Prevalence and Aetiology of Intervertebral Disc Degeneration

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Low back pain (LBP) is one of the most common disorders seen in general and orthopaedic practices. It is a significant cause of work related sick leave and results in loss of working hours to the detriment of all industrialized societies. One of the most common causes of severe LBP is degenerative disc disease which result in back pain and leg pain (sciatica), both can cause chronic disability and permanent neurological damage.

While degenerative disc disease (DDD) may be related to environmental risk factors (e.g. occupation and smoking), epidemiological studies have strongly supported a genetic predisposition. Moreover, aggrecan and specific vitamin D receptor gene polymorphisms have also been found associated with DDD. Recent strong evidence came from a study in a Finnish population with sciatica, in which some 30% of the individuals had a mutation in genes coding for two of the chains of collagen type IX, either COL9A2 or COL9A3. As the disease-associated changes in collagen IX genes were not found for over 70% of these patients, it is likely that other genes are involved. We therefore propose that the genetics of DDD is multifactorial, and is either due to the involvement of different genes or due to a combination of several minor mutations with low penetrance.

To determine the contribution of genetic factors to risk for DDD in the Southern Chinese population, we have performed a large-scale study in which over 1000 individuals between 18 and 55 years of age were recruited. Presence of DDD was objectively and specifically determined by Magnetic Resonance Imaging (MRI) and scored using the Schneidermans Classification. Blood was taken for DNA isolation.

To date, over 1000 volunteers underwent a whole spine MRI. The prevalence of lumbar disc degeneration was 67%, lumbar disc herniation was 30%, annular tears was 30%, Schmorl's nodes was 10% and ossified yellow ligament was 6%. When stratified by age, lumbar disc degeneration was present in 40% of the population between 18 and 30 years, and 85% of the population by 50 years of age.

In preparation for mutation screening, novel methods have been developed to adjust for age and levels of disease involvement. These have revealed unusual patterns of disease that were not previously recognised, including young individuals with severe multiple level involvement, skip lesions and isolated high lumbar spinal involvement.

This is the first large scale study in the world in which MRI was used to specifically diagnose DDD. The implications of these findings will be discussed. They form the basis for genetic studies which will be discussed in the next talk.