32.00

IMMUNITY IN DI GEORGE SYNDROME

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Syndrome diGeorge is relatively common congenital disorder with developmental defects including a/hypoplasia, or pathological migration of thymus, associated with deletion of contiguous genes on chromosome 22. Associated thymus pathology should theoretically cause the profound immunodeficiency. We then prospectively followed the cohort of children with confirmed 22q11 deletion. One to six repeated investigations were performed in 14 boys and 23 girls, age 4 days to 19 years. Due to the proposed role of thymus in T lymphocyte selection we observed, besides investigation of T lymphocytes and their function, the presence of double positive CD4+CD8+ and γ/δ T lymphocytes in the periphery. Low number of T lymphocytes was detected in majority of patients before the age of 2 years. Both spontaneous and PHA induced proliferation was unexpectedly increased in comparison with normal samples in tested children less then 2 years old. Both T cell numbers and function normalized thereafter in majority of patients. Double positive T cells were detected in one boy, together with transient positivity of antinuclear antibodies. γ/δ T cells were higher than 5% in 21% of children. In the 5 years long prospective study we did not yet observe serious clinical signs of immunodeficiency or autoimmunity in patients besides repeated respiratory infections that were successfully controlled by antibiotic therapy. All patients, classified as partial diGeorge syndrome, presented with delayed but gradual development of immune function on the background of impaired support of thymic tissue.
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