Diagnosis of Fabry disease in the UK

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Introduction

Fabry disease is a rare, life-limiting disease characterised by a deficiency of α-galactosidase A enzyme. There is a wide spectrum of disease that ranges from a severe form, with symptoms appearing in childhood, to milder forms presenting later in adulthood. Diagnosis is often delayed, and many individuals are only diagnosed following diagnosis in a family member. Presenting symptoms can be non-specific and not recognised as the consequence of Fabry disease.

Aim

The aim of our study was to determine the age at Fabry diagnosis and the other diagnoses that patients receive before Fabry is confirmed.

Methods

Age at diagnosis and date of birth was retrieved from the UK MPS Society's membership database in October 2021. Information on patient's prior diagnoses was collected at the MPS Society's Focus on Fabry Disease Expert and Patient Conference held in September 2019; and was collected via an anonymous questionnaire consisting of one multiple choice question, with space to record any other diagnoses not listed.

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Results

Prior diagnoses

Nine adult Fabry patients completed the questionnaire.

- Patients reported a mean of 2.9 different diagnoses before Fabry (range 0–9)
- One patient was diagnosed before symptoms appeared, due to a family history of Fabry, and had no prior diagnoses
- 18 different diagnoses were reported (Table 1)

The most common prior diagnoses were irritable bowel syndrome, depression, and fatigue (all 33%), followed by inflammatory bowel disease, vertigo, and protein in the urine (all 22%).

Table 1. Prior diagnoses received by Fabry patients

Arthritis	Cardiomyopathy	Depression
Eye problems (cornea)	Facial sweating	Fainting episodes
Fatigue	Growing pains	Hearing loss
Inflammatory bowel disease	Irritable bowel syndrome	Protein in urine
Rheumatoid arthritis	Skin lesions	Stroke
Tinnitus	Underactive thyroid	Vertigo

Age at diagnosis of Fabry

Date of birth and diagnosis data for 326 Fabry patients diagnosed between the years 2000 – 2020 were retrieved.

- The mean age at diagnosis was 29.8 ±20.1 years (median 32, range 0–83 years)

There was no difference in age at diagnosis between those diagnosed in 2000 to 2010 (mean 29.3±19.7, n=149) compared to 2011 to 2020 (mean 30.2±20.5, n=177) (Figures 1 & 2).

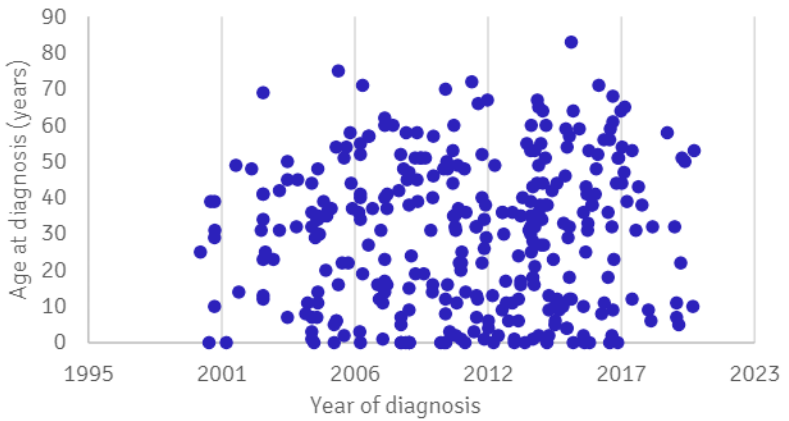

Figure 1. Age at diagnosis of Fabry by year of diagnosis

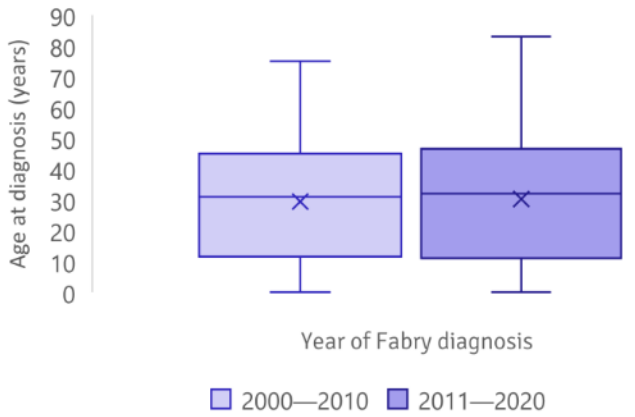

Figure 2. Age at diagnosis of Fabry by year of diagnosis

Their was no difference in the proportion of patients diagnosed under the age of ten (21%) in the years 2000–2010 compared to 2011–2020.

Conclusions

Our study highlights the fact that there has been no change in the age at diagnosis for Fabry in the UK over the last two decades, and that early symptoms of Fabry are frequently misdiagnosed as other conditions, particularly gastrointestinal issues.