Partial deletion of the long arm of chromosome 11: ten Japanese children

Abstract

The clinical features of partial deletion 11q were correlated with the size of the deleted region. Ten Japanese children with partial deletion of 11q were investigated. They were divided into three groups. Three patients in the first group had interstitial deletions and preserved subband q24.1. Six patients in the second group demonstrated terminal deletion of 11q including subband q24.1, with typical features of 11q- syndrome (Jacobsen syndrome). The third group included only one patient, who had terminal deletion of 11q without characteristics of typical 11q- syndrome. Prominent features of patients in the first group included severe mental and motor developmental delay, seizures, cleft lip and palate, and ophthalmological findings. Patients in the second group showed mild to moderate developmental delays without deterioration. Abnormalities in neuroimages, high intensity in the cerebral white matter in T2-weighted magnetic resonance (MR) images, and recurrent infections were not observed after the age of 7 years. The subject in the third group, with the smallest amount of deleted chromosome, did not show developmental delays, suggesting that some unknown genes related to developmental delays may be located adjacent to subband q24.1. Variation in the deleted parts of 11q resulted in different clinical features in each group.

Ono J, Hasegawa T, Sugama S, Sagehashi N, Hase Y, Oku K, Endo Y, Ohdo S, Ishikiriyama S, Tsukamoto H, Okada S. Partial deletion of the long arm of chromosome 11: ten Japanese children. Clin Genet. 1996 Dec;50(6):474-8. doi: 10.1111/j.1399-0004.1996.tb02715.x. PMID: 9147876.