

Immunological Abnormalities in Dyskeratosis Congenita

The recently described classical case of dyskeratosis congenita (DC)(1) lacked information as to whether the patient had features of immunodeficiency. The immune abnormalities described in X-linked DC include hypogammaglobulinemia (low IgM levels), severe B cell lymphopenia, low numbers of T cells, increased rate of apoptosis and marked reduction in cellular proliferation in short-term cultures(2,3). DC can occasionally present as T+B-NK- severe combined immunodeficiency (complete absence of B and NK or natural killer cells) in the most severe form, referred to as Hoyeraal-Hreidarsson syndrome(4). Recurrent infections and deaths secondary to *Pneumocystis* pneumonia and cytomegalovirus infections have been reported. Osteoporosis, liver and lung fibrosis are also seen and hence use of busulfan or excessive radiation are considered to be relative contraindications.

In situ hybridization techniques can detect short telomeres (cause of early ageing) even in clinically silent disease (disease anticipation)(5). X-linked DC is one of the conditions in the growing list of disorders of ribosome biogenesis and mutation testing (*DKC1* gene mutations in X-linked recessive DC) can be done for confirmation, but is only available in few referral centres. DC should be suspected in children presenting with aplastic anaemia and multi-organ problems, and clinicians should investigate for immunodeficiency (by measuring antibody levels and enumeration of lymphocyte subsets), so that recurrent infections are prevented before bone marrow transplantation.

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Reply

Immunoglobulin levels were normal in this child. He was not tested for other immune abnormalities described in X-linked dyskeratosis congenita as there was no history of recurrent infections. It has been noted that despite laboratory evidence of aberrant humoral immunity most of the patients do not have a long standing history of severe or recurrent infections(1).

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