

Patterns of treatment and genetic testing in transthyretin amyloid cardiomyopathy: a real-world, multinational survey

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INTRODUCTION

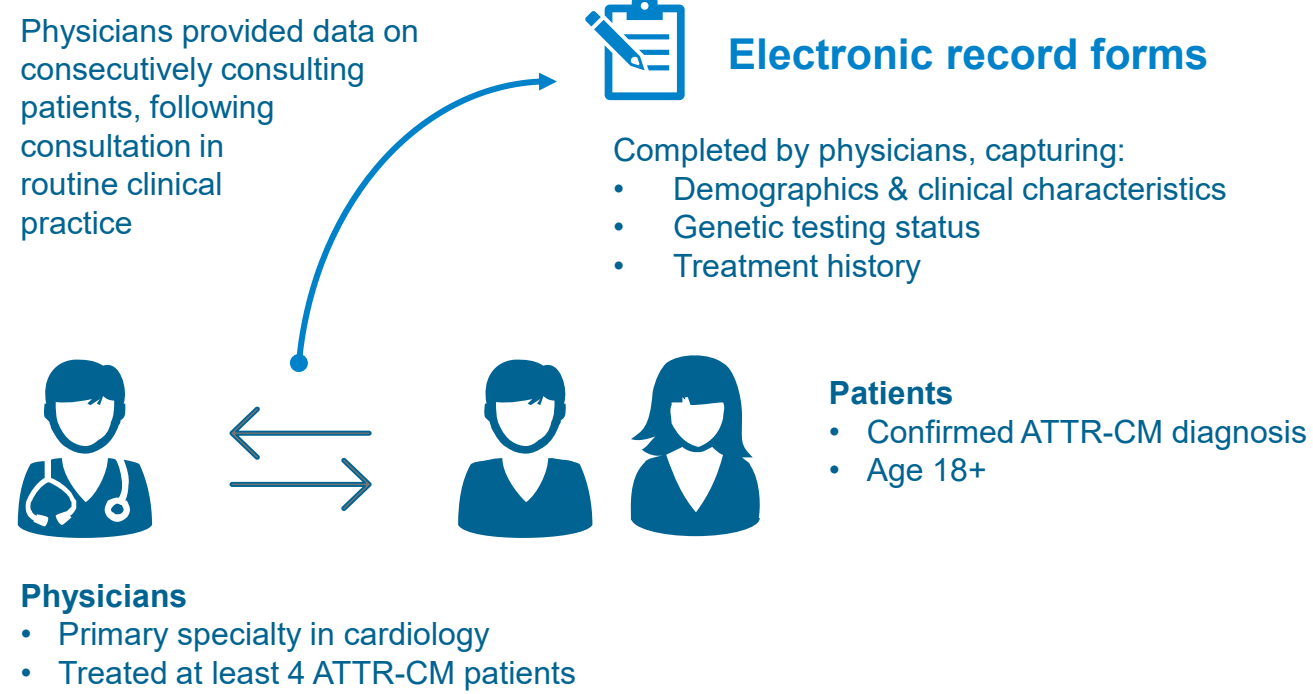
- Transthyretin amyloid cardiomyopathy (ATTR-CM) is caused by transthyretin (TTR) deposition in the myocardium, classed as hereditary (vATTR-CM) in the presence of TTR gene variants, or wild type (wtATTR-CM) with nonmutated TTR deposition.¹
- Genetic testing is key for the early intervention of vATTR-CM, with disease-modifying therapies (DMTs) such as TTR stabilisers, to slow progression and increase survival.^{2,3} However, real world data describing patterns of treatment and genetic testing for ATTR-CM patients are limited.

Objective

To describe treatment and genetic testing patterns in a multinational sample of ATTR-CM patients.

METHODS

- Secondary analyses were conducted using data from the Adelphi Real World ATTR Disease Specific Programme™ (DSP), a cross-sectional survey, with retrospective data collection, of physicians in Europe (France, Germany, Italy, Spain, and the United Kingdom), the United States, and Japan.
- The DSP methodology has been previously described,^{4,5} validated,⁶ and demonstrated to be representative and consistent over time.⁷
- Physicians reported demographics, New York Heart Association (NYHA) classification, treatment history and genetic testing for up to 10 consecutively consulting patients.
- Patients with a confirmed genetic variant were considered hereditary, and those with no variant were considered wild type. Missing data were not imputed.



RESULTS

- Overall, 181 physicians provided data for 858 patients with ATTR-CM. Patient characteristics are summarised in **Table 1**.
- Treatment patterns**
 - Overall, 73.4% of patients were prescribed a drug treatment at the time of survey, with 67.9% of all patients prescribed a DMT.
 - Patient treatment status and DMT prescription across countries is summarised in **Figure 1**.
 - Among patients prescribed a drug treatment, globally, 88.1% were prescribed tafamidis, 3.8% patisiran, and 1.6% diflunisal.
- Genetic testing**
 - Overall, 67.6% of patients had a genetic test performed, 64.4% after the onset of symptoms, and 3.2% before.
 - Patient genetic testing status across countries is summarised in **Figure 2**.
 - Genetic test results are summarised in **Figure 3**.

Table 1: Physician-reported patient characteristics				
Variable	All patients (n=858)	Europe (n=542)	US (n=204)	Japan (n=112)
Age (years), mean (SD)	71.1 (12.6)	73.1 (11.7)	63.9 (13.4)	74.9 (10.3)
Sex; male, n (%)	603 (70.3)	391 (72.1)	122 (59.8)	90 (80.4)
BMI (kg/m ²), mean (SD)	25.2 (3.8)	25.2 (3.2)	25.6 (5.1)	22.7 (2.8)
NYHA at survey, n (%)				
I	163 (19.0)	86 (15.9)	46 (22.5)	31 (27.7)
II	527 (61.4)	355 (65.5)	108 (52.9)	64 (57.1)
III	151 (17.6)	95 (17.5)	42 (20.6)	14 (12.5)
IV	17 (2.0)	6 (1.1)	8 (3.9)	3 (2.7)
Years since diagnosis, mean (SD)	1.8 (2.1)	1.8 (2.3)	1.9 (1.8)	1.5 (1.3)
Ethnicity ^a ; n (%)	n=653	n=431	n=204	n=0
White	526 (82.8)	404 (93.7)	122 (59.8)	-
Black or African American	79 (12.4)	20 (4.6)	59 (28.9)	-
Other ethnicity	34 (5.4)	10 (2.3)	24 (11.8)	-

^aEthnicity data not collected in France or Japan

Figure 1: Physician-reported patient treatment status in relation to ATTR-CM, at survey

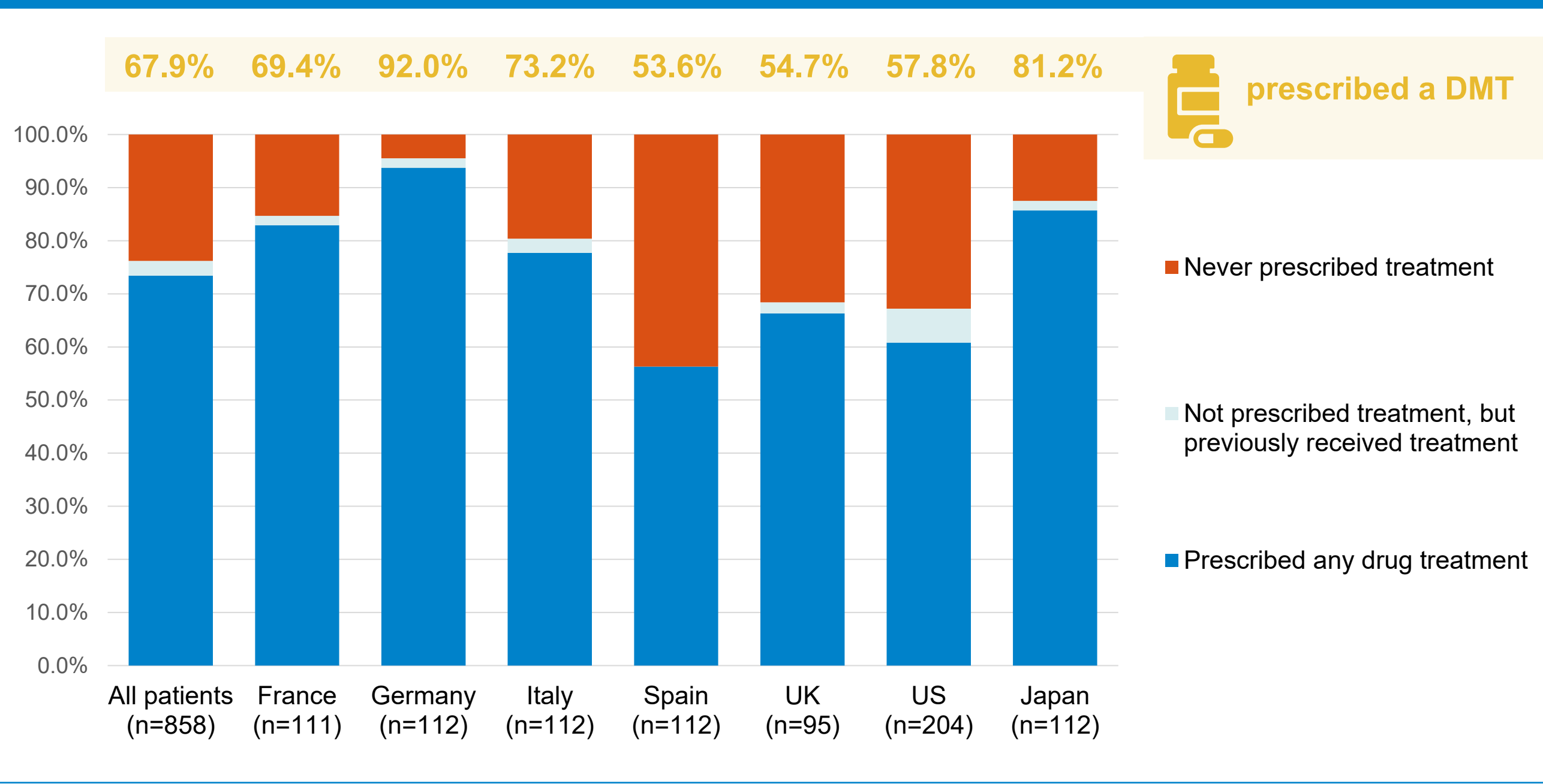
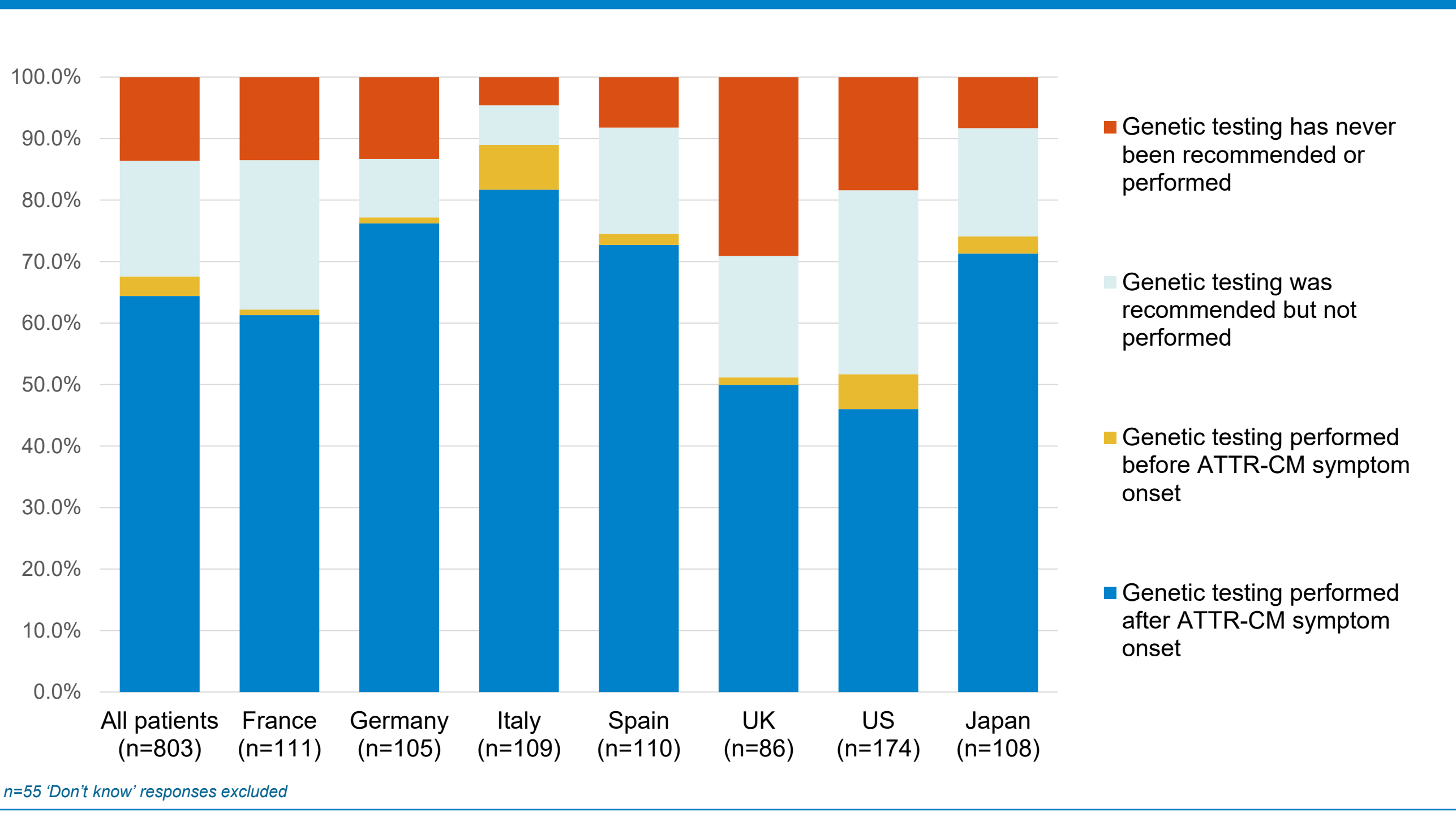


Figure 2: Physician-reported patient genetic testing status in relation to ATTR-CM



CONCLUSIONS

- Despite physicians reporting most patients as being NYHA class II or higher, around one-third of patients were not prescribed DMTs.
- Around one-third had not undergone genetic testing.
- This suggests some patients are not receiving standard-of-care, or treatment tailored to variant prognosis, potentially leading to sub-optimal management and health outcomes.

LIMITATIONS

Participants from the Adelphi Real World ATTR DSP do not constitute a true random sample; participation is influenced by willingness to complete the record forms.

DISCLOSURES

This study used data from the Adelphi Real World ATTR DSP, an independently conducted survey. The DSP is a wholly owned Adelphi Real World product. Pfizer were one of multiple subscribers to the DSP, and funded the analysis described here.

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Figure 3: Physician-reported genetic test results

