The epidemiology of Hereditary Spastic Paraplegia and associated common mental health outcomes in England and Northern Ireland

Introduction

Hereditary Spastic Paraplegia (HSP)

- A Rare neurogenetic condition causing progressive lower-limb spasticity and weakness
- Global prevalence: 3.6 per 100,000
- Previous studies suggest links with depression and anxiety

There is no study reporting the epidemiology of HSP and associated mental health outcomes in the UK.

Aim

To use routine primary care data to:

1. describe the prevalence and incidence of HSP (2000-2021)
2. study HSP's association with common mental health outcomes (depression and anxiety)

Methodology

Study Design: Retrospective population-based open cohort

Data Source: Clinical Practice Research Database (CPRD), a primary care database covering England & Northern Ireland

Study Period: 1st January 2000 to 31st December 2021

CPRD Aurum population (2000–2021)
31.3 million registered patients

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Identify HSP patients
Using SNOMED CT diagnostic codes

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Create matched cohort
Match each HSP patient 1:4 by: Age, sex and GP

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Extract outcomes
Diagnoses of depression and anxiety

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Statistical analyses
logistic regression (baseline) and Cox regression (incident cases)

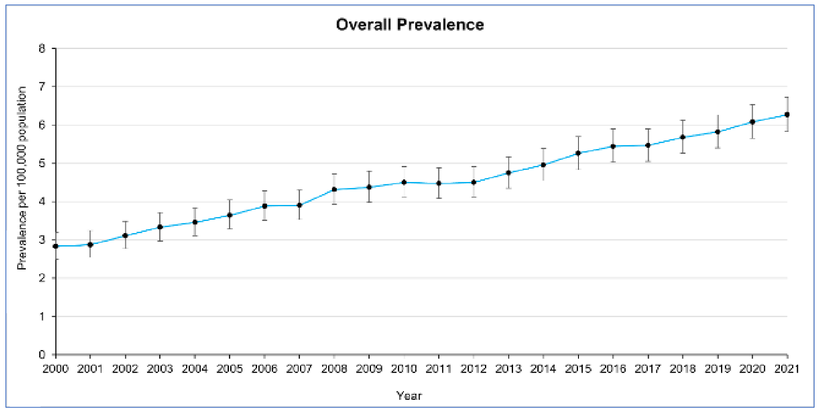
*adjusted for age at index date, sex, ethnicity, body mass index, smoking, and index of multiple deprivation.; missing data treated as a separate category.

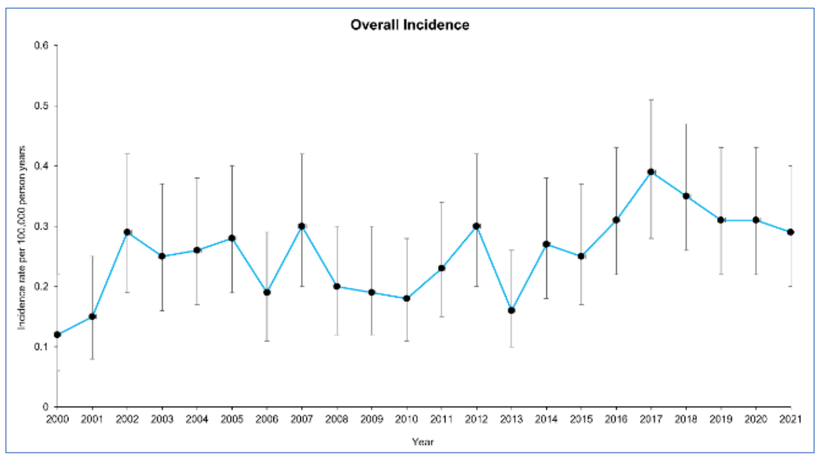
Results

Prevalence and Incidence

- HSP prevalence increased from **2.83 → 6.27 per 100,000** (2000–2021)
- Incidence remained stable at **0.29 per 100,000 person-years** in 2021

Graphs 1, 2: Annual HSP prevalence (per 100,000) and incidence rate (per 100,000 person years)





Mental Health at Baseline

- Higher odds of **depression** in people with HSP
  aOR **1.74** (95% CI 1.47–2.06)
- Higher odds of **anxiety**
  aOR **1.31** (95% CI 1.08–1.60)

Incident Mental Health Outcomes

- Increased risk of new **depression** diagnoses
  aHR **1.57** (95% CI 1.26–1.96)
- Increased risk of new **anxiety** diagnoses
  aHR **1.41** (95% CI 1.12–1.76)

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Discussion

Impact

- First UK-wide epidemiological description of Hereditary Spastic Paraplegia using routine primary care data
- Rising prevalence highlights the need to plan for increasing service demand
- Higher risks of depression and anxiety underline the importance of incorporating mental health support into HSP care pathways
- Demonstrates the value of routine primary care records for researching rare neurological conditions
- Findings can support commissioning, service planning, and patient advocacy organisations

Lessons learned & future directions

- Routine primary care data offers strong longitudinal follow-up but lacks HSP genotype information
- Diagnostic delay in rare diseases means the "first recorded HSP code" may not reflect true symptom onset
- Establishing clear temporality between HSP onset and mental health outcomes is challenging
- Future work should link primary care, hospital, and mortality datasets to better quantify the disease burden and improve service planning
- Early engagement with patient organisations strengthened the study design and interpretation

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