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# Rare Diseases and Orphan Drugs

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## Patients without diagnosis: A profile

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A rare disease is defined as a disease that affects less than 1 in 2,000 people in Europe. In Germany, about 4 million individuals have a rare disease, which therefore represents a major public health issue, especially for the patients without a diagnosis. The demand for experts in rare diseases is correspondingly large, with insufficient points of contact for affected patients. To address this unmet need and improve the management of patients without diagnosis, the Center for Rare Diseases (ZSEB) at the University Hospital of Bonn founded a subunit, the “Interdisciplinary Competence Unit for Patients without Diagnosis” (InterPoD) in 2012. Since 2014, we have investigated the medical history of 242 patients with no established diagnosis and collected detailed information about their somatic, psychological and social aspects of health by using a standardized questionnaire. The aim of the study is to characterize the patients’ collective data and retrospectively evaluating their data to create a patient profile. We determined the number and duration of medical assessments and investigations of patients without diagnosis before our evaluation through the InterPoD. We have also focused on determining the common accompanying symptoms or illnesses and have further investigated the most affected organ systems. Furthermore, we discuss how an unconfirmed diagnosis of a disease affects the patients’ social wellbeing and daily life with regard to symptoms of depressive disorders and anxiety syndromes.

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