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Single-cell transcriptomics reveals distinct pathophysiologic signature of the primary coenzyme Q10 nephropathy in pediatric SRNS patient

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Objectives : Coenzyme Q10 (COQ10) nephropathy is a well-known cause of hereditary steroid-resistant nephrotic syndrome (SRNS), primarily impacting podocytes. This study aimed to elucidate variations in individual cell-level gene expression in COQ10 nephropathy using single-cell transcriptomics.
Methods : We conducted single-cell sequencing of kidney biopsy specimen on a 5-year-old boy diagnosed with a COQ10 nephropathy caused by compound heterozygous COQ2 mutation. The analysis focused on the proportion of cell clusters, differentially expressed genes, changes in gene expression related to mitochondrial function and oxidative phosphorylation (OXPHOS), and pathway analysis.
Results : Our findings revealed a uniform downregulation of mitochondrial gene expression across various cell types in the context of these mutations. Notably, there was a specific increase in glycolysis-related gene expression in podocytes. The study also highlighted an altered immune cell population proportion attributed to the COQ2 gene mutation. Pathway analysis indicated a downregulation in OXPHOS and an upregulation of various synthesis pathways, particularly in podocytes.
Conclusions : The insights from this study enhance our understanding of the pathogenesis of COQ10 nephropathy and underscore potential therapeutic targets for primary COQ10 nephropathy.