EPH196

Characteristics, Comorbid GI Conditions, and Treatment Patterns Among Individuals Diagnosed with Rett Syndrome

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INTRODUCTION

- Rett syndrome (RTT) is a rare, neurodevelopmental disorder primarily affecting females[1] often resulting in early loss of acquired skills including hand use, walking, and verbal communication
- Disorders of gastrointestinal (GI) motility such as gastroesophageal reflux disease (GERD) and constipation are common for individuals diagnosed with RTT
- Reports of GI comorbidity prevalence in RTT from caregiver and physician questionnaires are substantially higher compared to estimates based upon analysis of administrative data. EHR data potentially include detail about comorbidities, especially clinical notes, not available in claims data, filling a gap in real-world evidence generation
- Few studies have examined the characteristics, clinical sequalae, medication and healthcare service utilization in response to GI comorbidities associated with RTT

OBJECTIVES

- Our objective was to understand the impact of GI comorbidities associated with RTT using EHR data

METHODS

Study Design

- Retrospective analysis of individuals diagnosed with Rett syndrome using EHR data from Nashville Biosciences[2]

Study Period

- The study data represent years 2017 to 2022 and includes encounter, problem list and pharmacy tables from Vanderbilt University Medical Center (VUMC) EHR

Study Population

- Eligible individuals had ≥1 encounter with an RTT diagnosis and were under the age of 30 at index (date of first RTT diagnosis)
- The presence of comorbidities and service utilization were measured during the 6-month pre- and 12-month post-index periods

Summary of Analytical Methods

- Data regarding GI conditions were abstracted from clinical notes and combined with structured EHR data to form the analytical dataset
- Chi-square and one-way ANOVA were used to test statistical differences for categorical and continuous variables, respectively. The alpha level for statistical significance was set at $p<0.05$

RESULTS

Table 1. Attrition Table for Selection of GI Notes

Step	Description	N	%
1	Patients with 1+ encounters associated with an RTT diagnosis	112	100.0%
2	Patient with at least 6 months of either diagnosis data or prescription records before the first RTT diagnosis	93	83.0%
3	Patients aged less than 30 years at their first RTT diagnosis	79	70.5%

Table 2. Demographic Characteristics among the 79 RTT Patients

	N(Mean)	%(SD)
Gender		
Male	13	16.5%
Female	66	83.5%
Age (years)	(10.2)	(7.4)
Race		
Asian	1	1.3%
Black	7	8.9%
White	51	64.6%
Unknown	20	25.3%
SD: Standard Deviation		

Table 3. Detection of GI comorbidities among the 79 RTT Patients

	N	%
Presence of GI comorbidity	62	78.5%
Presence of GI comorbidity by diagnosis code	34	43.0%
Presence of GI comorbidity found in a progress note (and not in diagnosis code or GI medication)	17	21.5%
Presence of GI comorbidity inferred by GI-related medication (and not in progress note or diagnosis code)	7	8.9%

Figure 1. Graphical Representation of GI Condition Identification within the EHR

- An additional 28 (35.4%) individuals with GI conditions were identified using clinical notes and medications that were not identified by coded diagnoses

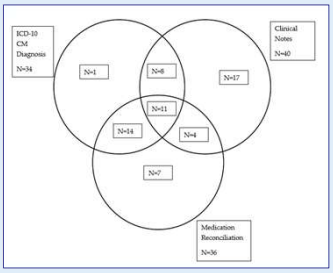

Table 4. Prevalence of GI Diagnoses in the Study Period among the 79 RTT Patients

	N	%
GI Comorbidity		
Dysphagia	31	39.2%
Constipation	29	36.7%
GERD	27	34.2%
Nausea/vomiting	15	19.0%
Diarrhea	4	5.1%
Gastritis	4	5.1%
Total (ICD-10 or Rx for GI)	56	70.9%
Rx for GI Symptoms	6	7.6%
ICD-10 Dx for GI Comorbidity	50	63.3%
GI: Gastrointestinal; GERD: Gastroesophageal reflux disease; Dx: Diagnosis; Rx: Medical Prescription		

Table 5. GI Diagnosis and Medication Use by Level of Care in the 18-Month Study Period

		Patients		Visits		GI Rx Present	
Diagnosis	Type of visit	(n)	%	(n)	Per Patient	(n)	%
Constipation	Inpatient	14	17.7%	23	1.64	14	100.0%
	Outpatient	22	27.8%	96	4.36	18	81.8%
	ED	2	2.5%	2	1.00	1	50.0%
Dysphagia	Inpatient	11	13.9%	35	3.18	7	63.6%
	Outpatient	26	32.9%	123	4.73	10	38.5%
	ED	0	0.0%	0	0.00	0	/
Nausea/Vomit	Inpatient	7	8.8%	18	2.57	5	71.4%
	Outpatient	8	10.1%	15	1.88	4	50.0%
	ED	2	2.5%	2	1	1	50.0%
GERD	Inpatient	13	16.5%	36	2.77	8	61.5%
	Outpatient	21	26.6%	92	4.38	12	57.1%
	ED	0	0.0%	0	0.00	0	/
Diarrhea	Inpatient	2	2.5%	2	1.00	1	50.0%
	Outpatient	2	2.5%	2	1.00	0	0.0%
	ED	0	0.0%	0	0.00	0	/
Gastritis	Inpatient	1	1.3%	1	1.00	0	0.0%
	Outpatient	3	3.8%	3	1.00	2	66.7%
	ED	0	0.0%	0	0.00	0	/
No GI Diagnosis but have GI Rx						6	
GI: Gastrointestinal, GERD: Gastroesophageal reflux disease; ED: Emergency Department							

CONCLUSIONS

- GI related comorbidities are prevalent among RTT cases as reflected in the problem list, IP and OP encounters, and pharmacy tables
- Encounter ICD-10 diagnoses for GI comorbidities appeared in 34 (43.0%) cases. Clinical notes and GI-related medications identified an additional 28 (35.4%) individuals with a GI comorbidity the absence of an ICD-10 code. A total of 62 (78.5%) cases within the study period had one or more GI comorbidities
- Constipation was the most common GI comorbidity (43.0%), followed by dysphagia (37.6%), and GERD (29.1%)
- EHR data including clinical notes and medications contain important information regarding comorbidities not available within coded data. Analyzing EHR data including clinician notes has the potential to fill an important gap in real-world evidence generation

LIMITATIONS

- The EHR data provides less comprehensive information regarding service utilization and treatment patterns

REFERENCES

1 The National Institute of Neurological Disorders and Stroke. Rett Syndrome Fact Sheet.
2 Nashville Biosciences.

ACKNOWLEDGMENTS

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DISCLOSURES

Drs. May and Kyle are employees of Acadia Pharmaceuticals, Inc. which sponsored the study. Dr. Fu is an employee of Vanderbilt University. Dr. Ruetsch and Ms. Yang are employees of Health Analytics, LLC which was funded to conduct the study.

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