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program
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Title: CLINICAL OBSERVATIONS IN SYSTEMIC SCLEROSIS

New results obtained in our study are as follows:

1. Out of the 246 SSc patients followed-up regularly in our institution, 177 (72%) patients developed significant clinical involvement of the alimentary tract. This was the first cohort study on gastrointestinal involvement of SSc in Hungary and even in Central-Eastern Europe.

2. In our present study, abnormalities of the anorectum and colon were observed only in 11% of our SSc patients. These results are not in accordance with other reports and suggest a particular importance on making non-invasive screening as well as extended diagnostic procedures in favour of early evaluation of gastrointestinal manifestations.

3. Associated diseases of the pancreas and biliary tract (e.g. PBC and Crohn's disease) were present in 10% of our SSc patients. A multidisciplinary collaboration is essential for early detection of these particular diseases and for designing an appropriate treatment protocol.

4. Our results suggest that the internal organ manifestations and the frequency of autoantibodies in juvenile onset SSc are far less pronounced in comparison to adult-onset SSc. Our study also suggests that the outcome of patients with jSSc may be better than that of patients with adult onset SSc.

5. In the present study, the genetic, serological and clinical characteristics of SSc-RA overlap syndrome were assessed in the to-date largest cohort of 22 patients. The clinical presentation of our SSc-RA patients showed a mixture of organ manifestations characteristic for SSc and RA and also a mixed serological pattern resembling both SSc and RA.

6. Our SSc-RA overlap patients carried both the SSc-associated HLA-DR3 and HLA-DR11 alleles, as well as the RA-related HLA-DR1 and HLA-DR4 alleles.

7. These data support that SSc-RA overlap syndrome may be a distinct genetic, serological and clinical entity.

8. In the present study we did not find significant differences in Hcy levels and MTHFR polymorphism between SSc patients as well as healthy controls lacking any vascular disease.

9. Significantly higher plasma homocysteine concentrations were observed in patients with macroangiopathy/thromboembolic events compared to patients without such clinical manifestations. Altogether 71% of patients with macrovascular disorders had either homozygous or heterozygous MTHFR variants. Our results suggest that besides other risk factors, hyperhomocysteinaemia and the polymorphism of MTHFR gene may be involved in the vascular damage associated with SSc.

10. Our results also suggest that macrovascular disease is not only an age-related feature in SSc but may also depend on disease duration, so it should be important to screen and follow these alterations.

Tárgyszavak: Szisztémás sclerosis, rheumatoid arthritis, juvenilis scleroderma, gastrointestinális manifesztáció, autoantitestek, homocystein szint, génpolimorfizmus

Keywords: Systemic sclerosis, rheumatoid arthritis, juvenile scleroderma, gastrointestinal manifestation, autoantibodies, homocysteine level, gene polymorphism