Kostmann's Syndrome and IgA Nephropathy: An Unknown Association

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Dear Sir,

Infantile congenital agranulocytosis was described by Kostmann [1] as an autosomal recessive disease characterized by severe chronic neutropenia, typically associated with a very high monocyte count. Serum immunoglobulins (Ig) are often increased. The disorder leads to an increase in sensitivity to bacterial infections and the mortality is high.

We report on a female patient who, as early as the neonate period, experienced recurrent infections with septicemia requiring massive antibiotherapy. Diagnosis of Kostmann's syndrome (KS) was made on deep neutropenia ($< 0.2 \times 10^9/l$) with high serum Ig (IgG 51 g/l, IgM 2 g/l, IgA 6 g/l), and monocytosis ($2.3 \times 10^9/l$). At the age of 4 years, the patient presented with acute renal failure and proteinuria. Serum IgA level was very high (9.5 g/l). Renal light microscopy showed intense mesangial hyperplasia and some subepithelial deposits. Immunofluorescence staining revealed massive mesangial IgA deposits, leading to the diagnosis of IgA nephropathy (IgAN). In the evolution, renal function improved, but proteinuria persisted. Prophylactic antibiotherapy made it possible to control most infections, but chronic renal failure (CRF) occurred and at the age of 20, chronic hemodialysis was started.

This is the first description of a KS associated with CRF and IgAN. Interestingly, the monocyte count of our patient was high during a long period of time. Studies of superoxide anion production and phagocytosis of monocytes and neutrophils showed an activation state. Interleukin 6 (IL6) production by monocytes was increased. Recently, the role of the monocyte system and IL6 secretion in the pathogenesis and progression of IgAN was

emphasized [2, 3]. We hypothesize that this association might be linked with the high monocyte activity observed in our patient.

References

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