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NEUROPSYCHOLOGY

Clinical Characteristics of a *PSEN1* Variant in the Argentine Cohort of the Dominantly Inherited Alzheimer Network

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Abstract

Background: In recent years, there has been an increase in the number of case reports of families with autosomal dominant Alzheimer's disease (ADAD) in Latin America (LA). Nevertheless, little is known about the clinical phenotypes and demographic characteristics of mutations in each country. Past literature has proved that variants in LA have different and unique characteristics than those reported in Asia, North America, and Europe. Clinical heterogeneity in ADAD supports the need to study clinical phenotypes, especially in preclinical stages, to develop cognitive and clinical markers of the disease.

We aimed to describe clinical and cognitive characteristics at baseline assessment of an Argentina *PSEN1* p.M146L family enrolled at DIAN.

Method: Fifteen Dian Participants living in Buenos Aires and its suburbs and in Taco Pozo, a rural and isolated community from the Province of Salta, positive for the *PSEN1* p.M146L mutation, were recruited. DNA was obtained from peripheral blood leukocytes according to standard protocols. In addition, exon 5 of the *PSEN1* gene was PCR-amplified, and Sanger sequencing was performed. NACC Uniform Data Set clinical assessment, including the 2.0 UDS Neuropsychological Battery, was administered. Two groups were used to compare cognitive data: Patients with CDR:0 (n:10) and patients with CDR>0 (n:5).

Result: The mean age at symptom onset reported in this cohort was 42,90 (SD: 4,62). Significant differences were found in several cognitive measures: Logical Memory Immediate, TMT B and Digit Backward (<0,001); MMSE, Category Fluency Animals, TMT A and Digit Symbol(<0,05). In neurological signs, paraplegia was reported in one of the patients.

Conclusion: As expected, differences in neuropsychological tests showed a prevalence of memory deficits, however, dysexecutive symptoms were also present for this

mutation. Paraplegia in *PSEN1* p.M146L has not been reported in the literature so far. Ongoing longitudinal work will be important in tracking changes in this cohort. Other characteristics, proper of underrepresented groups, such as socioeconomic status, education, race, and ethnicity, deserve appropriate follow-up. Future studies are planned, comparing urban and rural populations in this cohort with the same mutation.

	CDR = 0 (n:10)	CDR > 0 (n:5)	p value
Age	28,60 (4,60)	40,6 (4,39)	0,005
Education	13,90 (2,38)	12,00 (1,41)	ns
Female %	40%	70%	ns
AAO	43,00 (3,91)	42,5 (10,61)	ns
EYO	-14,44 (6,11)	-5,0 (15,56)	ns
MMSE	28,80 (0,92)	20,8 (5,26)	0,036
Logical Memory Inm	10,80 (3,33)	3,6 (1,14)	0,003
Digit Forward	6,60 (2,59)	5,2 (1,92)	ns
Digit Backward	5,50 (0,85)	2,2 (1,48)	0,001
Animals	21,60 (4,09)	13,6 (6,38)	0,037
TMT A	44,90 (14,01)	85,6 (40,66)	0,019
TMT B	106,30 (39,33)	271,6 (63,50)	0,004
Digit Symbol WAIS	43,10 (10,34)	16,8 (15,16)	0,012
Logical Memory Delayed	9,80 (3,39)	1,8 (1,64)	ns
OCCScore	6,40 (1,26)	5,4 (1,52)	ns
EDScore	3,20 (0,79)	4,0 (0,71)	ns
NPI	1,00 (1,70)	2,8 (2,17)	ns
FAQ	0	2,8 (2,95)	ns
GDS	2,80 (1,40)	6,00 (5,05)	ns