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Rare, but not insignificant

INVITERT KOMMENTAR

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International cooperation is crucial for the Norwegian health service to provide the most up-to-date and effective treatments for patients with rare diagnoses.

This edition of the Journal of the Norwegian Medical Association includes a study by Amland et al. on the incidence, clinical manifestations and treatment outcomes of Cushing's disease in Western Norway Regional Health Authority (1). The authors report an estimated annual incidence of 3.0 per million in the period 2010–22, which is higher than previously reported.

The study population is small, and statistical significance is clearly lacking, which is often the case in research on rare conditions. The challenges of small sample sizes are also highlighted in the Norwegian Ministry of Health and Care Services' *National Strategy for Rare Diagnoses* from 2021 (2). In the foreword, then Minister of Health Bent Høie noted that Norway is a small country with relatively few patients with each diagnosis and only a handful of specialists and researchers who are well acquainted with these conditions. He further stated that international cooperation is therefore needed on everything from diagnostics and treatment to research and innovation. The strategy's focus is on the disease-specific European Reference Networks (ERNs).

Through European cooperation, the ERNs aim to help ensure access to high-quality diagnostics and treatment for patients with rare and complex conditions, while also promoting knowledge-sharing among participating centres. Norwegian health authorities have decided that Norway will participate in the ERNs (3), and specialist

groups in Norway are affiliated with several networks at various levels. Norway is an associated partner in the ERN for endocrinology, but not a full member. The specialists from the university hospitals in Bergen and Oslo who currently represent Norway contribute at the national level by sharing acquired knowledge through regular focus meetings on specific issues, which in turn benefits the patients. This activity is in line with the national strategy, which proposes that ERNs serve as a model for national cooperation within rare diseases in Norway, thereby helping to increase knowledge.

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The need for a greater focus on rare diseases is clearly illustrated in the article on Cushing's disease, which describes challenges at every stage, from suspected diagnosis to treating relapses. In addition to the medical interventions, patients often endure long journeys and stays at tertiary centres far from home. Long distances and extended absences from home can have a major impact on a patient's family life, education and employment. Some of these challenges are country-specific and highlight the need for local and national studies.

Although national cooperation can facilitate knowledge-sharing and reduce unwarranted variation in health care in Norway, increasing the empirical base from two to four patients annually is not necessarily sufficient to successfully monitor the efficacy of new methods or the benefits of new diagnostic algorithms. In order for doctors in Norway to more effectively acquire the knowledge needed to give all patients the quality of health care they deserve, closer cooperation with major European centres through ERNs is essential. Valuable cooperation can include everything from joint studies and registries to international multidisciplinary team meetings, where Norway's specialists also share their results and experiences. Through systematic and thorough follow-up, outcomes from small clinical environments like those in Norway can gain international significance. This is clearly demonstrated in the article by Amland et al. One of the most advanced treatments for Cushing's disease, Gamma Knife radiosurgery, showed a surprisingly limited effect in their material compared with a previous multicentre study (4). This is an important finding that should be considered alongside follow-up and registry studies from clinical practice in other countries.

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In total, rare diseases (defined as conditions affecting fewer than 5 people in a population of 10,000) are not that rare (5). It is estimated that between 30,000 and 100,000 people in Norway (equivalent to the population of a medium-sized Norwegian town) are living with one of the 6000 to 8000 rare diagnoses registered in Orphanet (6). These individuals deserve diagnostic evaluation and follow-up based on up-to-date knowledge, just like any other patient in Norway, alongside continuous efforts to address knowledge gaps through international cooperation. Let us therefore hope that

health authorities continue to facilitate further research originating from environments such as the endocrinology department in Bergen. The recommendations outlined in the 2021 strategy must be followed up further to ensure that Norwegian specialists are represented in all ERNs, preferably as full members.

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