

Bristol & Bath guidelines for onward investigation/referral for adults with musculoskeletal hypermobility

Aim of this guideline

- To help clinicians decide whether an adult with musculoskeletal hypermobility requires further investigations or onward referral for identification of an underlying collagen abnormality.
- It is not designed to help guide management of symptoms such as pain or postural difficulties. For these we suggest considering physiotherapy or standard pain management approaches.

Summary

- The vast majority of adults with musculoskeletal hypermobility **do not require further investigations or onward referral**
- A very small number of adults with musculoskeletal hypermobility may require either
 1. an echo and reassurance if normal
 2. referral straight to clinical genetics at the BRI
 3. referral to the specialist Congenital Heart Disease clinic (CHDC) at the BRI

How to decide whether referral is needed in an adult with musculoskeletal hypermobility?

1. ask about the following features in their personal medical history

- aneurysm.....
 - dissection of artery.....
 - spontaneous pneumothorax.....
 - more than 7 long bone fractures.....
 - obstetric history of uterine rupture.....
- Referral to CHDC service or Clinical Genetics**
- Referral for echo to assess the aortic root and if abnormal refer onwards to CHDC service**
- Referral direct to Clinical Genetics**

2. ask about the following features in their family history (FH)

- FH of unexplained sudden death before aged 50
 - FH of thoracic aneurysm or dissection.....
 - FH of at least 2 relatives (one first degree) with aneurysm or dissection at any age..
 - more than 7 long bone fractures in a close relative.....
- Referral direct to CHDC service**
- Referral direct to Clinical Genetics**

3. identify if any of the following are present on physical examination

- Cardiac murmur.....
 - True cutis laxa (not soft skin) – only if really extreme or with other physical signs.....
- Referral for echo to assess the aortic root and if abnormal refer onwards to CHDC service**
- Referral direct to Clinical Genetics**

Referral direct to CHDC service

Adult Congenital Heart Disease Centre
Bristol Heart Institute
Bristol Royal Infirmary
Bristol
BS2 8HW

Referral direct to Clinical Genetics

Clinical Genetics Service
Dept Clinical Genetics
St Michael's Hospital

Tel 0117 342 5107

Referral for echo

Please use local services

Put on the referral form:
"Specialist echo required to assess aortic root. Possible genetic collagen abnormality such as Marfans Syndrome"