Are Diagnostic Criteria for General Joint Hypermobility and Benign Joint Hypermobility Syndrome Based on Reproducible and Valid Tests? A Review of the Literature

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ABSTRACT. ***Objective.*** In this review we focus on current knowledge of the reliability of tests and diagnostic criteria for generalized joint hypermobility (GJH) and benign joint hypermobility syndrome (BJHS).

Methods. Currently, The British Society of Rheumatology recommends the Beighton scoring system. With this approach, GJH is judged present when 4 or more of 9 tests are positive. Curiously, only one inter/intrarater reproducibility study is available and it uses a cutoff level of 6, rather than the Beighton-recommended 4 positive tests.

Results. Using a 6 cut level, intra- and interobserver kappa scores were 0.75 and 0.78, respectively. Beighton scoring recommendations have been correlated with a global joint mobility index as well as with 2 other scoring systems, the Carter and Wilkinson, and the Rotès-Quérol. All illustrate high concurrent validity with one another. For the recently proposed Brighton criteria diagnosing BJHS no reproducibility studies exist. In the latter, the recommendations reflect high nosographic sensitivity and specificity while predictive values for positive test scores are poor.

Conclusion. In general, the reproducibility of the various tests seems to be good, especially when performed by experienced rheumatologists. (First Release Jan 15 2007; J Rheumatol 2007;34:798–803)

Key Indexing Terms:
JOINT LAXITY HYPERMOBILITY RELIABILITY CRITERIA SYNDROMES

In 1967 Kirk, *et al*[1] drew attention to the possibility that musculoskeletal complaints in association with generalized joint hypermobility (GJH) are likely to represent what the authors label "hypermobility syndrome" (HS). With this in mind, 2 questions arise: (1) Does the syndrome really exist both clinically and pathologically? and (2) If it exists are there reliable tests and criteria that reproducibly diagnose the syndrome?

For some years, a special interest group within the British Society of Rheumatology has focused on joint hypermobility (JH), GJH, and HS, also called benign joint hypermobility syndrome (BJHS). "Benign" is the term used in contrast to more serious and potentially life-threatening musculoskeletal syndromes, such as Ehler-Danlos syndrome (EDS), Marfan syndrome, and osteogenesis imperfecta[2]. Currently, in some sectors, more attention is being paid to "local hypermobility" in relation to localizing musculoskeletal complaints[3]. General clinical knowledge of both GJH and BJHS seems consistently poor, as illustrated in 2 recently published surveys[4,5]. Therefore, distribution of pragmatically usable information and education on a wide front within the world rheumatologic community seems relevant. Before this occurs there needs to be an assurance that criterion-based clinical diagnostic methods are available. We reviewed the current state-of-the art knowledge regarding reliability, i.e., reproducibility and validity, of tests and diagnostic criteria. A second article focuses on JH epidemiology and suggested criteria for identifying BJHS[6].

MATERIALS AND METHODS

Four GJH clinical assessment methods are most common: the Beighton[7], Carter and Wilkinson[8], and Rotès-Quérol[9] methods, and a Global Joint Mobility Index[10]. For the BJHS approach, only one assessment method has been suggested — the Brighton recommendations[2].

Focal points in our review discuss JH, GJH, and BJHS diagnostic criteria, along with test/retest reproducibility and concurrent validity. To achieve this end, a key word search of PubMed, Cochrane Library, and PEDro was used to query the following: joint instability, hypermobility, joint luxation, back pain, shoulder injuries, sprain, children, age, sports injuries, and pregnancy. From the results, we reviewed hypermobility-related articles that used the above mentioned test methods.

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Defining terms

Normal joint movement variables. Rotès-Quérol[11] described the presence of normal joint ranges of motion (ROM) that vary within certain limits. However, according to the American Academy of Orthopedic Surgeons (AAOS) it is not possible to precisely determine mean joint mobility throughout the body[12]. As a result, the AAOS developed consensus-based estimates in degrees derived from statistical means based on reports from 4 committees of experts.

Localized or pauciarticular JH. Wood[13] comments that JH is a graded rather than an either/or phenomenon. Subsequently a consensus has developed that individual joint mobility follows a Gaussian distribution[14-17]. With this in mind, abnormal JH would reflect movements that deviate ± 2 standard deviation (SD) from the mean, i.e., the general, consensus-based estimate. For better or worse, we do not usually call for ROM measurements in degrees when testing for JH. Instead, Beighton tests (Figures 1A-E) that apply a dichotomous principle are widely used.

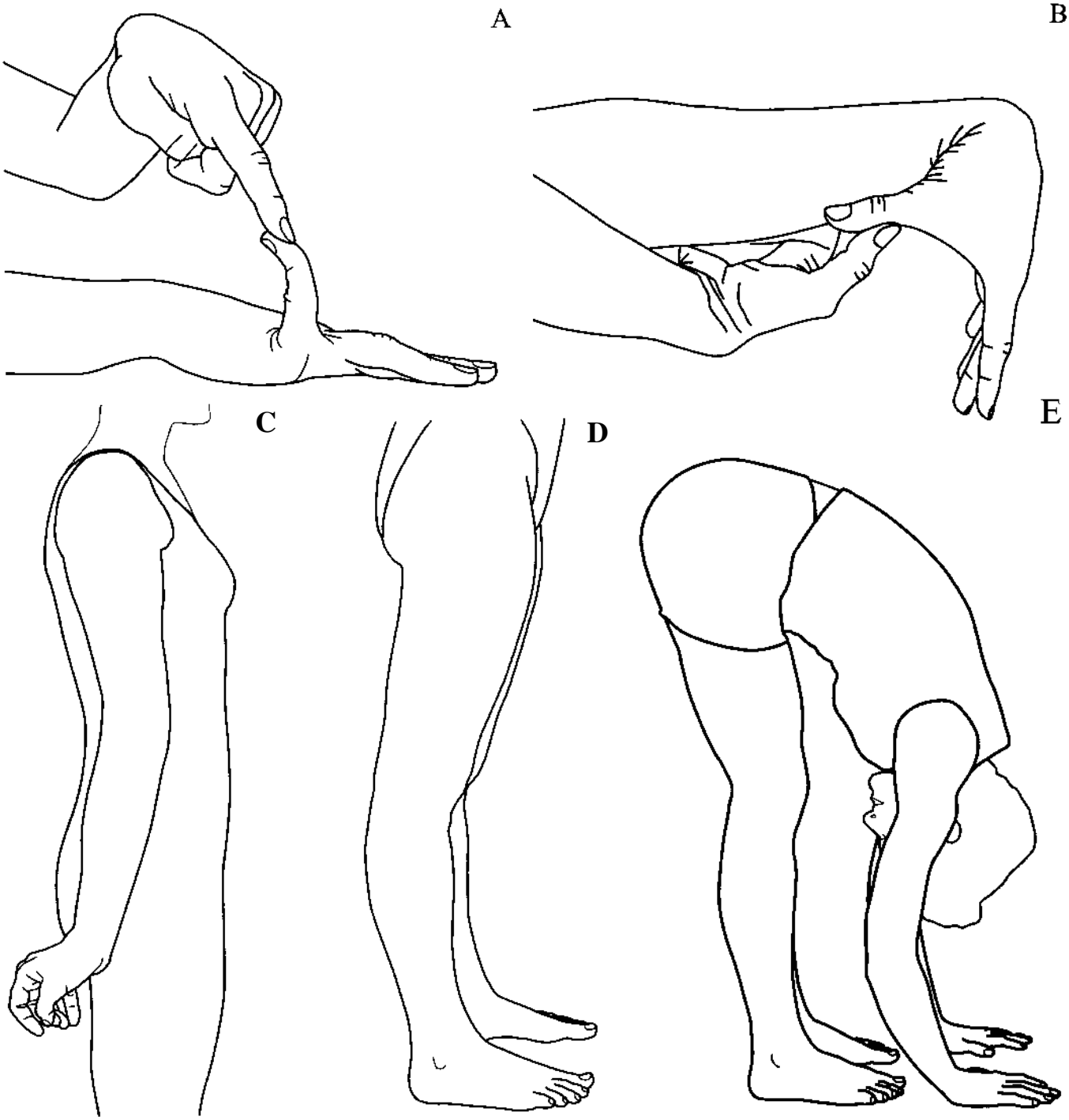

Figure 1. The Beighton tests for joint hypermobility. A. Passive dorsal flexion of the little fingers beyond 90°. B. Passive apposition of the thumbs to the flexor aspects of the forearm. C. Hyperextension of the elbows beyond 10°. D. Hyperextension of the knees beyond 10°. E. Forward flexion of the trunk, with the knees straight, so that the palms of the hands rested easily on the floor. The tests A-D are bilateral giving a score between 0 and 9.

Generalized joint hypermobility (GJH). Internationally there is no agreement on the definition of this entity. The various tests and test combinations used for GJH are described in Table 1. Some give a clear description of acceptable criteria and note that the diagnosis requires involvement of both upper and lower extremities[8]. Others are inexact in their definition but suggest that undue laxity of 3 of 5 joint pairs must be present for a positive diagnosis[1].

Beighton, *et al*[7,18] neither define nor use any criteria for GJH. The tests are used solely to describe the populations examined. They note that their EDS patient cohort frequently had joint luxation, and an increased incidence of osteoarthritis, when test scores were > 3 positive out of 5[18]. Using a revised 0–9 scoring system the authors note that 80% of female and 94% of male Tswana Africans (n = 1023) scored in the 0–2 ranges[7]. There is no discussion of a cutoff level for GJH.

The Rotès-Quérol, *et al* recommendations include more tests than described by other authors (Table 1). They also recommend different cutoff levels for children and adults[9].

Several papers, not mentioned in Table 1, offer a wide variety of assessment criteria[19-23], while others describe personal preferences[15,17,24-27].

There is no universal agreement for GJH criteria among authors using Beighton methods. Most define GJH as present when ≥ 4 of 9 tests are positive[28-31]. Others require ≥ 5 of 9 tests[32,33], while still others apply ≥ 6 of 9 tests[34]. Even among criterion-based recommendations there is a lack of consensus. For example, the Brighton BJHS recommendations uses ≥ 4 of 9 positive tests[2], compared to the Villefranche EDS recommendations that use ≥ 5 of 9[35].

Hypermobility syndrome (HS). The syndrome's name was revised in 1998, changing from HS to BJHS[2]. There have only been 2 attempts to define HS (Table 2). The more serious one suggests inclusion of both major and minor criteria[36].

Table 1. Review, in order of publication year, of the most often used tests for general joint hypermobility (GJH).

Test	Carter & Wilkinson[8]	Kirk, *et al*[1]	Beighton & Horan[18]	Rotès-Quérol, *et al*[9]	Beighton, *et al*[7]	Beighton, *et al*[35]	Grahame, *et al*[2]
Passive apposition of the thumb(s) to flexor aspects of the forearm(s)	+	+	+	+ (> 185°)	+	+	+
Passive hyperextension of fingers so that they lie parallel with the forearm extensor side	+	+					
Passive hyperextension in the elbow(s) > 10°	+	+	+	+	+	+	+
Passive hyperextension in the knee(s) > 10°	+	+	+	+ (> 5°)	+	+	+
Excessive passive dorsal flexion of ankle and excessive foot eversion	+	+					
Passive dorsiflexion of the little fingers beyond 90°, with forearm flat on a table			+		+	+	+
Passive dorsal flexion of 2nd finger so that the angle between the distal phalanx and the table is > 100°				+			
Forward flexion of the trunk, with knees straight, so that the palms of the hands rest easily on the floor			+	+	+	+	+
Shoulder external rotation > 90°				+			
Cervical rotation > 90° and cervical side flexion > 50°				+			
Hip abduction, bilateral > 90°				+			
Dorsal flexion in metatarsophalangeal joint >90°				+			
Lumbar lateral flexion with head and column below the horizontal plane				+			
Criterion for hypermobility	> 3 of 5 positive tests; in both upper and lower extremities	> 3 of 5 positive test pairs	A score from 0–5 to describe a population with EDS	Grade I: 0–2 Grade II: 3–5 Grade III: 6–7 Grade IV: 8–10	A score from 0–9 to describe a population of Tswana Africans	A score ≥ 5 of 9 positive tests indicates GJH	A score ≥ 4 of 9 positive tests indicates GJH

EDS: Ehlers-Danlos syndrome.

Table 2. Review of hypermobility syndrome definitions.

Publication	Test and Clinical Signs		Syndrome Criterion
Kirk JA, *et al*[1]	Carter & Wilkinson's tests with > 3 positive joint pairs. Musculoskeletal complaints (without any other systemic rheumatic disease)		None
Grahame R, *et al*[2]	Major criteria 1. Beighton score ≥ 4/9 (either currently or historically) 2. Arthralgia for longer than 3 months in 4 or more joints	Minor criteria 1. Beighton score of 1, 2, or 3/9 (0, 1, 2, or 3 if age 50+) 2. Arthralgia (≥ 3 mo) in 1–3 joints, or back pain (≥ 3 mo) or spondylosis, spondylolysis/spondylolisthesis 3. Dislocation/subluxation in more than one joint or in one joint on more than one occasion 4. Soft tissue rheumatism ≥ 3 lesions (e.g., epicondylitis, tenosynovitis, bursitis) 5. Marfanoid habitus (tall, slim, span/height ratio > 1.03, upper/lower segment ratio < 0.89, arachnodactyli (+ Steinberg/wrist signs) 6. Abnormal skin: striae or hyperextensibility, thin cutis or papyraceous scarring 7. Eye signs: drooping eyelids or myopia or antimongoloid slant 8. Varicose veins or hernia or uterine/rectal prolapse	BJHS is diagnosed in the presence of 2 major criteria OR 1 mjaor and 2 minor criteria OR 4 minor criteria 2 minor criteria will suffice where there is an unequivocally affected first-degree relative BJHS is excluded by presence of Marfan or EDS (other than the EDS hypermobility type) Criteria major 1 and minor 1 are mutually exclusive, as are major 2 and minor 2

RESULTS

Reliability of hypermobility tests

The majority of the available scientific papers dealing with hypermobility research are based on tests and procedures described in a few basic publications (Table 1). Importantly, none of the authors present a systematic analysis of intra- and inter-observer reproducibility along with studies of concurrent validity.

Reproducibility. Allander, *et al*[14] recognized significant inter-observer differences when examining ROM in the left metacarpophalangeal joint of the thumb and shoulder joints. Fairbank, *et al*[15] demonstrated high reproducibility for repeated ROM examinations in upper limb joints, but found low reproducibility when examining knee joint extension.

The interobserver variability of various hypermobility tests from 3 commonly used sets of tests was good to excellent with kappa values consistently between 0.68 and 0.93, except for 2 of the 11 Rotès-Quérol assessments[33]. Two of Beighton's tests and 4 of Rotès-Quérol's tests were slightly modified from their original descriptions.

In another study, interobserver variability between 2 experienced rheumatologists had kappa values between 0.44 and 0.82 for 4 Beighton tests[37]. In the same study a comparison between rheumatologists and laymen resulted in kappa values > 0.60 in only 2 tests, namely, the 2 tests characterized by an easily defined endpoint (Figure 1B, 1E).

Concurrent validity. No concurrent validity studies have been published. One study argues that a positive Beighton test is identical to a joint mobility + 3 SD beyond the mean ROM indicating that any positive test actually identifies a hypermobile joint[15].

Reliability of criteria for GJH

Rotès-Quérol, *et al*[9], in contrast to others[7,8,18], have outlined some GJH testing recommendations. They should: (1) be easy to measure; (2) be applied to joints in which the primary movement takes place in only one plane (elbow, knee, finger joints); (3) be extension movements because they are less affected by soft tissue interposition; (4) measure the angle of maximum mobility in degrees using lines and planes defined by the segments of the skeleton; and (5) have a cutoff level that identifies 20% of the general population as hypermobile.

Reproducibility. In 1996, Mikkelsson, *et al* published the first and only existing intra- and interobserver reliability article dealing with GJH diagnostic criteria[34]. Using Beighton scoring and an empirically applied cutoff level of ≥ 6 of 9 positives, kappa values were 0.75 (intraobserver) and 0.78 (interobserver).

Concurrent validity. The concurrent validities of the GJH cutoff points are not analyzed in the early publications[1,7,8,18]. Likewise, a GJH diagnostic cutoff point of ≥ 5 positive tests out of 9 in the Villefranche criteria is recommended without discussing the authors' reasoning[35].

In one article, Beighton tests using 0–9 scoring correlated well with a Global Index: $r = 0.81$ ($p < 0.001$). A cutoff level for GJH is not discussed[10].

When 2 experienced consultant level rheumatologists compared the Carter and Wilkinson protocol (3 of 5 tests) with the

Beighton, *et al* protocol, kappa values were in the 0.94 to 0.96 ranges, depending on whether the Beighton scoring cutoff was ≥ 4 or ≥ 5 of 9 tests[33]. When the Rotès-Quérol's scoring system — 5 or 6 of 11 tests — was compared with the 2 other scoring systems, kappa scores were less persuasive, but still above 0.60.

Reliability of criteria for BJHS

Reproducibility. Consensus-based BJHS diagnostic criteria were revised in 1998[2]. No reproducibility studies have been performed.

Validity. Criterion-based validity for diagnosing BJHS cannot pass a concurrent validity analysis because there is no clear and unambiguous gold standard that can be reliably measured. To date, no BJHS predictive validity study has been performed. In 1993, a special interest group under the British Society of Rheumatology reported on the comparative examinations of 43 persons with BJHS and 43 healthy controls[38]. Sensitivity and specificity for empirically agreed-upon diagnostic criteria were identical at 93%. The paper does not discuss the positive predictive value of negative findings in relation to positive findings, making their conclusions difficult to transfer to the clinical situation.

DISCUSSION

Currently, the Beighton tests and hypermobility scoring system has gained widespread international acceptance. How can this be, especially when detailed descriptions of test procedures have yet to be reported? Other concerns: Has there been any specific documentation for the choice of the individual joints and for the combination of joints for hypermobility testing? And if the tests are documented, are they reliable?

In 2 scoring system publications, the authors do not describe their methods with specific diagnostic details. Neither do they make arguments for their particular test selections[7,8]. Carter and Wilkinson[8] and Rotès-Quérol, *et al*[9] include upper and lower extremity joints. Only the Rotès-Quérol study discusses the reasoning that supports the recommendations. On the other hand, neither system has widespread current use.

Is there scientific evidence that the Beighton tests are sufficiently specific and sensitive to diagnose all abnormally hypermobile joints? According to Fairbank, *et al*, a positive Beighton test occurs when ROM exceeds mean + 3 SD[15]. This statistical standard implies that hypermobility is present in only 0.5% of the general population. In a biological context, however, abnormality is generally considered present when the measured parameter exceeds mean + 2 SD, which implies that 2.5% of the general population should be considered hypermobile. With this in mind, the question still to be answered is: Are the Beighton tests sensitive enough to diagnose all persons with abnormal hypermobile joints?

Beighton tests as well as the GJH testing seem to be reproducible in the hands of experienced examiners, in contrast to the situation when inexperienced rheumatologists or laymen perform the tests. However, low kappa values do not necessarily mean that tests are difficult to reproduce. It can reflect that the reproducibility trial is not optimized[39].

The reproducibility of BJHS diagnostic criteria is currently unknown.

As clinicians, we would like to know the predictive value of both positive and negative tests. These values can be calculated, applying Bayes' theorem[40], when the sensitivity and specificity figures of the tests and the overall prevalence of the condition are known. As an example: If the general overall prevalence of BJHS in an adult Caucasian population is 2–4%, and sensitivity and specificity of the criteria are 93% as published, then the Bayesian calculations result in a positive predictive value for a positive test in the range 21.3–35.6%. A positive predictive value for a negative test, on the other hand, falls in a range of 99.5–99.6%.

This means that making a positive diagnosis when examining the general population is a probable dilemma, but ruling out BJHS is quite easy.

Current JH testing procedures have been in place for many years and are to some extent reproducible in the hands of experienced rheumatologists. To date, however, the reproducibility of a specific GJH scoring system has been partially analyzed in only one study. We conclude therefore that: (1) Fundamental clinical research focusing on joint hypermobility can be improved by establishing a clear and unambiguous standard for test performances. (2) An internationally reproducible "consensus among experts" should be developed for age, sex, and racially dependent ROM, i.e., an agreed upon gold standard for normal joint mobility. (3) Such a gold standard will permit creation of a consensus-based cutoff level for general joint hypermobility. (4) In turn, the new methods will allow us to develop and validate all JH scoring systems.

Finally, HS or BJHS testing procedures and diagnostic criteria have been in place for a number of years. Because there are no universally accepted diagnostic standards, i.e. no gold standard, BJHS evaluations are a particular diagnostic problem. No symptom-based reproducibility or validity studies are available. Because there is no standard for evaluating signs and symptoms, future syndrome-related validity studies will have to be developed on the basis of construct validity using criteria presumed to be part of the syndromes. For these conditions, a process similar to that recommended above is also offered for consideration.

Results of the above-mentioned studies should be published consecutively. If they show that tests and criteria are reliable, they should be taught to medical students, residents, and, in particular, to rheumatologists.

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