Joint Hypermobility Syndrome Underdiagnosed

BY NANCY WALSH
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GLASGOW, SCOTLAND — Joint hypermobility syndrome is profoundly underdiagnosed and its impact underestimated despite its being one of the most common causes of widespread chronic pain—and indeed may be the most common rheumatic disorder, Dr. Rodney Grahame said at the annual meeting of the British Society for Rheumatology (BSR).

"I looked at a series of 500 unselected new patients referred to the rheumatology clinic at Willesden Community Hospital in London between 2003 and 2005, evaluating them for their rheumatic complaints but also to see how many fit the JHS [joint hypermobility syndrome] phenotype. What I found exceeded my expectations by several orders of magnitude—overall, 45% of patients fulfilled the criteria," Dr. Grahame said.

In contrast with his findings, "many of our rheumatology colleagues consider JHS to be rather trivial and relatively uncommon, and in a survey we did of all the BSR members, most said they diagnosed, on average, approximately 10 new cases each year, and half rated the impact on patients' lives as fairly minimal," he said (Rheumatology [Oxford] 2001;40:559-62).

Extrapolating from those data would suggest that if most rheumatologists are diagnosing only 10 cases per year, they are identifying only 4.5% of cases. "That means that in England, 103,568 cases are missed annually, as are 593,930 cases in the United States. These are appalling statistics," he said.

Evaluation of joint hypermobility traditionally is done using the Beighton scoring system, said Dr. Grahame, who was involved in the development of what are known as the revised Beighton criteria. The Beighton score identifies symptoms such as the ability to passively dorsiflex the fifth metacarpophalangeal joint to 90 degrees or more, to oppose the thumb to the volar aspect of the ipsilateral forearm, or to place the hands flat on the floor without bending the knees. This system is less than reliable in pauciarticular hypermobility, however, which is often the case in JHS. A common misconception is that hypermobility requires the involvement of four or more joints. In fact, only one joint need be hypermobile in JHS, he said.

Other typical presenting symptoms include acute or chronic pain and joint clicking. There may be a history of subluxations or dislocations, because the laxity of the ligaments leads to joint instability. Pain avoidance, typically beginning in childhood or adolescence, often leads to muscle deconditioning. Cutaneous findings include stretchability, paper-thin scars, and stretch marks. Ocular involvement can manifest with drooping eyelids and blue sclerae. Anxiety and other psychological disturbances such as phobias are common.

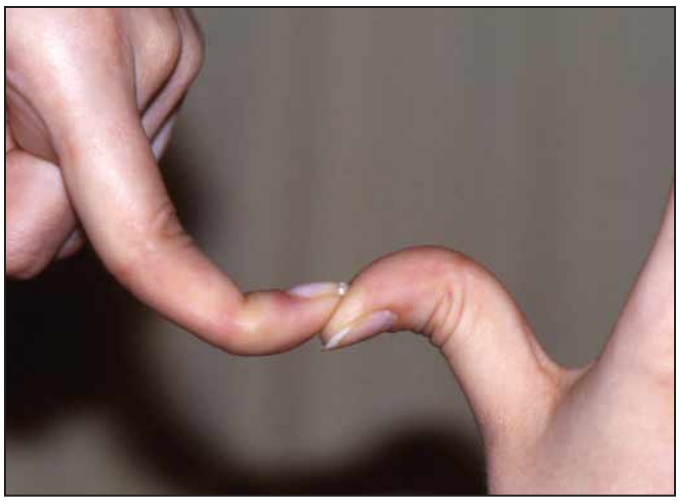
It has become increasingly clear that autonomic disturbances also play a significant role in the syndrome, according to Dr. Alan Hakim, another speaker at the meeting, who heads a hypermobility clinic at Whipps Cross University Hospital, London.

Three types of autonomic disturbances are predominant: syncopal, cardiorespiratory, and gastrointestinal. "It's phenomenal how many patients report presyncopal symptoms such as faintness and dizziness," Dr. Hakim said. In one series of 48 patients with JHS, 78% were found to have orthostatic hypotension, postural orthostatic tachycardia syndrome, or orthostatic intolerance (Am. J. Med. 2003;115:33-40).

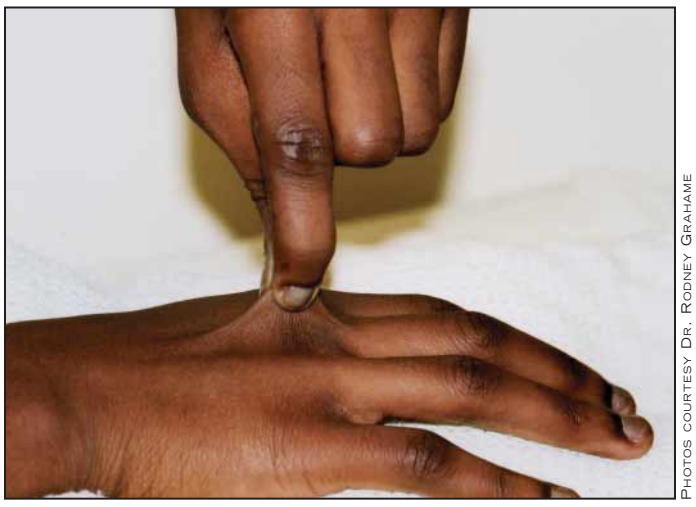
Cardiorespiratory findings include shortness of breath, while the gastrointestinal problems are similar to those seen in irritable bowel syndrome. Approximately 30% of patients report at least one autonomic disturbance, 20% have two, and 13%-14% report three autonomic disturbances, Dr. Hakim said.

Joint hypermobility syndrome is a genetically determined disorder of matrix proteins that is characterized by articular hyperextension, skin changes, marfanoid body habitus, and other manifestations such as hernias and varicose veins. It is considered benign, in that it does not significantly alter life expectancy, but affects quality of life and may be associated with frequent dislocations and early osteoarthritis and osteoporosis.

JHS manifests an autosomal dominant pattern of inheritance, with affected persons expressing varying degrees of joint laxity and other symptoms (Best Pract. Res. Clin. Rheumatol. 2003;17:989-1004). Women are affected approximately three times more commonly than men are. ■



Hyperextensibility of the finger joint is typical of hypermobility syndrome. A common misconception is that the diagnosis requires the involvement of four or more joints. In fact, only one joint need be hypermobile.



PHOTOS COURTESY DR. RODNEY GRAHAME

Excessive stretchiness of skin is associated with hypermobility syndrome. Paper-thin scars and stretch marks are other common cutaneous findings.