Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic /NEMO mutations

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Résumé en anglais	X-linked recessive ectodermal dysplasia with immunodeficiency is a rare primary immunodeficiency caused by hypomorphic mutations of the gene encoding the nuclear factor κB essential modulator (NEMO) protein. This condition displays enormous allelic, immunological, and clinical heterogeneity, and therapeutic decisions are difficult because NEMO operates in both hematopoietic and nonhematopoietic cells. Hematopoietic stem cell transplantation (HSCT) is potentially life-saving, but the small number of case reports available suggests it has been reserved for only the most severe cases. Here, we report the health status before HSCT, transplantation outcome, and clinical follow-up for a series of 29 patients from unrelated kindreds from 11 countries. Between them, these patients carry 23 different hypomorphic mutations. HSCT was performed from HLA-identical related donors (n = 7), HLA-matched unrelated donors (n = 12), HLA-mismatched unrelated donors (n = 8), and HLA-haploidentical related donors (n = 2). Engraftment was documented in 24 patients, and graft-versus-host disease in 13 patients. Up to 7 patients died 0.2 to 12 months after HSCT. The global survival rate after HSCT among NEMO-deficient children was 74% at a median follow-up after HSCT of 57 months (range, 4-108 months). Preexisting mycobacterial infection and colitis were associated with poor HSCT outcome. The underlying mutation does not appear to have any influence, as patients with the same mutation had different outcomes. Transplantation did not appear to cure colitis, possibly as a result of cell-intrinsic disorders of the epithelial barrier. Overall, HSCT can cure most clinical features of patients with a variety of mutations.
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