Rare disease registries: an initiative to establish vasculitis registry in Poland

Presently, in the majority of common diseases, therapeutic interventions are driven by the results of randomized controlled clinical trials. However, such an approach is almost impossible in diseases with very low prevalence. Yet, together, these rare diseases (less than 1/2000) pose a substantial health problem. This issue has been recognized by the European Union, which, in 2009, established the European Union Committee of Experts on Rare Diseases (EUCERD). The so called Rare Diseases Registries (RDRs) provide a major tool to gain more precise knowledge about the epidemiology of these diseases and to perform therapeutic research. Recommendations to create such registries have been recently published by the EUCERD.[1]

Although it is commonly perceived that the majority of rare diseases have a genetic background, many of them are acquired. Vasculitides belong to the latter group. Their prevalence is estimated at 1–9/100,000 and the annual incidence at about 20 patients per million.[2] Thus, in Poland, we can expect about 750 new cases every year. Vasculitides are a heterogeneous group of diseases with unknown etiology and the clinical spectrum ranging from life-threatening systemic involvement to minor isolated skin changes. Nomenclature and definitions of systemic vasculitides have been recently updated by the International Chapel Hill Consensus Conference.[3]

With time, updates will be necessary as our knowledge continues to expand. Owing to the rarity of these syndromes, a specialist expertise is required to establish diagnosis and institute the most effective available treatment. This resulted in an international initiative to develop new diagnostic and classification criteria.[4] Simultaneously, there is an unmet need to identify the subsets of patients at risk for relapses, more severe course of the disease, grumbling disease, end-stage organ failure, or infectious complications. Recently, a study by Lyons et al.[5] has pointed to the genetic differences between 2 types of antineutrophil cytoplasmic antibody-associated vasculitis (AAV), while the cluster analysis of a large cohort of AAV patients identified several clinically discrete subsets of AAV.[6]

To deal with these problems, large patient databases in the form of RDRs are definitely required. RDRs will also allow to identify specific biomarkers and prognostic factors to classify and predict the future course of different types of vasculitides and, especially, effective treatment schemes. Such registries already exist in Europe (e.g., French Vasculitis Study Group) and United States (Vasculitis Clinical Research Consortium).

The 2nd Department of Internal Medicine of the Jagiellonian University Medical College in Kraków, Poland, has been involved in research and management of vasculitis for many years now. We participate in one of the clinical trials of the European Vasculitis Society (EUVAS) and American College of Rheumatology/European League against Rheumatism study of Diagnostic and Classification Criteria in Vasculitis (DCVAS). We also serve as an informal regional vasculitis reference center covering the population of over 3 million people. Our mission is to provide high-quality clinical care together with scientific contribution to the field of vasculitis research. For these reasons, we have established a local vasculitis study group, called the Krakow Vasculitis Study Group (KRAKVAS). One of its major tasks is to set up a large vasculitis database for clinical and research purposes, which can also serve as the quality-of-care registry according to the European guidelines for RDRs. To this end, we have developed a web-based electronic database for continuous data storage and analysis (FIGURE). The software enables prospective central collection of anonymized clinical data with the local storage of patient-identifiable information. The project has been approved by the Jagiellonian University Ethics Committee. The registry, in which prevalent and incident cases of systemic vasculitides will be recruited, consist of 2 parts: one with a core data set with basic demographic and disease-related variables and the other with disease assessment scores (Birmingham Vasculitis Activity Score, Vasculitis Damage Index, and 36-Item Short Form Health Survey), data on immunosuppressive treatment, comorbidities, and mortality in a longitudinal follow-up section. Prospective data collection will be coupled with

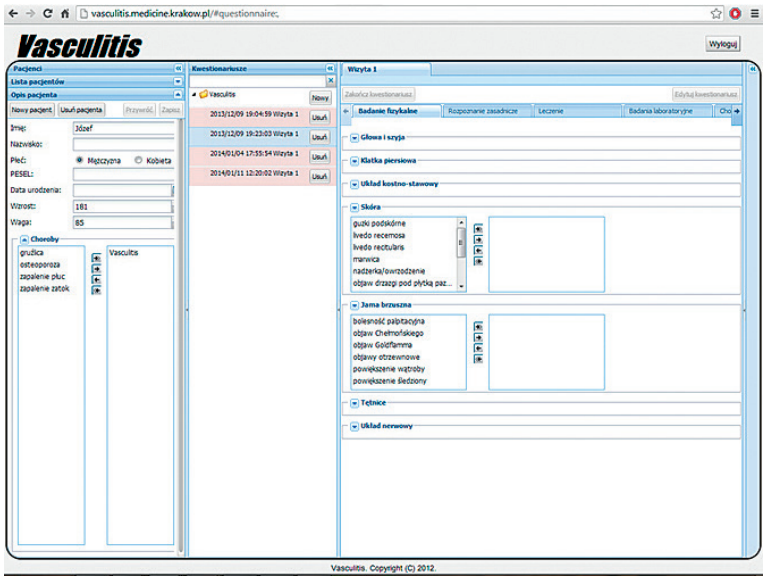

FIGURE Interface of the vasculitis database; the left-hand side of the menu: a searchable listbox of patients whose data have been entered into the database; the right-hand side of the menu: symptoms, laboratory test results, treatment, and history

DNA, plasma, serum, urine, and tissue samples biobanking. Efforts have been undertaken to secure the safety of data and multilevel access to the registry.

We hope that this registry will be a starting point for establishing a country-wide vasculitis registry in Poland through the cooperation with other national centers with experience in vasculitis management. Data collected anywhere in the country will be entered using a web-based application and stored in an anonymized format in the central database. Each participating center would have exclusive access to their data, and its common use for research purposes will be regulated by the formal agreement signed by each participating center. Such examples of successful cooperation at the national level are the Czech Vasculitis Registry and UK and Ireland Vasculitis Registry (UKIVAS), which so far have enrolled over 500 patients with systemic vasculitis each. The variables that will be recorded should be compatible with those existing datasets as this would allow to merge the collected data. There is an ongoing effort to develop a large international vasculitis registry and Poland should actively participate in this initiative. As already mentioned, such a large dataset will facilitate large-scale epidemiological, genetic, and therapeutic studies. It will also provide a means of linking data on unlicensed therapies, e.g., novel biologics.

In a long-term perspective, we can envision coupling of the information about history and course of the disease with artificial neural network attempting to plan and optimize treatment as well as predict future outcome of patients with vasculitis syndromes.

In a few weeks, we will be launching a website on vasculitis both for patients and health care professionals (www.zapalenianaczyn.pl). Thus, both groups will get a possibility to directly contact medical consultants and to join patient support groups, which may help them adjust to living with a chronic health problem. On the other hand, physicians and other health care professionals not dealing with vasculitides in their everyday practice will gain access to a valuable source of professional information.

In summary, our center has developed the local online electronic vasculitis database for data storage and analysis, which may improve diagnosis and treatment of various vasculitis syndromes. Its main future goal is to establish a platform for cooperation with other centers at the national level leading to the development of the national vasculitis registry.

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Conflict of interest The authors declare no conflict of interest.

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