Severe combined immuno-deficiency and trisomy D

Sir,

We report a case of severe combined immunodeficiency and trisomy 13–15 an association which we believe has not been reported before. Relevant clinical and laboratory findings were as follows. (1) Clinical data: typical malformations of trisomy D were present including alopecia and osseous lacunae of the cranium, low-set ears, cleft palate, microphthalmia, coloboma, persistence of ductus arteriosus, umbilical hernia, cryptorchidism, penile malformation, hexadactyly, and arthrogryposis. Severe recurrent respiratory, urinai, and gastrointestinal infections, conjunctivitis, and malnutrition were also observed during the 6 months that he survived. (2) Chromosomal and immunological data: trisomy D 47,XY, D+; increased Hb-F values; deficiency of circulating lymphocytes (360/mm³); T-lymphocyte deficit (absence of E-rosettes); deficiency of serum IgG and IgA and partial deficiency serum IgM; negative antibody production in response to antityphoid paratyphoid vaccine; absence of marrow plasma cells. (3) Pathological findings: thymic tissue was searched for, but not found in either normal or ectopic sites; scarce fibrotic lymph nodes were found only in lumbar aortic region, the nodes showed poor structural organization, an intense depletion of lymphocytes and an absence of plasma cells; the spleen was reduced in volume, lacked lymphatic follicles and was devoid of germinal centres. Feyer’s patches were not observed. Our case in summary was characterized by a malformation complex of trisomy D associated with a deficiency of both thymus-dependent and bursa-equivalent lymphoid systems.

Diagnosis of Di George’s (1968) or Nezelof’s syndromes (Nezelof et al., 1964), as well as of other previously described primary deficiencies of T cells (Rezza et al., 1974; Lawlor et al., 1974), were excluded since parathyroid glands were present, blood calcium and phosphorus were normal, and no signs of tetany were observed. Lymphoid cells were also absent in the B-dependent areas of lymph nodes, spleen, and bone marrow. In addition, a humoral immune defect was documented. These data support a diagnosis of combined immunodeficiency. Unfortunately, adenosine-deaminase (ADA) was not studied in our case, but probably was normal since ADA-deficient patients have a small thymus (Yount et al., 1974).

Our case is another argument in favour of the heterogeneity of the combined immunodeficiency syndrome, which probably includes several diseases, with different aetio-pathogenetic mechanisms. Our observation should induce clinicians and immunogeneticists to study patients with chromosome changes or with immunodeficiencies more carefully so as to gain further understanding of the inter-relation between genes and immune responses.

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REFERENCES


