F105 A Mutation Screening for Fabry Disease : Detection of Mutation in the α-galactosidase A Gene

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Fabry disease (FD) is an X-linked recessive lysosomal disorder caused by a deficiency of α-galactosidase A (EC 3.2.1.22 ; α-gal A), localized at Xq21.33. Besides onset of pain and paresthesias in the extremities, FD was diagnosed by absence of α-gal A activity. The α-gal A activity was spectrometrically analysed using an artificial substrate, 4-Methylumbelliferyl-α-D-galactoside. As expected, no α-gal A activity was detected in lymphocytes and lymphoblastoid cells from a FD individual, his mother and uncle. To screen the mutation in their α-gal A genes, we tried single-stranded conformational polymorphism (SSCP) and direct PCR sequencing for seven α-gal A exon and intron bounderies. But any detectable sequence alterations were not noticed. To identify the cause of FD in this family, the analysis of several putative regulatory elements in the region flanking the α-gal A gene is in progress.

F106 Modulation of X-inactivation state in a female mouse cell line without *Xist* expression

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The mouse *Xist* is expressed exclusively from the inactive X chromosome and may be implicated in maintenance of X inactivation as well as initiation. A little is, however, known about the mechanism which *Xist* leads to X-inactivation by. In this work, to ascertain the role of *Xist* for initiation and maintenance of X-inactivation, we used a mouse cell line HOBMSL2-HAT① which has one intact, active X and the other fragmented, inactive X. The cell line was originated from a female F_1 hybrid mouse derived by mating HPRT-deficient BM3 and HPRT-*A* AT29B mice. Its intact X was derived from the BM3 mouse whereas its fragmented X from AT29B. The breakpoint of the fragmented X was between DXMIT46 and DXBAY6 reflecting that *Xist* is deleted. Using this cell line, we have investigated the modulation of X-inactivation state, which induced by the deletion of *Xist*. That is, we have tested the change in the replicating pattern, the degree of global and site-specific methylation and the gene expression of the fragmented, inactive X under the condition of no *Xist* expression. The results will be presented.