Detection of microdeletion syndromes using Fluorescence in situ hybridization: the prospective experience in Thai population

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Background: Microdeletion syndromes are a collection of genetic disorders that are associated with very small deletions on certain chromosomes, which may not be detected by conventional karyotyping. However, these microdeletions can easily be seen by the Fluorescence in situ hybridization (FISH). The propose of this study was to detect the most common of microdeletion syndromes; DiGeorge/Velocardiofacial syndrome, Williams syndrome and Prader Willi/Angelman syndrome in Thai population.

Methods: Total of 427 cases with suspected microdeletion syndromes (DiGeorge/Velocardiofacial syndrome, Williams syndrome and Prader Willi/Angelman syndrome were 306, 67 and 54 cases, respectively) in Ramathibodi Hospital during 2010-2014 were detected by FISH using the specific probes including 22q11.2 (DiGeorge/velocardiofacial syndrome), 7q11.23 (Williams syndrome) and 15q11-q13 (Prader Willi/Angelman syndrome).

Results: Out of 427 samples tested, 84(19.7%) were found to be positive for various microdeletions. Among the 84 positive cases, 41(48.8%) had DiGeorge/velocardiofacial syndrome, 30(35.7%) had Williams syndrome and 13(15.5%) had Prader Willi/Angelman syndrome.

Conclusions: On the basis of these results, any of 3 microdeletion syndromes can be diagnosed by FISH. However, the sensitivity of these tests depends on the particular syndrome such as Prader-Willi/Angelman, the etiology of the syndrome is heterogeneous and microdeletions compose just only a portion of the cases.