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Patterns of and Experiences with Dysphagia in People with Hypermobile Ehlers Danlos Syndrome (hEDS) With or Without Dysautonomia

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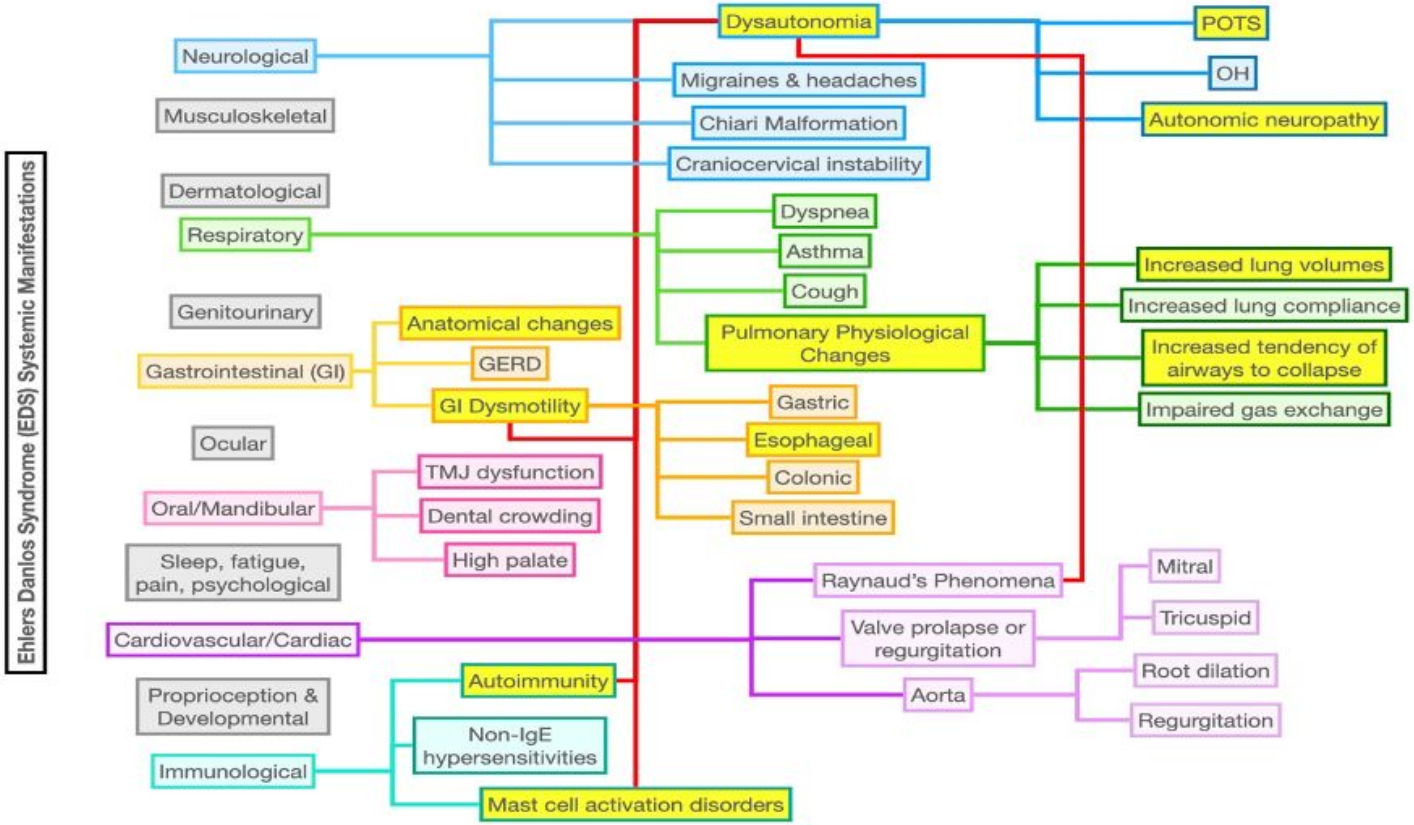
By: Karyssa Stonick, Dr. Deanna Britton, Emily Goble, Alison Wong, Dr. Alena Guggenheim, Dr. Donna Graville

Background

- **What is Ehlers Danlos Syndrome (EDS)?**
  - Why is the mascot a zebra?
  - General Overview
  - EDS is complex
  - Sub-types
- **Systemic Manifestations**
  - Comorbidities
  - Diagnostic challenge
- **EDS and Swallowing**
  - One prior study
- **EDS and Respiration**
  - Structural
  - Physiological



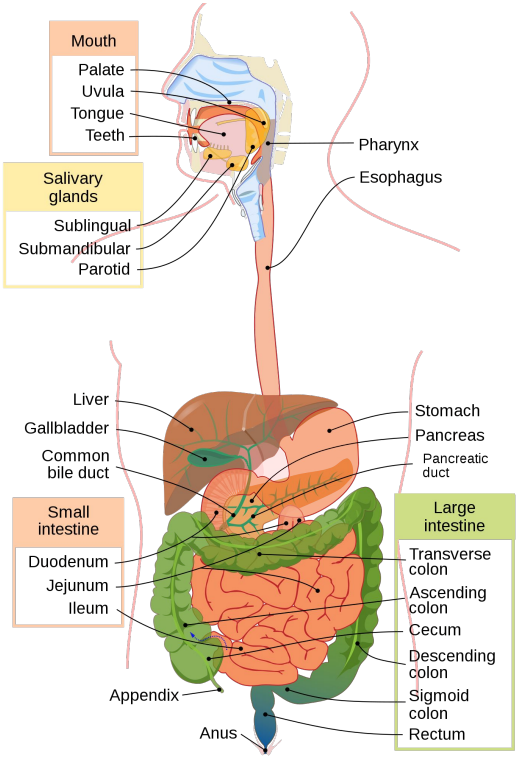
Note: The grey systems in the leftmost column are less relevant to this study, but not overall. The colored systems in the leftmost column are the ones that are potentially the most relevant to this study. The specific manifestations that are highlighted in yellow are hypothesized to be the most relevant to this study (Gazit et al., 2016; Gensemer et al., 2021; Grahame, 2016; Miklovic & Sieg, 2021; Song et al., 2020).

Prevalence of dysautonomia, dysphagia, & associated diagnoses in EDS at OHSU via *Cohort Discovery*

Primary Diagnosis	Primary Diagnosis Code	Number of patients	Secondary Diagnosis	Secondary Diagnosis Code	Number of patients	Percentage
EDS	**Q79.6**	**1070**	**Dysphagia**	**R13.1**	**159±3**	**14.9% ± 0.3%**
EDS	Q79.6	1070	Gastro-esophageal reflux disease (GERD)	K21	323±3	30.2% ± 0.3%
EDS	**Q79.6**	**1070**	**Dysautonomia**	**G90; G99.0; I95.1**	**452±3**	**42.2% ± 0.3%**
EDS & Dysautonomia	**Q79.6 G90; G99.0**	**452±3**	**Dysphagia**	**R13.1**	**83±3**	**18.4% ± 1.4%**
Dysautonomia	G90	7065	Dysphagia	R13.1	1224±3	17.3% ± 0.04%

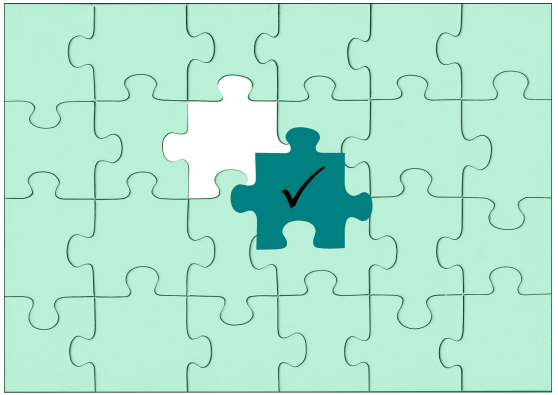
Objectives & Goals

- **Purpose**
  - Characterize experiences of swallowing
  - Population: hEDS patients with or without dysautonomia
- **Aim**
  - Identify patterns
  - Qualitative phenomenological approach
  - Derive themes
- **Long-term hypothesis**
  - Multifaceted impact on swallowing from EDS
  - Structural and physiological



Innovation

- **First** study of its kind
- **Fills gaps** identified in the literature
- **Improve assessment** and **access** to care for EDS patients
- **Add** to the **understanding** of EDS and comorbid dysautonomia impact on **swallowing**



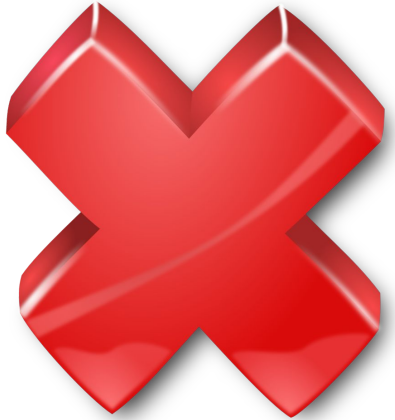
Inclusion Criteria

- **Diagnosis of hEDS** or probable hEDS by a physician based on the 2017 diagnostic criteria (Malfait et al., 2017)
- **English** as a primary language
- **Hearing** within functional limits
- **Access to a computer and internet** due to the virtual nature of the study
    - If computer and internet access is an issue, the researchers can help to problem-solve.

Exclusion Criteria

- **Vulnerable** populations (pregnant, incarcerated)
- **Moderate-severe cognitive impairment** self-reported by the patient
- **Age** <18 years or >70 years
- **Diagnosis** of a *subtype of EDS that is not hEDS*
- **Other diagnoses** *not associated with/related to EDS*

Because there is a wide range of conditions that are known to co-occur with EDS, we will not be excluding outright based on other diagnoses.



Methods

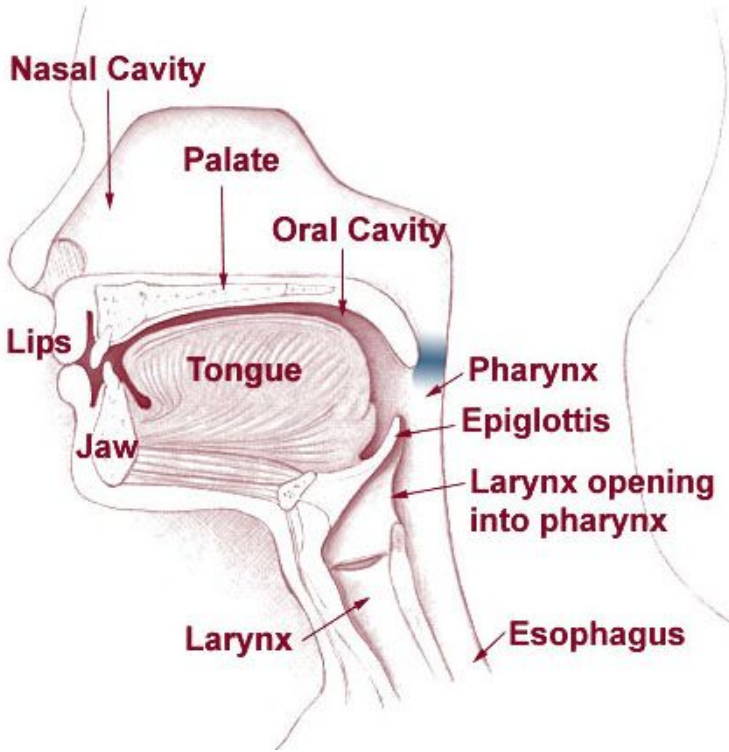
- **Study Design: Qualitative**
  - Questionnaires
    - *Eating Assessment Tool-10 (EAT-10)*
    - *Reflux Symptom Index (RSI)*
    - *Composite Autonomic Symptom Score (COMPASS-31)*
    - *Demographic*
  - Semi-structured interview (60-120 minutes)
  - Medical records review
- **Targeting 6-10 participants**
- **Data analysis with Atlas.ti 84 Web**
- **Comment and code transcripts**
- **Trustworthiness**
  - Validate analysis, member checking
- **Derive themes**

Current Status of the Project

- **9 participants** found eligible
- **Interviews** completed for **6 participants**
- **Participant Characteristics**
  - ***Age range***: 19-52
  - ***Genders***: 6 women, 2 they/them, 1 man
  - ***Diagnosis of Dysautonomia***: 7 of the 7 participants who have completed questionnaires
- **Currently —**
  - Recruiting 1 more participant
  - Transcribing interviews
  - Analyzing data with Atlas.ti 84 Web

Preliminary Trends

- Reports of experience most consistent with **oropharyngeal and esophageal dysphagia**
- Dysphagia **impacting quality of life**
- Dysphagia is more likely to be **intermittent/ waxing and waning often in association with EDS exacerbations**
- Occurring **in conjunction with other GI** signs, symptoms, and conditions
- **Medical gaslighting**
- ***Analysis is still ongoing***



Questions?

Thank you for your time!

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