Inaugural Patient-Reported Registry of Pediatric OMAS: Presentation, Diagnosis, and Treatment of 194 Patients

Sandra Jimenez Giraldo, MD , Michael Michaelis, BS, Lauren Kerr, BA, Christopher Cortina, MS, Bo Zhang, PhD, Mark P. Gorman, MD

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Background

- Due to its rarity, single-site studies of OMAS generally include small numbers of patients, limiting scope and generalizability
- Patient/parent-powered research registries offer an alternative approach to assemble larger data sets and generate meaningful data

Methods

- Retrospective cohort study of 194 patients with OMAS onset <18yo from 1990 - 2020
- Data in online surveys was entered between 2017 – 2020 by parents / guardians
- Overall study approved by the independent central Hummingbird Institutional Review Board
- Deidentified data transferred from the OMSLife Foundation to study authors at Boston Children's Hospital for analysis (IRB exempt, BCH IRB-P00031016)

TABLE 1.
Name, Source, Description, and Completion Rates of Surveys

Survey Name	Author	Description	Completed N (%)
Participant profile	NORD	Basic demographic information including sex, race, ethnicity, etc.	194 (100%)
OMS onset	OMSLife	Symptoms at onset, time to diagnosis, type of doctor diagnosing OMAS, etc.	166 (85.6%)
Treatment of OMS	OMSLife	Medications (routes, frequency, duration, side effects, etc.)	148 (76.3%)
Neuroblastoma	OMSLife	Timing of tumor detection and tumor type	133 (68.6%)
Other therapies	OMSLife	Participation in and results of speech, physical, behavioral, and occupational therapies	136 (70.1%)
Triggers	OMSLife	Tumors and their diagnostic evaluation, environmental triggers, etc.	128 (66.0%)
Medical and diagnostic data	NORD	Height, weight, tobacco, and alcohol consumption, etc.	129 (66.5%)
Treatment and return of symptoms	NORD	Medications, dietary issues, current status of patient, etc.	125 (64.4%)
Family medical history	OMSLife	Family history of autoimmune diseases	94 (48.5%)

TABLE 2.
Demographic Features of Survey Respondents

Parameter (Number of Respondents)	N (%)
Biological sex (n = 194)	
Male	89 (45.9)
Female	105 (54.1)
Tumor presence (n = 175)	
Yes	97 (55.4)
No	78 (44.6)
Race (n = 189)	
White	165 (87.3)
Black or African American	3 (1.6)
Asian	4 (2.1)
American Indian/Alaskan Native	1 (0.5)
Other	16 (8.5)
Ethnicity (n = 155)	
Hispanic or Latino	23 (14.8)
Not Hispanic or Latino	121 (78.1)
Unknown	11 (7.1)
Health insurance status (n = 188)	
Insured	181 (96.3)
Not insured	6 (3.2)
Unknown	1 (0.5)
Region (n = 194)	
United States	165 (85.1)
International (non-US)	29 (14.9)

Key findings – presentation

- Older age of onset in females (22 months [15 to 31] vs 18 [14 to 23], P = 0.0223) (overall n=166)
- High rate (31%) of parental autoimmunity which did not differ between tumor and non-tumor subgroups (overall n = 94) confirming results of prior smaller studies
- Nearly 40% of patients with OMAS did not have opsoclonus or myoclonus as one of the initial presenting symptoms (n = 166)
  - 8 patients (5%) never developed opsoclonus

Key findings - diagnosis

- Median time from symptom onset to a correct diagnosis was 25 days and from diagnosis to treatment was 7 days (n=166)
- Initial misdiagnosis at the time of OMAS onset occurred in 48.5% of cases, with most common misdiagnosis being acute cerebellar ataxia or postinfectious cerebellitis (71.3%) (overall n = 166)
- 23.1% of patients (overall n=78 with available data) had delayed tumor detection, including eight who had a tumor detected later than 12 months after the initial presentation. Routine follow-up scans and second opinion on initial set of scans both led to tumor detection

Key findings – treatment

- Of the 148 patients reporting treatment data, 18.2% received one, 12.2% received two, and 56.1% received three or more agents
- Most common medications received were steroids (79.7%), intravenous immunoglobulin (IVIG) (64.9%), and rituximab (52.7%)
- Only 23.5% of patients received behavioral therapy, which was perceived as beneficial in 90.6%

Conclusions

- Patient/parent-powered research is feasible in OMAS and generated the second largest published cohort to date
- High rate of parental autoimmunity suggests genetic predisposition for children with OMAS regardless of tumor presence
- Opsoclonus and myoclonus frequently absent at presentation, combined with high rate of initial misdiagnosis, point to need for increased education and definitive biomarker
- Delayed tumor detection was relatively common, suggesting need for improved methods and need for serial surveillance
- Most patients in the US receive multi-agent therapy
- Behavioral therapy is under-utilized despite being perceived as beneficial

Strengths

- Very large sample size (n=194) considering rarity of OMAS
- Findings comparable to published literature supporting validity of parent-entered data
- Ability to collect data from patients outside of large institutions and beyond individual institutional obstacles
- Ability to tailor future surveys for specific research criteria (i.e. sleep study)
- Now have 17 surveys on OMAS
- Quick turnaround in data collection

Limitations

- Low number of patients in some surveys – Data was collected in 2020 (third year of registry)
- Subjective report from parents without verification from clinician review or medical records
- Missing data for variable numbers of patients in different surveys
- However, comparability of results to clinic / hospital-based studies tempers these concerns
- Parents who do not speak English, have less education, and/or have less access to the internet may have been less likely to enroll in the study, which may limit generalizability (Next upgrade will include Spanish and other languages)

A 9 year history of the OMSLife Registry

- April 2016 - Awarded NORD grant
- January 2017 – In production w/ 9 surveys
- 2018-2024 – 7 additional surveys (16 total)
- March 2025 – 470 total participants
- Continued organic growth (2020 → 2024)
  - Participant Profile 194 → 279
  - Onset & Diagnosis 166 → 235
  - Treatment of OMS 148 → 211
  - Family Medical History 94 → 146
- Information from prior presentations/publications at

https://omslifefoundation.org/oms-registry/

- participant_profile
- onset_and_diagnosis
- oms_adult
- treatment_of_oms
- treatment_and_review_of_systems
- medical_and_diagnostic_data
- quality_of_life_pediatric
- quality_of_life_adult_part1
- quality_of_life_adult_part2
- therapies_for_oms
- oms_triggers
- neuroblastoma_and_precocious_puberty
- family_medical_history_survey
- teenager_and_young_adult_relapse_survey
- vaccinations_survey
- sleep_habits

Next steps

- Analyze cases with delayed tumor detection to determine if common features such as tumor location or imaging methods used
- Translated surveys – est Q4 2025
- Additional targeted surveys
  - Sleep
  - Behavior
  - Use of (OMAS consensus) medications
  - Long term outcomes (quality of life)
  - Trends since consensus statement was published
  - Adult onset
  - Others as requested

Questions?

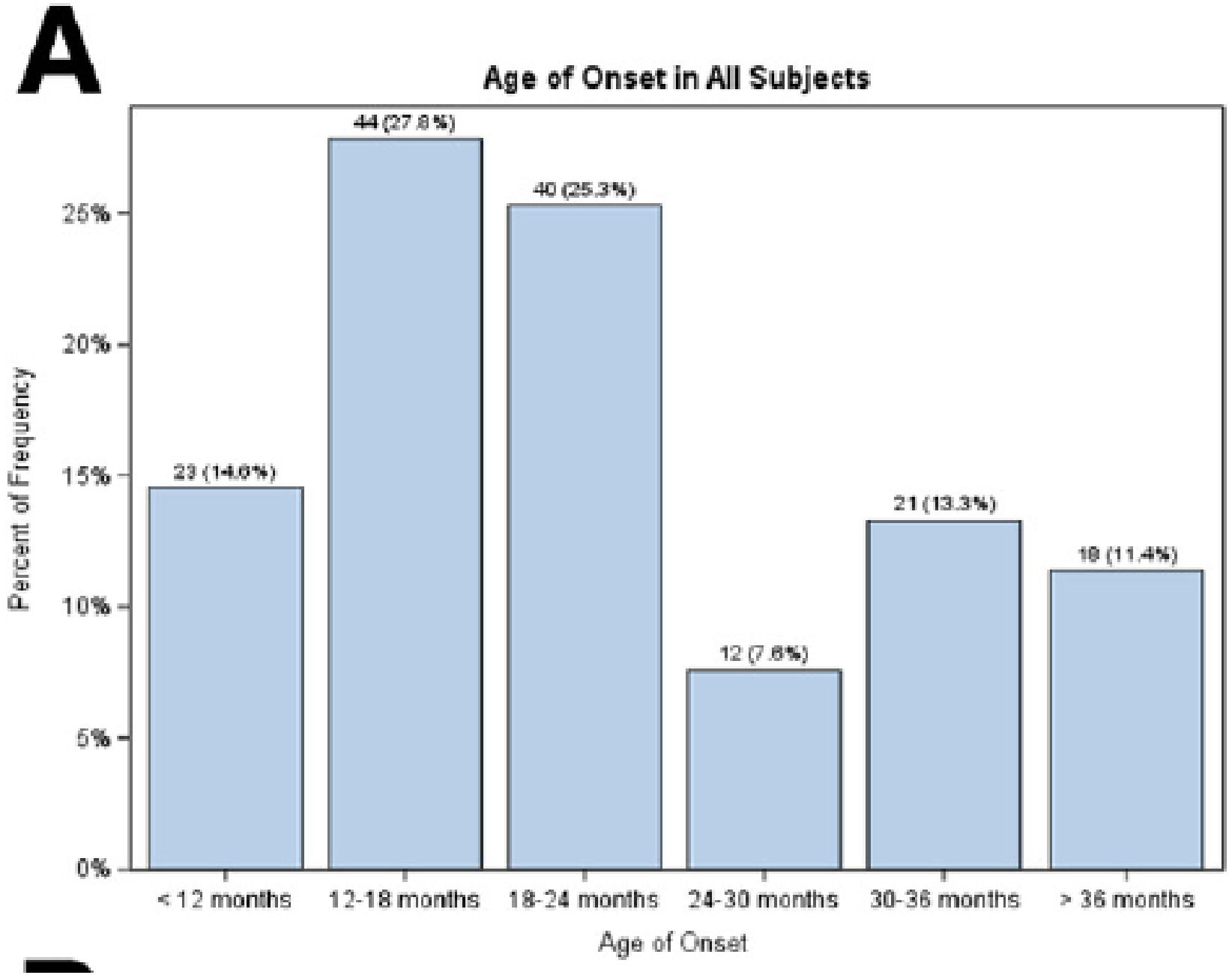
TABLE 3.
History of Autoimmune Disorders in First-Degree Relatives of 94 Patients With OMAS

Autoimmune Disease	N (%)
Autoimmune thyroid disease (Hashimoto thyroiditis, Graves disease)	17 (18.1)
Rheumatoid arthritis	4 (4.3)
Systemic lupus erythematosus	0
Insulin-dependent diabetes mellitus (type 1)	2 (2.1)
Inflammatory bowel disease	2 (2.1)
Psoriasis	7 (7.5)
Other	10 (10.8)
Addison disease	0
Celiac disease	1 (1)
Multiple sclerosis	1 (1)
Pernicious anemia	1 (1)
Vitiligo	2 (2.1)
Other (lichen planus, ulcerative colitis, autoimmune hepatitis, inflammatory polyarthritis, ankylosing spondylitis)	7 (7.6)
Any autoimmune disease	29 (30.9)

TABLE 4.
Age of OMAS Onset as a Function of Biological Sex and Tumor Status

Age Category at OMAS Onset				
Entire Cohort	Overall (n = 194)	Male (n = 89)	Female (n = 105)	*P* value (Wilcoxon rank-sum test)
Median (IQR)	18 (14-29)	18 (14-23)	22 (15-31)	0.0223
<12 months	23 (14.6)	10 (14.1)	13 (14.9)	
12-18 months	44 (27.8)	25 (35.2)	19 (21.8)	
18-24 months	40 (25.3)	20 (28.2)	20 (23.0)	
24-30 months	12 (7.6)	5 (7.0)	7 (8.1)	
30-36 months	21 (13.3)	5 (7.0)	16 (18.4)	
36 months-18 years	18 (11.4)	6 (8.5)	12 (13.8)	
Missing in age	36	18	18	
Tumor status reported	Overall (n = 175)	Male (n = 79)	Female (n = 96)	
Tumor status unknown	Overall (n = 19)	Male (n = 10)	Female (n = 9)	

A


Age of Onset in All Subjects
44 (27.8%)
40 (25.3%)
23 (14.6%)
21 (13.3%)
18 (11.4%)
12 (7.6%)
25%
20%
15%
10%
5%
0%
Percent of Frequency
< 12 months
12-18 months
18-24 months
24-30 months
30-36 months
> 36 months
Age of Onset

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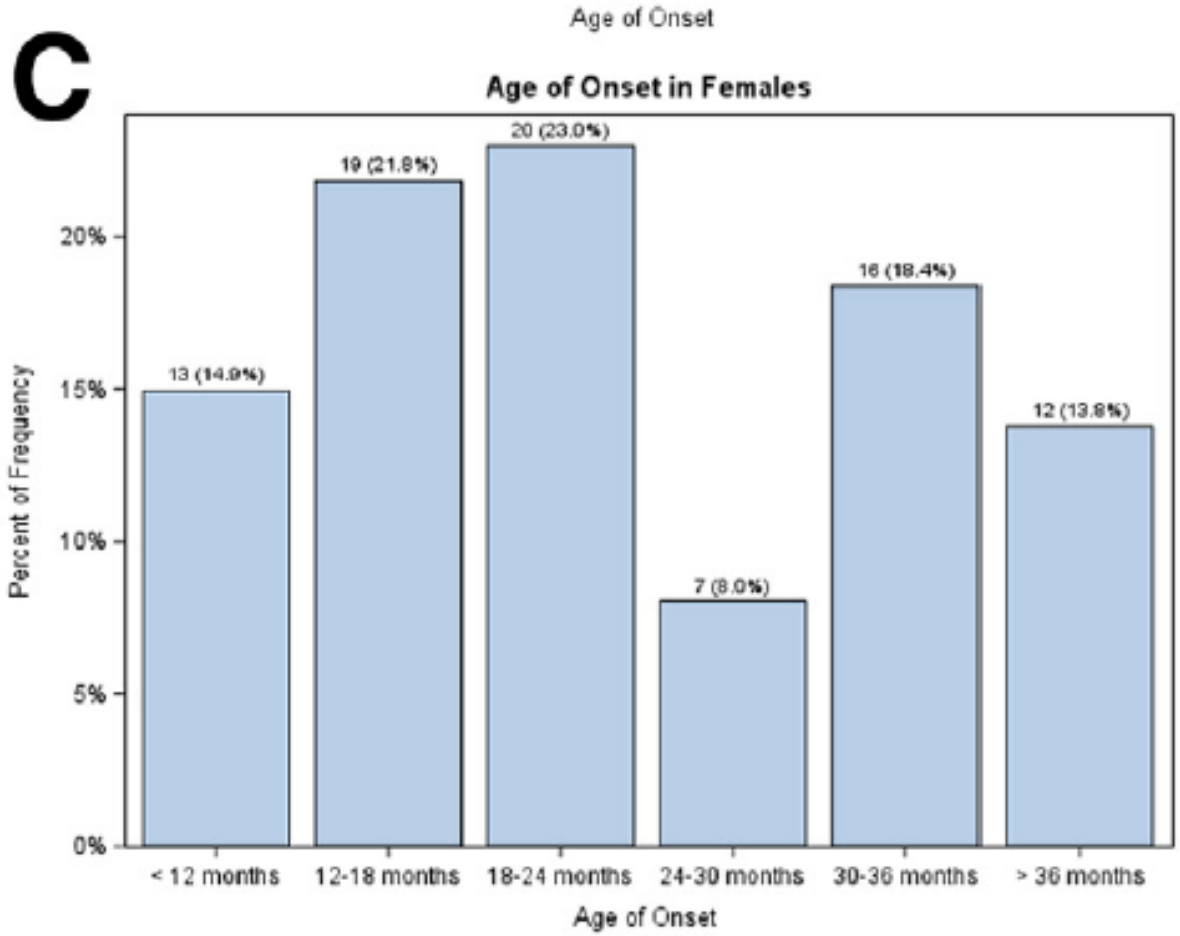
Age of Onset
Age of Onset in Females
Percent of Frequency
20%
15%
10%
5%
0%
13 (14.9%)
19 (21.8%)
20 (23.0%)
7 (8.0%)
16 (18.4%)
12 (13.8%)
< 12 months
12-18 months
18-24 months
24-30 months
30-36 months
> 36 months
Age of Onset

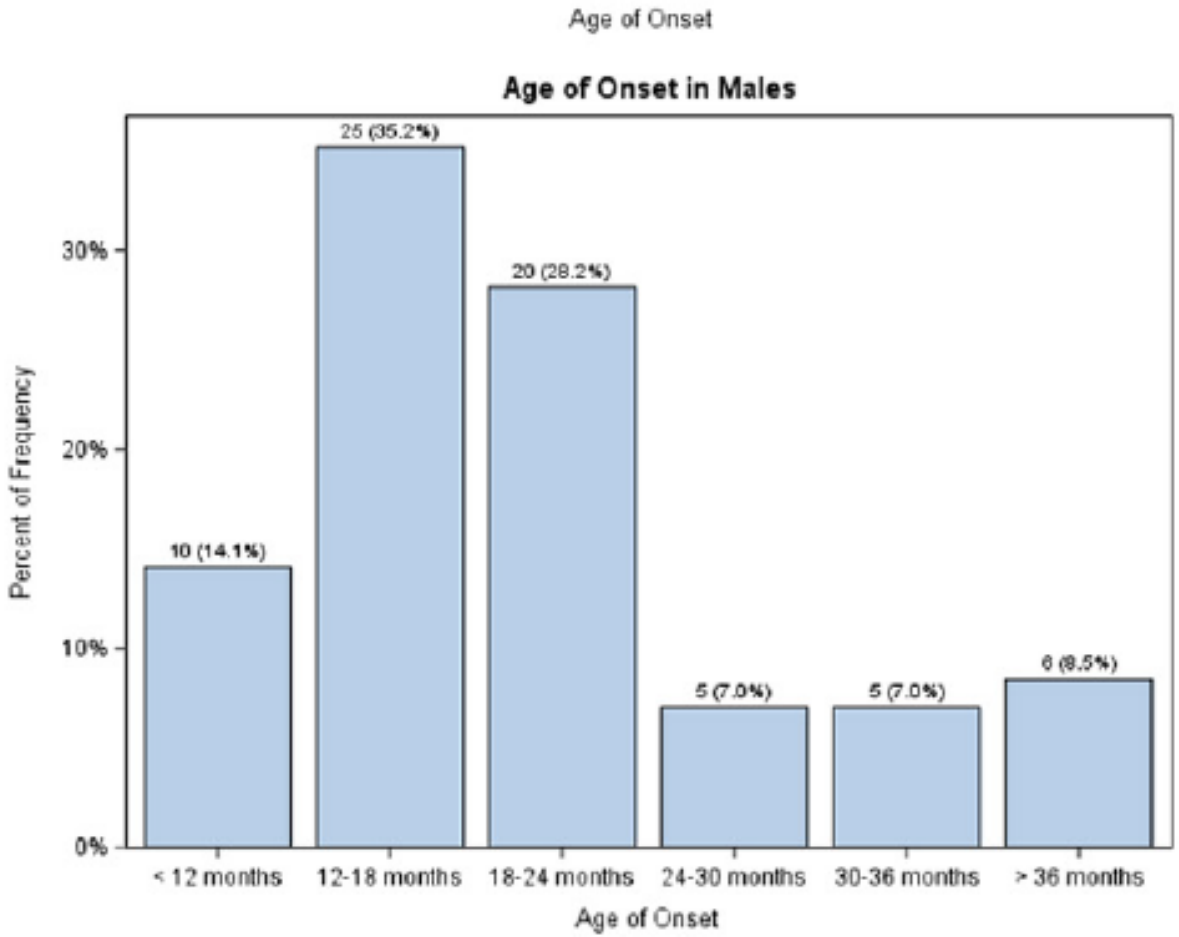
Age of Onset
Age of Onset in Males
Percent of Frequency
30%
20%
10%
0%
10 (14.1%)
25 (35.2%)
20 (28.2%)
5 (7.0%)
5 (7.0%)
6 (8.5%)
< 12 months
12-18 months
18-24 months
24-30 months
30-36 months
> 36 months
Age of Onset

TABLE 5.
Tumor Histology and Characteristics

Variable	N (%)
Tumor present (n = 97)	
Neuroblastoma	61 (70.9)
Ganglioneuroblastoma	14 (16.3)
Ganglioneuroma	9 (10.5)
Other	2 (2.3)
No response	11
Tumor stage (n = 78)	
Stage 1	31 (39.7)
Stage 2	19 (24.4)
Stage 3	7 (9.0)
Stage 4	4 (5.1)
Unknown	17 (21.8)
No response	19
Tumor discovered at time of diagnosis? (n = 78)	
Yes	60 (76.9)
No	18 (23.1)
No response	19
If No: time from OMAS onset to detection of tumor (n = 18)	
1-3 months from OMAS onset	4 (22.2)
3-6 months from OMAS onset	2 (11.1)
6-9 months from OMAS onset	1 (5.6)
9-12 months from OMAS onset	3 (16.7)
>12 months from OMAS onset	8 (44.4)
If No: method of tumor discovery (n = 18)	
Routine scans as part of OMAS follow-up	8 (44.4)
Second opinion on initial set of scans taken at diagnosis	3 (16.7)
Other/unclear from available data	7 (38.9)

TABLE 6.
Time From Onset to Correct Diagnosis and From Diagnosis to Treatment

Characteristics	Overall	Tumor	No Tumor	*P* Value (Wilcoxon Rank-Sum Test)
Time from onset to diagnosis				
Number of patients	164	90	74	0.4011
Median time (days) (IQR, range)	23 (7, 74) (7287, 3432)	18.5 (8, 61) (−7287, 988)	30 (6, 95) (−329, 3432)	
Time from onset to diagnosis excluding negative values				
Number of patients	159	89	70	0.1704
Median time (days) (IQR, range)	25 (8, 78) (0, 3432)	19 (8, 61) (0, 988)	31 (8, 108) (0, 3432)	
From diagnosis to treatment				
Number of patients	106	61	45	0.4317
Median time (days) (IQR, range)	1 (−6, 19) (−358, 5387)	2 (−6, 19) (−358, 676)	0 (−5, 23) (−299, 5387)	
From diagnosis to treatment excluding negative values				
Number of patients	75	43	32	0.2440
Median time (days) (IQR, range)	7 (0, 31) (0, 5387)	10 (1, 31) (0, 676)	3.5 (0, 30.5) (0, 5387)	

TABLE 7.
Pharmacologic Treatment Received in Patients With OMAS

Feature	N (%)		
	All Patients n = 148	Before 2013 n = 59*	2013 and After n = 84*
Treatment categories			
Missing agent information	20 (13.5)	8 (13.6)*	10 (11.9)*
One agent	27 (18.2)	10 (17)	17 (20.2)
Two agents	18 (12.2)	6 (10.2)	12 (14.3)
Three or more agents	83 (56.1)	35 (59.3)*	45 (53.6)*
Treatment agents			
Corticosteroids (prednisolone, dexamethasone, methylprednisolone) or corticotropins (ACTH)	118 (79.7)	46 (78)*	69 (82.1)*
IVIG	96 (64.9)	41 (69.5)*	53 (63.1)*
Cyclophosphamide	30 (20.3)	15 (25.4)*	13 (15.5)*
Rituximab	78 (52.7)	31 (52.5)*	44 (52.4)*
Mycophenolate mofetil	7 (4.7)	5 (8.5)*	1 (1.2)*