Genetic referrals for Ehlers Danlos syndrome (EDS) hypermobility type

	Major criteria	Minor criteria
Hypermobility (type III)	Generalised joint hypermobility Mild skin involvement	Recurring joint dislocations Chronic joint pain Autonomic dysfunction Positive family history

Associated features of **classic, vascular, kyphoscoliotic, arthrochalasia, dermatospraxis EDS**?

(see next page for clinical features)

Yes

Associated features of **Marfan syndrome spectrum?**

(see next page for clinical features)

No

No

Yes

Clinical Genetics referral not indicated for EDS hypermobility type

Refer to appropriate medical specialist for management if required

Request echocardiography and ophthalmic assessment

Refer to Clinical Genetics for genetic advice

Refer to other specialists for evaluation of heart/eye/joint/skin findings

Ehlers Danlos syndromes (EDS)

	Major criteria	Minor criteria	IP
Classic (Type I/II)	Skin hyperextensibility Widened atrophic scars Joint hypermobility	Easy bruising Smooth and velvety skin Molluscoid pseudotumours Subcutaneous spheroids Muscular hypotonia Complications of joint hypermobility Surgical complications Positive family history	**AD**
Vascular (type IV)	Excessive bruising Thin, translucent skin Arterial/intestinal/uterine fragility or rupture Characteristic facial appearance	Acrogeria (premature ageing of hands and feet) Early-onset varicose veins Hypermobility of small joints Tendon and muscle rupture Arteriovenous or carotid-cavernous sinus fistula Pneumo (haemo)thorax Positive family history, sudden death in close relative(s)	**AD**
Kyphoscoliotic (type VI)	Severe muscular hypotonia at birth Generalised joint laxity Kyphoscoliosis at birth Scleral fragility and rupture of the globe	Tissue fragility, including atrophic scars Easy bruising Arterial rupture Marfanoid habitus Microcornea Osteopenia	**AR**
Arthrochalasia (type VII A and B)	Severe generalised joint hypermobility with recurrent subluxations Congenital bilateral hip dislocation	Skin hyperextensibility Tissue fragility, including atrophic scars Easy bruising Muscular hypotonia Kyphoscoliosis Mild osteopenia	**AD**
Dermatosparaxis (type VIIc)	Severe skin fragility Sagging, redundant skin Excessive bruising	Soft, doughy skin texture Premature rupture of membranes Large herniae	**AR**

Marfan syndrome spectrum disorders

Marfan syndrome	Aortic root enlargement Ectopia lentis	Systemic features : reduced upper segment / lower segment AND increased arm span/height ratios, arachnodactyly with positive thumb/wrist sign, pectus carinatum/excavatum/asymmetric, scoliosis or thoracolumbar kyphosis, hindfoot deformity, flat feet, pneumothorax, scoliosis or thoracolumbar kyphosis, skin striae, reduced elbow extension , myopia, mitral valve prolapse, dural ectasia	**AD**
Loeys-Dietz syndrome	Aortic root aneurysm with dissection Generalized arterial tortuosity and aneurysms	Systemic features: arachnodactyly, pectus carinatum/excavatum/asymmetric, scoliosis, talipes equinovarus, soft and velvety skin, translucent skin, easy bruising, dural ectasia, highly arched palate/cleft palate, malar hypoplasia, micrognathia, retrognathia, hypertelorism, broad or bifid uvula	**AD**

Legend: IP, Inheritance Pattern; AD , Autosomal Dominant; AR, Autosomal recessive