

# New horizons for centres of expertise for rare diseases in Bulgaria

Eleonora Hristova<sup>1</sup>, Georgi Iskrov<sup>1</sup>, Rumen Stefanov<sup>1,2</sup>

<sup>1</sup>Department of Social Medicine and Public Health, Faculty of Public Health, Medical University of Plovdiv, Plovdiv, Bulgaria

<sup>2</sup>Institute for Rare Diseases, Plovdiv, Bulgaria

## ABSTRACT

**B**ulgaria was the second EU Member State and the first Eastern European country to adopt an official national plan for rare diseases in 2008. One of the plan's main priorities was to conduct a study on feasibility of reference centres for rare diseases in the country. The plan's funding was, however, greatly disrupted and this activity was never implemented.

Following the adoption of the EU Directive on the application of patients' rights in cross-border healthcare in 2011, Bulgarian health authorities started re-examining rare diseases national policy and formally adopted in 2014 a Ministerial Ordinance No.16 on the conditions and procedures for registration of rare diseases and designation of centres of expertise and reference networks for rare diseases.

This article aims to explore the evolution of the Bulgarian public health policy and practice on designating centres of expertise for rare diseases. The analysis explains the structure of the Commission on Rare Diseases, the development of the List of Rare Diseases, the designation of Centres of Expertise and the establishment of the National Registry for Rare Diseases in Bulgaria.

## KEYWORDS

rare diseases, health policy, centres of expertise, registries, Bulgaria

## CORRESPONDING AUTHOR:

Eleonora Hristova, MD  
Department of Social Medicine  
and Public Health  
Faculty of Public Health  
Medical University of Plovdiv  
15 A, Vasil Aprilov Blvd, 4002 Plovdiv, Bulgaria  
E-mail: eleonora.hristovaa@gmail.com

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## INTRODUCTION

Bulgaria was the second EU Member State after France and the first Eastern European country to adopt an official national plan for rare diseases in 2008. One of the plan's main priorities was to conduct a study on feasibility of reference centres for rare diseases in the country [1]. The plan's funding was, however, greatly disrupted and this activity was never implemented. Following the adoption of the EU Directive on the application of patients' rights in cross-border healthcare in 2011, Bulgarian health authorities started re-examining rare diseases national policy and formally adopted in 2014 a Ministerial Ordinance No.16 on the conditions and procedures for registration of rare diseases and designation of centres of expertise and reference networks for rare diseases [2].

Designating centres of expertise and establishing reference networks have been one of the pillars of rare disease policy at EU level. The scarce expertise and experience, as well as the lack of visibility and awareness of rare diseases have caused great inequalities in access to timely diagnosis, treatment and follow up of these patients across the different Member States. Patients' prolonged quest for adequate health care services for rare diseases has pushed the EU authorities to call for specific measures to designate centres of expertise at national level and to engage them into reference networks at European level [3-6].

This article aims to explore the evolution of the Bulgarian public health policy and practice on designating centres of expertise for rare diseases.

## MATERIAL AND METHODS

We performed a critical appraisal of the recently adopted Ordinance No.16 in regard to the EU Committee of Experts on Rare Diseases (EUCERD) recommendations, namely on quality criteria for centres of expertise for rare diseases in Member States (adopted in October 2011), on informed decisions based on the clinical added value of orphan medicinal products (adopted in September 2012), on European reference networks for rare diseases (adopted in January 2013), on rare disease patient registration and data collection (adopted in June 2013), and on the core indicators for rare diseases national plans and strategies (adopted in June 2013). Following our study's objective, an analytical framework was developed, comprising three overlaying issues: 1) to outline what is done and what needs to be done in the field of centres of expertise and registries for rare diseases in Bulgaria; 2) to synthesize important variables relevant to the rare disease national policy in Bulgaria; and 3) to unveil relationships between theory and practice.

## RESULTS AND DISCUSSION

Ministerial Ordinance No.16 was signed by the Bulgarian Minister of Health on July 30, 2014 and promulgated in State Gazette on August 12. This legislation was prepared by a Task Force at the Ministry of Health, established in 2013 and including health authorities, medical professionals and patient representatives. The Task Force benefited from the extensive work of previous task forces, as well as the current EU recommendations on rare diseases.

## COMMISSION ON RARE DISEASES

The Ordinance established the Commission on Rare Diseases at the Ministry of Health [2]. The Commission's structure and competences largely follow the model of the current European Commission Expert Group on Rare Diseases. Commission members are determined by the Minister of Health and include health authorities, medical professionals and patient representatives. The Commission has a mainly advisory role supporting national health authorities in field of rare disease policy. Commission's functions include adopting recommendations on disease dossiers for inclusion into the List of Rare Diseases and on applications for designation of centres of expertise and reference networks. The Commission monitors and evaluates activities of the designated centres of expertise and reference networks, as well as the activities by the National Registry of Rare Diseases. The Commission may provide recommendations on prevention, diagnosis, treatment, follow up and rehabilitation of rare diseases, as well as education and training of medical and non-medical professionals on rare diseases. The Commission collaborates with rare disease stakeholders at national, European and international level.

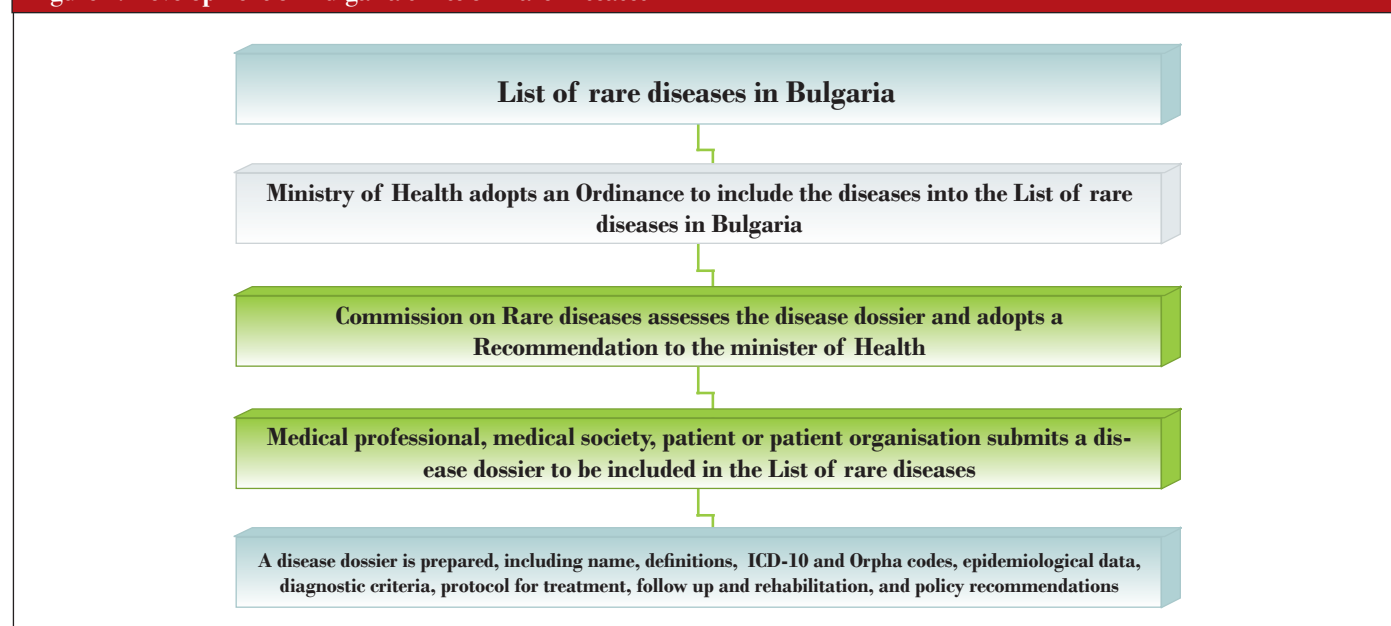
## LIST OF RARE DISEASES

The Ordinance laid down the grounds for a List of Rare Diseases [2]. Unlike Italy, where similar legislation was implemented along with a predefined list of rare disorders, the Bulgarian List will be established and updated on a current basis [7]. The Commission will examine the current list of disorders, whose outpatient medicinal treatment is covered by the mandatory health insurance, and will prepare an initial List of Rare Diseases by the second half of 2015. Bulgarian rare disease stakeholders can submit a dossier of a condition to be included in the List (Fig. 1).

The List of Rare Diseases itself is supposed to include disorders, for which expertise and experience are available in Bulgaria. The List will follow ICD-10 nomenclature. In case of a disorder lacking a specific ICD-10 code, Orpha codes will be applied. A disease dossier for inclusion should include name and definition, ICD-10 and Orpha codes, epidemiological data from both EU and Bulgaria, diagnostic criteria, protocol for treatment, follow up and rehabilitation, and policy recommendations. The Ordinance envisages the creation of a public database, containing all rare disease dossiers, which will serve as a reference portal for Bulgarian

rare disease stakeholders. The Commission is mandated to ensure scientific rigour and robustness of rare disease information that could be further used by the centres of expertise and reference networks.

**Figure 1. Development of Bulgaria's List of Rare Diseases**



### CENTRES OF EXPERTISE ON RARE DISEASES

Both inpatient and outpatient medical treatment facilities can apply for designation of expert centre for rare diseases (Fig. 2).

The Ordinance allows the possibility of two or more medical institutions to apply as a single centre of expertise established on a base of functional cooperation. This is important provision, as expert centres are mandated to provide diagnosis, treatment, follow up and rehabilitation of patients with rare diseases. Centres are explicitly required to bring together and to coordinate a multidisciplinary approach of a team of medical and non-medical specialists, in order to serve the specific medical, rehabilitation and palliative needs of rare disease patients and their families. The Ordinance put a substantial stress on the organisation of collaborations among medical treatment facilities to assure the continuity of care between childhood, adolescence and adulthood, as well as the continuity of care between all stages of the rare disease. Applying institutions are scrutinised to demonstrate high level of expertise and experience, documented by volume of referrals and second opinions, peer-reviewed publications, grants, teaching and training activities [6, 8-9]. Finally, all designated centres of expertise are required to participate in the National Registry for Rare Diseases.

**Figure 2. Designation of centres of expertise for rare diseases in Bulgaria**

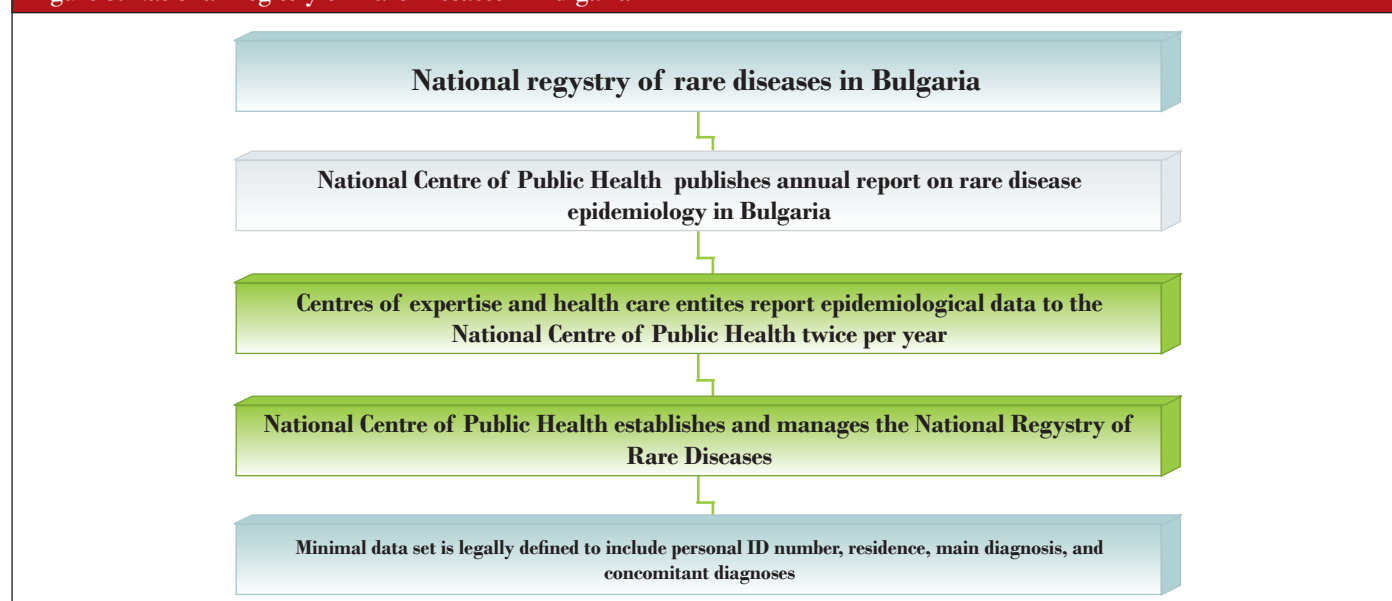


Period of designation is up to 5 years and may be renewed. Designated centres are expected to put in place quality of care control, including patient-reported outcomes. Centres are required to submit annual activity reports to the Commission, which performs an external evaluation at the end of the designation period.

### NATIONAL REGISTRY FOR RARE DISEASES

The final major provision of the Ordinance is the establishment of a National Registry for Rare Diseases (Fig. 3). The Registry will be managed and maintained by the Bulgarian National Centre for Public Health and Analyses (NCPHA) [2]. The minimum set of epidemiological data is synchronised with the European and national legislation on privacy protection [10]. All data is anonymised. All medical treatment facilities in Bulgaria, including designated centres of expertise, are mandated to submit epidemiological data to the Registry. Reported data should cover all rare conditions that are present in the List of Rare Diseases. Data will be updated twice per year. NCPHA is required to prepare and publish annual reports on the epidemiology of rare diseases in Bulgaria.

**Figure 3. National Registry of Rare Diseases in Bulgaria**



### BOTTLENECKS AND PROSPECTS IDENTIFIED

Designation of centres of expertise for rare diseases at national level is key to achieving an optimal use of the scarce knowledge, experience and resources for rare diseases [11]. The newly adopted Ordinance No.16 is expected to have an essential impact on the practical implementation of EU and national health policies. This legislation has a great potential to improve the quality and accessibility of health care services for patients with rare diseases and their families in Bulgaria. The official designation, financial support and ongoing evaluation of the centres of expertise will facilitate both patients and physicians in easily identifying and accessing specialised diagnosis, treatment and follow up. This optimised medical management should eventually lead to prolonged life expectancy and increased quality of life of rare disease patients in Bulgaria [12].

The most important challenge now is to ensure visibility and raise awareness among Bulgarian medical professionals and scientific societies about the opportunities offered by the Ordinance. The designation of centres of expertise mainly depends on willingness and motivation of the medical community to contribute for rare diseases. National health authorities need to consider implementing a package of incentives, including financial, for medical treatment facilities who are interested in expert centre designation and development of health care services for rare diseases [11]. Coordination of multidisciplinary teams and provision of continuous follow up should be supported by devoting additional resources to designated centres. Adopting a new national plan for rare disease in Bulgaria may address these important challenges and facilitate the implementation of the Ordinance.

### CONCLUSION

Designation of centres of expertise for rare diseases at national level is key to achieving an optimal use of the scarce knowledge, experience and resources for rare diseases. This process is a significant step for EU rare disease policy as well, as it lays down the basis for the next level of collaboration, the European reference networks for rare diseases. The newly adopted Ordinance No.16 has a great potential to improve the quality and accessibility of health care services for patients with rare diseases and their families in Bulgaria. The most important challenge now is to ensure visibility and raise awareness among Bulgarian medical professionals and scientific societies about the opportunities offered by the Ordinance. Coordination of multidisciplinary teams and provision of continuous follow up should be supported by devoting additional resources to designated centres. Adopting a new national plan for rare disease in Bulgaria may address these important challenges and facilitate the implementation of the Ordinance.

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