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FRAGILE X TESTING AND CARRIER SCREENING IN JAPAN

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Fragile X syndrome (FXS), which is the most common form of familial mental retardation, is caused by the expansion of the CGG repeat in the *FMR1* gene or GCC repeat in the *FMR2* gene on the X chromosome. We have developed a rapid molecular test and carried out diagnoses since 1993. We tested more than 1,000 samples and detected 41 samples from 20 families with the expansion in the *FMR1* gene and two samples in a family with expansion in the *FMR2* gene. Because we speculated the lower prevalence of FXS in Japanese, we analyzed a total of 946 normal Japanese (576 males and 370 females) and attempted to estimate the frequencies of the *FMR1* alleles. Within this population, we found that 1,155 alleles were in the normal range (less than 40 CGG repeats) and had a modal number of 27 repeats (35.75%). No carriers with premutations (55-200 CGG repeats) were observed in this normal population. We also identified six intermediate-sized alleles (40-54CGG repeats), with a reported incidence of 1 in 103 males and 1 in 324 females. However, this allele frequency was different from that previously reported for the Japanese population. We need more clinical studies on FXS and recently have organized the Japanese consortium. The outline of the consortium will be discussed.