Joint Hypermobility Syndrome Underdiagnosed

By Nancy Walsh
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Glasgow, Scotland — Joint hypermobility syndrome is profoundly underdiagnosed and its impact underesti- mated despite its being one of the most common causes of widespread chronic pain and indeed may be the most common rheumatic disorder. Dr. Rodney Gra- hame 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, evaluat- ing them for their rheumatic complaints but also to see how many fit the JHS (joint hypermobility syndrome) pheno- type. What I found exceeded my expecta- tions,” said Grahame, who was in- volved in the development of what are known as the revised Beighton criteria. “That means that if most rheumatologists are diag- nosing only 10 cases per year, they are iden- tifying only 4.5% of cases,” he said (Rheumatology [Oxford] 2001;40:559-62). Extrapolating from those data would sug- gest that if most rheumatologists are diag- nosing only 10 cases per year, they are iden- tifying only 4.5% of cases. “That means that in England, 103,568 cases are missed annu- ally, as are 593,930 cases in the United States. These are appalling statistics,” he said.

Evaluation of joint hypermobility tradi- tionally is done using the Beighton scoring system, said Dr. Grahame, who was in- volved 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 fifth metacarpophalangeal joint to 90 de- grees or more, and 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 hypermo- bility requires the involvement of four or more joints. In fact, only one joint need be hypermobile in JHS, he said.

Other typical presenting symptoms in- clude acute or chronic pain and joint clicking. There may be a history of subluxa- tions or dislocations, because the laxity of the ligaments leads to joint instability. Pain avoidance, typically beginning in child- hood or adolescence, often leads to mus- cle deconditioning. Cutaneous findings in- clude 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 au- tonomic disturbances also play a significant role in the syndrome, according to Dr. Alan Hakim, another speaker at the meet- ing, who heads a hypermobility clinic at Whipp’s Cross University Hospital, London. Three types of autonomic disturbances are predominant: syncopal, cardiorespira- tory, and gastrointestinal. “It’s phenome- nal how many patients report presyncopal symptoms such as faintness and dizzi- ness,” Hakim said. In one series of 48 patients with JHS, 78% were found to have orthostatic hypotension, postural or- thostatic tachycardia syndrome, or ortho- static intolerance (Am. J. Med. 2003;115:33-40).

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

Joint hypermobility syndrome is a ge- netically 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 sig- nificantly alter life expectancy, but affects quality of life and may be associated with frequent dislocations and early os- teoarthritis and osteoporosis.

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