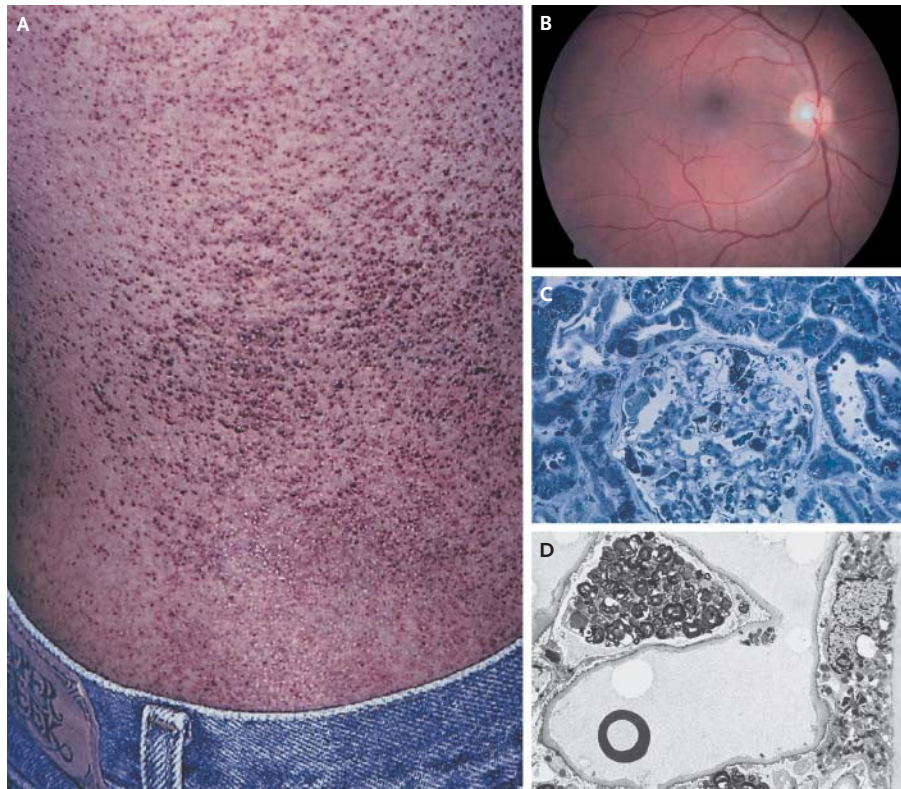


IMAGES IN CLINICAL MEDICINE

Fabry's Disease



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A 30-YEAR-OLD MAN HAD SEVERE NEUROPATHIC PAIN THAT AFFECTED his hands and feet. Examination of his skin showed angiokeratomas that were particularly prominent in the groin and on the back (Panel A) but that were also present on the lips and palms. Examination of the optic fundi showed tortuous retinal arterioles (Panel B). He had heat intolerance and decreased sweating. The diagnosis of Fabry's disease was confirmed by the detection of reduced α -galactosidase A activity in peripheral leukocytes. He received carbamazepine for the neuropathic pain. Testing of renal function when the patient was 38 years of age showed a serum creatinine concentration of 1.1 mg per deciliter (97.2 μ mol per liter), a creatinine clearance rate of 92 ml per minute per 1.73 m² of body-surface area, and a protein excretion rate of 3.9 g per day. Renal biopsy showed an accumulation of toluidine blue staining in cellular inclusions that was prominent in glomerular podocytes and also present in distal tubular epithelial cells and extraglomerular vascular cells (Panel C, $\times 80$). Electron-microscopical examination of the glomerulus revealed podocytes bearing electron-dense lamellar bodies that represented secondary lysosomes containing glycolipid (Panel D, $\times 3800$). The patient is being treated with α -galactosidase A administered by intravenous infusion every two weeks. His symptoms have improved, although his renal function has continued to deteriorate, with a serum creatinine concentration now of 2.2 mg per deciliter (194.5 μ mol per liter).

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