# Naval Medical Center Portsmouth
# Rheumatology Referral Guidelines

| **Diagnosis:** | **Joint Hypermobility Syndrome (formerly Ehlers-Danlos hypermobile type)** |
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| Clinic Name | Rheumatology (NMCP) |
| Clinic Phone Number | 757-953-2160 or 2161 |
| On Call Numbers | 757-860-5702 |

## 1. Indications for Specialty Care:

- Evaluation and management recommendations of a hereditary connective tissue disease, other than Joint hypermobility syndrome (JHS) or Ehler-Danlos Syndrome (EDS) hypermobile type.
- Refractory JHS or EDS hypermobile type patients with concern for another underlying contributing cause/disease to their symptomatology.
- Active Service Members undergoing PEB for a hereditary connective tissue disease.

*****Referral should not be given to merely reaffirm a previous diagnosis of a non-rheumatic disease. *****

## 2. Quality Consult Criteria

**When referring a patient, please include as much of the following information as possible (OK to cut and paste this into consult request)**

1. Provisional diagnosis
2. Duration of Problem
3. Prior treatments
4. Current treatments/medications
5. Diagnostic studies obtained (imaging, labs, other tests, etc.)
6. Primary reason for consult
7. Use of referral guidelines

## 3. Diagnosis Definitions

- Joint hypermobility (JH) is joint range movement exceeding what is considered normal for a specific joint. This normal range requires one to take into account an individual's age, gender and ethnicity. JH can be localized (affecting a few joints) or generalized (affecting multiple joints) (1).
- Generalized joint hypermobility (GJH) by itself does not necessarily indicate a disease nor lead to symptoms.
- GJH is defined by a score of ≥5 on the 9-point Beighton Scale (1, 2). See table below.

- Joint hypermobility syndrome (JHS), also termed benign hypermobility syndrome (BHS), is a connective tissue disorder characterized by chronic musculoskeletal pain due to joint hyperextensibility, for which a patient meets the Brighton Criteria (1-3).

- JHS resembles Ehlers-Danlos Syndrome (EDS) hypermobility type, and some experts consider it a milder variant, but currently this is debated and not fully elucidated.

## 4. Initial Diagnosis and Management

- The diagnosis of JHS is primarily clinical and can be established in the primary care setting, without subspecialty consultation. Patient presentation can be variable and currently there is no single pathophysiologic mechanism by which patients develop JHS. The most important aspect of diagnosing JHS, is distinguishing it from other forms of hereditary connective tissue diseases, such as EDS, Marfan's Syndrome (MS) and Osteogenesis Imperfecta (OI), as these other entities have the potential for life-threatening consequences (1-4).

- The primary role of evaluation is in the clinical examination as it screens for GJH, determines extent of disease burden and assists with excluding other connective tissue diseases (4). A screening 5-point questionnaire can assist the primary care provider on the assessment of JH (Table 1). At least 2 positive responses on the screening questionnaire have 84% sensitivity and 85% specificity for JHS, as characterized by the Brighton Criteria (4).

Table 1. 5-Point Screening Questionnaire (5)

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

Positive responses to 2 of these questions have a sensitivity of 84% and specificity of 85% for JHS, as characterized by the Brighton Criteria for JHS (4, 5).

- In addition to a general physical examination, a detailed musculoskeletal examination should be performed, including assessment of the degree of hypermobility of affected joints. This can be assessed and documented by the Beighton Score, which is a 9-point score and positive with ≥4 points (thumb apposition to the flexor aspect of the forearm, passive dorsiflexion of the MCP to 90 degrees, passive hyperextension of the elbow greater than 10 degrees, passive hyperextension of the knee greater than 10 degrees and forward flexion with hands flat on the floor and knees extended).

- Additionally, the patient's joints should be assessed for synovitis, deformity and redness. Patients with JHS should also be assessed for scoliosis, lordosis, pes planus, genu valgum, patellar subluxation, marfanoid habitus, varicose veins, rectal/uterine prolapse and thin, translucent, extensible skin. To diagnose JHS, the patient should meet the Brighton Criteria (Table 2), and other heritable connective tissue diseases should be excluded. There are no specific routine laboratories or imaging diagnostics and these should be performed on an individual basis. Specifically, the routine testing of serology is not recommended, unless the clinical history is suggestive of an underlying connective tissue disease, such as systemic lupus erythematosus.

- See Brighton Criteria as noted below.

| Table 2. Brighton Criteria (4) | |
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| **Major Criteria** | **Comment** |
| **Beighton Score ≥4** | |
| **Polyarthralgias** | **Joint pain for >3 months in ≥4 joints** |
| **Minor Criteria** | |
| **Beighton Score <4** | **Beighton score of 1,2 or 3 in <50 years of age**<br>**Beighton score of 0,1,2 or 3 in >50 years of age** |
| **Oligoarthralgias** | **Joint pain >3 months in 1-3 joints, or**<br>**Back pain for >3 months, or**<br>**Spondylosis, spondylolysis, spondylolisthesis** |
| **Dislocation or Subluxation** | **In >1 joint, or**<br>**In 1 joint on >1 occasion** |
| **Soft Tissue Lesions** | **≥3 soft tissue lesions (i.e. epicondylitis, tenosynovitis, bursitis** |
| **Marfanoid Habitus** | **Armspan > height (>1.03 ratio), or**<br>**Upper segment < lower segment (<0.89) ratio, or**<br>**Arachnodactyly** |
| **Skin Abnormalities** | **Skin striae, or**<br>**Hyperextensibility, or**<br>**Abnormal scarring** |
| **Ocular Signs** | **Drooping eyelids, or**<br>**Myopia, or**<br>**Antimongoloid slant** |
| **Varicose veins, Hernia, or Uterine/Rectal prolapse** | |
| **Mitral Valve Prolapse** | |
| **Exclusion – Presence of Marfan Syndrome or Ehlers-Danlos Syndrome (other than hypermobility type)** | |

- JHS is associated with a variety of other conditions to include musculoskeletal pain, fatigue, dysautonomia, headache, abdominal/pelvic pain and mood disorders. The musculoskeletal pain does not follow any typical pattern or distribution. The weight-bearing joints (ankle and knees) are more commonly affected and typically have non-inflammatory symptoms (worse with activity, limited morning stiffness) (4). Patients with JHS can have impaired proprioception, which may lead or exacerbate pain symptoms secondary to joint laxity or dislocations. Furthermore, recurrent dislocations can cause early secondary osteoarthritis, exacerbating pain symptoms.

- Again, the most important aspect of diagnosis is excluding other hereditable connective tissue diseases, which can have life-threatening consequences. There currently is limited evidence based data to support one specific therapy for patients with JHS. The most important intervention for patients with JHS is lifestyle modification. Patients should be advised to limit excessive joint movement, which can aggravate symptoms and potentiate further injury. Regular training is essential, but overtraining and excessive focus on joint flexibility may lead to injury. Bracing may be helpful for prevention of further injury and improve gait.

- Referral to physical therapy for joint protection strategies and tailored, personalized exercise regimens is recommended for the majority of patients. It is known that laxity of joints decrease with the normal aging process, and therefore some patients improve in symptoms without additional therapy.

- There is currently no data to support the use of opiates in the management of patients with JHS. Opiates carry the risk of dependency, addiction and potential for abuse, and therefore given their risk without benefit of use, are not recommended for the management of JHS and should be avoided.

## 5. Ongoing Management and Objectives

- While the evaluation is underway, management in clinically stable patients is usually supportive.
  - Supportive care with physical and occupational therapy is essential to maintaining functionality and avoiding further damage.

- Referral to a geneticist may be indicated if there is strong suspicion for EDS, MS, and OI as above.

- Associated conditions should be evaluated based on an individual patient basis. Specifically, it can be difficult, if not impossible, to differentiate JHS from other chronic pain syndromes such as fibromyalgia. Functional pain syndromes, sleep disorders and mood disorders are highly associated with central pain sensitization syndromes; therefore in patients with refractory cases these entities should be considered. Additional information can be obtained within the fibromyalgia referral guidelines.

## 6. Criteria for Return to Primary Care

- Diagnosis of JH established in the absence of autoimmune disease.

- Management questions do not require a re-referral and are preferably handled as a conversation (potentially electronically) between the referring provider and the consulting Rheumatologist.

- Patients with concomitant autoimmune disease and JH may be co-managed with Rheumatology.

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| Date Adopted or Last Reviewed: | 01 Feb 2018 | By | CDR Shauna O'Sullivan<br>LCDR Jeffrey Eickhoff<br>LCDR Terrence Kilfoil<br>LCDR Jason Weiner |
| Referral Guidelines require review every three years. | | | |

## 6. Resources:


- Syx D, De Wandele I, Rombaut L, Malfait F. Hypermobility, the Ehlers-Danlos syndromes and chronic pain. Clin Exp Rheumatol. 2017; 35 Suppl 107(5):116-22.
- Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, et al. The 2017 international classification of the Ehlers-Danlos syndromes. American journal of medical genetics Part C, Seminars in medical genetics. 2017;175(1):8-26.
- Colombi M, Dordoni C, Chiarelli N, Ritelli M. Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlers-danlos syndrome hypermobility type compared to other heritable connective tissue disorders. American journal of medical genetics Part C, Seminars in medical genetics. 2015; 169c (1):6-22.
- Kumar B, Lenert P. Joint Hypermobility Syndrome: Recognizing a Commonly Overlooked Cause of Chronic Pain. The American journal of medicine. 2017;130(6):640-7.
- Hakim A, Grahame R. Joint hypermobility. Best practice & research Clinical rheumatology. 2003; 17(6):989-1004.

American College of Rheumatology: https://www.rheumatology.org/I-Am-A/Patient-Caregiver/Diseases-Conditions/Hypermobility-Juvenile

- Joint hypermobility Syndrome UpToDate: https://www.uptodate.com/contents/joint-hypermobility-syndrome#!
- Hypermobility Syndrome association: http://hypermobility.org/help-advice/hypermobility-syndromes/the-brighton-score/