F109 Detection of the size of deletions at 7q11.23 in Korean Williams syndrome patients

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Williams syndrome(WS) is a multisystem developmental disorder caused by the deletion of contiguous genes at 7q11.23. Hemizygosity of elastin(*ELN*) and *LIM* kinase can account for the vascular, connective tissue abnormalities and mental retardation observed in WS patients, respectively. But the genes responsible for features including infantile hypercalcemia and dysmorphic facies remain to be identified. To investigate the size of deletions at the 7q11.23 in WS patients, we screened 10 patients by FISH and pulsed-field gel electrophoresis(PFGE). FISH was performed using 6 cosmid clones that located at the 500-kb region commonly deleted in WS. To detect altered restriction enzyme fragments in 10 WS patients, PFGE-southern blot hybridization analysis was performed with the cosmid probes. These results make the genopype-phenotype matching possible and, therefore, provide valuable informations to understand the WS pathogenesis.

F110 Isolation of BAC clones for rapid and precise detection of Williams syndrome and identification of causative gene(s) for WS

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Williams syndrome(WS) is a multisystem developmental disorder caused by the deletion of contiguous genes at 7q11.23 and is characterized by the vascular and connective tissue abnormalities, infantile hypercalcemia, dysmorphic face, and mental retardation. The gene that contributes to each of the features remains to be identified. In addition, the size of the genomic interval commonly deleted in WS individuals has not been established. In 10 Korean WS individuals, the microdeletion at 7q11.23 encompassing the elastin(*ELN*) gene was confirmed by PCR-based allelic typing and FISH analysis using a newly isolated BAC clone(244H3) as a probe, which corresponds to the *ELN* expressed sequence taqs(ESTs). Another BAC clone(592D8) corresponding to ESTs of *KIAA0038* gene was isolated from a BAC genomic DNA library and was localized at the critical region commonly deleted in WS individuals. Structural and functional studies for gene(s) included in 244H3 and 592D8 provide valuable informations for understanding the pathogenesis of WS.