

CHARACTERISATION OF LARYNGOLOGICAL PRESENTATIONS AND PATIENT REPORTED OUTCOME MEASURES IN PATIENTS WITH EHLERS-DANLOS SYNDROME

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BACKGROUND

Ehlers-Danlos syndrome (EDS) describes a group of inheritable connective tissue disorders involving defects in collagen and connective tissue synthesis. EDS can manifest in a range of ways with laryngological manifestations being common. Patient Reported Outcome Measures (PROMs) are often used to characterize the severity of symptoms. These include the Eating Assessment Tool (EAT-10), Reflux Symptom Index (RSI) and Voice Handicap Index (VHI). However, these tools have not been validated for patients with EDS (1)(2)(3). This study aims to characterize the laryngological presentations of EDS and provide a preliminary exploration of PROMs in this population.

METHODS

Retrospective data was collected from patients with a diagnosis of EDS who were seen in clinic by the senior author between 2005-2019. Patients without record of laryngological presenting symptoms were excluded. Details of patient demographics, date of first presentation to clinic, diagnosis, presenting symptoms and PROMs were collected. The PROMs used were the EAT-10, RSI and VHI.

RESULTS

In total 21 patients met the inclusion criteria: 3 males and 18 females, with a median age of 47 (range: 16-67) at first presentation to clinic. 9 patients first presented to clinic in 2019 representing a substantial increase on previous years. 20 patients had a diagnosis of EDS hypermobility, 1 patient had a diagnosis of EDS cutaneous. The commonest first presenting symptom were globus (7 patients), choking (5 patients) and hyoid subluxation (2 patients) (figure 1).

All symptoms reported by patients were grouped (table 1) and showed that symptoms related to swallow were commonest (15 patients) followed by voice (9 patients), hyo-laryngeal skeletal complex problems (6 patients), respiratory (5 patients), nasal (3 patients), vestibulocochlear (2 patients) and 5 patients reporting other symptoms. PROMs data were available for 8 patients with results showing a large variation – RSI (median 25.5, range: 0-33), EAT-10 (median: 21.5, range: 0-35), VHI (median: 21.5, range: 0-104).

CONCLUSION

The increase in the number of EDS patients seen in laryngology clinic in 2019 may suggest a wider trend of increasing numbers of EDS patients presenting to clinic and clinicians should be aware of this possibility. Swallowing symptoms were the most common presenting complaint in our case series. EDS patients commonly experience musculoskeletal issues which in the throat manifests as hyo-laryngeal skeletal complex problems as seen by the patients reporting such symptoms in our study. The large range in our PROMs data could suggest that EDS patients present with varying degrees of symptom severity. Future

studies with larger patient numbers are required to validate the PROMs used in this study for EDS patients.

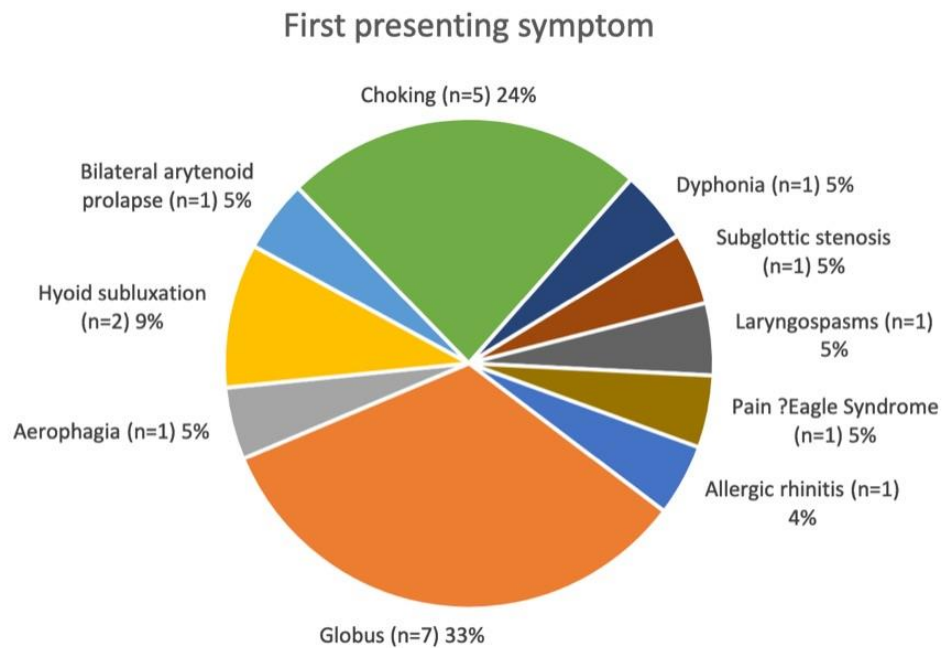


Figure 1 – Graph showing the first presenting symptom reported by patients presenting to the clinic.

All symptoms	Number of patients reporting symptoms in this group
Nasal – allergic rhinitis, sinusitis, nasal regurgitation	3
Vestibulocochlear – tinnitus, vertigo, clicking ears	2
Respiratory – obstructive sleep apnoea syndrome, coughing, dyspnoea	5
Swallow – dysphagia, globus, choking, reflux, odynophagia, aerophagia, dysmotility, hypercontractile oesophagus and cricopharyngeus	15
Voice – dysphonia, mutism	9
Hyo-laryngeal skeletal complex problems – hyoid subluxation, bilateral arytenoid prolapse, subluxation arytenoids, ?subluxation, laryngospasm	6
Other – temporomandibular joint syndrome, subglottic stenosis, pain ?Eagle syndrome, atlantoaxial subluxation spine, neck swelling, acute throat swelling ?anaphylaxis	5

Table 1 – Table showing all laryngological symptoms reported by patients.

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DECLARATIONS OF INTEREST

None