Several common differentially expressed genes between Kashin-Beck disease and Keshan disease

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Kashin-Beck disease (KBD) and Keshan disease (KD) are major endemic diseases in China. Postgraduate Xi Wang et al., under the guidance of Professor Xiong Guo from the Institute of Endemic Diseases of the Faculty of Public Health, Medicine College of Xi'an Jiaotong University, Key Laboratory of Environment and Gene Related Diseases in Ministry of Education, Key Laboratory of Trace Elements and Endemic Diseases of Health Ministry, set out to tackle these two endemic diseases. After several years of innovative research, they have made significant progress in determining the etiology and pathogenesis of these diseases at a molecular level; in particular, the identification of some common differentially expressed genes.

Their work, titled "Comparing gene expression profiles of Kashin-Beck and Keshan diseases occurring within the same endemic areas of China", was published in *SCIENCE CHINA Life Sciences*. 2013 Vol.56(9).

KBD and KD are distributed from the northeast to the southwest of China, where the selenium content is low in the soil. In China, there are 660000 KBD and 40000 KD patients, and approximately 30 million people are at risk. KBD is an endemic osteoarthropathy, the pathologic changes of KBD included significant alterations in chondrocyte phenotype, necrosis, and apoptosis, and abnormal terminal chondrocyte differentiation. The mainly pathologic changes of KD are multifocal myocardial necrosis and fibrosis that can result in cardiogenic shock and congestive heart failure. KD is an endemic myocardosis that happened in women and preschoolers. Since osteoarthritis and myocardium deformities, the most of KBD and KD patients will partially or completely lose their abilities to work even self-care, which seriously reduces their quality of life, and also bring heavy medical burden to society; the etiology and pathogenesis of KBD and KD remain unclear. However, both diseases happened in the same area of China. Moreover, the living conditions of KBD and KD patients are similar, for example, most patients live in remote rural areas and the areas of awful transportation, have a meager income, and a simply diet. There is little research conducted to compare KBD and KD gene expression profiles. Therefore, the two diseases may have a further relationship at the molecular biology level.

In this study, the Agilent Human 1A Oligo microarray was used to compare gene expression profiles of peripheral blood mononuclear cells (PBMCs) between KBD or KD patients and healthy controls, and identified the common genes differentially expressed in both diseases groups. One hundred and thirty-six differentially expressed genes (53 up-regulated and 83 down-regulated) were identified between KBD and normal controls. Moreover, comparing KD and normal controls, 3310 differentially expressed genes (3154 up-regulated and 156 down-regulated) were identified. Comparing all identified differentially expressed genes, 16 genes showed differential expression in both diseases, including nine with synchronous and seven with asynchronous expression. These 16 genes were subdivided into 11 categories, namely metabolism, cytochrome enzymes, transcription-related, G-protein-related, receptor, cytokine factor, ion channel transport protein, signal transduction, hematopoietic related, interleukin, and immune-related.

The distribution of KBD and KD is in the similar geographical regions, although the clinical presentations and target pathological focus are not same. The common differentially expressed genes identified in both

KBD and KD could be helpful to identify the potential mechanisms of the different organ lesions, caused by similar environmental risk factors, selenium deficiency. These findings make a great contribution towards clarifying the etiology and pathogenesis of KBD and KD.

The discovery of these common differentially expressed genes of KBD and KD was a collaborative effort involving many researchers from different institutes and universities. This research project was supported by a grant from the National Natural Science Foundation of China (30872192, 81273008) and a grant from Key Scientific and Technological Innovation Special Projects of Shaanxi "13115" of China (2009ZDKG-79). It is an important breakthrough in the field of endemic disease research. The researchers suggested that their work needs to be further validated and researched in different methods. These efforts will have significant impact on elucidating the etiology and pathogenesis of KBD and KD.

More information: WANG Xi, WANG Shuang, HE ShuLan, ZHANG Feng, TAN Wu Hong, LEI YanXia, YU Hanjie, LI Zheng, NING YuJie, XIANG YouZhang, GUO Xiong. Comparing gene expression profiles of Kashin-Beck and Keshan diseases occurring within the same endemic areas of China SCI CHINA Life Sci, 2013 Vol.56(9): 797-803. life.scichina.com:8082/sciCe/E … 07/s11427-013-4495-z

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