【Luncheon Symposium 11-2】

TSN Fabry Expert Consensus Approach for Screening, Diagnosis, and Multidisciplinary Management in Chronic Kidney Disease

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The prevalence of Fabry disease (FD) among males with chronic kidney disease (CKD) of unknown etiology in Taiwan is 0.6%. Despite this, FD is frequently overlooked in clinical settings. To address this issue, two consensus meetings were conducted in Taiwan—one in August 2022 and another in April 2023. The first meeting established screening criteria based on age, gender, family history, cardiac involvement, and symptoms. The second meeting, with a multidisciplinary team, developed treatment recommendations. The consensus emphasizes the importance of proactive data collection in dialysis units and outpatient follow-ups to enhance FD detection and management. The screening algorithm recommends incorporating FD screening into the diagnostic process for CKD patients, regardless of age. Priority is given to patients with a family history of FD, early stroke history, or classical FD symptoms. Comprehensive screening is also advised for CKD patients without obvious classical symptoms. Screening protocols for males include measuring α-galactosidase A enzyme activity, with reduced activity leading to further tests such as lyso-Gb3 level quantification and genetic analysis. For females, the protocol involves evaluating lyso-Gb3 plasma levels and genetic testing. FD, though often underestimated, is more prevalent than recognized and necessitates a multidisciplinary approach for timely diagnosis. Enhancing awareness and adopting a comprehensive approach are essential for improving patient outcomes.