Assessing Timely Diagnosis of Pediatric-Onset Opsoclonus Myoclonus Syndrome (OMS); Insight from the OMS Natural History Registry

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BACKGROUND/OBJECTIVE

Opsoclonus myoclonus syndrome (OMS) is an ultrarare disorder affecting as few as 1 in 5,000,000 people annually with onset typically between 1 and 3 years of age. Given the rarity, a primary challenge to care has been the lack of awareness and knowledge by healthcare stakeholders (physicians, insurance companies). To better understand the natural history and experience of patients with OMS, the OMSLife Foundation and the National Organization of Rare Disorders (NORD) launched the OMS Natural History Registry in 2017. Here we provide an update on the registry with a focus on time to diagnosis for pediatric-onset OMS.

METHODS

The registry is an aggregation of individual survey data with input from the patient, the patient caregiver, or both. Fifteen distinct surveys collect patient demographics and family history, symptoms, diagnosis and disease severity, therapies (behavioral, occupational, physical, speech), and medication details. Surveys are updated as applicable per patient / caregiver preference in response to email reminders. As of July 2023, 392 patients or caregivers from 22 countries have registered and 253 have contributed data to the OMS registry, making it the largest known collection of OMS patient data in the world. For this study, data were limited to 177 patients who had clinician-diagnosed, pediatric-onset OMS, known dates for symptom onset and diagnosis, and who completed profile/demographic, diagnosis, and symptom surveys. [FIGURE 1] Variable differences in time to diagnosis were assessed by Mann-Whitney U (2 categories) or Independent-Samples Kruskal-Wallis (>2 categories) with pairwise comparisons.

RESULTS

The majority of study patients were female (56%, 100), white (86%, 153), and with private health insurance (52%, 92). [FIGURE 2] Participants resided in 14 countries with 86% (153) in the US. Mean (median) age at symptom onset and diagnosis was 25 (20) and 29 (23) months, respectively. [FIGURE 3] Mean (median) time from symptom onset to diagnosis was 4.0 (0.9) months. Time to diagnosis was not significantly different based on Mitchell-Pike OMS severity classification at diagnosis, diagnosing specialty type, or presence/absence of fever, headache, ataxia, myoclonus, opsoclonus, sleep disturbances, temper tantrums, tremors, or vomiting at onset. [TABLE 1] For patients with symptom onset prior to 2011, time to diagnosis was significantly longer (mean [median] months = 7.2 [2.0]) than observed for patients with symptom onset post-2015 (mean [median] months = 2.1 [0.6], p=0.008).

CONCLUSIONS

The OMS registry developed by The OMSLife Foundation and NORD has proven to be a successful endeavor for this ultrarare disease, providing large samples of data for researchers and clinicians to use to identify care opportunities in OMS. Though in the beginning stages of utilizing this data, early trends such as shortening time to diagnosis in more recent years are evident. Additional insight is anticipated as the registry continues to grow in participants and type of data collected.

FIGURE 1: Patient Disposition

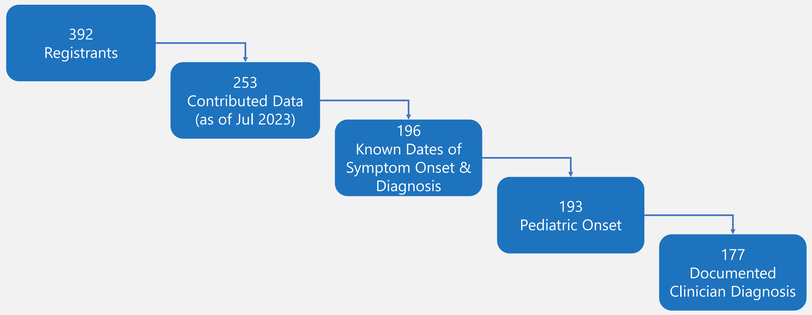

FIGURE 2: Study Population Demographics (n=177)

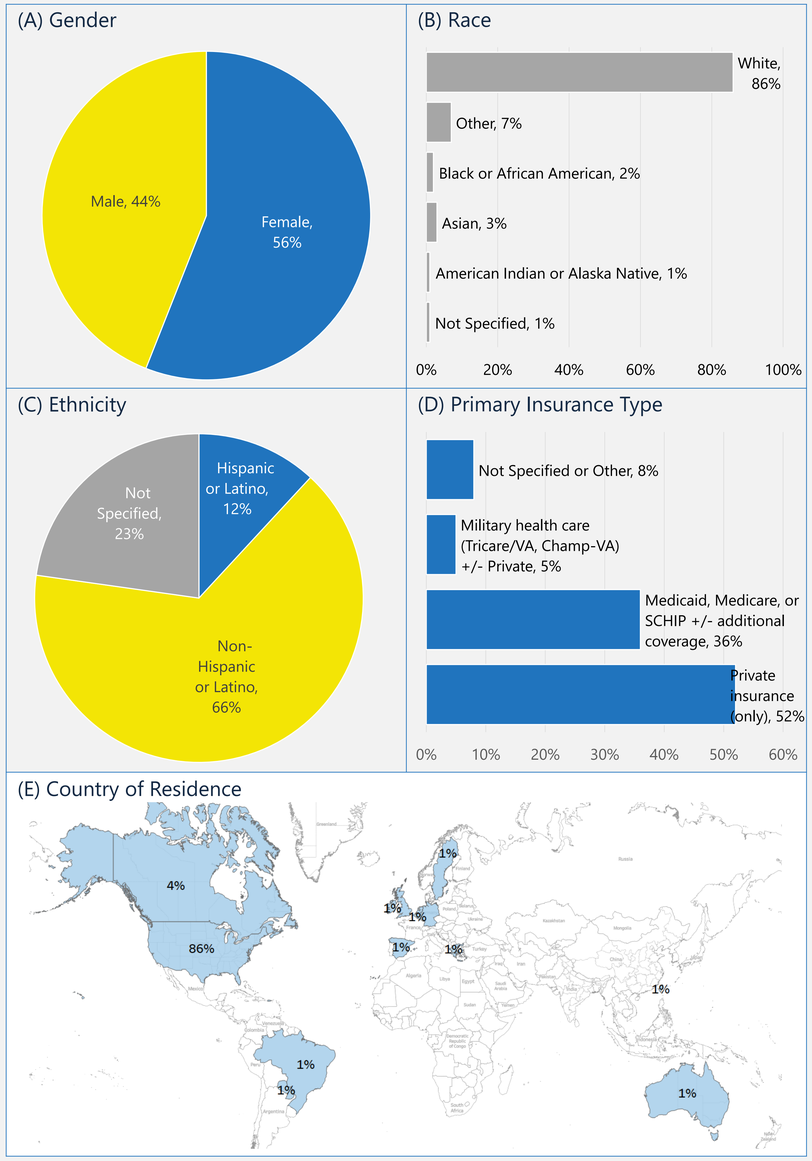

FIGURE 3: Age (months) at Symptom Onset and Diagnosis

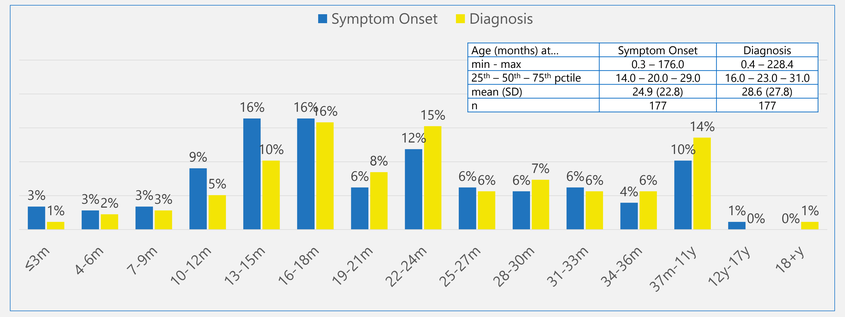

TABLE 1: Time (months) from OMS Onset to Diagnosis by Select Variables

Variable	Category	Months to Diagnosis from Symptom Onset						No. Pts	% Pts
		Mean	SD	Q1	Median	Q3	p		
Mitchell-Pike Severity	mild	4.5	11.5	0.1	0.5	3.9	0.212	22	12%
	moderate	3.9	9.0	0.5	1.0	3.9		71	40%
	severe	3.9	13.2	0.3	0.8	2.6		84	47%
Symptom Onset	<2011 (A)	7.2	18.6	0.6	2.0	5.8	0.008 AvB, 0.216 BvC, 0.522 AvC, 0.005	49	28%
	2011-2015 (B)	3.4	8.3	0.3	1.0	2.5		61	34%
	>2015 (C)	2.1	4.6	0.2	0.6	1.8		67	38%
Diagnosing Specialist	Neurologist	3.8	11.4	0.3	0.9	3.6	0.871	151	85%
	Other	4.9	11.9	0.4	0.9	2.0		26	15%
Fever at Onset	No	3.6	8.4	0.3	0.9	3.7	0.679	155	88%
	Yes	6.8	23.8	0.3	0.7	3.0		22	12%
Headache at Onset	No	3.4	10.1	0.3	0.9	3.0	0.430	162	92%
	Yes	9.8	20.8	0.4	1.2	7.1		15	8%
Ataxia at Onset	No	4.0	7.0	0.2	1.5	6.0	0.343	22	12%
	Yes	3.9	11.9	0.3	0.8	2.9		155	88%
Myoclonus at Onset	No	4.5	13.9	0.5	1.0	4.1	0.093	71	40%
	Yes	3.6	9.5	0.2	0.7	2.0		106	60%
Opsoclonus at Onset	No	4.3	13.9	0.4	1.0	3.7	0.180	70	40%
	Yes	3.7	9.5	0.2	0.8	3.0		107	60%
Sleep disturbance at Onset	No	2.7	6.8	0.2	0.7	2.6	0.058	90	51%
	Yes	5.2	14.7	0.4	1.0	3.8		87	49%
Temper tantrums at Onset	No	4.2	13.2	0.2	0.6	2.6	0.120	104	59%
	Yes	3.5	8.3	0.4	1.2	3.8		73	41%
Tremors at Onset	No	4.4	13.3	0.4	0.9	3.9	0.810	92	52%
	Yes	3.5	9.0	0.3	0.8	2.3		85	48%
Vomiting at Onset	No	3.9	11.6	0.3	0.9	3.1	0.671	137	77%
	Yes	4.2	10.8	0.4	0.9	2.8		40	23%
Total		4.0	11.4	0.3	0.9	3.1		177	100%

SM is an employee of Trio Health Analytics though Trio Health Analytics is not associated with this work.