Analysis of European National Policies and Programs to Advance Access to Care and Treatment

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Background

- The rare disease (RD) policy landscape is rapidly evolving, as evidenced by the increased international awareness and development of comprehensive policies/programs, precisely Rare Disease National Plans (NRDPs), to address access to care and treatment for rare disease patients globally.
- However, NRDPs vary considerably in terms of their content and focus, reflecting not only differences in national healthcare systems and infrastructure, supporting disease education and treatment, but also the influence of specific patient groups and clinical specialists.
- Despite progress in policy development, many RD patients still experience barriers in access to care and fewer than 10% receive disease-specific treatment.[1] Delayed diagnoses and limited access to resources preclude patients from receiving proper, timely care.
- As NRDPs evolve, patients and patient advocates continue to play an integral role in moving forward the implementation and adoption of the necessary programs for the better management of rare disease patients.

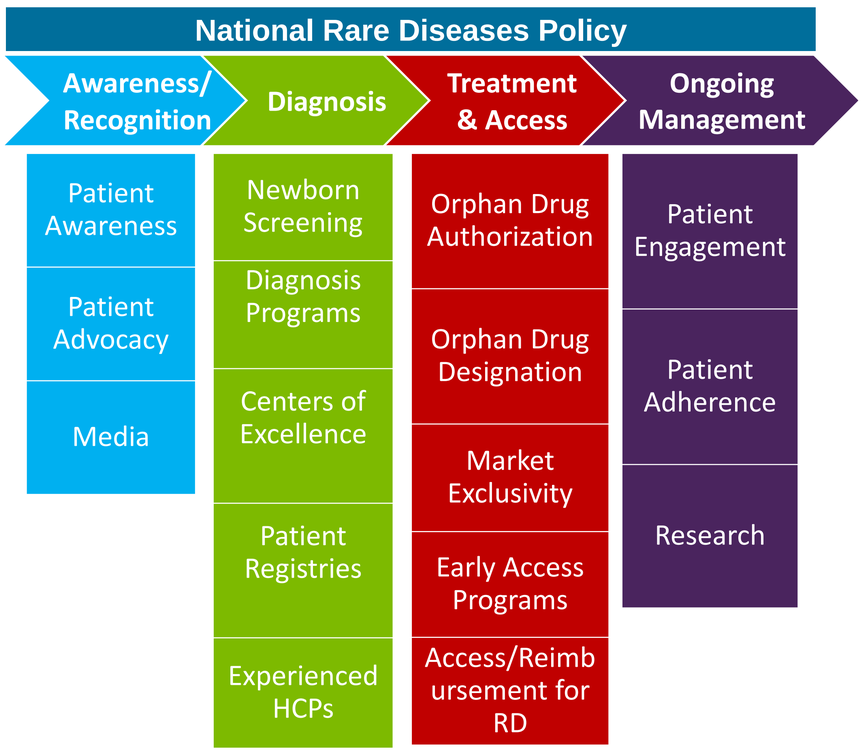
Aim

- The aims of the study were to:
  - Identify a framework to assist in understanding the global rare disease policy landscape
  - Determine key elements across diverse Rare Disease Programs and consider how these may relate to patient access to care and treatment through further policy development
- We analyze current policies and programs that could align with the key elements of comprehensive rare disease plans; these reflect the core needs of rare disease patients and translate into core programmatic aspects leading to improved care and treatment.

Methods

- Five countries, in Western and Eastern Europe, were selected for analysis.
- NRDPs were assessed using publicly available official documents and secondary research.
- Figure 1 outlines the key elements of NRDPs that were evaluated for each country based on the guidance published for rare disease policy.[2]
- Regional stakeholder insights were included and measured against current implementation of government policies and non-governmental organization programs.
- This study was limited in the scope of the countries considered and was not designed to assess the effect of specific policies on patient outcomes.

FIGURE 1: KEY POLICY COMPONENTS ALONG THE RARE DISEASE PATIENT JOURNEY



Results

- **National Rare Disease Policy (NRDP)** has been adopted with ongoing implementation in France, Germany and the UK. Bulgaria and Turkey have draft or full plans approved, but their plans have had limited implementation. Particularly in France, so far two NRDP have been implemented between 2005 and 2014, whereby the launch of a third is underway.
- **Newborn Screening Programs** could improve through **early diagnosis** and the timely and effective management of a fraction of rare diseases allowing for effective intervention early on. In all of the countries, screening programs for two or more rare conditions are generally performed at birth. More established screening programs are in place in Germany and the UK, whereas plans for a national funded early diagnosis program exist in Bulgaria.

Example: National program of neonatal screening for 14 conditions[3] exists in **Germany**.

- **Disease Registries** are important means of collecting disease, demographic, and treatment data. France is a best practice example with their Banque Nationale de Données Maladies Rares, a national organization collecting and organizing data from centres of excellence.[4] To help support the standardization and sharing of information across rare disease registries, the European Commission has initiated the establishment of an European Platform for Rare Disease Registries.[5] Thus, countries without well-developed such as Turkey, can participate at European level. Turkey has taken part in TREAT-NMD for neuromuscular disease and EUROCARE CF for cystic fibrosis registries.

TABLE 1: FINDINGS AGAINST THE SET OF POLICY COMPONENTS FOR RARE DISEASES

	DEVELOPED POLICIES			DEVELOPING POLICIES	
Rare Disease Policy Component	**France**	**Germany**	**UK**	**Bulgaria**	**Turkey**
Rare disease National Plans	✓	✓	✓	✓	X
Newborn Screening Programs	Moderate	Moderate	High	Moderate	Moderate
Patient Registries	High	Moderate	Moderate	Moderate	Moderate
Centers of Excellence	High	High	High	High	Moderate
Orphan Drug Designation	High	High	High	High	Moderate
Early Access Programs	High	High	High	Low	Moderate
Reimbursement for RD (designated funds)	Moderate	Moderate	High	Moderate	Low
Patient Engagement	High	High	High	High	Low
Research	High	High	High	Moderate	Moderate
Key Gaps	• Wide plan adopted and being implemented at the national level • Centralized national funding, not necessarily RD specific • Very good access to treatment through early access programs and distinct appraisal process for OD • Patient organizations play a central role			• NRDP adopted but with limited or stagnant implementation • Lack of designated funds for the coverage of RD treatments as well as early access programs • Increasing influence of patient organisations stimulates policy uptake	

Development Level Key: High Moderate Low

- **Centres of Excellence (COEs)** concentrate highly specialised and interdisciplinary healthcare professional skills allowing for improved disease management. In all countries, specialist care can be received but the number of COEs varies largely, with higher numbers in France, Germany and the UK as compared to those in Bulgaria and Turkey. Though, the situation in Bulgaria has improved, whereby 16 COEs were designated in 2016 according to nationally set criteria.[6]

Example: *~130 reference and 500 competence centers have been created throughout **France** since the second NRDP.[7] NHS **England** lists ~150 providers of highly specialized services & designated COEs are starting to be created (Birmingham Center for Rare Diseases).[8]*

- **Access to treatments** is facilitated through orphan drug designation, authorization, and early access programs. European Union Regulation (EC) No 141/2000, on orphan medicinal products applies to the UK, Germany, Bulgaria and France.[9] In addition, the UK and Germany have RD-specific attributions in the local reimbursement process – a health technology assessment (HTA) for Highly Specialised Technologies and orphan drug exemptions on the added benefit assessment, respectively.[10,11] In Bulgaria, special considerations are made for ODs during the HTA process, too.[12]

***Example: Scotland** designates a fund of £80 million per year for orphan drugs access.*[13]

- National campaigns are central to improving **patient's awareness and state support**. In the UK, France and Germany, established patient advocacy groups, both disease-specific and across diseases, organized and delivered a range of programs including education and awareness conferences and patient guides to RD research.[14] Their advocacy efforts spanned from support for legislative acts to receptions with government leaders.[15] In Bulgaria, where few other stakeholders are active, national RD patient networks implemented award-winning programs, educating and motivating the patient community. Efforts included a call for legislative action to support RD patient needs.[16]

Example: *In Bulgaria, patient advocacy groups played an integral part of launching 13 RD epidemiological registries.*[16]

- **Research** for RD has been relatively well funded in France, Germany and the UK, further strengthened by the EU Framework Programme for Research and Innovation, whereby between 2007 and 2013 over €620 million was granted to 120 collaborative RD research projects. There are fewer national initiatives to promote research and innovation for RD in Bulgaria and Turkey.

***Example: France** is funding 300 clinical research projects, including national and international collaborations.[17] In **Germany**, the Federal Ministry of Education and Research funds 12 research consortia, having invested €23+ million over the past three years with additional funding for initiatives such as the National Genome Research Network.*[10,17]

Conclusion

- Rare disease policies with well-developed patient engagement and support programs, excellent ongoing education and research, and targeted early diagnosis programs could improve patient access to therapies and care.
- Gaps exist between policy and practice. While a number of countries have regulations specific to rare disease and orphan drugs, implementation is limited and does not necessarily ensure access or care for patients.
- Furthermore, there has been ample progress in expanding neonatal screening programs to a higher number of rare diseases in most of the analyzed countries. Apart from France, limited progress has been achieved to ensure the comprehensive disease monitoring through patient registries. In contrast, only in two of the studied countries research and early access programs remain with limited attention to rare diseases.
- The role of patient organizations and advocacy groups has been instrumental in driving patient awareness and the prioritization of rare diseases on the national policy agenda, in countries with limited policy focus on rare diseases.
- The results of this survey provide a strategic framework that could act to structure the ongoing dialogue within and between countries. The results could underlie the efforts to define best practices in the management of rare diseases and harmonize cross-border efforts in improving patient care.
- Subsequent analyses are needed to assess the impact of policy on the implementation of actual programs and, ultimately, their effects on care.

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