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Epidemiology of progressive cerebellar ataxia in South Yorkshire, UK

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ABSTRACT

Background Cerebellar ataxias are a heterogeneous group of rare neurological disorders characterised by progressive gait instability, limb incoordination, speech and oculomotor impairment. UK prevalence estimates remain rather limited, with the most widely cited population-based study performed in South East Wales over two decades ago. The objective is to estimate the current prevalence of progressive cerebellar ataxia within South Yorkshire using a comprehensive NHS (National Health Service) tertiary ataxia service database and to compare findings with previously published UK data.

Methods A retrospective epidemiological analysis was undertaken using the Sheffield Ataxia Centre NHS clinical database. All alive patients aged over 18 years living in South Yorkshire with confirmed progressive cerebellar ataxia under active or historical follow-up were included, excluding static and structural ataxias where appropriate. Population sizes within South Yorkshire were derived from the 2024 Office for National Statistics (ONS) mid-year population estimates for everyone aged over 18 years and over 65 years.

Results A total of 1204 individuals with progressive cerebellar ataxia were identified as resident in South Yorkshire, corresponding to a frequency prevalence of approximately 106 per 100 000 population (1:942). Prevalence varied by local authority, ranging from 1:647 in Sheffield to 1:1677 in Doncaster.

Conclusions The prevalence of progressive cerebellar ataxia in South Yorkshire is substantially higher than previously reported UK figures, likely reflecting improvements in diagnostic capability, service centralisation and recognition of immune-mediated and new novel genetic ataxias. These figures have important implications for service planning and resource allocation within the NHS.

INTRODUCTION

Cerebellar ataxias are a diverse group of neurodegenerative, genetic and immune-mediated disorders characterised by progressive cerebellar dysfunction. Although individually rare, taken together they represent a major cause of neurological disability, often requiring lifelong specialist input and multidisciplinary care.

Robust population-based prevalence data for cerebellar ataxia are lacking. Available evidence derives mainly from condition-specific and regional studies, particularly of hereditary ataxias. A global systematic review of population-based studies estimated pooled prevalences of approximately 2.7 per 100 000 for autosomal dominant and 3.3 per 100 000 for autosomal recessive cerebellar ataxias, with significant variations via locations due to levels of specialty and how cases were identified and diagnosed.[1] Data on sporadic and late-onset ataxias are more limited. Cohort-based studies suggest prevalences in the range of 2–12 per 100 000, although these estimates are not population-derived and likely underestimate true disease burden.[2 3] Progressive ataxias are not consistently captured as a discrete category within global epidemiological frameworks such as WHO classifications or Global Burden of Disease estimates, reflecting ongoing under-recognition, clinical variation and inconsistent coding internationally.[4 5] While the International Classification of Disease (ICD) mentions ataxia and categorises early and late onset genetic ataxias, it does not comprise a full list which leads to a lack of coding accuracy.

UK epidemiological data remain limited. The most frequently cited population-based estimate derives from a 2004 study of late-onset cerebellar ataxia in South East Wales, which reported a combined prevalence of approximately 10 per 100 000 population.[6]

Since then, the recognition and diagnosis of ataxias have evolved substantially. Advances in neuroimaging, discovery of new genes following widespread availability of genetic testing (including whole genome sequencing) and improved recognition of immune-mediated ataxias have increased the specialist interest in the ataxias and the need for regular review in a clinic setup.

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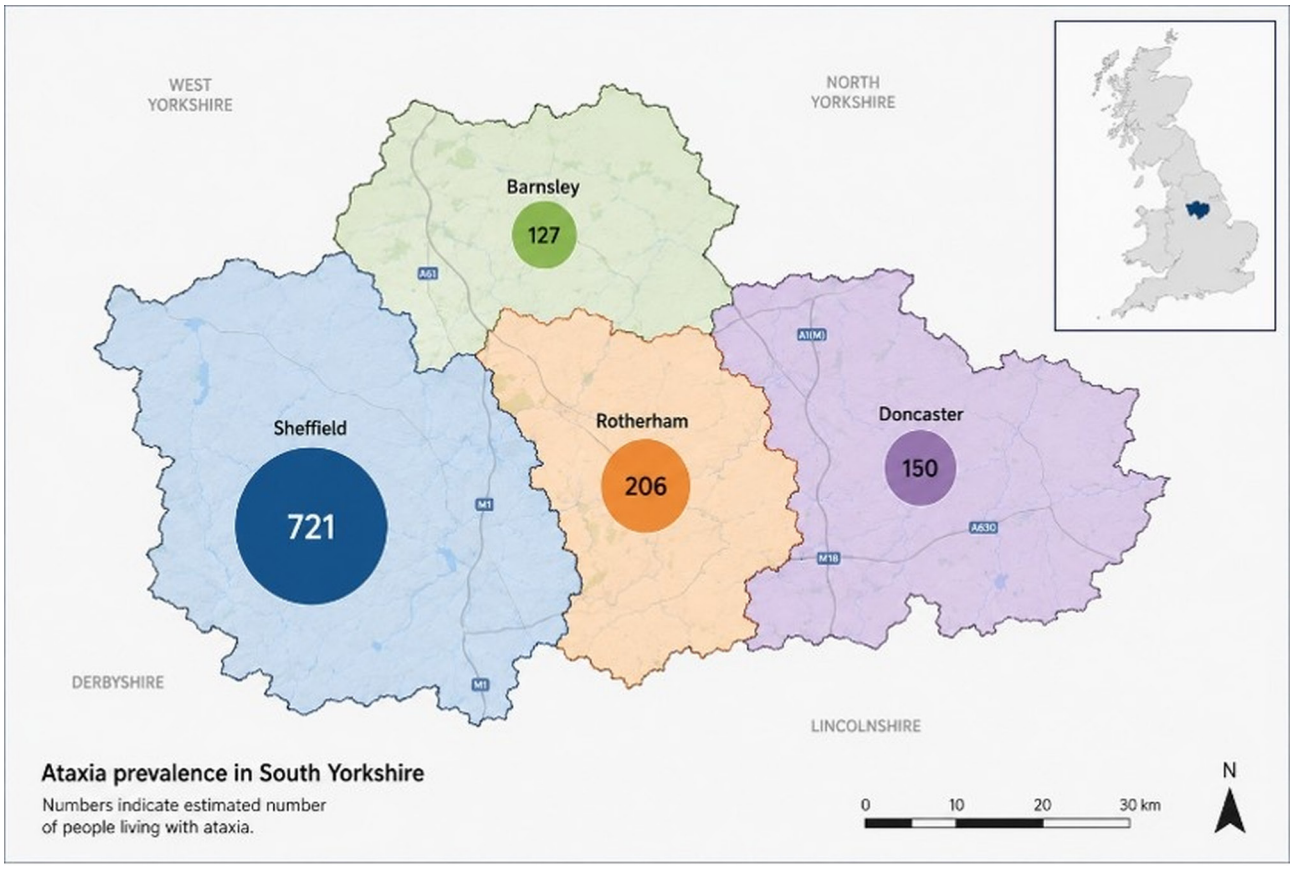


Figure 1 Ataxia prevalence in South Yorkshire by local authority area. Numbers indicate estimated prevalence. Map boundaries derived from Office for National Statistics Open Geography Portal boundary data; prevalence estimates calculated by the authors.

The Sheffield Ataxia Centre (SAC) is one of three national, tertiary NHS (National Health Service) services, providing diagnostic evaluation and ongoing management for patients with progressive cerebellar ataxia. The existence of specialist services covering a defined geographical region offers a unique opportunity to generate real-world prevalence data within the UK.

South Yorkshire is an urbanised region in the North of England comprising the local authorities of Sheffield, Rotherham, Barnsley and Doncaster. The region contains a heterogeneous population spanning urban and rural settings, with differences in demographic and socioeconomic characteristics that may contribute to variation in healthcare access and case ascertainment. Within South Yorkshire, the neurology services are centred in Sheffield with remote cover to each of the surrounding areas within the region. As a result, there is understanding that the vast majority of patients with a diagnosis of progressive cerebellar ataxia in South Yorkshire will be seen within the SAC.

METHODS

Study design and case ascertainment

A retrospective cross-sectional analysis was undertaken using the Sheffield Ataxia Centre NHS clinical database, which prospectively records all patients referred with suspected or confirmed ataxia as part of routine clinical care.

The database comprised 3265 individuals with progressive ataxia seen, evaluated and followed up at the SAC. All living patients with a progressive cerebellar ataxia resident within South Yorkshire constituted the final epidemiology-relevant cohort.

This included diagnoses of genetic ataxias both dominant and recessive, early and late-onset ataxias, acquired ataxias, immune-mediated cerebellar ataxias, neurodegenerative ataxias such as Cerebellar variant of multiple system atrophy (MSA-C) and those with recognised ataxia but with no cause identified as yet—idiopathic sporadic ataxias.

Population sizes

Population estimates for 2024 were obtained from the Office for National Statistics mid-year figures. The total adult population of South Yorkshire was 1 134 753, broken down to Sheffield (466 214), Rotherham (217 074), Doncaster (251 494) and Barnsley (199 971).[7]

Statistical analysis

Unadjusted proportion rates were calculated as the number of cases divided by the relevant population,

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expressed per 100 000 population and as ratios to gain the prevalence figures.

RESULTS

Overall prevalence

A total of 1204 adult individuals with progressive cerebellar ataxia were identified as residents of South Yorkshire. This corresponds to a figure of 106 per 100 000 population, equivalent to approximately 1 in 942 individuals (see figure 1).

Prevalence by local authority

Substantial geographical variation was observed across South Yorkshire:

- **Sheffield:** 721 cases; 154.7 per 100 000 (approximately 1 in 647).
- **Rotherham:** 206 cases; 94.9 per 100 000 (approximately 1 in 1054).
- **Barnsley:** 127 cases; 63.5 per 100 000 (approximately 1 in 1575).
- **Doncaster:** 150 cases; 59.6 per 100 000 (approximately 1 in 1677).

DISCUSSION

This study provides the most comprehensive recent UK prevalence for progressive cerebellar ataxia within a defined population. Using observed prevalence data from South Yorkshire (106 per 100 000 population) and applying this to the UK adult population, we estimate that 58 000 individuals may be living with progressive cerebellar ataxia nationally.

Even allowing for potential overestimation, these findings indicate that the burden of disease is likely to affect tens of thousands of people across the UK, substantially exceeding historical estimates derived from the early 2000s data. This prior UK-based population study reported crude prevalence figures of approximately 10–15 per 10 000 population for adult-onset cerebellar ataxia, figures that almost certainly reflect limited interest in the ataxias at the time, lack of regular follow-up because the ataxias were considered untreatable, limited genetic testing availability and under-recognition of immune-mediated ataxias.

The diagnosis of progressive ataxia has evolved significantly since the early 2000s, perhaps reflecting the new figures of high prevalence. The figure we found represents a pragmatic estimation rather than a definitive national prevalence. South Yorkshire has a large tertiary neuroscience centre with an accredited specialist ataxia centre, which is likely to strengthen diagnostic certainty within a well-established service. On the other hand, lower prevalence in other local authorities may be explained by the fact that some patients with ataxia are managed by the visiting neurologist and that patients may not wish to travel to Sheffield for review at the SAC. We also did not find any differences in the distribution of the different types of ataxias between South Yorkshire and the complete national cohort we look after.

Despite this, underdiagnosis and delayed referral remain well recognised challenges in rare neurological disorders, suggesting that prevalence in other regions may be underestimated rather than truly lower. These limitations, however, are more likely to lead to an underestimate rather than overestimate of the prevalence we highlight here.

The findings have important implications for service planning, particularly given the progressive nature of these disorders and the complexity of care required. Progressive cerebellar ataxias are associated with complex, lifelong disability, requiring coordinated neurological, rehabilitation, allied health and social care input. Underestimation of prevalence risks systematic underprovision of specialist services and limits accurate workforce and resource planning within neurology and clinical genetics. The improvement in diagnostic criteria for the immune-mediated ataxias means that patients could be missing out on the opportunities for accurate diagnosis and treatment options to limit progression of disease and symptom management.

Future work using national datasets such as NHS digital coding, genomic medicine service data and linked specialist centre registries will be essential to validate and refine national prevalence estimates. The present data provide a strong rationale for such coordinated national epidemiological work.

LIMITATIONS

These new estimates should however be interpreted cautiously. Limitations include reliance on crude prevalence estimates, potential referral bias and the absence of age-standardised rates. The inferred national figure represents a pragmatic estimation by extrapolating regional figures.

The presence of a specialist ataxia centre in Sheffield may influence the geographical distribution of patients due to service awareness within the serving NHS trust, resulting in higher referral from local (Sheffield) services. In our 30 years' experience of running a dedicated ataxia centre, we have not observed a tendency for patients migrating to Sheffield just to be closer to the specialist centre, accepting, however, that we have not specifically tracked patient movements.

CONCLUSIONS

Progressive cerebellar ataxia in South Yorkshire appears considerably more prevalent than previously recognised in UK studies. Conservative national estimate suggests that many tens of thousands of individuals may be affected across the UK. These findings underline the need for updated national epidemiological studies and fair access to specialist ataxia services within the NHS.

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Contributors EF and MH conceptualised the study. All patients were assessed by MH, PDS, EF, SD and EH working within the SAC at Sheffield Teaching Hospitals NHS Foundation Trust. EF produced the first draft. All authors read, provided input and approved the final manuscript.

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