Peninsula Clinical Genetics Service (PCGS – covers Devon and Cornwall)

Referral criteria for patients with hypermobility (includes referrals for assessment for hypermobile type Ehlers-Danlos syndrome (hEDS – also known as EDS type 3 in earlier classifications and joint hypermobility syndrome (JHS)).

Background

Hypermobility is very common in the general population. There is no genetic test available for hEDS as no major genetic cause has been identified. Hypermobility does tend to cluster in families and relatives often report similar problems. Symptoms can be troublesome and there are good online resources produced by the NHS (https://www.nhs.uk/conditions/joint-hypermobility-syndrome/) and the Ehlers-Danlos Syndrome Support Group (www.ehlers-danlos.org).

Referral criteria

PCGS is no longer routinely assessing patients with hypermobility or a suspected diagnosis of hEDS. The purpose of referral of hypermobile patients to PCGS is to identify the small subset of patients with **rare and more serious genetic conditions** which can be associated with hypermobility, such as Marfan and Loeys-Dietz syndromes, or the vascular form of Ehlers-Danlos (vEDS, or EDS type 4). The chance of diagnosing these in a person with isolated hypermobility is very small. We therefore only accept referrals if additional features are present.

Please write to us if there is a personal or family history of any of the following:

- Dissection or aneurysm of a blood vessel including aorta
- Bicuspid aortic valve or any widening of the aorta on echocardiogram
- Organ rupture
- Significant skin hyperextensibility or skin fragility (splitting of the dermis following relatively minor trauma)
- Facial milia
- Marfanoid habitus
- Pneumothorax
- Fragile sclerae
- Progressive scoliosis
- Dislocated lenses
- Detached retina
- Cleft palate or bifid uvula
- Craniosynostosis
- Severe gastrointestinal symptoms