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Taiwan Rare Kidney Disease Registry: Fabry disease

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The Taiwan Rare Kidney Disease Registry (TRKDR) aims to document and analyze rare kidney diseases, including Fabry disease, within Taiwan's unique medical framework. Built on the foundation of existing systems, such as the pre-ESRD upload platform, the TRKDR facilitates comprehensive data collection to enhance local and global research collaboration.

Fabry disease, caused by mutations in the GLA gene, is a systemic lysosomal storage disorder that affects the kidneys, heart, and nervous system. Insights from international registries like the Fabry Registry (FR), Fabry Outcome Survey (FOS) reveal diagnostic delays exceeding a decade and wide variability in clinical manifestations. The TRKDR addresses these challenges through advanced diagnostics, incorporating genetic testing and enzyme assays, alongside detailed data collection on clinical symptoms, biochemical markers, and imaging findings.

Preliminary results from Taiwan's pilot studies demonstrate the feasibility and effectiveness of the registry, providing valuable insights into Fabry nephropathy and its prevalence among renal biopsy cases. Taiwan-specific data align with international findings, highlighting comorbid conditions such as peripheral vestibular disorders and ischemic stroke in Fabry patients.

Aligned with global initiatives, the TRKDR fosters cross-national research and ensures data interoperability. Its long-term goals include increasing enrollment, improving early detection, and integrating findings into clinical practice and policy. By addressing diagnostic delays and optimizing management strategies, the registry aims to position Taiwan as a leader in rare kidney disease research, improving patient outcomes and contributing to global knowledge