Severe combined immuno-deficiency and trisomy D

Sir,

We report a case of severe combined immuno-deficiency 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 arthrogyrosis. Severe recurrent respiratory, urin ary, 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 others 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 aetiopathogenetic mechanisms. Our observation should induce clinicians and immunogeneticists to study patients with chromosomal 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


Dr. Meadow (1975) also implied that the attribution of acute nephritis to a preceding streptococcal infection in textbooks should perhaps be altered. This may well be so in Britain, but the textbooks are still right in Thailand.

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REFERENCES


Dr. S. R. Meadow comments:

It is interesting to hear that poststreptococcal nephritis is so common in Thailand. The evidence is impressive. The development of suitable techniques to show depressed serum complement levels has enabled Dr. Tanphaichitr and Dr. Chatasingh to confirm the presence of a streptococcal aetiology for their many cases of acute nephritis in a way that was not possible in Britain 20 years ago when acute nephritis in children was also common.

Meadow and Smithells remain unrepentant about having altered the second edition of their textbook to state that, 'Poststreptococcal nephritis is now uncommon in Britain'. But they will ensure that in the Thai edition this statement is qualified appropriately.

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