

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

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

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

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

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

Referral direct to CHDC service

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Referral direct to Clinical Genetics

Clinical Genetics
Dept Clinical Genetics
St Michael's Hospital

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"