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Cytogenetic and Molecular Findings in 46,XX Male

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A 46,XX male karyotype was detected in 36-year-old male who has normal external genitalia and azoospermia. The patient was married 3 years ago and considered to be primary hypogonadism on the level of endocrine function tests. He had markedly elevated serum FSH(42 mIU/ml), elevated serum LH(18m IU/ml) and decreased serum testosterone(1.9ng/ml). The volume of testes was small of 8ml. Most 46,XX males are attributable to an interchange of the pseudoautosomal region which contains the sex-determining region of the Y chromosome(SRY) gene with the X chromosome. Using XY dual probe fluorescence in situ hybridization(FISH) was performed and two X chromosome centromere signals were detected but Y chromosome heterochromatin signal was not shown. The polymerase chain reaction (PCR) was used to amplify a single-copy Y-specific sequences in SRY gene and DYS14 loci which are expressed in testicular tissue. All the PCR poducts were positive for DYS14 and SRY sequences. In this study we will discuss the genes which are related to sex determining and spermatogenesis on Y chromosome.

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Molecular Biological Diagnosis for Genetic Disease Using LA (Long and Accurate) PCR Method

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Recently, many genes responsible for genetic diseases have been identified and abnormal genes could be analyzed using southern hybridization and PCR techniques. However southern hybridization method is time consuming, takes too much labor, uses isotope and there are cost problems. PCR techniques has been difficult to amplify target genes which are greater than 5Kb. Nowadays, a new method was developed for effective amplification of longer DNA. It's called LA-PCR. This method is used to amplify up to 22Kb of human gDNA. Therefore, in the case of diagnosis for facioscapulohumeral muscular dystrophy(FSHD), it is more difficult to analyze the abnormal gene because of extremely GC rich and highly repeated region on FSHD gene. In this study, we developed the new technique for the analysis of FSHD gene using modified LA-PCR. The probes p13E-11 and pFR-1 detect DNA rearrangements associated with FSHD as under 28 kb DNA fragment in genomic southern analysis digested with *EcoR I* and the fragment contains 3.3 kb *Kpn I* tandem repeats(73% GC). This kind of DNA could not amplify usually but amplified the tandem repeats region up to 15 kb(4 repeats) using LA PCR modified by 7-deaza-dGTP. It may be useful for the diagnosis of triple repeats extension diseases(Fragile-X, DRPLA, Myotonic dystrophy, Huntington disease, etc.) and large fragment amplification of GC rich and/or tandem repeat region.