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Surveying treatment access and support for rare diseases in Europe

A Quantitative Cross-Country and Patient–Caregiver Comparative Analysis

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OBJECTIVES

This study aimed to:

1. Characterise treatment access experiences, medication collection burden and home delivery (HD) availability across five European countries
2. Compare outcomes between patients and caregivers using formal statistical testing
3. Identify statistically significant differences in key access indicators to inform patient-centred service design, orphan drug launch planning and policy recommendations

BACKGROUND

- Rare diseases affect approximately 30 million people across Europe, spanning over 6,000 distinct conditions[1]
- Despite advances in orphan drug development, patients face persistent barriers including geographic inequality, supply chain fragility and limited awareness of support services[2,3]
- The lived treatment experience remains inadequately characterised through the patient's own voice[3,4]
- Caregiving for rare diseases significantly reduces quality of life across all domains, with financial wellbeing moderating the burden–QoL relationship[5]

METHODS

In December 2025, Sciensus, a life sciences organisation specialising in patient access, engagement and insight solutions, and Rare Patient Voice (RPV), a patient recruiting agency, initiated a collaboration to capture patient and caregiver perspectives on treatment access, medication delivery, and day-to-day burden in rare disease across Europe.

Study design: Cross-sectional, mixed-methods online survey conducted across the UK, France, Germany, Italy and Spain. The survey examined treatment logistics (including travel and collection time), availability and use of home delivery, reliability of supply, emotional and financial impacts across different healthcare settings and access to medicines before official market authorisation (Early Access Programmes (EAP)).

Survey instrument: 20 structured and open-ended questions for patients/caregivers, plus 4 caregiver-specific questions. RPV ensured compliance with applicable data protection and privacy regulations and participants were entered into a raffle as an incentive for their time. Participants were recruited from a global panel covering 1500 diseases with rare disease participants analysed as a subgroup.

Inclusion criteria: Of 860 total respondents, 217 met European rare-disease prevalence criteria (<1 in 2,000) as patients or caregivers; unclear diagnoses were excluded.

Statistical analysis:

- Categorical variables: Pearson's chi-squared test (χ^2)
- Ordinal scales (wellbeing, delay): Mann–Whitney U (patient vs. caregiver); Kruskal–Wallis H (across countries)
- Significance threshold: $p<0.05$

RESULTS

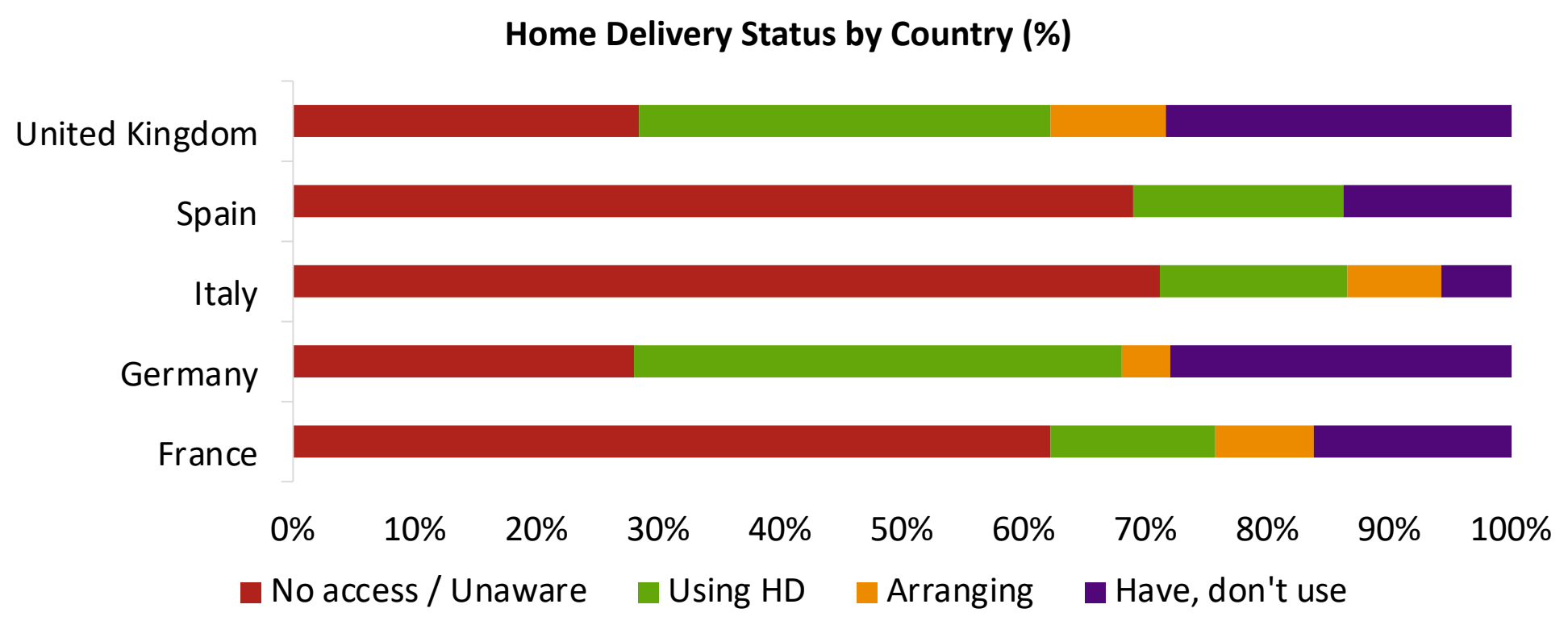
Sample characteristics

Characteristic	Overall (n=217)	Patients (n=161)	Caregivers (n=56)
Country: France	37 (19%)	30 (19%)	7 (13%)
Country: Germany	25 (12%)	19 (12%)	6 (11%)
Country: Italy	52 (18%)	29 (18%)	23 (41%)
Country: Spain	29 (15%)	24 (15%)	5 (13%)
Country: UK	74 (37%)	59 (37%)	15 (34%)
Gender Female	173 (80%)	126 (78%)	47 (84%)
Age 20-29 years	29 (13%)	29 (18%)	
Age 30-39 years	41 (19%)	30 (19%)	11 (20%)
Age 40-49 years	68 (31%)	46 (29%)	22 (39%)
Age 50-59 years	45 (21%)	31 (19%)	14 (25%)
Age 60-69 years	28 (13%)	20 (12%)	8 (14%)
Age 70+ years	6 (3%)	5 (3%)	1 (2%)

Home Delivery Access by Country

Significant cross-country variation in home delivery access (χ^2=39.57, df=12, p<0.001)

Country (n)	No access / Unaware	Using HD	Arranging	Have, don't use
France (37)	62%	14%	8%	16%
Germany (25)	28%	40%	4%	28%
Italy (52)	71%	15%	8%	6%
Spain (29)	69%	17%	0%	14%
United Kingdom (74)	28%	34%	9%	28%
Overall (217)	**50%**	**24%**	**7%**	**19%**

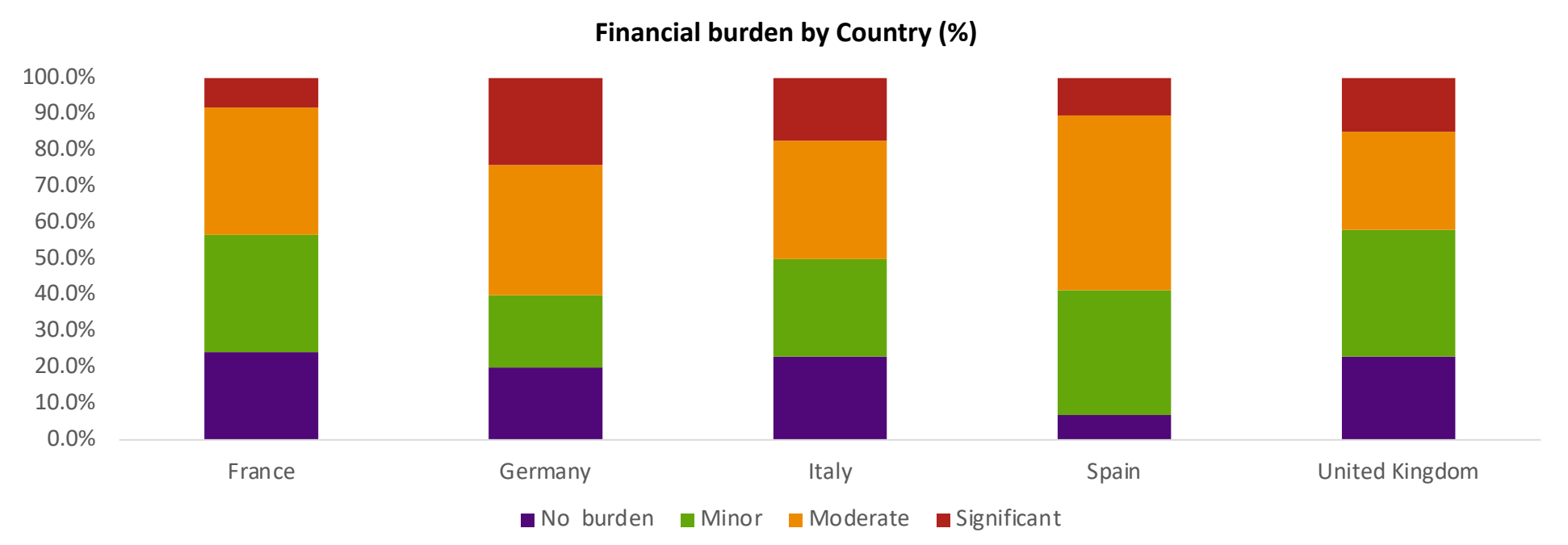


Home Delivery Access by Respondent Role

Significant difference in home delivery access between patients and caregivers (χ^2=13.62, df=3, p=0.004)

Caregivers were more likely to lack home delivery access or awareness (54%) than patients (48%). However, patients were more likely to have access but choose not to use it (22% vs 9%), suggesting different barriers: awareness/availability for caregivers vs suitability/preference for patients.

Financial Burden



Financial burden did not differ significantly by country of residence (χ^2 = 11.10, df = 12, p = 0.52; Kruskal-Wallis H = 4.04, p = 0.40). Germany had the highest rate of significant financial burden (24%).

Financial burden was significantly associated with collection time (χ^2=31.47, df=9, p<0.001; Spearman correlation: ρ = 0.287, p < 0.001)

There was a clear dose-response gradient. Among respondents reporting no financial burden, 15.6% spent more than one hour collecting medication, compared with 43.8% of those reporting significant burden. No respondent reporting zero financial burden spent more than two hours per trip, while 34.4% of those with significant burden did. These findings suggest that extended collection times translate directly into economic pressure, reinforcing the case for home delivery expansion and logistical streamlining.

Collection Time by Role

Caregivers spent significantly more time collecting medication than patients (χ^2=12.08, df=3, p=0.007)

Role	<1 hr	1–2 hr	2–4 hr	>4 hr
Patient (n=161)	75%	17%	3%	6%
Caregiver (n=56)	64%	13%	16%	7%

Caregivers were 5× more likely to spend 2–4 hours per trip (16% vs 3%), reflecting additional logistics of collecting medication on behalf of another person.

Wellbeing by Home Delivery Status

Respondents with home delivery reported significantly better wellbeing (mean 3.60 vs 3.05, Mann–Whitney U=5498, p=0.002) When grouped categorically, the distribution also differed significantly (χ^2=9.56, p=0.008)

Wellbeing Score (1=Neg. impacts … 5=Pos. supports)	With HD (n=53)	Without HD (n=164)
1 – Negatively impacts	3.8%	7.3%
2	11.3%	18.3%
3 – Neutral	37.7%	49.4%
4	15.1%	12.2%
5 – Positively supports	32.1%	12.8%
Negative (1–2)	**15.1%**	**25.6%**
Neutral (3)	**37.7%**	**49.4%**
Positive (4–5)	**47.2%**	**25.0%**
Mean (SD)	**3.60 (1.17)**	**3.05 (1.06)**

Respondents using home delivery were nearly twice as likely to report positive wellbeing (47% vs 25%) and substantially less likely to report negative wellbeing (15% vs 26%). This association supports the case that home delivery may reduce logistical strain and improve patient experience.

Delivery Delay Frequency & Wellbeing by Country

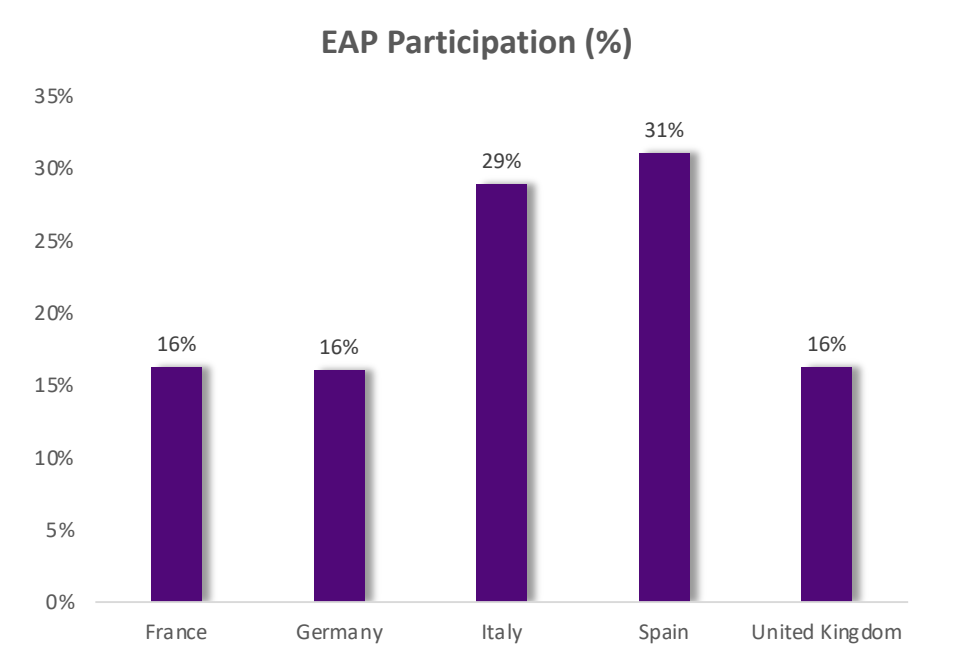
Country	Mean delay (1-5)	Frequent delays (4-5)	Mean wellbeing (1-5)	Negative wellbeing (1-2)
France	2.41	24%	3.41	24%
Germany	2.44	28%	3.04	16%
Italy	2.13	15%	3.12	21%
Spain	2.21	14%	2.79	31%
United Kingdom	2.68	27%	3.32	23%
Overall	**2.41**	**22%**	**3.18**	**23%**

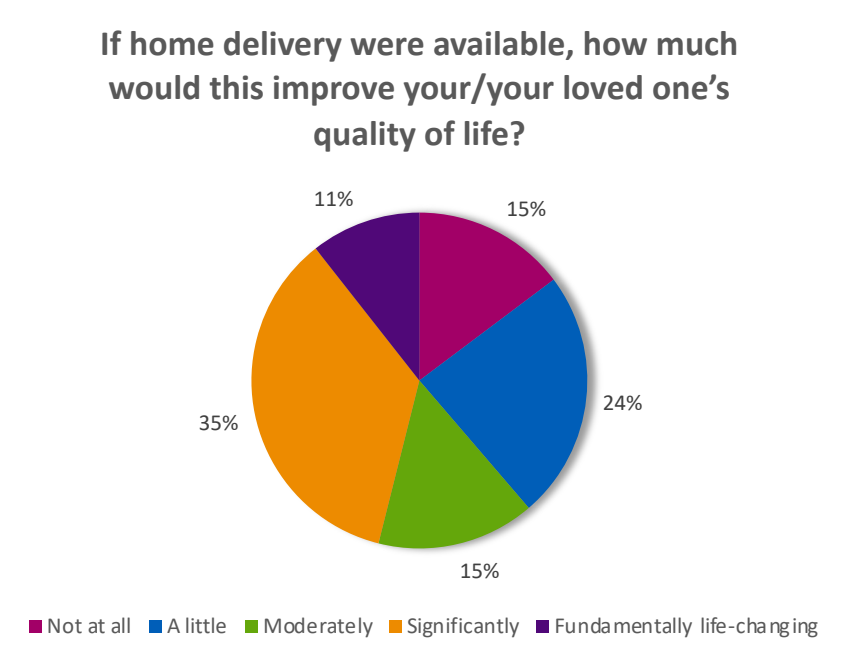
Cross-country comparisons using Kruskal-Wallis and chi-square tests revealed no statistically significant differences across the five countries for delay and wellbeing (H=5.86, p=0.210 and H=6.72, p=0.151). This suggests that the burden of treatment access is a shared, structural challenge across European healthcare systems rather than a country-specific phenomenon.

Patient vs. Caregiver Comparison

Measure	Patient (n=161)	Caregiver (n=56)	Test	p
Wellbeing (mean)	3.17	3.23	M-W U	0.725
Negative wellbeing (1–2)	24%	20%	Fisher's exact	0.582
Delay frequency (mean)	2.42	2.38	M-W U	0.794
Mod–Sig financial burden	45%	59%	χ^2	0.067
Home delivery status†	48/25/4/22	54/21/16/9	χ^2	0.004*
Collection time ≥1 hr	25%	36%	χ^2	0.007*

M-W U = Mann–Whitney U. * p<0.05. Caregivers reported numerically higher rates of moderate-to-significant financial burden (59% vs 45%), though this did not reach significance. † No access or unaware / Using HD / Arranging / Have but don't use (%). Difference driven by caregivers being more likely to be arranging HD (16% vs 4%) and patients more likely to have access but not use it (22% vs 9%).





EAP participation was low (21%) and did not differ significantly by country (χ^2=5.55, p=0.235 (NS)). Italy and Spain showed numerically higher rates (29–31%). If home delivery were available, 85% believed it would improve their quality of life (QoL). This suggests that beyond EAP, patients placed high hopes on homecare to improve their QoL.

Complementary Qualitative Themes

Knowledge deficit
Clinician unfamiliarity is leading barrier to timely diagnosis and treatment

Supply chain fragility
Stock-outs and backorders across all countries, creating clinical risk

Psychological distress
Isolation, anxiety, depression pervasive; psychological support absent

Geographic inequality
Rural patients travel to other regions or countries for specialist care

Caregiver crisis
Burnout, advocacy fatigue and financial strain dominate caregiver accounts

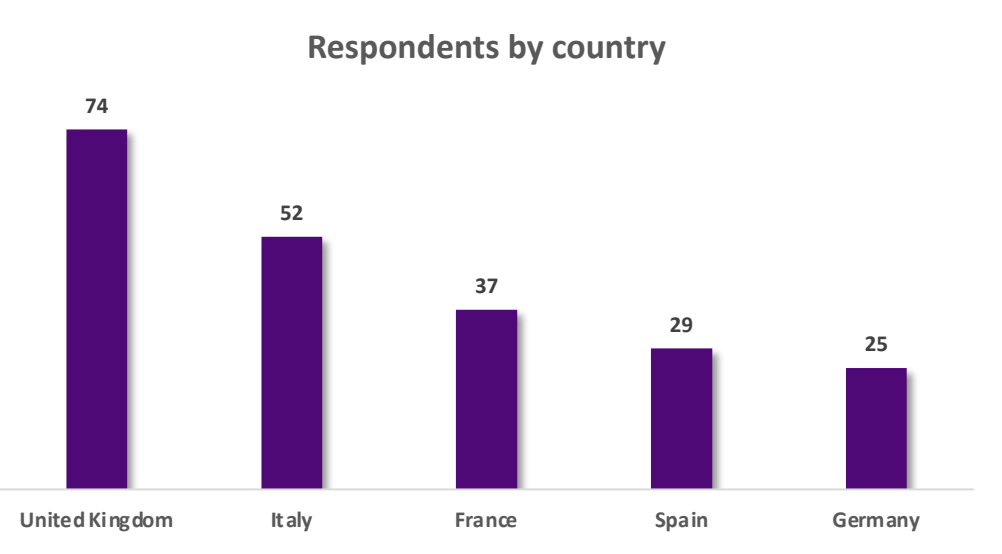
RARE DISEASES REPRESENTED

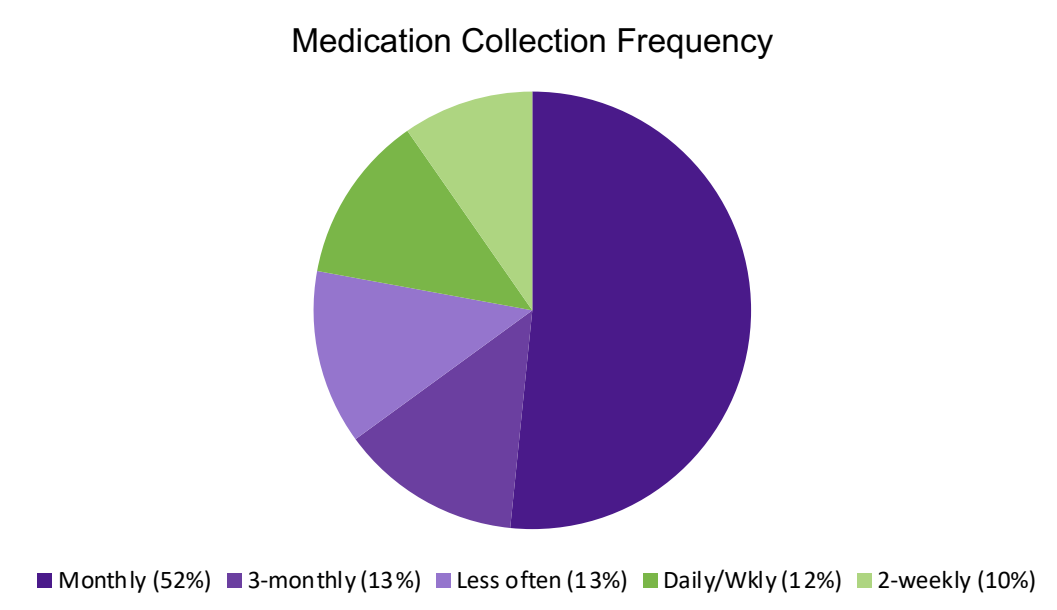
Rare Disease (n > 2)	n	%
Ehlers-Danlos syndrome	25	11.5
Myasthenia gravis	10	4.6
Narcolepsy	7	3.2
Sarcoma (bone & soft tissue)	7	3.2
Behçet's syndrome	6	2.8
Amyotrophic lateral sclerosis (ALS)	4	1.8
Eosinophilic granulomatosis with polyangiitis (EGPA)	4	1.8
Addison's disease	3	1.4
Adrenal insufficiency	3	1.4
Angelman syndrome	3	1.4
Functional neurological disorder	3	1.4
Idiopathic intracranial hypertension	3	1.4
IgA nephropathy	3	1.4
Mast cell activation syndrome	3	1.4
Spinal muscular atrophy	3	1.4
Other conditions (n<3 each)	130	60.0

Over 150 distinct rare diseases were reported, illustrating the fragmented, long-tail nature of rare disease populations. Connective tissue, neuromuscular and neurological disorders dominated.

SAMPLE DEMOGRAPHICS

217 Respondents met criteria	**74%** Patients (n=161)	**26%** Caregivers (n=56)
80% Female	**40–49** Median age range	**150+** Distinct diseases





Medication sources (multiple allowed): community pharmacy 42%, local/regional hospital 39%, national specialist centre 34%, home delivery 21%. Most respondents (59%) self-collected; 23% relied on a caregiver.

CONCLUSIONS

Access is workable for many, but a meaningful minority face recurring logistical burden: 28% spend more than an hour collecting treatment and 12% spend more than two hours.

Home delivery is a clear opportunity: more than half of respondents either lack access or are unaware of it, yet 85% believe it would improve quality of life. The marked cross-country variation (p<0.001) points to structural healthcare system differences, with Italy (71%) and Spain (69%) showing the highest unmet need. Critically, respondents with home delivery reported significantly better wellbeing than those without (p=0.002), with nearly double the rate of positive wellbeing (47% vs 25%).

Experience and impact are uneven: variability in service reliability and indirect costs leaves a notable subset reporting delays, financial strain, and neutral/negative wellbeing effects — supporting the case for standardisation, clearer communication, and targeted support.

Additional significant findings: caregivers face longer collection times (p=0.007); financial burden increases with collection time (p<0.001). These findings reinforce the case for expanding home delivery as a lever to reduce both logistical and economic burden.

Key priorities: expand home delivery in Southern Europe; improve clinician education; strengthen psychological support; reduce supply disruptions. These have direct implications for orphan drug launch planning, EAP design and patient support services.

Limitations

Self-selected sample; cross-sectional design limits causal inference; unequal country sample sizes may reduce power for subgroup comparisons; translation of open-ended responses may introduce interpretation variability; some expected cell counts <5 in chi-squared tests.

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Disclosures
RA Huml, M Loiseau, N Gregory, K Duncalf, H Mossallem, S Raouf, W Hart are employed by, former employees of, or have been contracted by Sciensus Pharma Services Limited.
P Cusick, J Harscheid are employees of Rare Patient Voice, Towson, MD, USA.

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PAR6800526-B Implementation date 12/05/2026