

Clinical Burden of Advanced Autosomal Dominant Alzheimer's Disease in Home-Based Care

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Introduction

Autosomal Dominant Alzheimer's Disease (ADAD) is a rare early-onset condition with a severe clinical course, typically progressing to dementia by late 40s. In advanced stages (Functional Assessment Staging Test (FAST) 6–7, care shifts to the home setting, where families must manage complex medical and caregiving challenges, including seizures, infections, and malnutrition. Despite this high burden, these events remain poorly characterized in ADAD populations.

Objective

To describe the clinical profile, acute events, and complications among individuals carrying pathogenic *PSEN1* genetic variants receiving home-based care.

Methods

A cross-sectional study including 75 patients enrolled in the Grupo de Neurociencias de Antioquia (GNA) home-care program (2025–2026) was conducted. A standardized protocol covering functional scales, clinical assessment, comorbidities, and acute events was applied. Data was extracted from clinical records and analyzed descriptively. Missing data were present in some subgroup analyses due to incomplete records.

Conclusions

This study highlights the complex clinical burden of advanced ADAD in the home care setting. Characterized by high rates of seizures, infections, malnutrition, and functional dependency (geriatric syndromes) despite relatively young age. These findings underscore the need for multidimensional, home-based care models tailored to this population. Given the progressive nature of disease, care strategies should prioritize quality of life, symptom management, and end-of-life support.

Acknowledgments

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Results

Table 1. Sociodemographic, genetic, and clinical characteristics of the population (n=75)

Sex, n (%)	
Female	43 (57,3)
Male	32 (42,6)
Age at visit (years), mean (SD)	
56,93 (6,59)	
Genetic variant, n (%)	
E280A	68 (90,66)
I416T	4 (5,33)
Unknown	3 (4)
Survival, n (%)	
Alive	58 (77,33)
Dead	17 (22,66)
Socioeconomic strata, n (%)	
1	9 (12)
2	39 (52,00)
3	23 (30,66)
4	2 (2,66)
5	1 (1,33)
6	1 (1,33)
Residence, n (%)	
Urban	62 (82,66)
Rural	13 (17,33)
Health coverage, n (%)	
Government subsidized	37 (49,33)
Basic health insurance	38 (50,66)
Barthel Index (n =73), mean (SD)	
28 (35,13)	
FAST Score (n =74), n (%)	
4	5 (6,75)
5	1 (1,35)
6	22 (29,72)
7	46 (62,16)

Figure 1. Frequency of acute clinical events (n = 75)

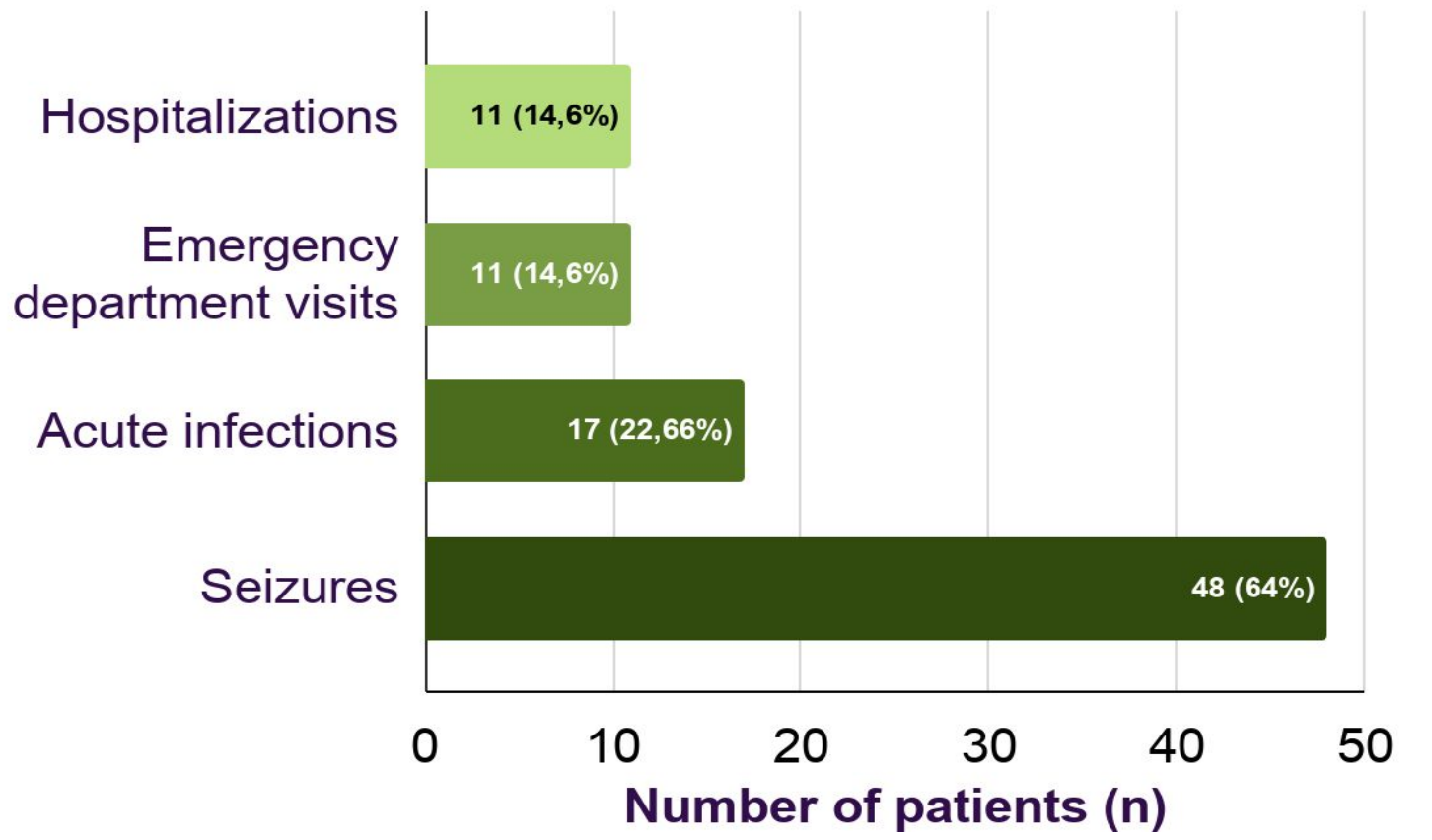


Figure 2. Frequency of chronic complications (n = 75)

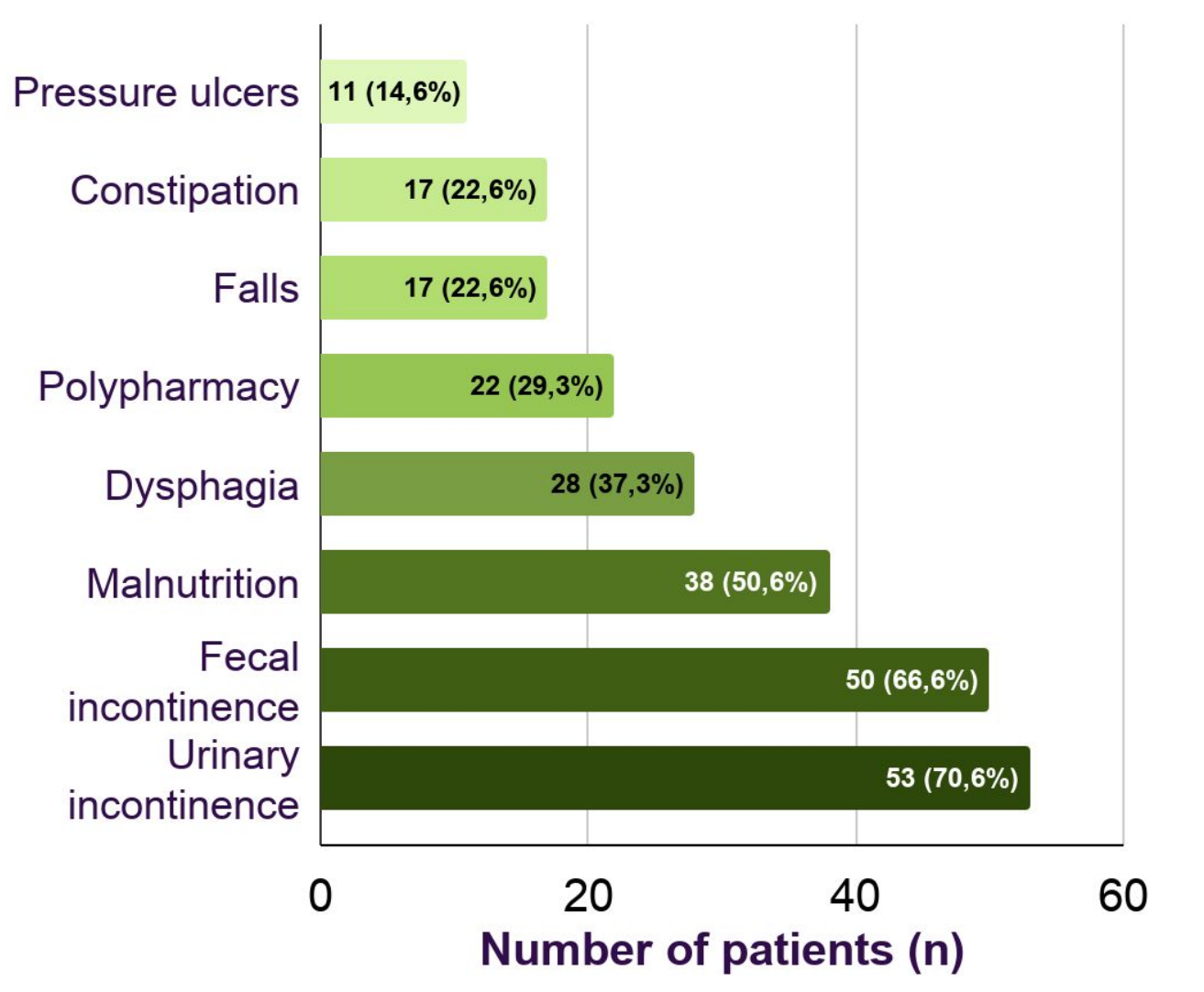
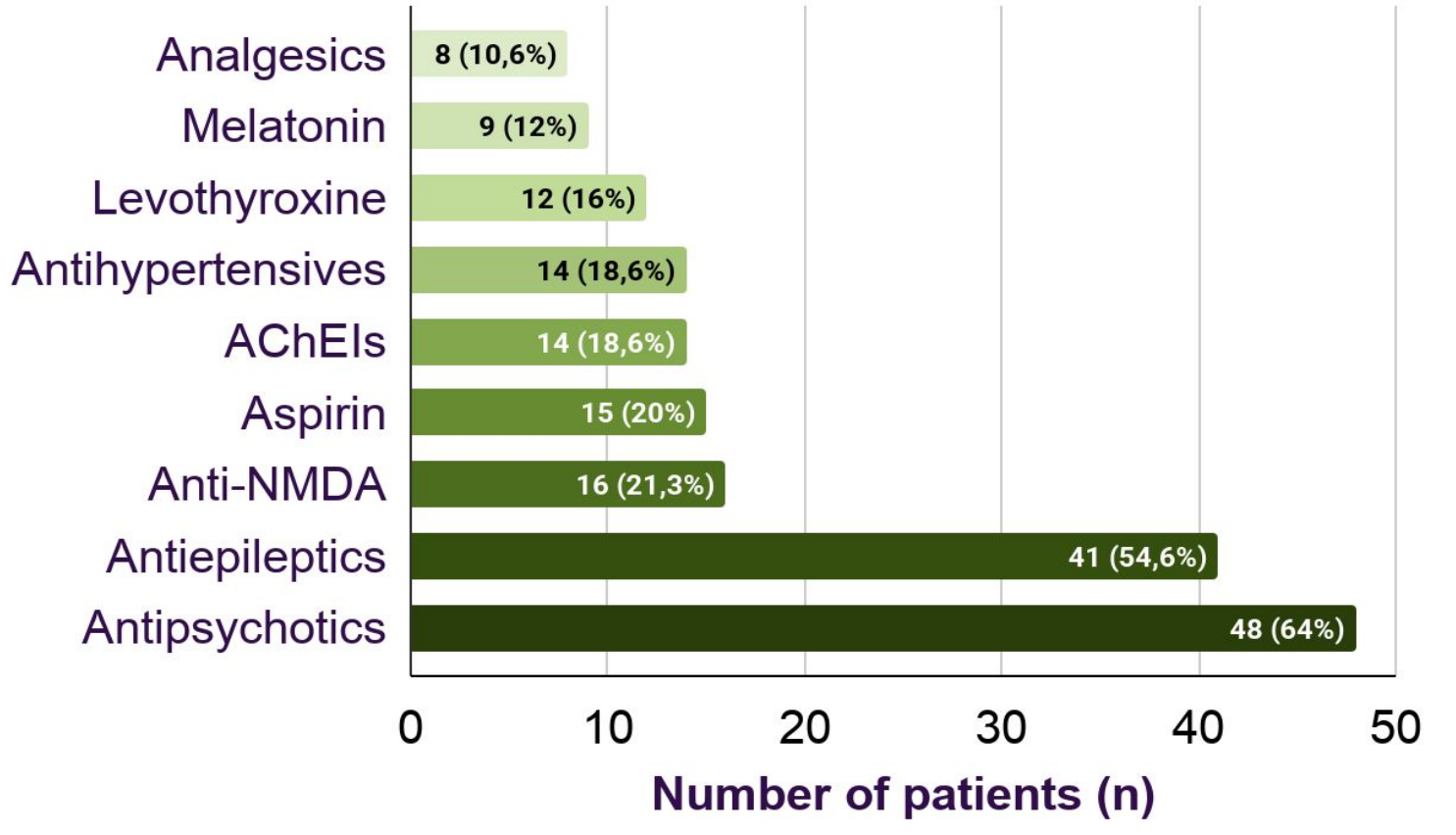


Figure 3. Frequency of medication use (n = 75)



AChEIs = Acetylcholinesterase inhibitors

Figure 4. Frequency of neuropsychiatric symptoms reported by NPI-Q (n = 61)

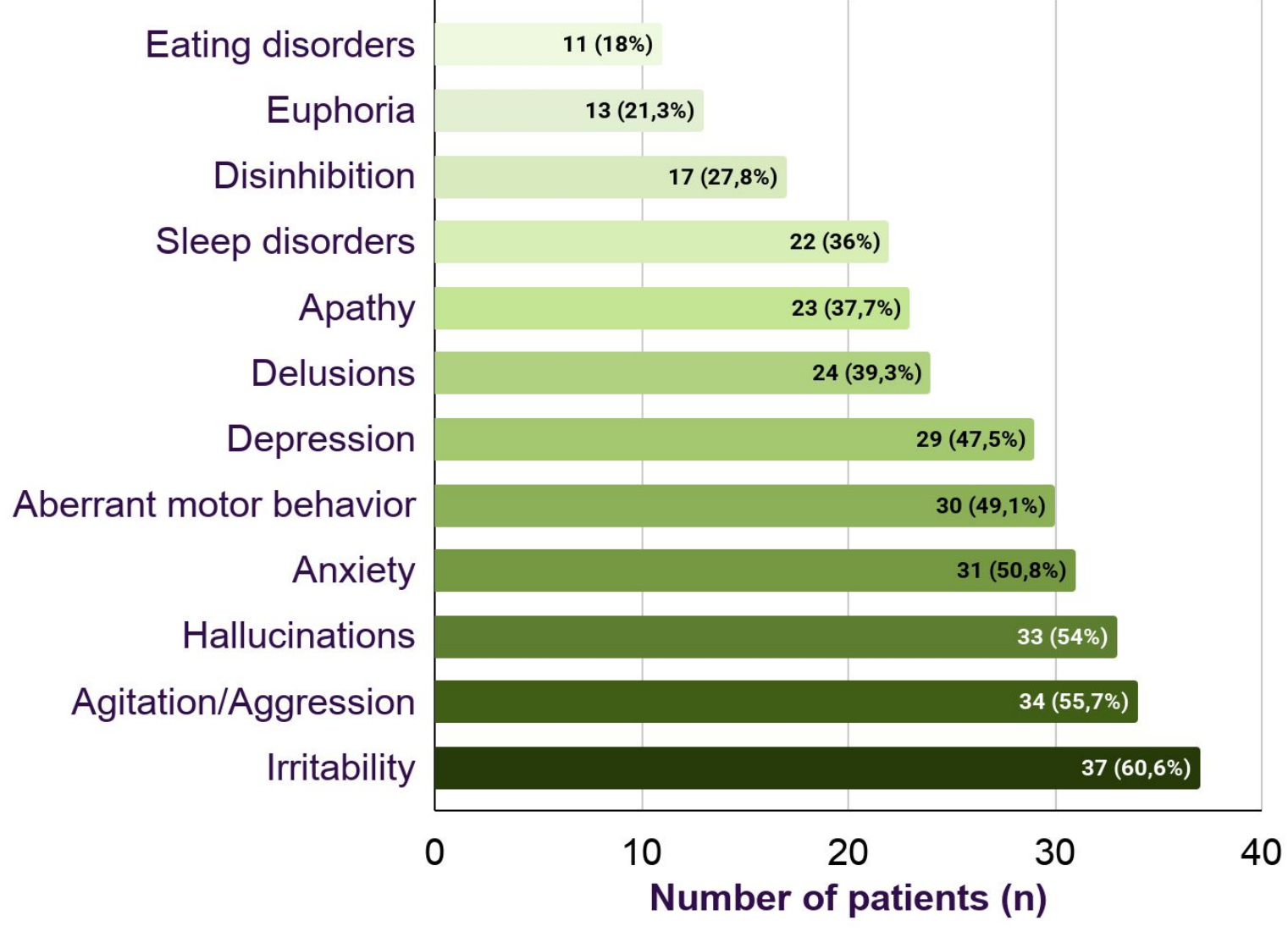


Table 1 outlines the sociodemographical characteristics of the sample. Most patients (90.6%) were *PSEN1*-E280A carriers and in the terminal stages of the disease (62.1% at Functional Assessment Staging (FAST) 7. Seizures were the most common acute event (64%), followed by infections (22.6%) (**Fig. 1**). The main chronic complications included urinary (70.7%) and faecal incontinence (66.7%) (**Fig. 2**). Patients used an average of 3.8 medications, 29.3% met polypharmacy criteria, and antipsychotics were the most frequently prescribed medication (64%) (**Fig. 3**). Nutritional screening by Mini Nutritional Assessment Short Form (MNA-SF) (n=65) showed severe impairment (mean 5.6), with 58.5% malnourished. Neuropsychiatric symptoms evaluated by the Neuropsychiatric Inventory Questionnaire (NPI-Q) (n=61) were common: irritability/lability (60.6%), agitation/aggression (55.7%), and hallucinations (54.1%) (**Fig. 4**). Clinical scales showed relatively preserved quality of life (Quality of Life in Late-Stage Dementia (QUALID), n=57: 24.4 ±10.42), mild risk of pressure ulcers (BRADEN, n=65: 17.18 ±6.35), and low pain levels (PAINAD, n=65: 1.41 ±1.91).

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GNA RESEARCH CAPABILITIES

