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Letter to the medical community

The Imperative of Publishing Data on Rare Diseases like PANS-H63D-Multisystemic Instability Syndrome

With this letter, we would like to express our opinion on the groundbreaking study recently conducted by basic scientists describing the multisystemic PANS-H63D instability syndrome. This extremely rare systemic disorder is manifested by the coincidence of PANS (pediatric acute neuropsychiatric syndrome), H63D syndrome, and dysautonomias in adulthood due to secondary mitochondriopathies caused by PANS and NTBI (the critical substance in H63D syndrome). The urgent need to disseminate this information is clear evidence of the widespread disregard for rare or "orphan" diseases in clinical medicine and medical science.

The Detrimental Impact of Scientific Neglect

Rare diseases often struggle to move beyond the stage of case reports or case series in scientific literature. This unfortunate reality is the result of a lack of interest among researchers, scarcity of research funding, and an insufficient market to attract pharmaceutical companies. However, this creates a vicious cycle: the less attention a condition receives, the fewer resources are allocated to study it, leading to a profound lack of understanding and therapeutic options.

Early Recognition and Cultural Relevance

It is worth mentioning that a syndrome (Oshtoran Syndrome) bearing striking similarities to the recently described PANS-H63D-Multisystemic Instability Syndrome was reported by Dr. Zafarian years ago (doi.org/10.5281/zenodo.7109840). Remarkably, despite its limited initial documentation, Dr. Zafarian's findings have percolated into popular culture. This signifies that although the medical community was slow to recognize its relevance, the phenomenon

resonated on a societal level. Importantly, the H63D Research Consortium has now undertaken the task to give this newly described systemic syndrome the attention it deserves. They are mobilizing both logistical and personnel resources to drive research on this important subject.

The Role of Scholarly Publishing

In such a landscape, it becomes imperative that whatever data is collected should meet scientific standards and be disseminated as widely as possible, ideally in well-regarded electronic publishing (ePub) journals or repositories after rigorous peer review. The aggregation of such data, although seemingly sparse, lays the foundation for future research and offers clinicians much-needed insights into diagnosis and treatment options. Given the scarcity of patients, each individual case assumes a level of significance that may not apply to more common diseases, making each publication an invaluable contribution to medical knowledge.

Medical Practice: An Art Based on Science and Evidence

The adage 'lege artis,' meaning 'according to the rules of the art,' signifies the essence of medical practice. Medicine is an art grounded in science and evidence. While it is easy to defer the importance of rare diseases, arguing that time will reveal their significance through the 'gatekeeping' journals, such a standpoint is neither scientific nor ethical.

Ethical Imperatives

As healthcare professionals, we have an ethical obligation to assist every individual, regardless of how rare their condition might be. In this context, sharing knowledge—especially in a non-profit manner—is a seminal first step in fulfilling this ethical mandate. The recent publication on PANS-H63D-Multisystemic Instability Syndrome, therefore, deserves recognition not only for its scientific contribution but also for its ethical stance.

It is an indictment of our collective professional integrity to ignore or defer attention to such conditions. We must remember that while medicine is an art that should be practiced 'lege artis,' it is also an art that must be practiced with compassion and a deep-rooted sense of responsibility towards each and every patient.

Yours Sincerely,

Riku Honda
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