# Summary of investigations - hEDS/HSD

The following extract is taken from:
Understanding hypermobile Ehlers-Danlos Syndrome and Hypermobility Spectrum Disorder

Redcliff-House Publications

NB/ The original article, on which the following extract is based, was written for the Hypermobility Syndromes Association by Dr A.J. Hakim
(hypermobility.org - 'An Update for Clinicians')
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## Chapter 3
## Diagnosis and Management

# Summary of investigations - HSD and hEDS

**This section has been written by Dr Alan Hakim and has been included in this book with the kind permission of the Hypermobility Syndromes Association. It is aimed primarily at clinicians (Hakim A.J 2017c (HMSA).**

A patients' concerns may be protean [ever-changing]. A long list of investigations and treatments is inappropriate for a summary of this nature. Detail regarding specific concerns can be found on the Hypermobility Syndromes Association website (hypermobility.org - 'An Update for Clinicians') and in the cited reference literature at the end of this page. Many aspects of care should involve guidance over self-management, and likely include physical treatments, medicines, and therapies, often running in parallel and managed in a multidisciplinary way.

The more common areas of investigation include:

### Musculoskeletal and fatigue blood tests:

- If there is any concern that joint and/or muscle pain may be due to an inflammatory or autoimmune disorder then the relevant blood tests should be undertaken.
- Blood tests may be required to exclude haematologic, endocrine, and metabolic causes for fatigue.

### Neuro-muscular Imaging:

- Radiographs, Ultrasound, MRI : imaging of joints / soft tissue may help to determine whether mechanical or inflammatory damage is present, impingement at the joint or of a nerve has arisen, or whether subluxation/listhesis etc. is occurring.
- Neuropathic concerns might require central nervous system imaging; peripheral tests including NCS/ EMG.

### Echocardiography

Echocardiography should be carried out if there is any concern on examination, or as part of the diagnostic work up for hEDS and other HDCT.

### Bowel and urogynaecologic investigations:

- Tests for helicobacter, coeliac, bacterial over-growth.
- Upper or lower GI endoscopy and functional bowel tests.
- Urodynamics and cystoscopy might be required to delineate a problem, as might hysteroscopy.

### The more common areas of management include (recent reviews cited in brackets and shown at the end of this page):

- Physical therapies (Engelbert et al, 2017).
- Pain Management (Chopra et al. 2017).
- Anxiety and Mood management (Bulbena et al. 2017).
- Fatigue (Hakim et al. 2017b)
- Reflux, nausea, and sluggish bowel (Fikree et al. 2017)
- Cardiovascular autonomic dysfunction (Hakim et al. 2017b)
- Management of gynaecological concerns

Very recent literature reviews in this field of medicine detail the current understanding of the associations with hypermobility-related disorders (in particular HSD and hEDS) and the treatment options available. These are cited in the references below:

Bulbena A, Baeza-Velasco C, Bulbena-Cabré A, Pailhez G, Critchley H, Chopra P, Mallorquí-Bagué N, Frank C, Porges S. Psychiatric and psychological aspects in the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet. 2017 Feb 10. doi: 10.1002/ajmg.c.31544. [Epub ahead of print] PubMed PMID: 28186381.

Castori M, Tinkle B, Levy H, Grahame R, Malfait F, Hakim A. A framework for the classification of joint hypermobility and related conditions. Am J Med Genet C Semin Med Genet. 2017 Feb 1. doi: 10.1002/ajmg.c.31539. [Epub ahead of print] PubMed PMID: 28145606.

Chopra P, Tinkle B, Hamonet C, Brock I, Gompel A, Bulbena A, Francomano C. Pain management in the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet.2017 Feb 10. doi: 10.1002/ajmg.c.31554. [Epub ahead of print] PubMed PMID: 28186390.

Engelbert RH, Juul-Kristensen B, Pacey V, de Wandele I, Smeenk S, Woinarosky N, Sabo S, Scheper MC, Russek L, Simmonds JV. 2017. The evidence-based rationale for physical therapy treatment of children, adolescents, and adults diagnosed with joint hypermobility syndrome/hypermobile Ehlers Danlos syndrome. Am J Med Genet Part C Semin Med Genet 175C:158–167.

Fikree A, Chelimsky G, Collins H, Kovacic K, Aziz Q. Gastrointestinal involvement in the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet. 2017 Feb 10. doi: 10.1002/ajmg.c.31546. [Epub ahead of print] PubMed PMID: 28186368.

Francomano C and Bloom L. 2017. http://ehlers-danlos.com/2017-eds-international-classification-webinar/

Hakim A. 2017c (HMSA) - Hypermobility Disorders; an update for clinicians. hypermobility.org/update-for-clinicians/ March 16th 2017

Hakim A, De Wandele I, O'Callaghan C, Pocinki A, Rowe P. Chronic fatigue in Ehlers-Danlos syndrome-hypermobile type. Am J Med Genet C Semin Med Genet. 2017 Feb 10. doi: 10.1002/ajmg.c.31542. [Epub ahead of print] PubMed PMID: 28186393.

Hakim A, O'Callaghan C, De Wandele I, Stiles L, Pocinki A, Rowe P. Cardiovascular autonomic dysfunction in Ehlers-Danlos syndrome-hypermobile type. Am J Med Genet C Semin Med Genet. 2017 Feb 4. doi: 10.1002/ajmg.c.31543. [Epub ahead of print] PubMed PMID: 28160388.

Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, Bloom L, Bowen JM, Brady AF, Burrows NP, Castori M, Cohen H, Colombi M, Demirdas S, De Backer J, De Paepe A, Fournel-Gigleux S, Frank M, Ghali N, Giunta C, Grahame R, Hakim A, Jeunemaitre X, Johnson D, Juul-Kristensen B, Kapferer-Seebacher I, Kazkaz H, Kosho T, Lavallee ME, Levy H, Mendoza-Londono R, Pepin M, Pope FM, Reinstein E, Robert L, Rohrbach M, Sanders L, Sobey GJ, Van Damme T, Vandersteen A, van Mourik C, Voermans N, Wheeldon N, Zschocke J, Tinkle B. 2017. The 2017 international classification of the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet 175C:8–26.

Tinkle B, Castori M, Berglund B, Cohen H, Grahame R, Kazkaz H, Levy H. Hypermobile Ehlers-Danlos syndrome (a.k.a. Ehlers-Danlos syndrome Type III and Ehlers-Danlos syndrome hypermobility type): Clinical description and natural history. Am J Med Genet C Semin Med Genet. 2017 Feb 1. doi:10.1002/ajmg.c.31538. [Epub ahead of print] PubMed PMID: 28145611.

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