The Impact of Prader-Willi Syndrome (PWS) on Caregivers and the Healthcare System: A Burden of Illness Study Design

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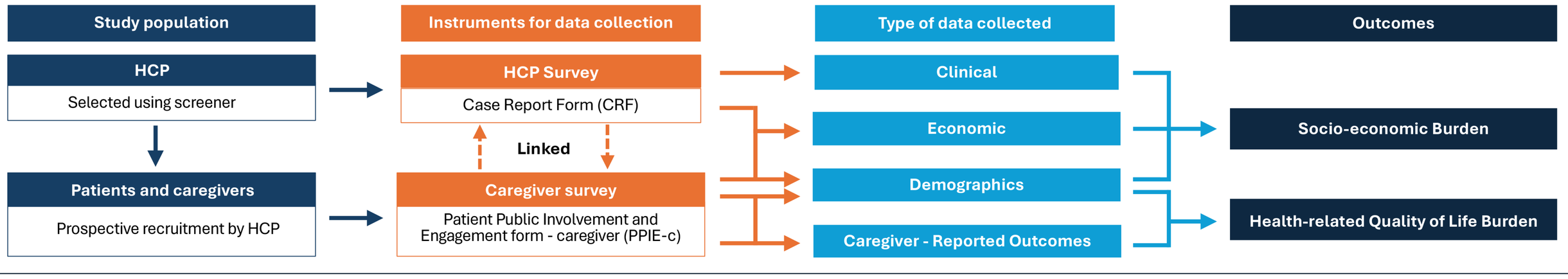
Study Design Schematic



STUDY DESIGN

To assess the burden of PWS, a retrospective review will be conducted by consenting approximately 80 healthcare professionals (HCPs). HCPs who qualify and consent to participate will complete case record forms (CRFs) for eligible patients under their care. The target sample is 330 CRFs. The CRF will collect data on PWS disease history, management, and burden during the past 12 months from the date of consultation with the HCP. Aspects captured by the CRF will include socio-demographics, symptoms, disease history, comorbidities, management, interventions, and care requirements.

HCPs will invite caregivers of these patients to complete corresponding Patient Public Involvement & Engagement caregiver forms (PPIE-c). The PPIE-c will capture health-related quality of life (HRQoL) through EuroQoL 5-Dimensions 5-levels (EQ-5D-5L); the Work Productivity and Activity Impairment (WPAI) Questionnaire; the Hyperphagia Questionnaire for Clinical Trials (HQ-CT), the Zarit Burden Interview (ZBI) and the Food Safe Zone (FSZ) Questionnaire. The PPIE-c will help provide insights into the wider impact of PWS on caregivers and families. All CRF and PPIE-c data will be collected anonymously.

The mean per-patient cost of PWS will be calculated based on a comprehensive resource utilisation analysis incorporating unit cost estimates for direct medical and non-medical, and indirect resource requirements. Key factors to be evaluated include hospitalisations, HCP visits, treatments, PWS-related transportation, specialised equipment, education status, and dietary modifications.

INTRODUCTION

Prader-Willi syndrome (PWS) is a rare, genetic, neurobehavioural-metabolic disorder defined by hyperphagia and behavioural/psychological complications.[1,2] PWS is associated with a significant global health and socio-economic burden. A new generation of therapies has the potential to dramatically improve health outcomes and reduce the burden of care. However, robust data on the 'real life' burden and cost of PWS to both patients and their caregivers and families is needed to understand the impact of existing interventions, as well as the potential gains of new therapies.

To provide a comprehensive understanding of the health and socio-economic burden of PWS, we will carry out a micro-costing (a detailed approach to costing that involves measuring all resources used for a specific health service or intervention) burden-of-illness (BOI) study across the USA, UK, France, Germany, and Italy.

Countries Included in the Burden-of-Illness (BOI) Study

USA UK France Germany Italy

The study aims to provide robust evidence to quantify the burden of illness of PWS for the first time and to provide valuable inputs for the PWS health economic model. It will assess the extent of the unmet need, the clinical impact of living with PWS, resource utilisation, the cost of living with the disease, and its humanistic burden. The data generated will provide a deeper understanding of how PWS affects caregivers and families, translating clinical classifications into valuable advocacy tools for the PWS community. The study will begin in Q1 2025 and is estimated to be complete by Q3 2025.

Primary Study Objectives

To assess the clinical burden of PWS
- Measure key variables associated with the clinical outcomes of PWS
- Measure treatment patterns in the management of PWS

To assess the economic burden associated with the management of PWS
- Measure the direct and indirect costs incurred by patients and caregivers and the healthcare setting

To assess the humanistic burden associated with PWS
- Measure the impact on patient and caregiver quality of life

HCP Inclusion Criteria

- Primary speciality: To include endocrinology (adult or paediatric), paediatrics, and psychiatry
- Qualified in their medical speciality for at least three years
- Personally responsible for the management of adult and/or paediatric patients (aged 4 and over) with PWS.

Patient Inclusion Criteria

- Patients with PWS who are:
  - Aged 4 years or over
  - Diagnosed with PWS at least 12 months prior to index date
- Patients who are residents of care homes must have routine consultations with their HCP.

Caregiver Inclusion Criteria

- Adult caregivers (18 years of age and over) who care for patients with PWS who are:
  - Aged 4 years or over
  - Diagnosed with PWS at least 12 months prior to index date
- Able to provide informed consent as next of kin/legal representative
- Able to understand the Informed Consent Form and questionnaire in the local language of the country in question

REFERENCES

1. Butler, MG, Miller, JL, Forster, JL. Prader-Willi syndrome – Clinical genetics, diagnosis and phases in Prader-Willi syndrome. Am. J. Med.. Curr Pediatr Rev 2019; 15(4):207-244. 2. Miller, JL, Lynn, CH, Driscoll, DC, et al.. Nutritional phases in Prader-Willi syndrome. Am J Med Genet Part A 2011; 155:1040–1049.

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