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Letter to the Editor

Improving the Diagnosis and Treatment of Mental Health Problems of People Living with Rare Diseases Requires Comprehensive Considerations

Josef Finsterer
Department of Neurology, Neurology & Neurophysiology Center, Vienna, Austria

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Corresponding Author: **Josef Finsterer**

We were interested to read the article by Richardson *et al*. about a study conducted by a panel of experts representing various stakeholders that looked at changes in insurance plan design or coverage options to improve the diagnosis and treatment of mental health problems in people with rare diseases [1]. It has been noted that people with rare diseases face particular mental health challenges because their conditions are not well understood, may be misdiagnosed, have limited treatment options, are economically burdened by increased medical needs, are dependent on caregivers, and are subject to work disruption [1]. The combination of these factors, as well as health insurance coverage, create a unique mental health situation for PLWRD and make it necessary to prioritize mental health treatment for this patient population [1]. The study is excellent, but some points should be discussed.

The first point is that mental health in PLWRD is highly dependent on the type of rare disease a PLWRD suffers from. Patients with rare hereditary neuropathies may simply suffer from sensory, autonomic or motor problems without being severely disabled, and therefore be able to lead an almost normal life without severe mental impairment. In contrast, patients with a rare form of Leigh syndrome may have a rapidly progressive course accompanied by a variety of medical problems that can increase rapidly, multiply exponentially and lead to severe health problems or even early death. In addition, patients carrying a pathogenic *L2HGDH* variant may have epilepsy, dysarthria and developmental delays, but experience no psychological or psychiatric problems due to their disease and have an almost normal life course with adequate education and vocational training.

The second point is that the frequency and severity of mental health problems is highly dependent on whether or not the brain is involved in the underlying rare disease. PLWRD with organic cerebral disease are more likely to have a psychiatric or psychological complication than patients without cerebral involvement in the rare disease. Therefore, depending on their socialization and education, patients with intact, unimpaired brain function and normal cognition may more easily develop coping strategies and establish a supportive social or medical network than patients with organic cerebral involvement. Cerebral disorders that are often associated with depression, anxiety, impaired drive, insomnia, obsessive-compulsive disorder or psychosis include epilepsy, demyelinating diseases, vascular complications and neurodegeneration.

The third point is that the causes of mental health problems in PLWRD are very diverse. Mental health problems may be due to stigmatization, inadequate coping strategies, years of misdiagnosis of the rare disease, lack of effective treatment for the underlying disease, social withdrawal, financial problems, uncertainty about the future, encephalopathy due to the underlying disease, dependence on parents, family members, caregivers and the healthcare system, unemployment, daily unpredictability of physical health, or lack of social networks. Therefore, diagnosis, therapeutic management and costs can be highly dependent on the underlying cause.

The fourth point is that the extent and nature of the mental health problems also depends on the current medication, either for the underlying rare disease or the secondary somatic, psychiatric or psychological complications. It must also be considered that often it is not the primary rare disease but a comorbidity that may be responsible for the mental health problem.

In summary, this interesting study has limitations that relativize the results and their interpretation. Addressing these limitations could strengthen the conclusions and support the message of the study. Improving mental health in PLWRD requires extensive thought and political will.

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