

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

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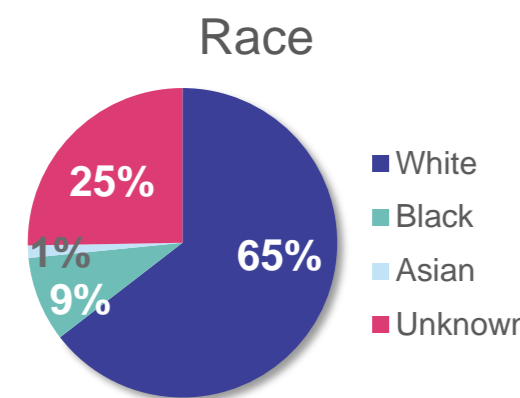
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EXECUTIVE SUMMARY



RTT Population Characteristics

79 Eligible RTT patients
Predominantly female
Mean age: 10.2 years



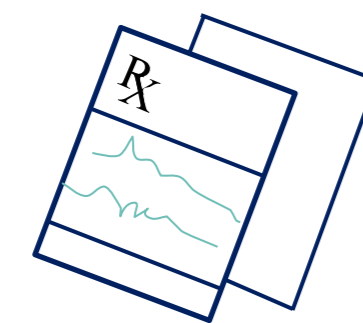
Top 3 GI Diagnosis

39% Dysphagia
37% Constipation
34% GERD

GI Diagnosis

Out of the 79 RTT Patients :

- 50 Had an ICD-10 Diagnosis for a GI Comorbidity
 - 39 Had at least 1 GI Rx and at least 1 GI Dx
 - 27 of the 39 had a specific GI medication following a GI Dx
 - 11 Had no GI Rx and at least 1 GI Dx
- 6 Had at least 1 GI Rx and no GI Dx
- 23 Had no GI Rx and no GI Dx



GI HCRU

- 123 Outpatient visits for 26 patients for Dysphagia
- 90 Outpatient visits for 22 patients for Constipation
- 92 Outpatient visits for 21 patients for GERD

39 (78%) cases reported taking a medication for their GI Symptoms

INTRODUCTION

- Rett syndrome (RTT) is a rare, neurodevelopmental disorder primarily affecting females¹ 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
- Secondary analysis of administrative data is among the most efficient methods for studying healthcare resource utilization, however, GI comorbidities might not always be included on inpatient (IP) or outpatient (OP) claims forms. E.H.R. data potentially include more detail about comorbidities and over the counter (OTC) pharmacy utilization
- Few studies have examined the characteristics, clinical sequelae, 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 E.H.R. data

METHODS

Study Design

- A retrospective analysis of individuals diagnosed with Rett syndrome using E.H.R. data from Nashville Biosciences²

Study Period

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

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

- 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

Table 1. Attrition Table for Selection of GI Notes

Step	Description	N	%
1	Patients with at least 1 encounter associated with a diagnosis of RTT Syndrome	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 under 30 years old at their first RTT Diagnoses	79	70.5%

RESULTS

Table 2. Demographic Characteristics of RTT Patients

	Total Population n=79	
	N(Mean)	%(SD)
Gender		
Male	13	16.5%
Female	66	83.5%
Age (Mean, SD)	(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. Patients with GI Medication (Rx) and/or Diagnosis (Dx) in the 6-Month Baseline and Follow-Up Period.

	GI Medication Present	GI Diagnosis Present	
		Yes	No
	Yes	39	6
	No	11	23

GI: Gastrointestinal

Table 4. Prevalence of GI Diagnoses in the Study Period

	N= 79	
GI Comorbidity	N	%
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%
ICD-10 Dx for GI Comorbidity	50	63.3%
Rx for GI Symptoms	6	7.6%
Total (ICD-10 or RX for GI)	56	70.9%

GI: Gastrointestinal; GERD: Gastroesophageal reflux disease; Dx: Diagnosis; Rx: Medical Prescription

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

Diagnosis	Type of visit	6-Month Baseline-Follow-Up Period				Have GI Medication (# of Patient)	% Have GI Medication
		Visits	Visit/Patient	Patients	%Patients		
Constipation	Inpatient	23	1.64	14	17.7%	14	100.0%
	Outpatient	96	4.36	22	27.8%	18	81.8%
	Emergency Room	2	1.00	2	2.5%	1	50.0%
Dysphagia	Inpatient	35	3.18	11	13.9%	7	63.6%
	Outpatient	123	4.73	26	32.9%	10	38.5%
	Emergency Room	0	0.00	0	0.0%	0	/
Nausea/Vomit	Inpatient	18	2.57	7	8.8%	5	71.4%
	Outpatient	15	1.88	8	10.1%	4	50.0%
	Emergency Room	2	1	2	2.5%	1	50.0%
GERD	Inpatient	36	2.77	13	16.5%	8	61.5%
	Outpatient	92	4.38	21	26.6%	12	57.1%
	Emergency Room	0	0.00	0	0.0%	0	/
Diarrhea	Inpatient	2	1.00	2	2.5%	1	50.0%
	Outpatient	2	1.00	2	2.5%	0	0.0%
	Emergency Room	0	0.00	0	0.0%	0	/
Gastritis	Inpatient	1	1.00	1	1.3%	0	0.0%
	Outpatient	3	1.00	3	3.8%	2	66.7%
	Emergency Room	0	0.00	0	0.0%	0	/
No GI Diagnosis but have GI Rx						6	

GI: Gastrointestinal, GERD: Gastroesophageal reflux disease

CONCLUSIONS

- GI related comorbidities are prevalent among RTT cases as reflected in the problem list, IP and OP encounters, and pharmacy tables
- ICD-10 diagnoses indicating GI comorbidities appeared in 50 (63.3%) cases while another 6 (7.6%) cases had an Rx for GI symptoms in the absence of an ICD-10 Dx of GI comorbidity. A total of 56 (70.9%) cases within the study period had either a GI comorbidity or medication for GI symptoms
- Of those with an ICD-10 GI diagnosis, 39 (78.0%) reported taking medications for those issues
- Dysphagia was documented most frequently in OP visits and less frequently in IP or ED visits
- When documenting GI issues among patients with RTT, it is important to examine both ICD-10 diagnoses as well as medications indicated for GI issues. E.H.R. data are more well-suited for this analysis than would be claims data as not all the diagnoses on a patients' problem list and none of the OTC medications will be represented in claims data

LIMITATIONS

- The E.H.R. data provides less comprehensive information regarding service utilization and treatment patterns.

REFERENCES

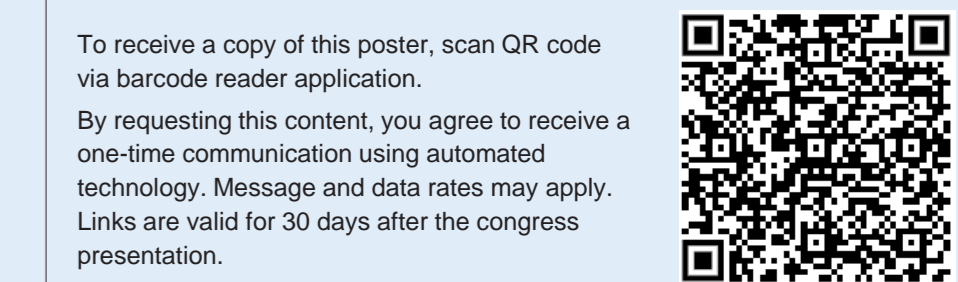
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DISCLOSURES

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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