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EXPERT OPINION

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The diagnostic gap – an expert opinion

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Many individuals with rare diseases of different types experience gaps between symptoms, diagnosis and treatment when in need of care. This is known in patient groups but not always known in society. A common experience is that no one is knowledgeable about the rare disease so the patient must, at every visit to healthcare, describe the disease and its consequences, which is not easy if you are in need of care.

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1. What does the diagnostic gap mean?

The term 'diagnostic' might lead the readers to think of a diagnostic test. In this editorial, however, the focus is on the process where a patient meets the physician in order to get a correct diagnosis. When people start searching for a diagnosis for their medical (or other) problems, they encounter the gap. The first step is to find a doctor who can clarify the diagnosis. This may take several years, causing uncertainty and stress. According to the EurordisCare2 survey, 25% of patients are reported waiting between 5 and 30 years from the time of their first symptoms to a confirmatory diagnosis of their disease [1], which is showed in other studies as well [2]. Next, while waiting for a diagnosis, treatment is not available, or if it exists it may be the wrong one, causing frustration for the individual. Also, treatment is not free of charge; it may be costly for travel, medication and other individual needs. Personal economy is, thus, stressed by this. Because of the lack of a diagnosis, experiences such as humiliation in school, working life or healthcare are common [2]. To stand up for your rights as a patient or citizen is not easy in this situation [3]. Parents of a child who has a rare diagnosis need support for themselves and the child in order to manage their daily lives [4]. Sometimes different caregivers do not communicate with each other and that also adds to the difficulties of the patient. If there is a patient organization to contact, option is to meet others in the same situation and get personal support [5].

So, three reasons can be seen to cause this gap in the society: there is a lack of diagnosis, a lack of knowledge about rare diagnoses and their consequences and a lack of communication between professionals.

2. What can be done about the gap?

The first possible solution is to raise the issue of overall knowledge about rare diseases in society as well as in the healthcare sector [1,4]. As part of their strategic partnership, EURORDIS and NORD share a common vision to empower patient advocates with rare diseases worldwide. The purpose of this is to raise awareness in the scientific community, which includes researchers, pharmaceutical companies and regulators about the important issues surrounding rare diseases [5]. This is crucial and should be a continuing effort in educational systems. All students and healthcare professionals must be included, since rare diseases often have multifaceted problems [6]. Here are two examples: If your shoulder joints are hypermobile

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and can easily dislocate, you cannot walk with crutches if you have a fractured leg in a cast. And when you have cognitive problems and need dental care but do not open your mouth because you are afraid and have been mistreated as a child, you might need to sleep with anesthesia during treatment. These are just some short descriptions of the complexity of problems in rare patient groups.

There is a need for more research into the implementation of research results in the development of care which is a much-required procedure [7]. In future, patients will demand to participate as partners in their care much more than today. To make this possible, it is necessary to change the thinking of both healthcare professionals and patients. This is already underway but it is just the beginning. Patients design their own tests of the drug they are using, they learn how to manage their dialysis procedure themselves and they construct databases to evaluate their treatment [8,9]. The structure today is that healthcare professionals design how to deliver care to the patient as the receiver. In the near future, the patient wants a partnership in design, shared responsibility and evaluation of care. Better standards of care are essential to construct, and if care for the rare diseases is improving, the more common diseases will have more room in healthcare [10].

Internet communities have grown in the past few years and many patients are active in them. Groups around common interests are formed and people with the same area of interest can discuss things. Here, the patient organizations have done a lot of important accomplishments with educational materials and meetings between patients and professionals. The communities have several advantages. People ask questions, respond to each other, share tips about how they have solved similar problems and so on. It is one way to get information out to others and discuss worries and other such emotions and a way to empower the patients to take up their focus in a wider network which is a good point. Since rare diseases are chronic, are difficult to manage and can be disregarded by research, different ways to empower these individuals are necessary to construct [11]. According to the World Bank, empowerment is a process of increasing the capacity of individuals or groups to make choices and to transform those choices into desired actions and outcomes [12].

The primary aim with International Classification of Diseases is to make it possible to classify and statistically describe diseases and other health problems for epidemiology, health management and clinical purposes [13]. Besides traditional diagnoses, the classification must embrace a spectrum of symptoms, unnatural finds, difficulties and social aspects.

3. What is needed by research?

One area that must be more active is the promotion of support for research into rare diseases. Looking at the research investments today, the diseases that are common in the population, such as diabetes, stroke and dementia, are funded. This is understandable when you look at the possibility for everyone to get these diseases. But most of the rare diseases are inherited and 'if you do not have it from birth you will not get it'. This is one fact behind the distribution of funding research in healthcare. If you are a researcher and want to focus on a rare disease, to have your research funded will be very difficult. Also, finding other researchers that you can work with is difficult because they know about the difficulties to get this type of research funded. Therefore, even though they may be interested, professionals choose to engage in more common diseases where funding is available.

In order to limit the diagnostic gap, there are several ways to act. Research into rare diseases ought to be highly prioritized. Estimations are made that the figures of rare diseases are around 6500 [14]. Population studies are needed and then it would be obvious that rare diseases are not rare in numbers. When you look at any country, you will find a great many affected people in the population. Every disease might be rare but, when you take all diseases into account, you will find more affected individuals than you might think.

Another important factor is that longitudinal studies into rare diseases are lacking. Many studies are cross-sectional and results show the situation at the present time, but say nothing about the next 5 – 10 years when thinking of chronic pain, disability and such. For delivering a prognosis to a patient in healthcare, this is not sufficient. The patient often wants to discuss with the doctor how to manage symptoms in daily life but, since no research is found on that, the doctor cannot give the patient the correct advice. Here, the family is an important party, needing information to be able to support the suffering person [15]. Patient-oriented outcomes are important to focus on in clinical trials. However, this type of research may be difficult to arrange but is surely possible with international cooperation. That way many patients can participate in research.

4. How about the future?

One weakness is that many rare disease organizations are not active in research projects. The reason for this is that they are never asked to participate. A logistical and financial problem is that people are living in different parts of the country. Most groups do welcome research, but better funding for research with a focus on rare diseases is crucial.

The ultimate goal for people with rare diseases in general is to be able to get a diagnosis when they need it. They know what foot they are standing on when the diagnosis is clear and they can ask for treatment, when it exists. But as previously mentioned, patients want to be more than just receiving care: they want to be partners. And to make that possible, there is a need for a change of views and attitudes, both in patients and in personnel. If so, patients want to be involved in designing, planning, implementing and evaluating their care. This collective empowerment will create new solutions and structures that will add value not only to patients but also to professions and society.

The care needs to be correct and one way is to construct quality registries for the rare diseases so that evaluation of care is possible. When a patient has the diagnosis, the next step is to manage daily life with that information. Here is a magic threshold. If there is no treatment or program, can the patient and the family cope as well as trust healthcare? Most rare diseases have no program for their care so the professionals have to start from zero and thus risk delivering the wrong treatment. Guidelines for care built on patient experiences can be of help.

The political policy must change so that the rare diseases are considered like any other disease. An expert center with professionals that can work together around the patient is absolutely necessary and that is supported by all EU countries [5]. If you take proper care of the rare diseases, there will be room for other patients with more common diseases in healthcare.

The rare diseases need research that is designed to find the reason behind the disease, to examine the difficulties that are experienced, to try out and test treatment opportunities and to evaluate individual experiences of daily life. A big challenge is to find the groups that can participate in research and that is possible with international cooperation. In many countries, patient groups are established, but in order to start research a setting must be established together with the groups. It is also important to see that these patient groups must be compensated the same as other groups in clinical trials. Just because their interest and focus is voluntary, it does not mean they can participate without costs.

A lot of effort and funding is directed to research on molecules and genes. This is, of course, a predisposition to drug development, but since many rare groups are never going to have drug treatment, they are left out. Quality registers may be the way forward to get information in healthcare for the professionals so they can be of help to the patients. A weakness is that, if a register is to become effective, there must be an understanding and commitment in all healthcare professionals to participate with patient data.

When the rare diseases are better known in healthcare, management of the patients might reach to a higher standard. To get there, the research into rare diseases must be published, evaluated and also implemented in care. Education about how to implement the research is required to make it function in healthcare. The leaders in healthcare have a responsibility to support these efforts, but all professionals must take it on as well. Then, quality care is possible even for patient groups with rare diseases.

Declaration of interest

The author has no relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript. This includes employment, consultancies, honoraria, stock ownership or options, expert testimony, grants or patents received or pending or royalties.

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