

Rare disease as a shared societal responsibility: A patient advocate’s experience and the policy landscape for change

Amanda Hansson Hedblom¹, Erika Stariha², Gerdi Strydom¹ ¹Valid Insight, Macclesfield, United Kingdom; ²SATB2 Europe

Background and objective

≤5 in 10,000

people affected is the EU threshold for a rare disease¹

6,000+

rare diseases, which together affect...^{2,3}

27–36M

...people in the EU^{2,3}

Due to the rarity of these conditions, caregivers and patients often face a much greater burden than those with non-rare diseases when organising and funding their care.^{4,5} Clinical guidelines, specialist training, and referral pathways may be lacking if not initiated and developed by families and patient advocates.^{4,6,7}

The objective of this research was to evaluate the alignment between the lived caregiver burden in rare diseases and current European policy initiatives.

Methods

A mixed qualitative-descriptive approach was applied in three steps

1 Structured interview

- Caregiver and patient advocate Erika Stariha was interviewed on the 8th of June 2026, via Microsoft Teams
- She is the parent of a child with SATB2-associated syndrome and co-founder of SATB2 Europe. The family lives in Slovenia
- Unmet need themes were identified from the transcript and mapped to three phases of the care journey (pre-diagnosis, at diagnosis, post-diagnosis) plus one cross-cutting category

2 Targeted review

- A targeted review was conducted, focused on grey literature and policy documents
- The aim was to identify EU and UK initiatives relevant to the unmet need themes
- The original search was conducted in June 2026, and the status of each initiative was checked on the 30th of September, 2026

3 Mapping and rating

Each unmet need theme was mapped against each initiative on a four-level scale:

Direct

the initiative directly addresses the need

Partial

the coverage is tangential or incomplete

Weak

the initiative is relevant in principle, but does not materially address the need

No link

no link to the need

Overall rating per need: **Partially addressed** if a law, policy or funded programme rates Direct or Partial; otherwise **Largely unaddressed**. Time-limited links alone do not count for long-term funding.

Policy initiatives reviewed

Binding EU law
EHDS and genomics infrastructure (EU regulation)⁸
EU medicines framework⁹

Adopted policy, non-binding
Council Recommendation 2009 and national plans: non-binding EU recommendation (2009/C 151/02) that led to national rare disease plans¹⁰
EU mental health and carer policy¹¹
Global frameworks: WHO and UN resolutions, IRDiRC research goals and UDNI¹²

Funded programmes and infrastructure
ERNs: 24 virtual networks of expert centres for rare diseases¹³
JARDIN: EU-funded project embedding the ERNs in national health systems¹⁴
Orphanet and ORPHAcodes: knowledge base and coding system¹⁵
ERDERA: Horizon Europe research partnership, building on Solve-RD, including reanalysis of unsolved cases¹⁶
EU funding: EU4Health, MFF 2028–2034¹⁷

Not yet policy
EU Rare Disease Action Plan (called for, not yet proposed)¹⁸
EURORDIS: European Organisation for Rare Diseases¹⁹

Results

Unmet needs and how policy responds

6 of 9 needs partially addressed

3 of 9 largely unaddressed

From the interview · Step 1			From the review and mapping · Steps 2–3	
Unmet need		Lived caregiver burden	How current policy responds	Overall rating
Pre-diagnosis	1 Fragmented coordination and unequal access to diagnostics	It took seven years to obtain the SATB2 diagnosis, despite tests being available.	National plans, the ERNs, JARDIN, ERDERA and IRDiRC provide frameworks and goals for diagnostic coordination, but there is no mechanism at the point of care which coordinates laboratories and clinicians or guarantees equal access to sequencing.	Partially addressed
	2 No systematic reanalysis of undiagnosed patients	Whole exome sequencing became available in Slovenia in late 2016. As there was no system to identify undiagnosed patients and reanalyse their data, diagnosis was delayed until 2020.	ERDERA reanalyses data in research, and Orphanet codes for undiagnosed patients, but no health system is obliged to reanalyse data or re-invite undiagnosed patients on a regular cycle.	Partially addressed
	3 No psychological support during diagnostic uncertainty	The family had no psychological support during the years of uncertainty ahead of diagnosis.	While EURORDIS documents the burden, no initiative provides funds or requires psychological support for families of undiagnosed patients.	Largely unaddressed
At diagnosis	4 No standard handover to networks, community and families	Families often find specialist networks, patient organisations or other families with the same diagnosis on their own, for example, through social media.	EURORDIS and ERDERA offer networks, and the ERNs, JARDIN and Orphanet provide directories, but none is linked to the point of diagnosis. No mechanism ensures families are introduced to these networks or directories at diagnosis.	Largely unaddressed
	5 Inconsistent genetic counselling	The quality of genetic counselling varies. Staff shortages can lead to months-long delays between a test result and a diagnosis, and some results arrive by post with no accompanying explanation.	The ERNs, JARDIN and Orphanet give access to genetics expertise and expert-centre directories, and EURORDIS advocates for standards in how a diagnosis is communicated, but no initiative addresses counselling capacity, the delay before a counselled diagnosis, or results sent without explanation.	Partially addressed
Post-diagnosis	6 Caregiver as default coordinator; no functional expert referral	The caregiver becomes the care coordinator and the main source of information.	The ERNs and CPMS provide a route for expert consultation, but limitations include underutilisation by clinicians, limited funding, and the fact that many ultra-rare conditions have no expert centre.	Partially addressed
	7 Clinical practice guidelines absent or family-led	For many ultra-rare conditions, there is no expert centre and no clinical practice guideline. For SATB2-associated syndrome, families are driving the guideline work, which is labour-intensive and unpaid.	The ERNs develop guidelines for certain conditions, but most ultra-rare conditions still lack them.	Partially addressed
	8 Advocacy out of reach for families with the highest need	Effective advocacy requires language skills, professional skills, time, money, a stable family, and a sufficiently healthy child. The families with the greatest clinical need are least likely to have these.	EURORDIS training and EU carer policy support families, but none specifically address the barriers posed by language, skills, time, money, and health.	Partially addressed
All phases	9 Project-based funding; foundations pay for public services	EU funding is project-based, meaning it may build infrastructure, but then there is no long-term funding. Patient foundations pay for services that public systems should provide.	EU4Health funds the ERNs, Orphanet and JARDIN for fixed periods, but there is no permanent funding, and the main grants end in 2027.	Largely unaddressed

Coverage by initiative

Step 3 mapping: each initiative rated against each unmet need

Direct

Partial

Weak

No link

Phase	Unmet need	EHDS and genomics	Medicines framework	Council Rec. and national plans	Mental health and carer policy	Global frameworks	ERNs	JARDIN	ERDERA	Orphanet	EU funding	EU Action Plan (called for)	EURORDIS
Pre-diagnosis	1 Diagnostic coordination and access	Weak	–	Partial	–	Partial	Partial	Partial	Partial	–	–	Partial	–
	2 Reanalysis of undiagnosed patients	–	–	–	–	Partial	–	–	Direct	Direct	–	–	–
	3 Psychological support	–	–	–	Weak	Weak	Weak	–	–	–	–	Weak	Weak
At diagnosis	4 Handover to networks and families	–	–	–	–	–	–	–	Weak	–	–	–	Weak
	5 Genetic counselling	–	–	–	–	–	Partial	Partial	–	Partial	–	–	Weak
Post-diagnosis	6 Caregiver as coordinator	Partial	–	Partial	Weak	–	Partial	Partial	–	–	–	Partial	–
	7 Clinical guidelines	–	–	–	–	Weak	Partial	–	Weak	–	–	–	–
	8 Advocacy for highest-need families	–	Weak	–	Partial	–	–	Weak	–	–	–	Weak	Partial
All phases	9 Project-based funding	–	–	Weak	–	Weak	Weak	Partial	–	Weak	Partial	Partial	Weak

Conclusions

There is a gap between the ambitions of rare disease policy and the lived experience of caregivers. The largest unmet need remains for psychological support before diagnosis and in care coordination handover.

Europe has research programmes and data infrastructure that could help resolve undiagnosed cases, but there is no care-system mechanism to ensure that undiagnosed patients benefit from them.

The ERNs are structurally positioned to address most unmet needs through expert centres, the consultation platform, and the guideline mandate. However, sustained and sufficient EU-level funding is required to achieve this.

