spectra 120

Rare diseases

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Federal government commitment to people with rare diseases

In Switzerland some 500 000 people suffer from a rare disease. Between 7 000 and 8 000 such diseases exist. There are so many of them that they account for a quarter of illnesses worldwide and are thus as frequent as one of the most widespread diseases. However, the lower the number of people suffering from a particular disease, the less we know about its causes, symptoms and treatment options.

A disease is deemed to be rare when fewer than five out of every 10 000 people suffer from it. But they are usually even much rarer than that. They are characterised by the low number of patients with the same rare disease and by their supraregional distribution, which complicates diagnosis and treatment and the conduct of studies. Very few physicians specialise in rare diseases and they tend to be geographically far apart. Access to good treatment and care facilities is not always self-evident.

Paradox: rare diseases are rare only in isolation; viewed as a group, they are widespread.

Patients often feel left to deal with their condition on their own. They usually do not know that, even though the number of people with the same disease is vanishingly small, many are faced with very similar problems in their everyday lives. Awareness of the metaphenomenon of rare diseases is lacking at all levels.

National Rare Disease Policy

To support the patients affected, their family members and caregivers, the Swiss government approved the National Rare Disease Policy (NRDP) on 15 October 2014 and the corresponding implementation plan on 13 May 2015. The aim had been to implement the measures set down in the NRDP by the end of 2017. This deadline proved to be too short as it took longer than planned to implement the key measure for designating the reference centres and care networks. Thanks to the National Rare Disease Coordination (kosek) body that was set up by the stakeholders in June 2017 and is now the lead agency in designating the reference centres and care networks, work can now proceed on the activities that depend on this step.

The main objectives of the National Rare Disease Policy are as follows:

- diagnosis within a reasonable timeframe;
- securing high-quality care throughout the entire course of the disease;
- supporting and strengthening the resources available to patients and their family members;
- securing socio-professional support for patients in administrative matters;
- improving general conditions for research on rare diseases and for providing information on current studies;
- international integration of Swiss institutions in the fields of research, diagnosis, treatment and care.

Low patient numbers make it difficult to document treatment methods and long-term effects, which in turn can cause problems with providing the evidence required for reimbursement purposes.

For the reasons stated, it was obvious from the outset that implementation of the policy would face a number of challenges. Generally speaking, the data available is scanty. We often know little about the frequency and spontaneous course of the individual diseases. Research results – particularly failures – are not always communicated in a transparent way. Low patient numbers make it difficult to document treatment methods and long-term effects, which in turn can cause problems with providing the evidence required for reimbursement purposes. Then there are the often complex realities of care provision, the shortage of specialist know-how that parallels the rarity of the particular disease, and the lack of clarity on responsibility for coordinating treatment and care.

Implementing the national policy

All stakeholders affected by the national policy (federal government, cantons, patients, physicians, health workers, researchers, representatives of social insurers, and others) had already been involved in drafting the policy and are playing a very active part in its implementation. Under the leadership of the FOPH, progress has therefore been achieved at many levels, thereby creating a foundation for the stated objective of improving care of the patients and family members affected.

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A key role will be played by the National Rare Disease Coordination body (kosek) set up in June 2017, which is the lead agency in designating the reference centres, care networks and information platforms. Many other measures laid down in the policy are directly or indirectly connected with these structures since the aim is to bundle scientific, medical and socio-professional expertise within them. The first pilot project scheduled for 2018 is in the planning phase.

Preliminary work on a Swiss Register of Rare Diseases is also far advanced. Such a register will greatly broaden our knowledge of rare diseases. In particular, it will reveal whether the figures derived to date from extrapolations are realistic. Furthermore, we will learn more about the spontaneous course of such diseases and the effects of treatment. The patients affected will receive information on international studies that are of relevance to them. In addition, we will gain new insights into appropriate care.

Questions regarding reimbursement of costs can prove particularly complex for patients with rare diseases. In children, the issue is whether the disease is deemed to be a birth defect. If it is, disability insurance covers the costs incurred by treatment up to the age of 20. The List of Birth Defects is also being revised and adapted in connection with the National Rare Disease Policy and the revision of the law on disability insurance. In the context of health insurance, the modifications that came into force in March 2017 at ordinance level and the availability of standardised application for reimbursement forms for physicians and hospitals are designed to clarify and streamline reimbursement procedures. The focus is on applications that require an individual case review by the health insurer and its independent medical advisory service.

The Swiss Catalogue of Learning Objectives for Undergraduate Medical Training, on which medical students can be tested from 2020 onwards, includes a section on rare diseases.

Of particular importance are the basic and further training and continuing education of physicians and other medical and non-medical professionals who have contact with the patients affected. The Swiss Catalogue of Learning Objectives for Undergraduate Medical Training, on which medical students can be tested from 2020 onwards, includes a section on rare diseases. Measures to raise awareness of the phenomenon of rare diseases are planned for 2018 and 2019. Individuals and institutions that can offer socio-professional support will

also be included. A key element here is cooperation between the stakeholders involved in implementing the policy. Particularly valuable is the experience-based knowledge that patients and self-help organisations can offer; this too will be incorporated into the process.

Implementation to be extended

Implementation work to date has shown that international networking on rare diseases is indispensable for Switzerland. This concerns research as well as diagnostics, treatment and care. The policy has therefore been extended to include a subproject designed to promote international networking of Swiss experts.

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In order to fulfil the federal government's mandate, work on implementing the National Rare Disease Policy will be continued and completed between 1 January 2018 and 31 December 2019. What is more, quality of care for rare diseases will continue to be an ongoing topic of discussion. However, the FOPH's current assessment is that, thanks to the foundation work scheduled for completion by the end of 2019 and the structures set up, the participating stakeholders can take over this task themselves and will no longer depend on the federal government for coordination.

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Genetic testing: when does the statutory health insurance (SHI) cover the costs, when does it not?

Genetic testing. Many rare diseases are of genetic origin, i.e. they can be inherited. Changes in an individual's genetic make-up can now be demonstrated in the blood. Genetic analyses have to be accompanied by genetic counselling. However, the statutory health insurance (SHI) does not cover the often considerable cost of genetic testing in all cases.

For people with rare diseases and for their families, it can be important to know whether the disease is due to a change in their genetic make-up. This link can now be demonstrated for many such conditions. But because genetic diagnosis can be time-consuming and therefore expensive, it is important to know whether the SHI covers the costs of such investigations.

The SHI is designed primarily to reimburse the costs of diagnosis and treatment in cases of illness. This framework was laid down by parliament. As a result, the SHI does not necessarily cover the cost of diagnostic tests that are not directly connected to specific treatment. A distinction has to be drawn between the following sets of circumstances:

1. A hereditary disease is suspected. In such cases the costs are covered if the genetic diagnostic tests have consequences for the further diagnosis or treatment. If the tests are conducted for another purpose, for instance to "name" an illness without this resulting in a change of treatment, or in connection with a patient's life planning, then the conditions for a disease risk ("therapeutic consequence") or an obligation on the part of health insurers to reimburse the costs are not given. In cases in which doubts exist, the physicians commissioning the tests must explain the disease risk and the consequences of the tests to the insurers' independent medical referees. These subsequently advise the insurers, who then decide whether or not to cover the costs. If the decision is negative, the insured person can take legal action against it.

2. The relatives of somebody with a rare disease want to know whether they, too, are at risk. The costs of such investigations are reimbursed only if they are explicitly listed in the Health Care Benefits Regulation (KLV). This is currently the case only for a number of hereditary forms of cancer and a single metabolic disease (porphyria). Extending the list to cover other rare diseases is under consideration. A disease is included in the list only if, as a result of the investigation, prophylactic measures can be taken that prevent or delay the onset of the disease or have a beneficial effect on its course.

3. A further possible objective of genetic testing is to determine whether relatives of people with a rare disease are healthy carriers of a genetic mutation (for instance, cystic fibrosis). Such tests can have a significant impact on family planning. They do not fall within the scope of the SHI, however, as they focus on life planning rather than on treatment or on beneficially affecting the course of a disease.

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At first hand

When the talk is of "rare diseases", we do not usually feel directly addressed: anything "rare" just affects others. But it is only the individual diseases that are rare. As a group, they are as common as diabetes. I too realised this only when we began drafting the National Rare Disease Policy.

The task reminded me of a number of encounters I have had in recent years, most tellingly with the father of a young boy the same age as my son. He told me how he and his wife had realised that their son was not developing like other children of his age. He spoke of the enormous efforts it took to have him referred to a specialist facility. And even then it was ages before a diagnosis was made. The doctors eventually informed them that their child was suffering from an extremely rare metabolic disorder. At a stroke the whole family's life was transformed into a daily obstacle race. The mother had to give up her job in order to devote herself entirely to looking after the boy. The frequent doctor's visits and treatment sessions took up a great deal of their time. Further stress was caused by uncertainty as to whether their medical insurance would cover the necessary treatments and by the lack of clarity regarding any support to which the family was entitled. Cases as this, which on closer scrutiny many of us can identify with, underscore the importance of the National Rare Disease Policy.

They also reveal the exceptionally large number of people and institutions – family members, physicians, health professionals, teachers, social workers, health insurance providers, patient advocacy organisations and many others – that play a role in the care of any patient suffering from a rare disease. I am therefore delighted that so many stakeholders are involved in implementing the policy. Their great level of commitment is impressive.

Such commitment reassures me that, when we finish implementing the National Rare Disease Policy, we will be able to greatly improve the life and everyday existence of the patients and family members affected.

Thomas Christen
Head of Health and Accident Insurance Directorate

National policy – participation in international networks

International. In order to remain involved internationally in research, diagnosis, treatment and management of rare diseases, Swiss centres of excellence rely on international partnerships. A new "International Networking" subproject in the context of the National Rare Disease Policy has been developed to help provide Swiss centres an anchor for their international integration.

On 9 March 2017, the first 24 European Reference Networks (ERNs) for rare diseases were formally launched in Vilnius. More than 300 hospitals and over 900 highly specialised teams are participating in these networks, opening a new era of EU-wide cooperation in the area of rare diseases. The aim is to help physicians and researchers to exchange data and expertise across borders in the EU, and to facilitate patients' access to specialised diagnosis and treatment of highly complex diseases. The ERNs were created on the basis of the EU Directive on the application of patients' rights in cross-border health care (2011/24/EU). Based on delegated legislative acts, the EU Commission has laid down the criteria for recognition of the ERNs: they must regroup at least 10 service providers from no less than 8 member states. Each institution must have been previously

certified as a reference centre by their state.

Switzerland seeks a connection

Unfortunately, it is currently impossible for competence centres from non-EU countries such as Switzerland to participate formally as partners in the European Reference Networks. Specialised Swiss centres that already have regular international contacts are following developments with great interest – but also with concern, as shown by the number of interventions at various events during the past year. Service providers and patient representatives fear that Switzerland will no more be involved in European developments, and they expect the Swiss Confederation to act.

New "International Networking" subproject

To address these difficulties, the two responsible federal agenicies, the Federal Office of Public Health FOPH and the State Secretariat for Education, Research and Innovation SERI, have set up a working group. Its purpose is to find ways to facilitate the involvement of Swiss partners in international and European collaborations in the context of the National Rare Disease Policy. Key stakeholders such as kosek, ProRaris, Orphanet and the Swiss Clinical Trial Organisation are also involved in the subproject. As a basis for analysing the situation and assessing the needs, a survey was first conducted among specialised institutions and experts in Switzerland. A second patient organisation survey is planned for spring 2018. The working group will then use the findings of the surveys in a targeted manner in order to gather better information on access to transnational networks (in Europe and elsewhere), and try to establish further contacts at an intergovernmental level. Relevant information will be provided to stakeholders in a suitable form. However, given the lack of legal basis for Switzerland in relation to EU mechanisms in the field of health, the hopes for formal access should not be set too high. Swiss research and health care institutions as well as patient organisations will therefore be even more dependent on developing and maintaining innovative forms of collaboration with European and international partners.

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Improving awareness of rare diseases

Interview. The subject of the 8th international Rare Disease Day in Switzerland this year was "I have a diagnosis. What happens now?" Jean-Marc Nuoffer, Chief Physician in Metabolic Analytics and head of the Interdisciplinary Metabolism Team at the Inselspital in Bern, told us what it means to go for a long time with no diagnosis and what is urgently needed to improve matters.

spectra: The theme of Rare Disease Day this year was "I have a diagnosis. What happens now?" Why is having a diagnosis particularly important for people who have a rare disease?

Jean-Marc Nuoffer: It is important for anyone who is ill to know what their disease is. The problem is that the diagnostic odyssey and associated uncertainties and fears can often last for years if you have a rare disease. Having a diagnosis puts an end to that, since it means doctors can start giving disease-specific advice and care and can avoid inappropriate treatments. A diagnosis is essential for specific treatment, enrolment in clinical trials and the development of new treatments. For patients, it means being able to share experiences with other people who have the same condition. A correct diagnosis of the cause of any disease is essential for the development of treatments. The rarer a disease is, the more important this becomes.

It can often take a long time until patients and their families find out what the disease is. What needs to be done to speed up the process of diagnosis?

There's no powerful lobby. We need to improve awareness of rare diseases among the public and health-care professionals. Diagnoses of idiopathic diseases – diseases with no tangible cause, in other words – should be avoided, and rare diseases should be evaluated by specialists. The decision on whether to carry out accredited genetic investigations should be taken by rare disease specialists and not by insurance companies.

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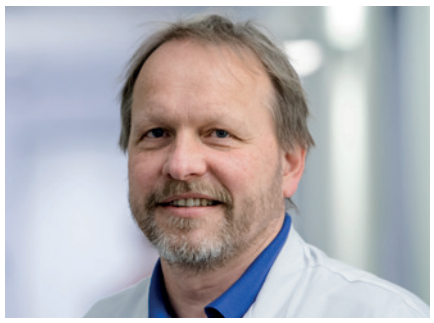
How well organised is psychosocial support for patients?

Support is often inadequate, and getting reimbursement for services is a battle – like it is for diagnosis. People with rare diseases often feel misunderstood; insurance companies will only pay for a large number of services in response to applications and appeals. The administrative work involved is enormous, regional rules vary and are not transparent. This is another challenge for everyone – on top of the problems associated with the disease itself and the burden that families have to bear. We need better coordination between care at centres and on-the-spot support.

Where could there still be gaps, and why?

New rare diseases are being described every year, and new treatments are constantly being developed. Our health-care system should be able to keep pace with that. The current legal framework, with its positive lists and administrative



PD Dr. med. Jean-Marc Nuoffer

routes, is not helpful in enabling it to do so. The National Rare Disease Policy needs to be fully implemented and re-evaluated. We need national reference centres with specific competences. However, in my opinion the financial side of the National Policy is still inadequate. Also, patients need to be given access to international clinical networks.

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