Symposium II:
S2-1
Integrated Approach toward Bone and Joint Diseases using Human and Mouse Genetics

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One of the challenges in the “post-genome sequence” era is to utilize the genome information to the research associated diseases in bone and joint, including disease and osteoporosis. These diseases are serious concern for the world health and economy, as exemplified by the WHO campaign of “Bone and Joint Decade” (2001-2010); however, most of their etiology are unknown and their pathogenesis are unclear, resulting in lack of effective and fundamental treatment.

Recent advance in molecular genetics and genome medicine has revealed that genetic factors play a critical role in etiology and pathogenesis of these common bone and joint diseases. Identification of the genetic factors (i.e., susceptibility genes) is the first, mandatory step toward the innovative treatment and “order-made” medicine. To identify susceptibility genes, we have been performing systemic large-scale association studies followed by linkage–disequilibrium mapping in various diseases. Though these projects, we have found genes for OA, ASPN [1], GDF5 [2] and Deya [3], which are supported by functional evidence and replication in different ethnic populations, as well as genes for lumbar disc disease, CILP [4], COL1A1 [5], TBSP2 [6] and MMP9 [6]. Identification of these genes gave us many insights into the molecular mechanism of the diseases, which would lead to the logical invention of innovative treatment.

In this talk, I explain the detail of our approach for the common diseases, using OA study as an example.

References: