Poster #IR026

No single record tells the whole story: the fragmented diagnosis and treatment journey for patients with Ehlers-Danlos syndrome

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Background/Synopsis

- Ehlers-Danlos syndrome (EDS) is a rare group of heritable connective-tissue disorders. The disorder involves co-occurrence of dysautonomia/postural orthostatic tachycardia syndrome (POTS), mast cell activation syndrome (MCAS), gastrointestinal dysmotility, and chronic pain.[2,3]
- Patients seek medical care across cardiology, allergy/immunology, gastroenterology, neurology, rheumatology, and physical therapy. This specialized care is often dispersed across unaffiliated health systems.[1]
- Single-system real-world evidence (RWE) studies capture only a partial patient journey.
- The Novellia platform links patient-authorized EHR records across health systems to quantify care fragmentation and enable complete characterization of EDS care patterns

Research Objectives

- Examine how fragmentation across health systems limits real-world evidence research on Ehlers-Danlos syndrome (EDS)
- Examine whether linking multiple EHR records restores a complete view of diagnosis and treatment.

Novellia Data Platform

STEP 1

Join

Patients authorize Novellia and consent to allowing their de-identified data to be used for research

STEP 2

Collect

Novellia's proprietary approach instantly aggregates 20+ years of EMR data

athena health · MyChart · Cerner · Quest Diagnostics



- De-identify, harmonize, and standardize data for analysis
- Create research ready datasets

Novellia for Research

Patients can manage their medical care in one place:

- Organize medical records
- Track symptoms
- Log medication use

Novellia for Patients

Methods

- EDS patients were identified using ICD-10 Q79.6x, ICD-9 756.83, SNOMED codes, or diagnosis descriptions containing "Ehlers-Danlos"
- Fragmentation was quantified per patient across distinct health systems, care sites, providers, encounters, ICD-category breadth, record span, and diagnostic odyssey (first record to first EDS-specific code). A single-system simulation estimated the share of each multi-system patient's total linked record held by their most complete system alone, quantifying what any one institution would miss
- Group comparisons used Mann-Whitney U tests with rank-biserial correlation and bootstrap 95% CIs; multivariable OLS models adjusted for age, sex, record span, encounter volume, and diagnostic breadth. RA served as the primary matched-acuity comparator
- Care structure was characterized by a concurrency index (fraction of active span with ≥2 systems overlapping) and k-means clustering on 10 care-structure features to identify patient archetypes

Patient & Clinical Characteristics

Characteristic	EDS (n=494)	Rheumatoid Arthritis (n=2,965)
Demographics		
Age, years, median [IQR]	38 [30–46]	54 [44–64]
Sex, n (%)		
Female	440 (89.1%)	1,920 (64.8%)
Male	41 (8.3%)	932 (31.4%)
Other / unknown	13 (2.6%)	113 (3.8%)
Record unification		
Health systems linked (attempted), median [IQR]	3 [2–5]	2 [1–4]
Health systems delivering data, median [IQR]	2 [1–3]	1 [1–2]
≥2 health systems delivering data, n (%)	309 (62.6%)	1,411 (47.6%)
Care utilization		
Total encounters, median [IQR]	206 [94–402]	156 [73–311]
Encounters per active year, median [IQR][b]	21.9 [11.1–37.8]	16.3 [8.6–29.6]

The EDS Diagnostic Odyssey

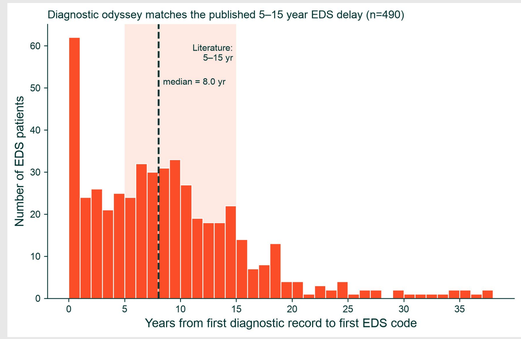


Figure 5. Years from first diagnostic record to first EDS-specific code (n=490). Median diagnostic odyssey in the EDS cohort is 8.0 years, with short measured odysseys concentrated single system patients, indicating incomplete records understand time to diagnosis.

Results

494 participants with Ehlers-Danlos syndrome were included in a fragmentation analysis. The single most complete system held a median of 68% of the linked record; nearly half (48%) of multi-system patients lost >1/3 of their record to any single-system view.

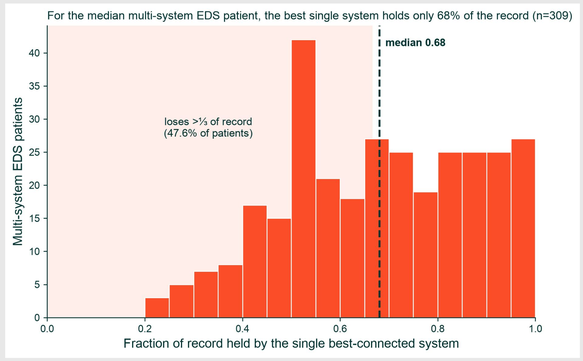


Figure 1. What a single health system misses. Fraction of a multi-system EDS patient's record held by their single best-connected system (n=309); median 0.68, so nearly a third of the record is invisible to any one system.

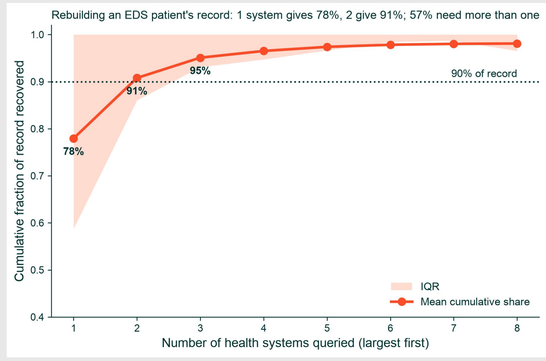
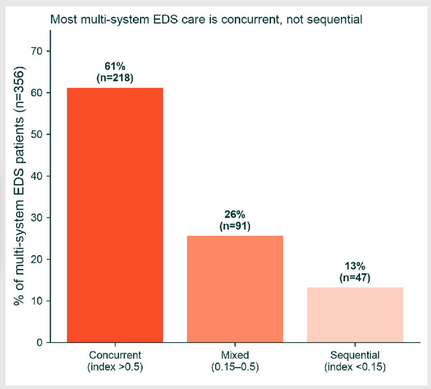


Figure 2. Reconstructing the record across systems. Mean cumulative fraction of a patient's record recovered as systems are added largest-first (shaded IQR). One system recovers 78%, two 91%; 57% need more than one system to reach 90%.

Reconstructing 90% of a patient's record required a median of 2 systems; 83% needed >1 system and 35% needed ≥3, with no single dominant system substituting for linkage. Median diagnostic odyssey was 8.0 years.



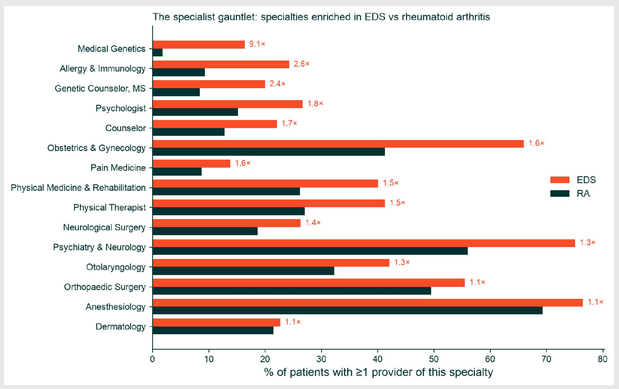


Figure 3. (Left) Multi-system care is predominantly concurrent. Journey-class split among 356 multi-system EDS patients (61% concurrent, 26% mixed, 13% sequential)

Figure 4. (Right) The specialist gauntlet. Specialties ranked by enrichment in EDS vs rheumatoid arthritis. Medical Genetics 9.1x, Allergy & Immunology 2.6x.

Compared to RA, EDS patients spanned more specialties (20 vs 16, p<1e-12) and saw more care sites and encounters; the excess in independent systems and care sites persisted after adjustment (p<0.001). About 60% presented a multisystem comorbidity picture split across >=2 systems. Multi-system care was predominantly concurrent (61% ran systems in parallel; 13% sequential), and the cohort resolved into three care-structure archetypes (concurrent multi-system, non-concurrent high-intensity, non-concurrent low-intensity).

Conclusion

- EDS care is fragmented across unaffiliated health systems, and single-system records under captures the patient journey, omitting ~1/3 of the typical multi-system patient's record and biasing measures such as the diagnostic odyssey
- Compared to another complex disease, RA, EDS patients spanned more specialties, more care sites and encounters
- EDS care is predominantly concurrent across systems, and the fragmentation manifests as redundant, vendor-heterogeneous records rather than cleanly divided specialist roles
- Multi-system linked data make this structure measurable and enable more complete characterization of EDS care patterns than any single-source dataset, supporting study of diagnostic delay, comorbidity burden, and care coordination.

Rare Disease Patients Use Novellia to Manage and Advocate for Their Care

Patient names are pseudonyms to preserve patient anonymity

"You have to be your own advocate the most." Todd has rare condition, Addison's Disease, which required >3 hospital visits before finding a diagnosis. He manages ~10 daily medications across 3 distinct time windows (5am, 6am, 7pm), using Novellia's Medication Adherence.

"Your backend engineers... were the only ones that actually got all my data." Michelle has MCAS, and found that Novellia was the only platform capable of retrieving her full health history, after ChatGPT Health and other services failed.

"What you provide is pretty much like us taking back our control." Sheila previously relied on handwritten notes for clinical visits that were barely acknowledged by doctors.

Acknowledgments

The study thanks the EDS patients that consented to provide their valuable data for research.

Disclosures

All authors are employed by Novellia.

References

1. Jones K, Black C, Cogan J, Callen E. Resource utilization and multidisciplinary care needs for patients with Ehlers–Danlos syndrome. *Mol Genet Genomic Med.* 2022;10(11):e2057. doi:10.1002/mgg3.2057
2. Aziz Q, Harris LA, Goodman BP, Simrén M, Shin A. AGA clinical practice update on GI manifestations and autonomic or immune dysfunction in hypermobile Ehlers-Danlos syndrome: expert review. *Clin Gastroenterol Hepatol.* 2025;23(8):1291-1302. doi:10.1016/j.cgh.2025.02.015
3. Wong S, Hasan S, Parducci C, Riley BA. The gastrointestinal effects amongst Ehlers-Danlos syndrome, mast cell activation syndrome and postural orthostatic tachycardia syndrome. *AIMS Allergy Immunol.* 2022;6(2):19-24. doi:10.3934/Allergy.2022004

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