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Study Group



A Large-Scale Analysis of NMOSD in Brazil: A Nationwide Study of 872 Patients from the NMO-RG Database

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INTRODUCTION

Previous studies on neuromyelitis optica spectrum disorder (NMOSD) in Brazil suggest that, compared to other Western countries, the disease has a higher prevalence, with attacks leading to more severe disability. However, these studies remain limited, based on small cohorts and typically restricted to specific geographical regions. The Neuromyelitis Optica Registry (NMO-RG) was recently introduced to encourage collaborative research among investigators. Consequently, a large cohort was gathered, encompassing patients from diverse geographical areas and genetic backgrounds, allowing for a more comprehensive characterization of the disease.

METHODS

We analyzed demographic and clinical data from NMOSD patients fulfilling the 2015 Diagnostic Criteria, registered in the NMO-RG Database by November 30, 2024.

RESULTS

A total of 872 patients were included with median age at presentation of 33 (range 1-79) years; 85% were female. The most common ethnicities were Caucasian (43.2%), Mixed (36.9%), and African-descent (18.7%), with smaller proportions of Asian and Indigenous backgrounds (1.4%), reflecting the country's diverse genetic heritage. The most frequent presenting symptoms were myelitis (49.6%), optic neuritis (48.7%), area postrema syndrome (APS) (16.3%), simultaneous ON and myelitis (10.9%), and brainstem symptoms (BSS) (9.5%). Aquaporin-4 IgG was positive in 73.1%, with brainstem symptoms more common in seronegative cases ($p=0.03$). Cerebrospinal fluid-specific oligoclonal bands were detected in 14.6%. Mean disease duration was 8.8 ± 8.2 years, and the mean annual relapse rate was 0.79 ± 1.20 . At last visit, mean EDSS were 4.62 ± 2.34 (seropositive) vs. 4.05 ± 2.32 (seronegative) ($p=0.004$). Patients with EDSS ≥ 6.0 had a median disease duration of 8.8 (range 0.1-53.3) years.

Table. Comparison of demographic and clinical characteristics of NMOSD according to AQP4-Ab serostatus

	Total	AQP4-Ab positivity	AQP4-Ab negativity	P
Age at onset, yr	n=872 n=865	n=637 n=630	n=187 n=187	
Median [IQR]	33.0 [23.0-43.0]	34.0 [24.0-45.0]	31.0 [22.0-42.0]	NS
Range	1.0-79.0	1.0-79.0	6.0-72.0	
Mean±SD	33.8±14.4	34.4±14.8	32.6±13.6	
Female gender, no./total no. (%)	735/866 (84.9)	554/632 (87.7)	141/186 (75.8)	<0.001
Ethnicity, no./total no. (%)				NS
Caucasian	328/764 (42.9)	239/553 (43.2)	73/163 (44.8)	
Mixed	282/764 (36.9)	200/553 (36.2)	59/163 (36.2)	
African descent	143/764 (18.7)	104/553 (18.8)	30/163 (18.4)	
Asian/Other	11/764 (1.4)	10/553 (1.8)	1/163 (0.6)	
Presenting symptoms, no./total no. (%)				
Optic neuritis (ON)	419/861 (48.7)	306/627 (48.8)	90/186 (48.4)	NS
Myelitis (MY)	427/861 (49.6)	318/627 (50.7)	81/186 (43.5)	0.095
Area postrema syndrome (APS)	140/861 (16.3)	101/627 (16.1)	27/186 (14.5)	NS
Brainstem (BSS)	82/861 (9.5)	50/726 (8.0)	25/186 (13.4)	0.030
Diencephalon	8/861 (0.9)	4/627 (0.6)	4/186 (2.2)	0.086
Cerebral	18/861 (2.1)	14/627 (2.2)	4/186 (2.2)	NS
Optic neuritis and myelitis	94/861 (10.9)	65/627 (10.4)	21/186 (11.3)	NS
Exclusive ON+MY	68/861 (7.9)	46/627 (7.3)	19/186 (10.2)	NS
Exclusive ON	294/861 (34.1)	216/627 (34.4)	63/186 (33.9)	NS
Exclusive MY	290/861 (33.7)	222/627 (35.4)	52/186 (28.0)	0.064
Exclusive APS	52/861 (6.0)	35/627 (5.6)	16/186 (8.6)	NS
Exclusive BS	29/861 (3.4)	16/627 (2.6)	12/186 (6.5)	0.019
General Symptoms, no./total no. (%)				
No general symptoms	286/751 (38.1)	192/516 (37.2)	82/187 (43.9)	NS
Pain	302/751 (40.2)	212/516 (41.1)	71/187 (38.0)	NS
Pruritus	23/751 (3.1)	20/516 (3.9)	3/187 (1.6)	NS
Fatigue	134/751 (17.8)	87/516 (16.9)	34/187 (18.2)	NS
Anxiety/depression	206/751 (27.4)	137/516 (26.6)	51/187 (27.3)	NS
Cognitive symptoms	43/751 (5.7)	29/516 (5.6)	9/187 (4.8)	NS
Annual relapse rate, relapses/yr	n=862	n=628	n=186	
Median [IQR]	0.40 [0.19 - 0.80]	0.38 [0.18-0.75]	0.44 [0.20-1.00]	NS
Range	0.00 - 10.00	0.00-10.00	0.00-6.00	
Mean±SD	0.79±1.20	0.79±1.25	0.82±1.10	
EDSS at last visit	n=830	n=603	n=182	
Median [IQR]	4.00 [3.00 - 6.50]	4.00 [3.00-6.50]	4.00 [2.0-6.00]	0.004
Range	0.00 - 10.00	0.00 - 10.00	0.00 - 10.00	
Mean±SD	4.59±2.40	4.61±2.34	4.04±2.32	
AQP4-Ab positivity, no./total no. (%)	637/824 (73.1)	NA	NA	NA
OCB/high IgG index, no./total no. (%)	70/478 (14.6)	48/356 (13.5)	18/104 (17.3)	NS
Disease duration (months)	n=865	n=630	n=187	
Median [IQR]	79.0 [31.5 - 153.0]	84.0 [35.1-156.0]	66.0 [26.0-132.0]	0.063
Range	1.0 - 639.0	1.0-637.0	1.0-639.0	
Mean±SD	105.6±98.8	108.8±97.8	97.5±102.5	

CONCLUSION

This study comprises one of the largest single-country NMOSD cohort worldwide. Our findings underscore the importance of the NMO-RG for advancing collaboration and highlight unique features of NMOSD in Brazil, paving the way for targeted interventions that address this population's specific needs.

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