Z601 Characterization of CeMEF-2 in *Caenorhabditis elegans*

Kyu Yeong Choi and Joohong Ahnn
Department of Life Science, Kwangju Institute of Science and Technology Kwangju, 506-712, Korea

MEF-2(myocyte enhancer factor-2) is known as a myogenic and neurogenic transcription factor in mammals, amphibians, and drosophila. CeMEF-2, a *C. elegans* homolog of MEF-2, was cloned and mapped to the right side of *unc-29* (M. Krause et al., in preparation). We are currently investigating the expression pattern and the role of CeMEF-2 in *C. elegans*.

Northern analysis showed 3 different size transcripts; 1.9 kb, 1.65 kb, and 1.2 kb. In order to investigate the nature of transcripts, we performed Genomic Southern analysis. Southern analysis confirmed that single gene for mef-2 exists in *C. elegans*. Northern analysis also showed that Cemef-2 RNA is expressed throughout the developmental stages although its RNA level is much lower than other genes that we have studied.

We succeeded in overexpression of CeMEF2 N-terminus fused with GST protein in *E. coli,* and the production of polyclonal antibody in a rabbit. When we performed Western blotting with *C. elegans* extract, this polyclonal antibody detected an band of 43 kDa. By immunostaining wild type worms, we detected signals between two pharyngeal bulbs with a Akalinephosphatase conjugated secondary antibody and fluorescence labelled secondary antibody. This pattern was also observed by the experiment in which gfp was expressed with CeMEF-2 upstream sequence. Therefore, we believe that CeMEF-2 is expressed in neuronal cells in nerve ring.

Z602 Partial deletion mutation in LDL receptor gene in Korean patient with FH

Ji-Hyun Bae[1] · Sung-Han Kim[2] · Jin-A Shin[1] · Un-Kyung Kim[1] · Jae-Jin Chae[1] · Young-Bae Park[3] · Hyo-Soo Kim[3] · Yong Namkoong[4] · Chung-Choo Lee[2]
Department of [1]Molecular Biology and [2]Biology, Seoul National University, Seoul 151-742, Korea, [3]Department of Internal Medicine, Seoul National University Hospital, Seoul 110-744, Korea, [4]Department of Biology, Kangnung National University, Kangnung 210-703, Korea

Familial hypercholesterolemia(FH) is a common inherited disorder with autosomal dominant inheritance, characterized by a selective increase of low density lipoprotein(LDL) in plasma, giving rising to tendon and skin xanthomatosis and to premature atherosclerosis. The genetic defect in FH resides in the gene coding for the cell-surface receptor for LDL, located on the short arm of chromosome 19. 48 unrelated Korean heterozygotes for FH were screened to assess the frequency and nature of major structural rearrangement at the LDL receptor gene. The availability of a suitable PCR assay provides a rapid and reliable alternative to Southern hybridization for the detection of various large rearrangements. For the amplification of the entire LDL receptor gene, spanning approximately 45-kb, we have designed overlapping five large PCR fragments. 5.71-kb deletion mutation(FH3, FH6 and FH311) extending from intron 8 to intron 12 was identified by long-PCR and detailed restriction mapping. Sequence analysis shows that the deletion breakpoint was shared with the same oriented Alu sequences in each intron. This suggest that the deletion was caused by unequal crossing over event following misparing of two Alu sequence on different chromatids during meiosis. This mutation is presumed to be a null allele, since the deletion shifts the reading frame and results in a truncated proteins that terminate in exon 13. As reproted previously, most of deletion mutations in Korean FH patients were related to an Alu sequence in intron 8 and the deletion break points are found within specific sequence, 27-bp in length. This supports the hypothesis that this region might have some intrinsic instability, and act as one of the important facters in large recombinational rearrangements.