Scleroderma and Associated Cancer Due to the Same Mutation

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By Norman Bauman [2]

New in the non-rheumatology journals: A single mutation linked to both scleroderma and associated cancers. Is unified treatment a possibility? Also: A neurologist reviews the neuropathic vasculitides.

Source: Rheumatology Network

Association of the Autoimmune Disease Scleroderma with an Immunologic Response to Cancer

Science, December 5, 2013

Cancer researchers report that a mutation that causes cancer also seems to trigger scleroderma. Treating the cancer may cure the scleroderma too.
The investigators found that scleroderma patients with anti-RPC1 autoantibodies had tumors with mutations in the POLR3A gene, which codes for RPC1. Scleroderma patients with anti-RPC1 autoantibodies get cancer more quickly after being diagnosed than those without these autoantibodies.
Ten of 16 patients had breast cancer; the rest had lung, ovarian, colorectal, or anal cancer, with one B cell lymphoma.
The antibodies to the mutated RPC1 in the tumor also attack the normal RPC1 outside the tumor.
Patients without the POLR3A mutations didn’t have anti-RPC1 autoantibodies, and developed cancer much later.
The authors hypothesize that the POLR3A mutation causes cancer, which may be occult, and also triggers scleroderma. The symptomatic cancer appears soon after.
They note that sometimes when patients with scleroderma and cancer are treated for cancer, the scleroderma also disappears. They suggest that treating the cancer may have cured the scleroderma too.

Vasculitic neuropathies

Lancet Neurology, January 2014
Vasculitic neuropathies result from the inflammatory destruction of nerve blood vessels, from rheumatological, viral, diabetic, and other causes.

This review for neurologists classifies the vasculitic neuropathies into two groups - nerve large arteriole vasculitis and nerve microvasculitis - comparing this classification with the Peripheral Nerve Society Task Force and the Chapel Hill Consensus Conference classifications.
Written from the perspective of a neurologist, assessing the conditions according to the vessels affected, the article reviews the primary and secondary systemic vasculitides, and local vasculitides, including diabetes and infectious diseases.
It discusses the histology of myelinated fiber loss and perineural thickening, and the vascular pathology of neuropathologies. The article also enumerates how neurologists assess the clinical response to therapy: Using predetermined neurological endpoints, including muscle power, deep-tendon reflexes, sensory thresholds, functional rating scores, and electrodiagnostic testing.

Case 38-2013 — A 30-Year-Old Man with Fever and Lymphadenopathy

New England Journal of Medicine, December 12, 2013
Fever and Lymphadenopathy

Now@NEJM, December 13, 2013
The patient in this report presented with Kikuchi-Fujimoto disease, which can be impossible to distinguish from systemic lupus erythematosus (SLE) on biopsy.
Serology nails the diagnosis. This patient had normal rheumatoid factor, antinuclear antibodies and complement level. Sarcoidosis was also eliminated.
In a North American or European patient, cancer or benign tumors would lead the differential, followed by infections, SLE and sarcoidosis. However, this patient was born in India, and recently visited India. In an Asian context, rare conditions such Kikuchi-Fujimoto disease and Kimura’s disease are also a consideration.
Letter: Regulatory T cell proliferative potential is impaired in human autoimmune disease

Nature Medicine, December 8, 2013

A mechanism has come to light that may account for the progressive loss of regulatory T cells ($T_{reg}$) cells in autoimmune diseases. Altered IL-2 (interleukin-2) secretion and disturbances in signaling via the STAT5 pathway triggered by the IL-2 receptor caused impairment in $T_{reg}$ proliferation after stimulation of the T-cell receptor in relapsing-remitting multiple sclerosis.

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