Underdiagnosed: Joint Hypermobility Syndrome

BY KERRI WACHTER
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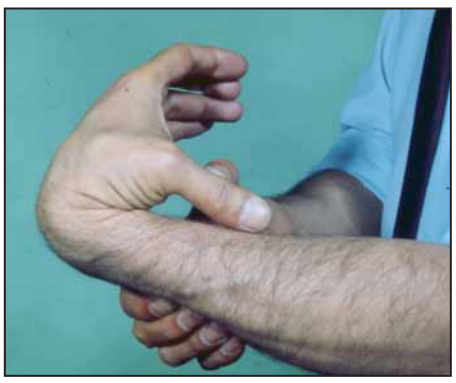
DESTIN, FLA. — Rheumatologists are probably missing the diagnosis of joint hypermobility syndrome on a regular basis, Alan J. Hakim, M.D., proposed at a rheumatology meeting sponsored by Virginia Commonwealth University.

Joint hypermobility syndrome (JHS) shares a number of features with related disorders, making it difficult to identify, but the correct diagnosis will make all the difference in patients' lives, said Dr. Hakim, a rheumatologist at Whipps Cross University Hospital in London.

JHS is thought to be related to Marfan syndrome, Ehlers-Danlos syndrome, and osteogenesis imperfecta. In addition, JHS mimics fibromyalgia and chronic fatigue syndrome, and it can be difficult to distinguish JHS from these other conditions, he said.

JHS can involve a variety of musculoskeletal symptoms, from chronic pain and fatigue to soft-tissue and visceral injury, all in the presence of general joint laxity.



JHS can involve various symptoms, all in the presence of general joint laxity.

PHOTOS COURTESY DR. ALAN J. HAKIM

Skin abnormalities such as excessive stretching of the skin are typical.

Of the musculoskeletal symptoms, chronic noninflammatory joint pain or spinal pain is a key complaint. Other musculoskeletal symptoms include dislocation or subluxation of joints; ligament, tendon, or muscle overuse injuries; and deconditioning as a result of kinesphobia.

The other key clinical components include skin abnormalities such as excessive stretching of the skin and abnormal scarring, and psychosocial problems. This overall symptom complex can manifest in childhood, adolescence, or even early adulthood.

The prevalence of JHS in the general population ranges from 10% to 30%; the disorder is three times more common in women.

JHS is more commonly seen in people of African and Asian ethnicity than in whites. There appears to be a strong genetic component, with approximately a 75% heritability rate of the phenotype.

Psychologic aspects are typical, because patients tend to feel anxious or depressed as a result of their chronic pain and disability.

Panic disorders and phobias are four times more common in JHS patients. They may not be able work, adding to their sense of isolation. They avoid relationships and social activities. Sexual difficulties and reproductive concerns are common.

These patients may feel frustrated with a medical system that has been unable to correctly diagnose them, Dr. Hakim explained.

Cardiorespiratory and bowel disturbances are also frequent and often disabling.

Related symptoms include palpitations, chest discomfort, lightheadedness, and presyncope. Bowel disturbances include nausea, dyspepsia, constipation, and diarrhea. All these symptoms are strongly associated with fatigue and anxiety.

There also appear to be proprioception and autonomic–nervous system disturbances.

Proprioceptive deficits have been shown to create a vicious cycle, whereby diminished proprioception leads to altered neuromuscular control, causing altered mechanical loading, which leads to joint

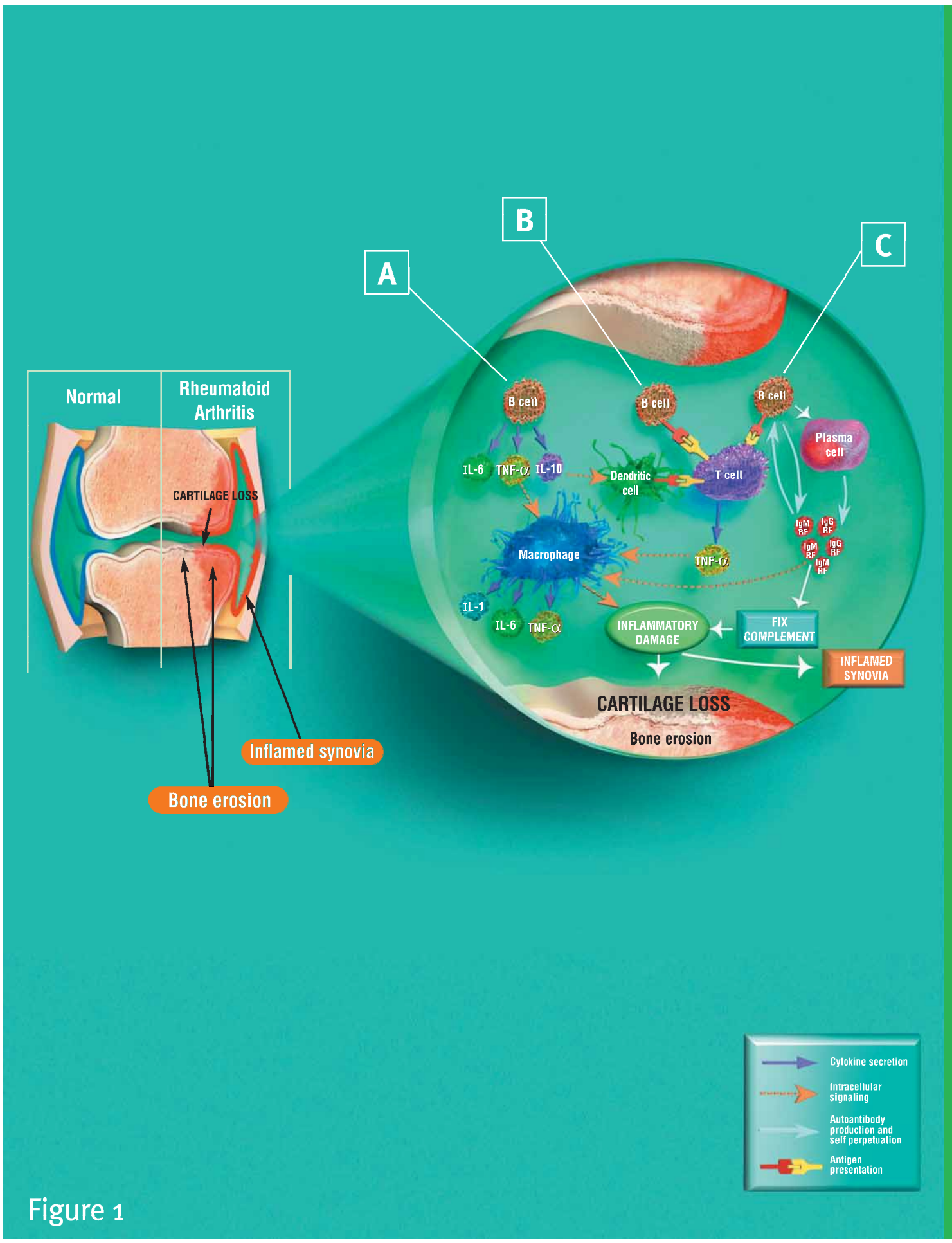


Figure 1

capsule and ligament damage, which in turn creates more of a tendency toward poor proprioception.

The Beighton nine-point scoring system, the conventional means of detecting hypermobility, assesses a patient's ability to perform five maneuvers on the right and left side of the body.

Maneuvers include passive dorsiflexion of the fifth metacarpophalangeal joint to at least 90 degrees, opposition of the thumb to the volar aspect of the ipsilateral forearm, hyperextension of the elbow to at least 10 degrees, hyperextension of the knee to at least 10 degrees, and the ability to place the hands flat on the floor without bending the knees.

More recently, Dr. Hakim and colleagues have developed a simple five-item questionnaire to use as an adjunct for screening individuals with diffuse or localized musculoskeletal symptoms, in whom no clear-cut degenerative or inflammatory disease can be found. (See box.) The questionnaire is 80%-85% sensitive and 80%-90% specific, Dr. Hakim said.

Caring for these patients likewise requires a multidisciplinary approach, involving the patient, a physiotherapist, an occupational therapist, a psychologist, a nurse specialist, and a physician specialist, Dr. Hakim said at the meeting, also sponsored by the International Society for Clinical Densitometry.

The goals of rehabilitation are to reassure and educate these patients; develop core stability; enhance joint stability and proprioception; restore normal mobility, which for these patients may still mean hypermobility; reverse deconditioning, by improving fitness and stamina; and develop behavioral strategies for coping and pain control.

Acute pain often responds to simple analgesics, NSAIDs, and local steroid injections. Chronic pain is more challenging to treat, in that anecdotal evidence suggests that simple analgesics are ineffective. Alternatives include serotonin/norepinephrine reuptake inhibitors, amitriptyline, tramadol, and gabapentin, Dr. Hakim said.

Cognitive behavior therapy has been shown to be helpful for improving quality of life and reducing pain and depression severity.

Physiotherapy alone does not appear effective, with JHS patients often reporting failure of this treatment.

In a study of 100 patients with back pain who participated in a rehabilitation program, Dr. Hakim and a colleague retrospectively assessed patients for JHS. Patients who had been diagnosed with JHS were then matched for age and gender with patients not diagnosed with the disorder. Although both groups showed the same ability to walk prior to rehabilitation, those with JHS showed much less improvement immediately following the program and up to 3 months afterward.

The ideal JHS program would focus on developing core and peripheral stability, improving general posture, and improving proprioception.

The ability to stand from a sitting position improved to a lesser extent in JHS patients, compared with those without the disorder.

However, the JHS patients had returned to baseline at 3 months' follow-up. The same was true for the ability to step up. Likewise, pain scores had not improved in the JHS group at 3 months, and those without the disorder showed marked improvement.

But physiotherapy can still be an important component of JHS treatment, he said.

The ideal program would focus on developing core and peripheral stability, improving general posture, and improving proprioception. The pace of physiotherapy should take tissue fragility into account, because injuries in JHS patients can take longer to heal, and many patients need special care to overcome their fear of movement. ■

Five Questions Identify JHS

1. Can you now (or could you ever) place your hands flat on the floor without bending your knees?
2. Can you now (or could you ever) bend your thumb to touch your forearm?
3. As a child, did you amuse your friends by contorting your body into strange shapes, or could you do the splits?
4. As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
5. Do you consider yourself double-jointed?

Source: Dr. Hakim