Advances in Assessment of Hypermobility Related Disorders

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Abstract

There has been increasing recognition in recent years of the prevalence and impact of symptoms which extend beyond the musculoskeletal system on the lives of people with hypermobility related disorders. This has led researchers to develop more comprehensive assessment tools to help direct and monitor treatment. This article presents some of the latest assessment and diagnostic developments and their implications for practice from a physical therapy perspective.

Key words

Hypermobility, Assessment tools

Introduction

Over the past decade, there has been increasing recognition that joint hypermobility (JH) is a risk factor for pain, as it may predispose to altered movement patterns, muscle imbalance and is associated with overload injuries, as well as joint dislocations, subluxations with subsequent lesions of ligaments, sprains of surrounding soft tissues and elongation or irritation of peripheral nerves (Syx et al., 2017, Tobias et al., 2013). Furthermore, research has also revealed that symptoms can also extend beyond the musculoskeletal system (Tinkle, 2020). Up to two thirds of patients with symptomatic JH also report multisystemic symptoms (Hakim and Grahame, 2004), including functional gastrointestinal symptoms, cardiovascular dysautonomia, urogenital problems, fatigue and anxiety which impact on disability and quality of life (Bulbena et al., 1993, Zarate et al., 2010, Mastoroudes et al., 2013a, Mastoroudes et al., 2013b, De Wandele et al., 2014, De Wandele et al., 2016, Fikree et al., 2017). Researchers and clinicians have therefore recognized the need for more extensive examination tools to assess the extent and impact of hypermobility and associated conditions

in order to direct treatment and also to monitor change. This paper provides and overview of recent developments from a physical therapy perspective.

Hypermobility musculoskeletal assessment and diagnostic tools

The two most common tools for identifying generalized hypermobility are The Beighton score (Beighton et al., 1973) and the 5-part hypermobility questionnaire (5HQ) (Hakim and Grahame, 2003). The Beighton score comprises 9 maneuvers involving the elbow, 5th metacarpophalangeal, thumb/wrist, and knee joints and the lower lumbar spine/pelvis region. It was originally designed as an epidemiological tool to screen for Generalized Joint Hypermobility (GJH), with a cut point of ≥ 4 defining GJH. Recent cohort research has recommended a variable cut-off for the age and gender (Singh et al., 2017; Chan et al., 2018). Current recommendations are ≥ 6 joints for children aged 6-17, ≥ 5 points for those aged 18 – 50 years and >4 points for persons older than 50 years. The current guidelines allow an additional point to be added to the Beighton score if there are two or more affirmative responses to the 5HQ (Malfait et al., 2017). An advantage of the Beighton score is that it is quick and requires a goniometer only when joint range is unclear. The 5HQ is a sensitive and specific tool which is particularly useful in adults whose joints may have stiffened with age. An answer of 'yes' to two or more of the questions gives a very high prediction of the presence of GJH; sensitivity and specificity have been reported between 71–84% and 77–89% respectively (Hakim and Grahame, 2003, Moraes et al., 2011).

In order to provide greater detail and to guide treatment, more comprehensive multidirectional peripheral joint hypermobility and laxity assessment tools have been developed for use by therapists. The Lower Limb Assessment Score (LLAS) was developed initially for use in children by Ferrari and colleagues (Ferrari et al., 2005) and more recently has been validated for adults and in football players (Meyer et al., 2017, Paul Johnson et al., 2019). The assessment comprises 12-item tests covering the major joints of the lower limbs including the hip, knee, ankle, midfoot, forefoot and 1st metatarsophalangeal. A cut-off score of ≥7/12 is used to determine generalized and lower limb specific hypermobility. The LLAS has been shown to have excellent intra and inter-rater reliability and is able to discriminate between different extents of lower limb hypermobility with sensitivity of 68% and specificity of 86% (Meyer et al., 2017).

The Upper Limb Hypermobility Assessment Tool (ULHAT) was devised by Nicholson and Chan in 2018 to complement the LLAS (Nicholson and Chan, 2018). This tool also involves 12-item multiplanar tests and includes the glenohumeral joint, elbow, distal radioulnar, thumb, and metacarpophalangeal joints. The ULHAT has been validated for use in adult

populations. Excellent inter-rater reliability has been established (ICC = 0.92) and the cut-off score of ≥7/12 has been shown to give sensitivity 84% and specificity 77% (Nicholson and Chan, 2018).

Further to this, researchers have been exploring the potential role of strain elastography (SEL), an ultrasound-based system designed to examine the elasticity of human structures by providing color-coded images (Alsiri et al., 2019). The hypothesis behind this, is that genetic variants involved in the altered connective tissue in hypermobility-related disorders are thought to reduce the mechanical rigidity of the soft tissues in the musculoskeletal system, and this laxity may be identified using SEL. Other factors could lead to significant softening of the musculoskeletal system in people with Hypermobility Spectrum Disorders (HSD) and Ehlers Danlos syndromes (EDS), including muscular weakness, reduced muscle-tendon stiffness, the pain-muscular inhibition cycle, and reduced activity (Rombaut et al., 2012). Initial research findings show encouraging results. Using three SEL image analysis approaches, people with HSD showed significant reductions in the elasticity of muscular and tendinous structures including the biceps brachii and brachioradialis muscles, and patellar and Achilles tendons (Alsiri et al., 2019). People with HSD complained of multiple joint pain which demonstrated a moderate correlation with musculoskeletal elasticity. SEL may therefore provide a supplementary tool for diagnosing HSDs, monitoring, and assessing the effectiveness of physical therapy, strengthening programs and other management regimes.

Functional assessments

While joint range and tissue elasticity are important to assess from a body structure and function perspective, functional assessments are needed to help further understand how movement and balance is impacted. Sophisticated laboratory studies of perturbation reveal that there is a delay in muscle activation in people with symptomatic hypermobility which may contribute to the impaired balance and control and reduced strength and stability observed by clinicians (Bates et al., 2021, Keer and Simmonds, 2011). Gait studies reveal altered kinematics in children (Fatoye et al., 2011, Rombaut et al., 2011). Adults with hypermobile EDS (hEDS) have been observed to walk more slowly and with shorter step and stride length and have a greater number of falls compared to healthy controls (Rombaut et al., 2011).

Given the association between hypermobility related disorders and Developmental Coordination Disorder (DCD) in both children and adults (Castori and Voermans, 2014, Clark and Whitall, 2011, Moore et al., 2019), tools such as the Developmental Coordination Disorder Questionnaire (DCDQ) which has very good sensitivity and specificity for identifying

probable DCD in children can be used for initial screening (Wilson et al., 2009), while the newly developed Functional Difficulties Questionnaire can be used in adults (Clark, Dorey & Williams, 2016). These tools alongside other functional assessments can be used to direct the focus and approach to rehabilitation. For example, a referral to occupational therapy may be beneficial to help children and adults to develop skills for day-to-day activities and well-being. Physical therapists may focus on movement planning, balance and coordination. Ergonomic adaptions may be required, such as grips on pens, modified eating utensils and Velcro straps instead of shoelaces.

Within the athletic domain, the LLAS in combination with the Lower Extremity Grading Scale (LEGS) can be been used to drill down to assess lower limb neuromuscular control (Smith et al., 2018). The LEGS assessment includes three functional tests; the Y-balance Test for measuring dynamic balance, the Drop Vertical Jump Test for jump-landing mechanics and the Triple Crossover Hop for Distance Test (Figure 1). An example of how these assessments can be combined, is a study undertaken in a group of young Greek gymnasts by Athanasoglou and colleagues in 2020 (Athanasoglou et al., 2020). The prevalence of lower limb hypermobility using the LLAS in the gymnasts was found to be 32%. Gymnasts with lower limb hypermobility had significantly lower mean neuromuscular performance scores compared to those without hypermobility ($p = 0.016$). Further analysis into the subcomponents of the LEGS assessment, revealed that although there was no difference in dynamic balance, lower limb strength and control, measured by the Y-balance and Triple Crossover Hop for Distance test respectively, hypermobile gymnasts demonstrated significantly lower mean scores in the Drop Vertical Jump Test ($p < 0.001$). The identification of the altered landing mechanics in the jump test can be used by coaches and physical therapists to inform targeted injury prevention, functional strength and conditioning programmes.

Patient reported impact and quality of life assessment tools

To assess and monitor the impact of joint hypermobility on quality of life, the 55 item Bristol Impact of Hypermobility (BIOH) questionnaire has been designed and has been validated in adult populations. The BIOH has strong psychometric properties including test-retest reliability and discriminative validity (Palmer et al., 2017, Palmer et al., 2020).

While the BIOH is helpful for exploring musculoskeletal implications, to improve insight into the broad symptom profile and to help direct multidisciplinary care and monitor treatment, researchers have developed ‘The Spider’, a 25-item questionnaire tool involving 8 domains aims to evaluate the impact of the more common multisystem symptoms associated with

HSD and hEDS. The completed Spider produces a radar graph shaped like a spider's web, that provides a visual overview of a patient's symptom profile (Figure 2). In clinics, this screening tool will potentially allow health care professionals to quickly identify which problems should be assessed and treated as a priority within the multidisciplinary team. The IQOLA guidelines for cross-cultural validation of health-related quality of life questionnaires are being used to produce a robust tool. Face and content validity have been undertaken with expert clinicians and patients (De Wandele et al., 2020). Initial research in young people aged 12-18 years has established good convergent validity for the pain ($r = 0.7$, $p=0.01$), fatigue ($r=0.74$, $p=0.01$), anxiety ($r= 0.6$, $p=0.01$) and depression ($p=0.8$, $p = 0.01$) and very good discriminative validity has als been established (Ewer & Simmonds 2021; Tang et al., 2020). Further research is underway to validate the other domains in both adolescent and adult groups.

Conclusion

With the increasing recognition of the prevalence and impact of symptoms which extend beyond the musculoskeletal system on the lives of people with hypermobility-related disorders researchers are developing more comprehensive assessment tools to help understand problems and to direct and monitor treatment. Some of these assessments will remain in the domain of researchers while others have excellent potential for clinical utility.

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Conflict of Interest

No conflicts of interest

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