

Genetic Testing and its Impact on the Workplace

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Recently, the big news in health care has been all about the amazing advances in genetics. I must admit, my knowledge of this field is a bit dated. I remember taking a class in genetics as an undergraduate, way back in the middle of the last century. At that time, the field was just beginning to gain momentum. Much of the work—at least the work that I knew about—was limited to the fruit fly and the common garden pea. Since then, tremendous headway has been made in this promising field. With the recent press releases about the Human Genome Project, the development of a precise gene map, and cloning, genetics is constantly in the news. My guess is that for the next decade or so, every condition known to man will somehow be blamed on genetics, or at the very least, be shown to have a genetic predisposition. This interest in finding a genetic predisposition has already made it to the workplace and may have a significant impact on the way in which we view various work-related disorders.

A recent article in the *Los Angeles Times* (February 25, 2001) described the case of genetic testing at Burlington Northern Railroad. It appears that, based on work done at the University of Washington (Chance, 1999), someone at Burlington Northern got the bright idea that carpal tunnel syndrome (CTS) had a genetic marker that had been identified. Apparently, in an attempt to demonstrate that CTS was not solely a work-related injury, Burlington Northern began genetic testing to screen workers for the presence of this marker. According to Dr. Phillip Chance, the discoverer of the genetic marker, it was actually associated with a condition known as "hereditary neuropathy with liability to pressure-sensitive peripheral palsies" or (HNPP), not specifically to CTS. In the *Times* article, Chance states that testing for the marker would have little or no value in screening workers for CTS. Nevertheless, Burlington Northern plunged ahead with their program of testing. According to the article, a railroad worker in Nebraska began to experience numbness in the tips of his fingers and ultimately had carpal tunnel release surgery. In the follow-up period, the company instructed the worker to report to a doctor for "additional, objective medical information to determine whether or not the condition is work-related." The employee's wife, a registered nurse, became suspicious about the type of tests that would be performed and objected. (She couldn't understand why the doctor would need to take blood samples to determine if the CTS was work-related). In a case comparable to that of Erin Brockovich, the worker and his wife hired an attorney and filed suit against the railroad for EEOC violations. To make a long story short, the railroad has since dropped its practice of genetic screening for CTS.

The *Times* article attracted my attention for a number of reasons. At a course I recently attended, one of the instructors was describing the biomechanics of the intervertebral disc. During the discussion, he made a comment that caught my attention. He stated that 71 percent of disc degeneration was genetically based. He quoted a study by Sambrook, et al. (1999) as a reference. Since I hadn't read the article, I couldn't object too strongly at the time, but it did make me think. Perhaps because of this comment, I am a bit more sensitive to the issue of genetic predispositions. I did a bit of homework and read the article by Sambrook, et al. In an attempt to determine the extent of genetic influences on disc degeneration, the authors compared MRI features of degenerative disc disease in the cervical and

lumbar spine of 172 monozygotic and 154 dizygotic twins. An overall score for disc degeneration was calculated, and the results were then adjusted for age, weight, height, smoking, occupational manual work, and exercise. Disc height and bulge were shown to be highly heritable in both the cervical and lumbar sites, and osteophytes were heritable in the lumbar spine. The authors suggest an important genetic influence on variation in intervertebral disc degeneration. However, they also state that variation in disc signal is largely influenced by environmental factors shared by twins. Was the link due to genetic factors or environmental issues? I must admit, I'm not as convinced by the article as my instructor.

In a similar study using magnetic resonance imaging and plain film radiography, Matsui, et al. (1998) investigated whether a family history of lumbar disc herniation is a risk factor for disc degeneration. According to the authors, the study provided evidence that a family history of operated lumbar disc herniation has a significant implication in lumbar degenerative disc disease. They state that there may be a genetic factor in the development of lumbar disc herniation as an expression of disc degeneration.

Other studies have also attempted to identify a link between heredity and back pain. Postacchini, et al. (1988) surveyed the parents, siblings and children of 284 patients complaining of discogenic lowback pain (Group I); 114 patients that had undergone surgery for lumbar disc herniation (Group II); and 280 that had never complained of lowback pain (Group III). Of the families in Group I and Group II, 35 and 37 percent, respectively, had at least one member with a history of discogenic back pain, and five and 10 percent, respectively, had one or two members who had undergone disc surgery. Of the asymptomatic subjects in Group III, only 12 percent had at least one or more affected relatives, and one percent had a relative who had undergone disc surgery.

Of the affected families, 41 percent had two or more members with a history of back pain. As one might expect, the proportion of symptomatic relatives in the affected families was higher among sedentary workers and motor vehicle drivers than among heavy or light manual workers. Nevertheless, the authors conclude that there is a strong familial predisposition to discogenic low-back pain, and suggest that the etiology of degenerative disc disease is related to genetic and environmental factors.

Richardson, et al (1997) performed a retrospective case-control study to test the hypothesis that there is a familial predisposition to lumbar disc pain and injury. In this study, the immediate relatives of patients who had surgically proven lumbar disc herniations or repetitive upper-extremity overuse syndromes were given a questionnaire that had been tested previously and found to reliably identify discogenic lower back pain. The authors state that analysis identified familial grouping and a history of lifting as the only variables associated with a positive response on the questionnaire. They conclude there is a familial predisposition toward lumbar disc pain and injury.

While these studies appear to indicate that there is a familial link to the presence of disc degeneration and resulting back pain, they do not, in my opinion, demonstrate that this familial predisposition is genetic. Some contend that it is both environmental and psychosocial. In one interesting study, a previously disabled family member is presented as a psychosocial determinant in the subsequent disability of the patient. Eighty-three disabled patients presented for an independent medical examination. All were seeking a workers' compensation settlement or continued workers' compensation benefits. Forty-four of the 83 had at least one disabled family member. These authors suggest a significant correlation between the "disabled" support system and subsequent disability of the patient (Stutts and Kasdan, 1993). In other words, the "pattern" of familial disability appeared to

have a psychosocial origin rather than a genetic one. However, given the recent interest in genetics, one could also argue that there was a predisposition to disability in these individuals.

I don't mean to imply that genetic factors are not important in the development of a number of diseases. Obviously, there are conditions such as sickle cell anemia where genetic factors are the primary consideration. In others, genetic factors may be important, but they may play only a complicating role. Still others, additional factors that are genetically based, such as anthropometry (e.g., wrist size and structure), may contribute to the development of disorders such as CTS.

But I'd like to proceed cautiously, and not throw everything else that we have learned in the past number of years out the window. As an example, it is well established that individuals that take control of their conditions typically do better than those who relinquish control. The idea that the problems that we face are genetically linked may provide some individuals with a feeling of helplessness and further contribute to the idea that they are victims of circumstances.

It is important to remember that most of the conditions that we see in practice, and in the workplace, are caused by the interaction of a variety of factors. For most conditions, genetics is probably not the "magic bullet" that some are seeking. I must admit, I'm not as convinced by the article as my instructor.

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