Typical Western blotting pattern of human immunodeficiency virus type 1 (HIV-1) antibody of controls and patients (R-P, reference serum positive for anti-HIV-1 antibody, R-N, reference serum negative for anti-HIV-1 antibody, H-T, healthy control positive for anti-P25 antibody, SY, patient positive for anti-P25 and anti-P55 antibodies, SLE-1, SLE patient (LAaCL positive, negative for anti-P52 and anti-P68 antibodies, SLE-2; SLE patient with aCL (C-2), positive for anti-P25 and anti-P52 antibodies).

and 4/23 (17%) aCL/LA-negative. None of the subjects’ sera that was reactive to HIV-1 and HTLV-1 viral proteins showed a typical reactive pattern diagnostic of true HIV-1 and HTLV-1.

Talal et al. reported that 22/61 patients with SLE were positive for p24 (syndromically with p25 in this paper) antibody and suggested that there was a close relation between HIV-1 and conserved idiotype antibody. They also found that anti-Sm antibody in this clinical setting showed cross-reaction with p24. Bick et al. confirmed that HIV-1 hybridising sequences are frequently present in patients with SLE. Furthermore, Ranki et al reported that a few patients with connective tissue diseases, including SLE, had antibodies reactive to the gag protein (p24, p55) of HIV-1.

New method for detection of C34-T mutation in the AMPD1 gene causing myoadenylate deaminase deficiency

Myoadenylate deaminase (MAD) is the muscle isoform of AMP deaminase (EC 3.5.4.6). MAD deficiency is the most common enzyme defect in human skeletal muscle and seems to cause a metabolic myopathy which is found in 2-5% of all muscle biopsies. It therefore might be one of the most common causes of exercise-induced myalgias and early fatigue.

MAD is encoded by the AMPD1 gene. Recently, the nonsense mutation C34-T (exon 2) was reported in exon 2 of this gene. The mutation was detected in 30 cases of denaturing temperature, 44°C annealing temperature, and 72°C extension temperature, each for one minute. The PCR product contains 10 mM Tris-HCl, 50 mM KCl, 1.5 mM MgCl2, 0.001% (v/w) gelatin (Perkin Elmer Cetus PCR reaction buffer), 200 μM of each dNTP, 150 ng of each primer, 2.5 U AmpliTaq DNA polymerase (Perkin Elmer Cetus) and 10 ng of genomic DNA.

After PCR, 1/10 volume of 100 mM Tris-HCl pH 7.5, 100 mM MgCl2 and 10 mM diithiothreitol (L-buffer from USB, Cleveland, Ohio) are added to the reaction volume. The PCR product is mixed with 10 U NsPI (USB, Cleveland, Ohio) and incubated at 37°C for one hour. After incubation, 1 μl of the mixture is separated by electrophoresis in a 2.5% agarose gel followed by ethidium bromide staining. The 2 base pair fragment is hardly detectable. Only the bands of 99 and 119 base pairs are seen clearly (figure).
DNA of 11 patients with MAD deficiency and 100 randomly selected subjects were analysed for the C34-T mutation using both methods and always demonstrated identical results. The mismatch in primer 1 did not prevent sufficient PCR amplification in any experiment.

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