Variante clinica "renale" della malattia di Anderson-Fabry

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"Kidney" variant of Anderson-Fabry disease

With an estimated incidence of about 1:40.000, Anderson-Fabry disease (AFd) is a rare inborn X-linked recessive glycosphingolipid storage disorder, that is caused by deficient activity of the lysosomal enzyme α-galactosidase A (α-gal A). Affected hemizygotes with no detectable plasmatic α-gal A activ ity and with the typical clinical manifestation of AFd present a variety of molecular disorders including large and small gene rearrangements, missense and nonsense mutations and splicing defects. However, AF patients show protean clinical features and attempts to predict genotype-phenotype correlations are premature because most patients exhibit private mutations. The deficient lysosomal activity of α-gal. A induces progressive globotriaosylceramide accumulation within endothelial and smooth muscle cells of the vascular system, myocardial cells, cells of the autonomic nervous system, tubular epithelial cells, and mesangial, endothelial and ephitelial cells of the glomeruli.
It is well known that AF patients may show "atypical" olinical variants with predominant kidney or cardiac involvement. We report and comment on the clinical history of a 20-year-old male patient with AFd characterized by evident kidney involvement without a complete phenotype of the metabolic disease. (Giorn It Nefrol 2001; 18: 216-30)

KEY WORDS: Glycosphingolipidosis, Anderson-Fabry disease, Lysosomal α-galactosidase A, Kidney pathology, Electron microscopy of the kidney