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Austrian National Action Plan for Rare Diseases

Executive Summary
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Current situation

Rare diseases are defined by their frequency in the whole population (prevalence). In the European Union, a disease is considered rare if its prevalence is less than 1:2000.

For the Austrian population, this means that up to 4,200 people may be affected by a certain disease for it to be considered „rare“. Typically, however, significantly fewer people suffer from a specific disorder. In many instances, only single cases have been described.

Even though the diseases themselves are rare, the spectrum of individual disorders is very wide. As of December 2014, „Orphanet“, the worldwide reference data base for rare diseases, lists 5,833 different entities. The European Commission estimates a total of up to 8,000 rare disorders in Europe.

Rare diseases are defined not only epidemiologically by their low prevalence, but also clinically as chronic, complex (i. e. affecting multiple organs or organ systems), life threatening and/or permanently debilitating conditions. This means that the term „rare disease“ comprises a wide spectrum of possible clinical appearances. But not only the individual clinical appearance and complexity of the respective disease poses a challenge for patients, relatives, physicians, and other health care professionals. It is the principle of rareness itself that causes a significant number of problems.

Challenges in the context of rare diseases

Multiple challenges in the field of rare diseases were identified by an empirical survey on the situation of rare disease patients in Austria commissioned by the Austrian Ministry of Health in 2011 (Voigtländer et al. 2012):

- Awareness of and common knowledge regarding rare diseases are generally lacking among health care professionals, in health care politics, and in the general population.
- Expertise und specialized institutions are not well known and/or not visible.
- Shortcomings in the coordination of clinical care (barriers of access to diagnostic and care facilities), deficits in the interface management and in the cooperation between in- and outpatient facilities exist.
- Current possibilities of documentation within the health care system are insufficient with respect to rare diseases. Due to the lack of a central case registration and data collection system, there is presently no valid information on the epidemiology of rare diseases in Austria. There are deficits in the establishment, as well as the long-term support of reliable central sources of information with regard to rare diseases.
- Definition of standards and quality assurance are frequently missing in diagnostics, clinical care, therapy, and rehabilitation.

- For the treatment of individual rare diseases, specifically developed therapeutics, so-called Orphan drugs, are available. However, these are often very expensive due to the high development costs for a comparably low number of patients.
- Deficits with respect to research and development, as well as diagnostics, therapy, and screening of rare diseases prevail (e. g. long latency periods before patients receive a clinically reliable diagnosis).
- Services and accomplishments of patient organizations are not sufficiently well known; their expertise is not sufficiently acknowledged and/or made use of.

Development and objectives of the national plan

The ultimate goal of the Austrian National Action Plan for Rare Diseases is to improve the lives of all rare disease patients – involving their families as well as their professional and social environment – irrespective of age, gender, extent of disability or socio-economic status.

The plan was developed by the National Coordination Centre for Rare Diseases CCRD (*Nationale Koordinationsstelle für seltene Erkrankungen, NKSE*) on behalf of the Austrian Ministry of Health, in cooperation with two advisory committees, the expert group on rare diseases (from 2014 advisory committee for rare diseases), and the strategic platform for rare diseases.

Starting points for the national plan were European requirements (e. g. recommendations, guidelines), the national empirical survey „Seltene Erkrankungen in Österreich“ („Rare diseases in Austria, Voigtländer et al 2012), the structured exchange with national experts, and current national developments such as the definition of health targets (*Rahmen-Gesundheitsziele*), the health care reform or the Child and Youth Health Strategy (*Kinder- und Jugendgesundheitsstrategie*).

Structure, contents, and key aspects

The Austrian National Action Plan for Rare Diseases comprises nine core areas (also referred to as „fields of action“ in the document), which take into account European recommendations, as well as national requirements:

- » 1: Mapping of rare diseases in the health care- and social system
- » 2: Improvement of medical/clinical care for rare disease patients
- » 3: Improvement of diagnostics of rare diseases
- » 4: Improvement of therapy and access to therapies for rare disease patients
- » 5: Promotion of research in the field of rare diseases
- » 6: Improvement of general knowledge with respect to and awareness of rare diseases
- » 7: Improvement of epidemiology in the context of rare diseases
- » 8: Constitution of permanent advisory bodies for rare diseases at the Ministry of Health
- » 9: Acknowledgement of the merits of patient organizations for rare diseases

The Austrian National Action Plan for Rare Diseases combines plan and strategy. For each field of action, objectives and according measures were defined based on the assessment of the current situation. Exemplary objectives are detailed below.

Rare diseases/Rare disease patients should be made more visible. To register rare disease cases, the national plan provides that a comprehensive epidemiological platform for patients with rare diseases in Austria is established. The development will be coordinated with current national and international activities in order to guarantee the interoperability with already existing systems. To be able to document the costs of medical services provided for rare disease patients, the national plan defines the introduction of a documentation/coding system for rare diseases as one of its primary goals.

With respect to health service provision, the national plan focuses on improved coordination of clinical care by means of the designation of specialized centers for rare diseases (centers of expertise), and their subsequent interconnectedness within Austria as well as within the relevant European Reference Networks (ERN). The national plan does not aim to establish new structures, but to concentrate existing expertise – meeting certain standards and quality criteria – in supra-regional centers. Intensified cooperation should add to a maximally efficient use of highly specialized expertise and resources, ultimately leading to an improvement of diagnostics, therapy, and clinical care of rare diseases. In addition, the national plan envisages the optimization of the continuous access to Orphan Drugs.

Rare disease patients should receive a clinically reliable diagnosis faster. The national plan postulates – in addition to the improvement of clinical care – the implementation of nationwide diagnostic standards and the concentration of diagnostic expertise.

To overcome the deficits in knowledge with respect to rare diseases, to promote the awareness of rare diseases and to improve access to objective and reliable information, the national plan defines specific measures, amongst others in the field of primary care. Primary care physicians play an outstanding role as „first contact within the health care system“. Patient safety and health competence should be strengthened by objective, quality assured and target group-specific information, thereby indirectly facilitating the access to the „best point of service“.

Patient organizations greatly contribute to the care of patients with rare diseases. The national plan therefore aims to make the accomplishments and services of patient organizations more visible and to acknowledge the experience and expertise of patients and their relatives, as well as to make use of this expertise in the context of national activities and developments. Representatives of patient organizations played a major role in the development of the national plan.

Implementation and monitoring

The implementation process of the National Action Plan, as well as its success will be monitored by specific indicators for each field of action. Indicators are defined in accordance with European developments and actions. Variety and complexity of the measures stated in the national plan require the definition of indicators in the course of the implementation process of each respective measure. This allows for the consideration of current developments and potentially necessary adaptations.