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RELATIONSHIP OF KIDNEY AND ANDERSON-FABRY DISEASE

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Background. Anderson-Fabry disease is a multi-systemic, X-linked lysosomal storage illness that results in lysosomal accumulations of neutral glycosphingolipids and globotriaosylceramide Gb-3 due to impaired alpha-galactosidase A activity. The typical skin lesion found in Fabry disease is angiokeratoma corporis diffusum, which is associated with renal dysfunction, particularly proteinuria. Renal failure is a serious side effect of this illness. Fabry nephropathy lesions appear and progress in children, but the disease is often undetectable by normal clinical tests. As the late beginning of enzyme replacement therapy may not stop growing renal impairment, early and timely diagnosis of Fabry nephropathy is critical. Due to the difficulty in diagnosing Fabry disease in children and the lack of a sensitive non-invasive biomarker of early Fabry nephropathy, this may be difficult. Although accurate glomerular filtration rate (GFR) measurement and regular proteinuria and microalbuminuria assessments are beneficial, they are not sensitive enough to detect early kidney abnormalities.

Aim: the purpose of this study is to highlight the morphological aspects of renal microscopic changes in Anderson-Fabry disease patients using data from the Fabry Outcome Survey (FOS) survey conducted by MacDermot KD., Holmes A., and Mehta A. of the Takeda-Shire foundation (2021).

Materials and methods. This study is based upon The "Fabry Outcome Survey (FOS)," a clinical observational research that took place in Europe, Australia, Canada, Brazil, and Argentina from 2017 to 2021, drew in 4000 participants. In the Survey, 26% of the adult patients had an estimated GFR of less than 60 ml/min/1.73 with chronic kidney disease stages between 3–5. Twenty-two percent individuals were on dialysis, and twenty-six percent had a kidney transplant. Mild proteinuria and globotriaosylceramide-containing urine sediment are among the complications which lead to end-stage renal disease (ESRD) in late adolescence in some situations.

Results and discussion. The researchers discovered zebra bodies, intralysosomal inclusions, and myelin bodies, as well as lipid vacuoles, in the cytoplasm of podocytes, whose small processes were mostly reduced in the patients. The mesangial space was expanded in many cases, the mesangiocytes proliferated, fatty vacuoles were found in the cytoplasm, and immune complex deposits were found intramembranously and paramesangially. Immunohistochemistry research revealed fixations of IgG, kappa, and lambda immunoglobulin chains on the glomerular basement membrane of focal granular pattern in each case. Three patients exhibited fibrinogen fixation, two exhibited IgM fixation, and one had IgA and complement component 3 fixation. The number and shape of the cellular inclusions differed greatly between cell types. The largest quantities of Gb3 were found in podocytes and distal tubular epithelial cells, while proximal tubular epithelial cells were essentially unaffected. Inclusions appeared as tiny, black, densely beaded granules in certain cell types and as bigger complex laminated entities in others. With the course of the illness, there was also a significant buildup of Gb3 in the renal cortex.

Conclusion. Thus, cross-sectional studies show that renal manifestations appear early in life in a considerable number of children, many women, and almost all males with Fabry disease. Nearly all men and some female patients develop ESRD as a result of these complications. Patients with Fabry disease are more likely to develop cortical and parapelvic kidney cysts. The frequency of cortical and parapelvic kidney cysts is also higher. Most hemizygotes develop ESRD around the age of 50 due to progressive worsening of renal function. Fabry disease patients have a lower dialysis survival rate than individuals with other kidney diseases; nevertheless, kidney transplantation is effective in improving patient survival rates.