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EFFECTS OF INTERRUPTION OF ERT IN RENAL FUNCTION IN FABRY DISEASE PATIENTS

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Fabry disease (FD) is an X-linked lysosomal disorder due to the deficiency of  $\alpha$ -galactosidase A that causes storage of globotriaosylceramide (Gb3). Disease progression leads to vascular disease secondary to the involvement of kidney, heart and the central nervous system. The current treatment for FD is Enzyme Replacement Therapy (ERT), which prevents the deposition in the kidney and heart, or reverts, at least partially, the vascular pathophysiology. We are reporting our experience regarding to the interruption of ERT in one group of FD patients (n=5). We have been treating 4 male and 1 female patients from 3 different families, for over 8 years. During this period patients had two intervals of ERT interruption (18 and 8 months), when glomerular filtration rate (GFR-Cr EDTA) was analyzed. Data shows that a patient, the oldest one, showed important decline in renal function after ERT interruption. We believe this is a unique opportunity to show whether ERT interruption might cause worsening of renal function in FD patients