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Awareness of Fabry disease among non-Fabry specialists: opportunities for education

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Declaration: This research was conducted in accordance with the British Healthcare Business Intelligence Association's Legal & Ethical Guidelines for Market Research. The survey was non-interventional, and all participants signed a consent form.

Background

- Fabry disease has a prevalence of 1 in 37,000 to 117,000 births for classic Fabry disease, with the prevalence of atypical Fabry disease being 1 in 1,400 to 1 in 3,900 in some regions.[1] It is caused by mutations in *α-galactosidase A* gene resulting in deficient enzyme activity and accumulation of globotriaosylceramide within lysosomes disrupting normal cell function[1,2]
- The disease is highly heterogenous and presents a broad spectrum of symptoms which vary significantly across patients[3]
- Despite its severity, the variability in symptoms, combined with the rarity of the condition, frequently leads to diagnostic delays and misdiagnoses[3]
- Awareness and education regarding Fabry disease remain limited among healthcare professionals (HCPs), with non-specialists in Fabry disease identified as needing additional support[4]

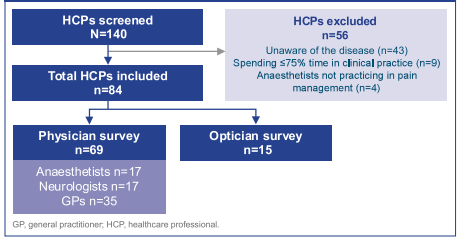
Objective

- To evaluate the level of awareness, understanding and ability of non-Fabry specialists in the UK to recognise Fabry disease and to explore their unmet medical education needs.

Methods

- This study presents the results of an online survey (5–16 September 2024) by non-Fabry specialists in the UK. 140 HCPs were recruited and screened through a market research panel **(Figure 1)**
- HCPs were included in the study if they were anaesthetists, general practitioners (GPs), neurologists, or opticians who have heard of Fabry disease; spend ≥75% of professional time in clinical practice; and for anaesthetists, must work in chronic pain medicine/pain management clinics
- Two surveys were used for this research, one for physicians (anaesthetists, GPs, and neurologists) another for opticians. Respondents provided consent

Figure 1. Study participants



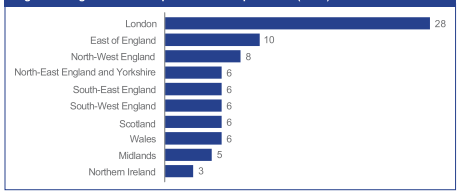
GP, general practitioner; HCP, healthcare professional.

Results

Respondent characteristics

- This study included 84 non-Fabry specialists; 69 physicians and 15 opticians **(Figure 1)**, from the UK **(Figure 2)**
- Of the 84 HCPs who had heard of Fabry disease, 89% (75/84) had never seen/encountered a Fabry patient in their practice. Eight physicians and 1 optician had experience with a Fabry patient, the majority (56%, 5/9) of whom had seen the patient within the last year

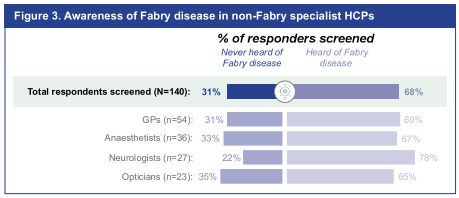
Figure 2. Region of current practice for respondents (n=84)



Awareness and knowledge of Fabry disease

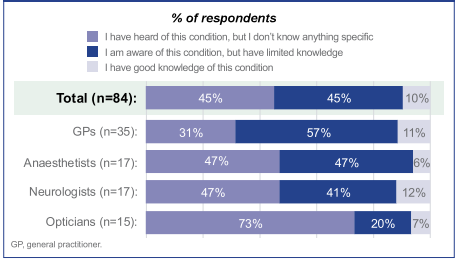
- Non-Fabry specialists have limited awareness and knowledge of Fabry disease
- Of the total HCPs screened, 31% were unaware of Fabry disease. Of these, opticians were most likely to be unaware of the disease **(Figure 3)**

Figure 3. Awareness of Fabry disease in non-Fabry specialist HCPs



- Of the 84 HCPs who were aware of Fabry disease, the majority (90%) reported having limited or no specific knowledge of the disease **(Figure 4)**

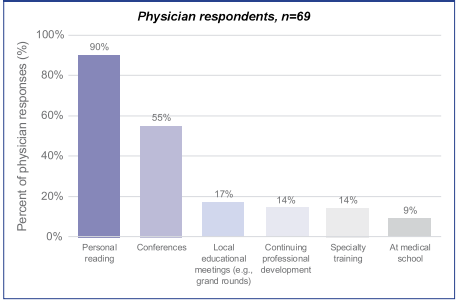
Figure 4. Knowledge of Fabry disease in non-Fabry specialist HCPs



GP, general practitioner.

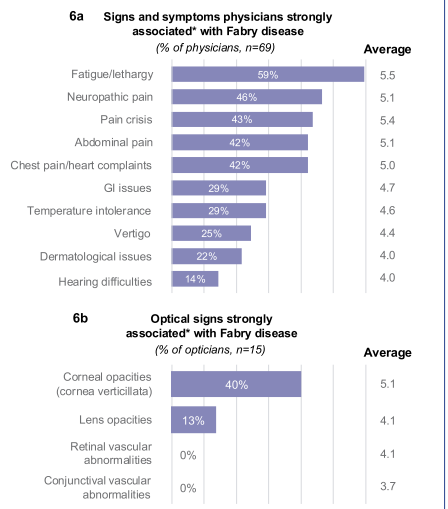
- Formal training on Fabry disease is minimal, with only 9% and 14% of physicians recalling having received education from medical school or specialty training respectively. The majority of physicians (90%) depend on personal reading for information **(Figure 5)**

Figure 5. Sources of education on Fabry disease for non-Fabry specialist physicians



- Physicians associate Fabry disease most commonly with fatigue/lethargy, while dermatological issues and hearing difficulties were the least recognised symptoms **(Figure 6a)**. For opticians, corneal opacities was considered the most common optical sign of Fabry disease **(Figure 6b)**

Figure 6. Signs and symptoms of Fabry disease (prompted) a) physicians b) opticians



*Scoring 6–7 on a 7-point rating scale (1=weak association and 7=strong association). HCPs were asked how strongly, if at all, they associated each of the symptoms with Fabry disease.
GI, gastrointestinal.

- Unprompted, HCPs cite pain (71%), neurological (33%), and cardiovascular (29%) signs and symptoms as associated with Fabry disease

Confidence in identifying, diagnosing diagnosing and managing Fabry disease

- Respondents reported low confidence (scoring 1–3 on a 7-point confidence scale) in identifying signs and symptoms and diagnosing Fabry disease **(Table 1)**
- Specifically, physicians reported low confidence (scoring 1–3 on a 7-point confidence scale) in answering patient questions on prognosis (49%), management (43%) and genetics/ inheritance and impact on family planning (62%)

Table 1. Areas of low confidence in the identification and diagnosis of Fabry disease

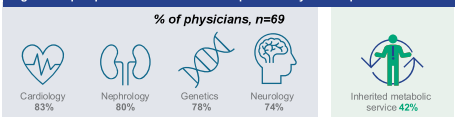
	HCPs (n=84) who scored 1 – 3 on a 7-point confidence scale* (%)	Mean score (7-point scale*)
Deciding which diagnostic tests are most appropriate	48	3.8
Identifying signs of the condition	56	3.7
Identifying symptoms of the condition	51	3.7
Answering patients' questions on the condition	47	3.7
Informing patients about their condition at diagnosis	52	3.6
Making the formal diagnosis	63	3.3

HCP, healthcare professional.*7-point confidence scale: 1=not at all confident to 7 = extremely confident

Referrals and management of Fabry disease

- Only 30% of HCPs surveyed felt confident in knowing which speciality to involve and/or refer to when a Fabry diagnosis is suspected
- Physicians were most likely to refer suspected Fabry cases to cardiology (83%) followed by nephrology (80%) **(Figure 7)**
- Inherited metabolic service is the seventh most likely specialty where suspected Fabry patients are referred to, with only 42% of non-specialist physicians referring patients here **(Figure 7)**
- 83% of physicians consult with/refer to other HCPs when encountering suspected Fabry disease

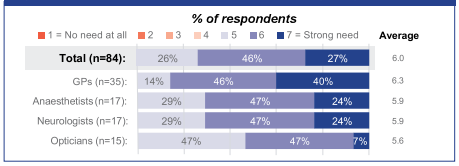
Figure 7. Top 4 specialities referred to for suspected Fabry disease patientsdisease



Non-specialists seek improved Fabry knowledge

- There is a very high interest among non-Fabry specialists to learn more about the disease (79%) particularly from GPs
- The HCPs identified a clear need (73%) for greater support/ education to help them identify Fabry disease, with the highest demand among GPs (86%) and lowest among opticians (54%) **(Figure 8)**

Figure 8. HCPs need for support/education to help identify Fabry patients



- Physicians see a strong need to increase and/or improve the level of education on Fabry during specialty training, as well as via professional development and in medical school
- The priority education needs suggested by physicians and opticians to support identification of patients with Fabry is patient characteristics/signs/symptoms specifically optical signs for the opticians **(Table 2)**

Table 2. Spontaneous reporting of education needs by non-Fabry specialists

HCP education needs	Physicians (%)	Opticians (%)
Patient characteristics/signs/symptoms	67	40
Treatment paradigm/standard of care	43	–
Diagnostic tools and protocols	35	40
Guidance on patient management	30	30
Genetics guidance	22	–
Disease awareness for physicians	22	–
Differential diagnosis/misdiagnosis	14	–
Guidance on referral/specialities to involve	10	–
Public disease awareness	7	–

HCP, healthcare professional.

Disseminating Fabry education

- Most physicians relied on key opinion leaders (93% of physicians), medical journals (90%), conferences (87%) and educational courses (86%) as their primary source of information on Fabry disease
- The top two preferred channels for new information on Fabry disease were medical journals and educational courses for 65% and 64% of physicians respectively

Conclusions

- There is a need for improved education and awareness of Fabry disease among non-Fabry specialists, specifically around identifiable signs and symptoms. The current lack of understanding may be a key factor contributing to a slower diagnosis
- Physicians lack confidence in the diagnosis and management of Fabry disease, and the insufficient referral by UK physicians to the inherited metabolic service highlights the necessity for further education to ensure timely management of patients
- Enhanced education and training targeted to non-Fabry specialists could improve disease recognition, timely referrals and lead to a shorter diagnosis journey, improving patient outcomes

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