

## Abstract 89

### MOLECULAR GENETIC ANALYSIS OF MENTAL RETARDATION IN JAPANESE COHORT

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Japan Mental Retardation Research Consortium (JMRRRC) has organized a research system at National Institute of Neuroscience (NCNP) for genetic analysis of mental retardation. We developed a research repository for MR and now the registration is increasing in number. As of the end of May in 2009, 276 pedigrees consisted of 132 familial and 144 sporadic cases have been registered. We have already analyzed for known 19 X-linked genetic defects such as genes for fragile X, MECP2, ARX, and so on. The examined cases (about 150 index cases) with positive results include 1 with *FTSJ1*, 1 with *ZNF41*, 1 with *ATRX*, 1 with *PAK3*, 1 with *ARHGEF6*, 1 with *AGTR2*, 3 with *FMR1* expansion, 1 with repeat expansion in *ARX* gene. Our preliminary results showed that the frequency of positive cases was also low as in previous reports from the Occidental countries. We also analyzed chromosomal abnormalities by our house-made X-tiling array CGH, and found 7 with duplication and 2 with deletion of specific regions in X chromosome. The X-tiling array CGH study is promising to find the new copy number aberrations. But it needs additional functional studies. We try to find the way that systematic genetic analysis should be a useful means for medical care and genetic counseling.