

# Congenital spine disorders

## *An enlightened approach to treatment*

By Charles Burton, M.D., F.A.C.S.

There is little disagreement that the greatest single medical advance of the 20th century was the introduction of antibiotics. It is also clear that the greatest single medical advance of the 21st century will involve our improved understanding of human genetics and the application of this information to the prevention and treatment of disease. Unfortunately, at this time genetic (genomic) analysis essentially has been limited to the fields of forensic medicine and to curiosities such as identifying Thomas Jefferson's descendants, or validating the claims of Lemba tribesman in southern Africa and the Bene Israelis of Bombay, India, to provide proof that they

are direct descendents of Moses' elder brother Aaron.

Awareness of the need for better genomic information in diagnosing and treating illness

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is in its infancy. Only recently has media attention focused on the issue of sudden death in young athletes due to hidden congenital heart defects. Yet, from a public health standpoint, congenital back problems represent a less emotional but much

more significant liability in our society in terms of cost and resulting disability.

Other than health care expenditures related to psychologic, psychiatric, and drug-related disease, low back problems represent the single greatest expenditure in our health care system today. Researchers from Duke University Medical Center recently published results of an extensive analysis of data from 1998, and concluded that in that year, back problems

accounted for \$90 billion, with \$26 billion directly attributable to back pain therapy.

Despite the fact that back problems account for a massive outpouring of financial resources, the medical community's understanding of congenital spine disorders seems to be unfocused and confused. Few radiologists or physicians appear to consider this as a diagnosis when they review lumbar imaging studies or make inquiries regarding cohorts in the family when taking patient histories of individuals with long-term back problems.

### Research on congenital spine disorders

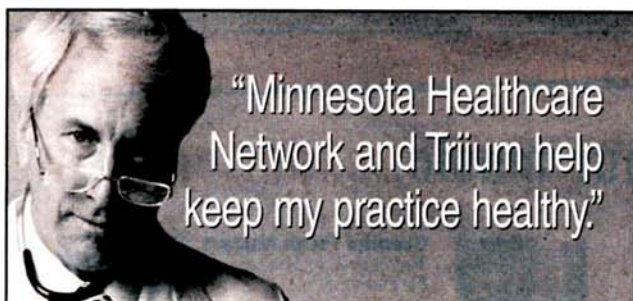
The first modern study regarding congenital spine disorders based on MRI and CT scan data was published in the journal *Spine* in 1994. The study identified a group of similar anatomic imaging abnormalities in patients with low back pain, and the researchers coined the term *juvenile discogenic disease* (JDD) as a means of describing the diffuse abnormalities that were associated with advanced disc degeneration and a variety of associated endplate abnormalities producing disability.

Some of the endplate abnormalities seen in JDD were similar to those originally described by Holger Werfel

Scheuermann (1877-1960), a Danish orthopedic surgeon and radiologist, who first described thoraco-lumbar endplate deformities associated with discal wedging and associated kyphosis in 1921. Some of the abnormalities also resembled Schmorl's nodes, abnormalities first described by Christian G. Schmorl (1861-1932), a German pathologist, in 1927. Schmorl attributed these nodes to endplate deformities produced by the herniation of disc material into the vertebral body rather than due to familial disorders.

In the 1994 *Spine* study, 9 percent of patients presenting for CT and MRI studies due to back pain were found to have JDD. A decade later, some of the authors and their colleagues found that as many as 40 percent to 50 percent of patients presenting with low back pain had JDD and other significant underlying diffuse congenital spine abnormalities. [These findings were evident because of advanced MRI examination and more experienced MRI readers.] The researchers found a high level of correlation with other existing congenital abnormalities, the most evident of which are scoliosis, spondylolysis, spondylolisthesis, congenitally small spinal canal, tropism of the facet joints, asymmetric and molded vertebrae, spina bifida, conjoined nerve roots, and intraosseous hemangiomas.

As MRI technology has advanced, it has become evident to spine specialists that JDD only represents one form of congenital anatomic and metabolic disorders that are seen on a regular basis. It is also evident that more specific diagnoses, in the future, will be determined by genetic cell analysis, not by imaging studies. As awareness and understanding of congenital spine disorders have increased, it is now not unusual for health care professionals to suspect their existence simply by reviewing plain x-rays of the spine. In the future, we may be able to determine JDD from genetic cell analysis at birth.



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## Diagnosis and treatment

There is no question that JDD is an important medical diagnosis because it is common and often leads to disabling pain and neurologic impairment with advancing age. JDD and associated congenital spine abnormalities frequently appear among members of the same family. If these disorders are diagnosed at an early age, existing prevention programs can moderate their progression. Preventive programs focus on elimination of smoking, exercise, intermittent regular spinal traction, and improvement in muscle tone, strength, and flexibility.

Another important reason for understanding JDD and related genomic spine disorders is our current understanding that spinal medicine is producing patients with failed back surgery at an alarming rate. It has become fashionable at a number of spine centers throughout the U.S. to treat patients who have multi-level degenerative spine disease with multi-level discography followed by multi-level rigid instrumented pedicle screw-and-rod fusions. This practice is being brought into question today by even the most ardent supporters of pedicle screw fusion.

In 1981, the author was involved in an international study which determined that the most common reason for poor results following back surgery was failure to adequately diagnose and treat spinal nerve compression due to lateral spinal stenosis. An updated review in 1991 reiterated this conclusion. Remarkably, and unfortunately, in 2006 this observation remains valid. Lateral spinal stenosis is a common sequela of congenital spine disorders and not uncommonly results in serious progressive neurologic problems.

Fortunately for patients, congenital problems typically progress only slowly; and, because of the remarkable adaptive capabilities of the human nervous system, compromised exiting nerve roots can gradually be reduced in volume (as much as 50 percent) without a loss of function. It is when an event occurs (such as a fall) to upset

## Resources

Additional information about congenital spine disorders is available from the following sources:

### Articles

- Luo X., Pietrobon R., Sun S.X., Liu G.G., Hey L. "Estimates and patterns of direct health care expenditures among individuals with back pain in the United States." *Spine* 2004, 1:29(1): 79-86.
- Heithoff K.B., Gundry C.R., Burton C.V., Winter R.B. "Juvenile discogenic disease." *Spine* 1994, 1:19(3): 335-40.
- Burton C.V., Kirkaldy-Willis W.H., Yong-Hing K., Heithoff K.B. "Causes of failure of surgery on the lumbar spine." *Clin Orthop* 1981, 157: 191-198.
- Burton C.V. "Causes of failure of surgery on the lumbar spine: 10-year follow-up." *Mt Sinai J Med* 1991, 58(2): 183-7.

### Web sites

- [www.burtonreport.com](http://www.burtonreport.com)
- [www.spine-health.com](http://www.spine-health.com)

this fine balance that clinical symptoms such as pain, lumbar claudication, or weakness occur.

It is now clear to the few spine specialists and spine surgeons familiar with congenital spine disease that this is an important clinical entity demanding greater physician awareness. It has also become clear to many that the multi-level rigid instrumented fusions currently being performed in this patient population as a treatment for back pain are ill advised. When these patients require surgical intervention, surgery must be based on legitimate indications and should be performed only when nonsurgical therapeutic means have failed. Human spines were created with physiologic flexibility. Rigid multi-level fusion does not represent the only surgical option for a patient. Other reasonable and more physiologic surgical choices now exist.

There was a time when all hip, knee, and ankle problems were fixed by fusing these joints. With the advent of artificial joints, this paradigm suddenly changed. Though this important change in orthopedic philosophy has not yet occurred in spine surgery, we have seen the beginnings of awareness that flexible stabilization is a more physiologic answer than rigid fusion. The term "arthroplasty" has been introduced to define this concept, and CPT coding has removed the term "fusion" and replaced it with "arthrodesis" (from the Greek *arthro*, meaning "joint," and

*desis*, meaning "to bind") as a means of recognizing the full spectrum of technologies.

## Reducing costs and disability

There can no longer be any question that the time has come for all physicians, particularly those in primary health care, to develop expertise in the diagnosis, prevention, and treatment of congenital spinal problems. It has been said by some that disc

degeneration is not a "disease." Mitosis is not a disease either, but when the mitosis process goes awry and creates disability and incapacitation (cancer), it becomes a disease. The same is true for the spine. Combined with inadequate disc nutrition and structural weakness, and potentiated by the insults and injuries of life, JDD can lead to patient disability. With proper interventional treatment, however, surgery can often be avoided.

The current cost of treating and operating on back problems is exorbitant. Congenital spine conditions are now more prevalent because of our aging patient population, in whom these problems surface more frequently. By adopting a more enlightened approach to treating congenital spine conditions, physicians could dramatically cut health care costs and prevent increasing disability from these conditions. ☐

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