Interview: Joy Ladurner – Co-Director, NKSE, Austria

"Rare diseases affect – in total - a large population. However, individual rare diseases are very heterogeneous and people affected by these as well as their families are confronted with numerous challenges."

04.12.2017

Tags: Austria, NKSE, CCRD, Rare Disease, Healthcare, R&D

Joy Ladurner, co-director of the NKSE (National Coordination Center for Rare Diseases) in Austria, discusses the rationale behind Austria's prioritization of rare diseases and its leading role in the field.

Austria launched a national action plan for rare diseases, the NAP.se in 2015. NKSE has played an instrumental role in the development of this plan. Could you start us off by introducing NKSE's *raison d'etre*?

The National Coordination Center for Rare Diseases was established at Gesundheit Österreich GmbH (GÖG), the national public health institute, in 2011. We work for the Federal Ministry of Health and Women's Affairs and cooperate very closely with the Medical University of Vienna, e.g. with Orphanet, the internationally recognized database for rare diseases. One of our main responsibilities is to improve the provision of care for persons affected by rare diseases in Austria. We do this for instance by bringing together the different stakeholders in the field of rare diseases to coordinate national strategies and activities. Often NKSE drafts initial concepts for strategies, which are then presented for discussion to the stakeholders.

Stakeholders work in two committees, an expert committee and a strategic committee. The expert committee brings together a broad range of experts: clinicians of various medical specialties, patients, representatives of public institutions (ministries, social health insurance institutions and the regions), professional associations (e.g. Chamber of Physicians), hospital carriers, representatives from the field of ethics and from the pharmaceutical industry. The strategic committee includes the main payers of the Austrian health care system (representatives of the federal state, the regions and the social insurance institutions) as well as a representative of GÖG, as NKSE is located at GÖG.

The first task of NKSE was to describe the status of rare diseases in Austria. Colleagues of mine undertook a big effort in conducting a survey among all sorts of institutions and persons related to rare diseases and published a report in 2012. The second major piece of work was the compilation of the national action plan for rare diseases. It is based on 4 pillars: European requirements (recommendation), the study of 2012, the structured exchange with Austrian experts and other national agendas which were relevant at the time.

Why did Austria choose to prioritize the area of rare diseases?

Recognizing the importance of rare diseases did not start with the national action plan nor with establishing NKSE. Ministers of Health were already well aware of this topic and had already addressed it in a subcommittee of the Austrian Health Council (Oberster Sanitätsrat) between 2009 and 2010. In 2011 NKSE was established, demonstrating the Ministry of Health’s strong commitment to improve the care of persons with rare diseases in a structured and continuous way. Rare diseases affect – in total – a large population. However, individual rare diseases are very heterogeneous and people affected by these as well as their families are confronted with numerous challenges. A very common question we are often asked is “What is different when comparing (persons affected by) common and rare diseases? It is the notion of rareness and all the factors resulting thereof.

The national action plan is a joint publication of several ministries, which is quite unusual in Austria, showing the broad commitment for the topic. It is a collaboration of the Federal Ministry of Health, The Federal Ministry of Labour, Social Affairs and Consumer Protection and the Federal Ministry of Science, Research and Economy.

What has been achieved and what still needs to be done?

One of the key achievements of NKSE has definitely been the development of the national action plan. The process brought together the relevant actors in the field, who defined the priority areas for rare diseases in Austria. The action plan comprises nice core areas (field of action). It combines plan and strategy: for each field of action, objectives and according measures were defined based on the current situation (EU and nationally).

Deciding on a national strategy created a common understanding and commitment among the actors. Both the process and the result thereof were crucial. They promoted awareness and understanding for rare diseases and encouraged networking and communication.

[Featured_in]

A key target of NAP.se is to identify and bundle/concentrate clinical expertise in the field of rare diseases in Austria in designated centres of expertise. As a first step towards the achievement of this target NKSE undertook a mapping of clinical expertise in Austria in very close collaboration with Orphanet Austria. The result thereof is displayed in the Orphanet database. Austria has a lot of expertise for rare diseases, a large number of highly dedicated experts. The mapping enabled us to identify their expertise and to make it visible in Orphanet. The next steps were to find out which of the candidates claiming expertise could meet the criteria to become “centers of expertise” as defined in NAP.se. and to define the process to designate these centres. Quality criteria were defined (based on European recommendations, adapted to the Austrian setting) and possible candidates invited to undertake a self-assessment in order to find out whether could fulfil these. Candidates claiming to meet all criteria afterwards underwent a structured prioritization process in order to finally decide, which of the institutions/departments should be designated at what point in time.

Other achievements in connection with NAP.se-targets concern the following topics: coding of rare diseases (assessment of options, collaboration in the Rare Disease Joint Action), diagnostics (definition of quality criteria), following discussions on EU-level in the designated committees and transferring the results to the national environment i.e. integrating the results in national processes, continuous networking on a national level (in the committees mentioned above), maintenance of Orphanet, orphan medicinal products (performance of a survey), self help (e.g. development of a patient information card, improvement of websites, structured communication with social health insurance, etc.). Furthermore, numerous activities were undertaken by various stakeholders to promote awareness for rare diseases (presentations, publications, etc.).

Can you tell us a bit about these centers of expertise?

The main aim of these centers is to offer and demonstrate sustainable high-quality expertise in their relevant field of expertise. It is not possible to designate centers of expertise for every illness or illness group in Austria; We need to collaborate within Austria and on a European level, via European Reference Networks (ERN). Austria has a transparent process for the designation of centres of expertise, which was developed by NKSE (based on the concept outlined in NAP.se) and approved by all relevant actors (experts, funding agents, political level) in a lengthy process (involving 2 pilot-centres). So far, two centers of expertise have been designated; one in Vienna for pediatric oncology, and one for genetic skin diseases, specifically epidermolysis bullosa in Salzburg. Six more institutions/departments will hopefully be designated in 2018, the aim being to enable these to take part in the second call for European Reference Networks (ERN), planned to be about mid-2018.

There seems to be a lack of public funding for rare diseases. What does becoming a center of expertise imply for these departments?

Public funding for rare diseases will never be sufficient for those working in the area of rare diseases. The Federal state has invested a considerable amount of money so far by funding (the work of) NKSE. Moreover, a large number of health services are provided based on social health insurance services, whereby about 99% of the Austrian population is protected by social health insurance.

Being a centre of expertise means making (existing) expertise visible, in Austria and on an EU-level. It shows that centres meet clearly defined standards and have sustainable structures. Becoming a centre of expertise does not imply the creation of new/additional structures. The designation of centres of expertise is a highly innovative process. This makes it very difficult to forecast what a designation may imply, e.g. with regard to reputation, patient numbers, etc.

[related_story]

Sustainable care structures for the population require – most of all – the commitment of the funding agent/s. For entering the designation process an institution/department, next to demonstrating its clinical expertise, therefore also needs the approval of the three main actors in the Austrian health care system (the federal state; the social insurance institutions and the regions). Another key actor is the responsible hospital carrier. Three different types of centers of expertise exist according to NAP.se,; so far Austria has been focusing on the designation of B type centres.

Tell us a bit more about this dialogue with all the different stakeholders and the challenges in promoting (the work of) NKSE.

The committees NKSE works with involve experts of a considerable range of organizations. Bringing these together for discussion has increased the members’ i.e. the organizations’ general awareness for rare diseases as well as promoting mutual understanding (different agendas, perspectives, reasoning). Involving e.g. experts by own experience (patients) is crucial to learn about day to day challenges when dealing with a rare disease. Patients on the other hand learn about the framework conditions of social insurance, the regions, the industry or generally about political decision making. Communicating results of the work of the committees is challenging and cannot be done by the Ministry of Health and Women’s Affairs and/or by NKSE only. All stakeholders are needed.

What are your top priorities now and what role can Austria play in advancing the diagnoses, treatment and awareness of rare diseases in Europe and globally?

One of our immediate priorities is to designate national centers of expertise for rare diseases. Another is to integrate the Austrian expertise in the European Reference Networks. We believe that by linking expertise on rare diseases within Austria, across Europe and globally, patients can benefit most. Austrian experts can make a big contribution to this global achievement.

It is however important to be aware that most of the care of persons with rare diseases will, also in the future, not take place in centres of rare diseases (only), but in the care structures close to their place of residence (expertise should travel, not patients). This means that awareness for rare diseases has to be increased across all levels of care and also in the field of social services and research.

It will be vital to offer a perspective to persons who have not yet been diagnosed. This is intended by establishing so-called expertise clusters (type A) according to NAP.se.

Other priorities are to improve the documentation (registers, coding) of rare diseases and to provide sustainable structures for self-help organizations.

See more interviews