CASE #3

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The patient was a 2½ year old male who first presented at age 7 months with a short history of fever, rash and vomiting. He was found to have hepatosplenomegaly and thrombocytopnnia (platelets 20,000). No etiology, including viral (the patient's father had concomitant infectious mononucleosis) was found, and fever, organomegaly and thrombocytopneia all responded to corticosteroids. Over the next several months, he had several similar episodes, all responsive to steroids. At age 18 months , he was found to have phenotypic type I hyperlipoproteinemia, with chylomicronemia and elevated triglycerides (670 mg/dl). At age 23 months , he was first noted to have progressive lower extremity weakness, with induration of muscles and greatly elevated creatine phosphokinase (5060 units, MM isoenzyme). Electromyogram revealed decreased insertional activity, occasional fibrillations and positive waves, but no motor unit potentials either at rest or with stimulation. Muscle biopsy (quadriceps) exhibited replacement of muscle by mononuclear cells. Several immunologic abnormalities were observed: peripheral blood lymphocytes (PBL) readily went into extended culture, a property generally seen with viral infection; however, no virus could be recovered. In addition, PBL in co-culture almost completely suppressed allogeneic donor T cell function (blast transformation) and B cell function (immunoglobulin production). T gamma lymphocytes ("supressor cells") were elevated in peripheral blood (22.5%) and in lymph node (24%). Serum immunoglobulins were normal, but a positive FANA was reported. The child was treated with cyclophosphamide, prednisone, methotrexate and vincristine, with 1000 rads given also to the spleen. He developed respiratory insufficiency and died several months later, with necrotizing staphylococcal pneumonia.

AUTOPSY FINDINGS: Non-agressive type lymphocytic infiltrates in portal areas of the liver, in the spleen, in the lymphatic tissue, and in the lamina propria of the small bowel. Myocardium not involved. Diffuse, severe lymphocytic infiltration in skeletal muscles (including those of esophagus and about prostate), with near total replacement in lower extremities and myositic-like picture in upper extremities. (Brain not examined, but spinal cord and peripheral nerves not involved.) Submitted sections, stained with hematoxylin and eosin, are from pectoralis or intercostal (A), and psoas or gastrocnemius (B).

POINTS FOR DISCUSSION:

1. *Can this disease be considered to be a lymphoproliferative disorder?*
2. *Why is there apparent specifity of the disease for skeletal muscle?*
3. *Why have the lymphocytic cells persisted in the skeletal muscles of the lower extremities.*
4. *Can viruses produce a similar picture?*