All in the family: Lower back disease may be in your genes

February 2 2011

– Symptomatic lumbar disc disease, a condition caused by degeneration or herniation of the discs of the lower spine, may be inherited, according to a new study published in the *Journal of Bone and Joint Surgery* (JBJS).

"Previous studies, including studies of twin siblings and subsequent genetic marker studies, have suggested a genetic predisposition for the development of symptomatic lumbar disc disease but have been limited by a small number of patients," noted study author Alpesh A. Patel, MD FACS, assistant professor of orthopaedic surgery at the University of Utah School of Medicine. "The results of this study provide evidence based on a population of more than 2 million people, indicating that there likely is a genetic component in the development of this disease. Additionally, the factors that differentiate a symptomatic disc from a non-painful disc may also be affected by genetics."

Study Details:

- The researchers used data contained in the Utah Population Database, a public information repository containing health and genealogic data of more than 2 million Utah residents, examining health and family records of 1,264 individuals with lumbar disc disease, defined as either lumbar disc degeneration or lumbar disc herniation.

- To measure how closely patients were related, the researchers used the Genealogical Index of Familiaity, which compares the average relatedness of affected individuals with expected relatedness in the general population. Relatedness is measured by generations or degrees:
    - first-degree relatives (or immediate family) including parents, offspring and siblings;
    - second-degree including grandchildren, grandparents, uncles, aunts, nieces, nephews, and half-siblings; and
    - third-degree comprising great-grandchildren, great-grandparents, great-aunts and great-uncles, grandnieces and grandnephews and first cousins.
    - In this study, only patients with at least three generations of genealogical data in the database were included.
- The researchers also determined and calculated the Relative Risk (RR) for relatives. This measure defines the risk of lumbar disc disease among family members of patients compared to individuals without disease.
  Important Findings:

- Individuals with lumbar disc disease were more likely to have family members with disc disease.
- Relative risk for lumbar disc disease was significantly elevated in both close and distant relatives.
- The combination of the two findings, given the large patient population, strongly supports a genetic basis to symptomatic lumbar disc disease.

"Although excess risk in the immediate family might indicate evidence of a genetic contribution, it could also simply indicate shared environment risks or household exposure that may be contributing to the disease," Dr. Patel noted. "Conversely, excess risks in second and third-degree relatives strongly support a genetic contribution to disease, given the measurable genetic sharing in these more distant relatives and the relative absence of shared household risks."

"There are limitations to our study. We could not measure disease severity or response to treatment. Furthermore, the population of Utah is genetically representative of a US or North European background. As such, this study does not prove a purely genetic basis for disease but suggests that it may play an important role." Dr. Patel noted. "With additional data, this hypothesis can be tested with larger sample sizes."

According to data from the American Academy of Orthopaedic Surgeons (AAOS), back pain is a common problem, and in 2008, attracted more than 12 million physician visits. Dr. Patel said identifying the factors that contribute to the disease can have far-reaching implications.

"Lumbar disc disease is likely due to a number of factors, including mechanical stresses to the spine, age-dependent disc degeneration, biochemical factors and genetics," he said. "This study identified an inheritable predisposition to the development of symptomatic lumbar disc disease and also identified high-risk families in the Utah population, which can be studied to identify genes responsible for this predisposition. Identification of these specific genes may help in the future development of drugs or other interventions to prevent and/or treat lumbar disc disease in the public at large."

Provided by American Academy of Orthopaedic Surgeons