E70.0 Classical phenylketonuria
  - E70.1 Other hyperphenylalaninaemias
- PKU patients who were alive and ≥16 years old on January 1, 2018, were included.
- For each eligible PKU patient, up to five controls without PKU were drawn from the database via direct exact matching without replacement on age, gender, and region.
- The demographic characteristics age and gender were analyzed on January 1, 2018.
- Outcomes were analyzed for the year 2018.
- The healthcare costs were defined from the third-party payer perspective. They reflect the costs reimbursed by the statutory health insurance in France.

Acknowledgments

The study, data analysis, writing, editing, and poster production was funded by BioMarin Europe Limited (BioMarin).
PO and AJ are full-time employees of BioMarin. JBA, SC, and FM received expert honoraria from BioMarin. JA, 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.

Results

Figure 1. Third-party payer healthcare costs (€) of PKU patients and controls in 2018

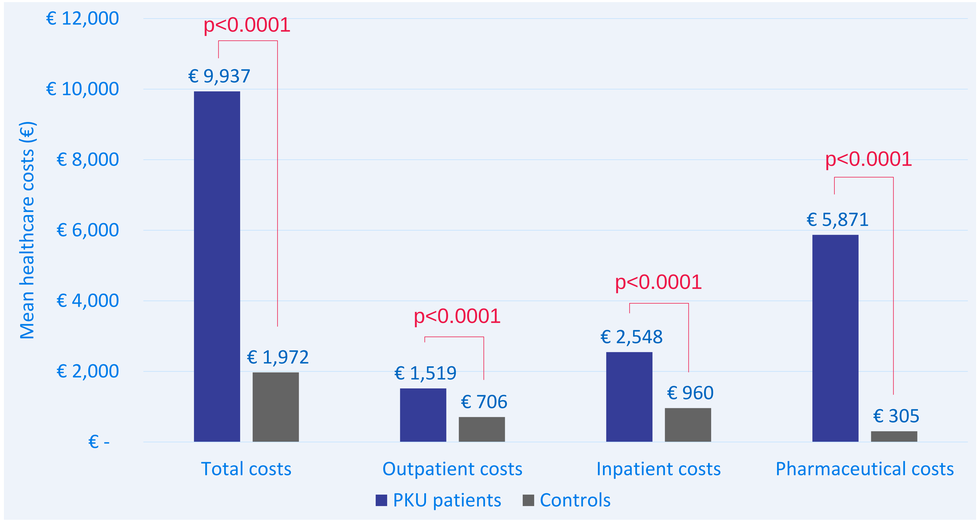


Figure 2. Healthcare costs differences

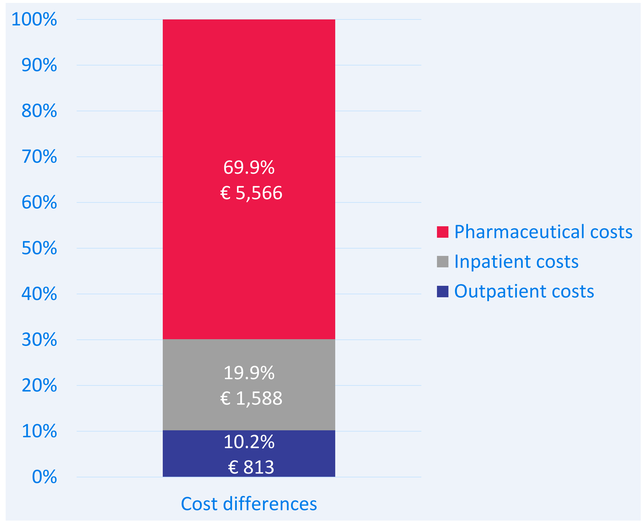
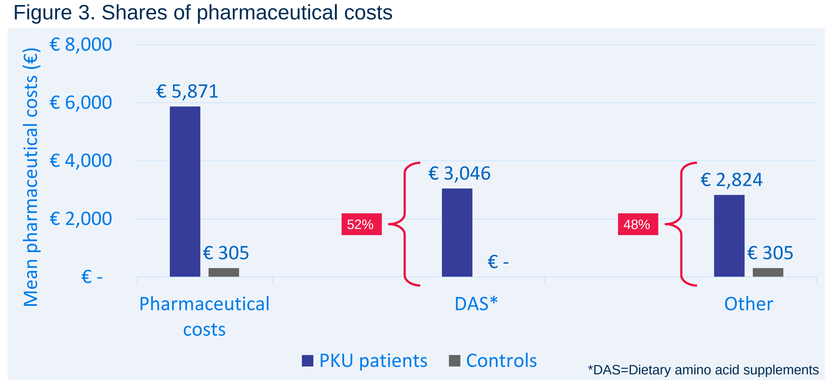


Figure 3. Shares of pharmaceutical costs



- Overall, 1,911 PKU patients aged 16 years or older were matched to 9,505 controls (mean age 41.2 years, 56.2% female). The PKU cohort included patients born before and after implementation of newborn screening in France in 1972.
- On average, total healthcare costs were five times higher in PKU patients compared to non-PKU controls (€9,937 vs €1,972; p<0.0001) (Figure 1).
- Pharmaceutical (69.9%) and inpatient costs (19.9%) accounted for most of the overall cost difference (€7,965) (Figure 2).
- Around half of the pharmaceutical costs (51.9%) in the PKU cohort were attributable to dietary amino acid supplements (DAS) (Figure 3).
- More PKU patients were hospitalized at least once in 2018 (29.3% vs 18.1%; p<0.0001), with more stays (3.3 vs 1.8 hospitalizations; p<0.0001) and longer stays (7.3 vs 4.6 days; p=0.0007) (Table 1).

Table 1. Hospitalizations in 2018

	PKU patients	Controls	p-value
Hospitalization	559 (29.3%)	1,718 (18.1%)	<0.0001
Frequency (if at least one hospitalization)			
Mean (±SD)	3.3 (±13.7)	1.8 (±3.3)	<0.0001
Min / Median / Max	1.0 / 1.0 / 163.0	1.0 / 1.0 / 44.0	
Quantile 25 / Quantile 75	1.0 / 2.0	1.0 / 2.0	
Length of stay (if at least one hospitalization)			
Mean (±SD)	7.3 (±23.6)	4.6 (±14.3)	0.0007
Min / Median / Max	0.0 / 2.0 / 353.0	0.0 / 1.0 / 359.0	
Quantile 25 / Quantile 75	0.0 / 7.0	0.0 / 4.0	

Conclusions

- This study utilized the French database SNDS including data from over 66 million French inhabitants, leading to the largest PKU population analyzed.
- The study indicated higher healthcare costs and resource use for PKU patients in France compared with the general population.
- These incremental costs were mainly driven by pharmaceutical (including DAS) and inpatient costs.
- Claims data analyses are subject to inherent limitations, as claims data are collected for reimbursement purposes. Clinical parameters, self-treatment or over-the-counter medications are not available.