

# Hereditary Disorders of Connective Tissue in the Emergency Department: A Case Series, Discussion and Proposed Screening Tool

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## Abstract

There are over 200 Heritable Disorder of Connective Tissue (HCTDs). These conditions are heterogenous and can present with a multitude of pernicious signs and symptoms. HCTDs conditions are not commonly associated with emergency medicine and their relevance to emergency department presentations frequently goes unrecognised. There is a paucity of formal data on presentations relating to these conditions in emergency medicine, as only life-threatening complications are currently recognised. There is a lack of formal training in medicine on how to recognise the clinical signs and history that could represent a possible HCTD, especially in emergency department contexts. The authors present 7 case studies of patients with known and possible HCTDs who presented with a wide variety of complaints. These included chest pain, syncope, spontaneous arterial dissection, spontaneous pneumothorax, severe dysmenorrhea and dysphagia. The presenting complaints are discussed with relevance to HCTDs and a proposed clinical tool for identification of HCTDs in the Emergency Department. This tool requires validation for use in clinical settings. Further research is required to establish the frequency of problems relating to HCTDs in emergency medicine. Education on HCTDs is recommended. Testing of the clinimetric properties of the proposed screening tools for identification of HCTDs in the emergency department is required.

**Keywords:** Heritable Disorders, Connective Tissue, Dysmenorrhea, Dysphagia

## Introduction

There are over 200 Heritable Disorders of Connective Tissue (HCTDs). The true prevalence of these conditions remains unknown although Generalised Joint Hypermobility (GJH) which can represent an underlying HCTD is reported in 2% to 67% of the general paediatric and adult population globally [1-5].

HCTDs are a group of diseases not commonly associated with presentations to the emergency department. Typically, these extremely heterogeneous disorders are managed in outpatient rheumatology settings. However, from clinical experience working in emergency medicine the primary author reports from clinical experience working in emergency medicine these patients frequently present to emergency departments with a diverse variety of problems ranging from acute pain crises, POTS-related tachycardia/syncope and various psychiatric presentations to life threatening emergencies such as pneumothorax, acute aortic syndromes such as dissections, unprovoked VTEs and rupture of hollow viscera. Despite this, there exists very limited to no formal literature on attendance rates of such conditions in emergency care contexts. There are no formal guidelines on how to assess such patients for an underlying HCTD. Informal and formal literature

is prevalent with patient experiences describing inadequate care in hospital contexts [6-10]. Additionally, there is often a long diagnostic delay with conditions of connective tissue weakness, sometimes taking over 10 years with many patients struggling to receive any diagnosis at all [7,11]. As these patients frequently present to emergency departments seeking help, emergency doctors are on the frontline of interactions this patient cohort presents, and therefore in a position of being able to screen for HCTDs which could potentially hasten referral and diagnoses, however a lack of formal education in this area prevents many doctors from recognising the signs and symptoms of HCTDs [8,11].

These factors support the necessity for better understanding patients who present with signs and symptoms suggestive of connective tissue weakness that are contributing to presenting complaints. Research into effective screening tools in an emergency department context is lacking in formal literature. This requires urgent attention.

Over a 4-week period in May and June of 2023 several patients with known HCTDs and suspected HCTDs presented to the Emergency Department with a variety of complaints and were seen by the

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authors. A selection of these patients was selected and agreed to be part of this case series. Their presentations are discussed here.

## Case Presentations

### Case Report 1: Chest Pain in a Patient with Loeys-Dietz Syndrome

A Caucasian female in her early fifties presented to the Emergency Department with sudden onset central chest pain whilst she was out shopping. The pain was pleuritic in nature and persisted despite administration of aspirin.

The patient had a complex medical history including type 4 Loeys-Dietz Syndrome with a history of prior brain aneurysms, one of which had spontaneously ruptured, requiring clipping in 2009.

The patient also reported hypertension, asthma, Gastroesophageal Reflux Disease (GORD), and poorly controlled Rheumatoid Arthritis (RA) and early onset osteoarthritis (OA) secondary to generalised joint hypermobility. RA combined with OA had resulted in widespread joint hypomobility. The patient's regular medications included amlodipine, diclofenac, esomeprazole and she had recently been recommenced on upatacitinib. Previously her treating rheumatologist had ceased upatacitinib in late 2022 as it had been thought to be causing intermittent severe chest pain.

On examination the patient appeared systemically well. She was noted to be slightly overweight. There were no obvious dysmorphic features on initial inspection. Vital signs were within normal parameters. Heart sounds were dual with no murmur. Breath sounds were normal bilaterally. The abdomen was soft and non-tender. There was no pedal oedema present.

Serial ECGs were performed on the patient which did not demonstrate evidence of ischemia. Serial troponins were negative. Standard bloods were unremarkable. Due to the patient's underlying diagnosis a decision was made to perform a CT aortogram. This was negative for aortic dissection.

The patient was ascertained as safe for discharge from an Emergency Department perspective and advised to follow up with her cardiologist for an echocardiogram and her rheumatologist for consideration of review of upatacitinib for management of her RA.

### Case Report 2: Syncope in a Patient with Generalised Joint Hypermobility

A Caucasian male in his early twenties presented to the Emergency Department following a syncopal episode at work. He had been moving items from a freezer room to a hot oven and had felt lightheaded before suddenly losing consciousness and collapsing on the ground. He regained a normal conscious state within a minute of the event and had no signs or symptoms suggestive of a post ictal state. The patient reported 3 similar episodes over the previous 18 months but was otherwise well. He had no other significant medical history and considered himself a healthy person for his age. On examination he had normal vital signs and was afebrile. Heart sounds were dual with no murmurs noted. Breath sounds were normal bilaterally. His abdomen was soft and non-tender. No abnormalities were noted on examination of the patient's lower legs. Given the history of repeated syncopal episodes, the primary author decided to screen the patient for signs of a HCTD. He was found to have extremely hypermobile digits (See Figure 1) and scored 9/9 on the Beighton Score. He was also found to have hyperextensible skin to 1.5cm on the dorsum of the patient's hands, blue sclera, widened scars and polaris keratosis (See Figure 1). The patient did not exhibit signs of postural hypotension, or tachycardia on changes in position. ECG, CXR, bloods and CT brain were all normal. A diagnosis of Hypermobility Spectrum Disorder was considered likely. The patient was deemed safe for discharge from an Emergency Department point of view and advised, if he had further episodes he should present to his GP for referral to a cardiologist and rheumatology for screening for hypermobile EDS, or one of the other HCTDs.

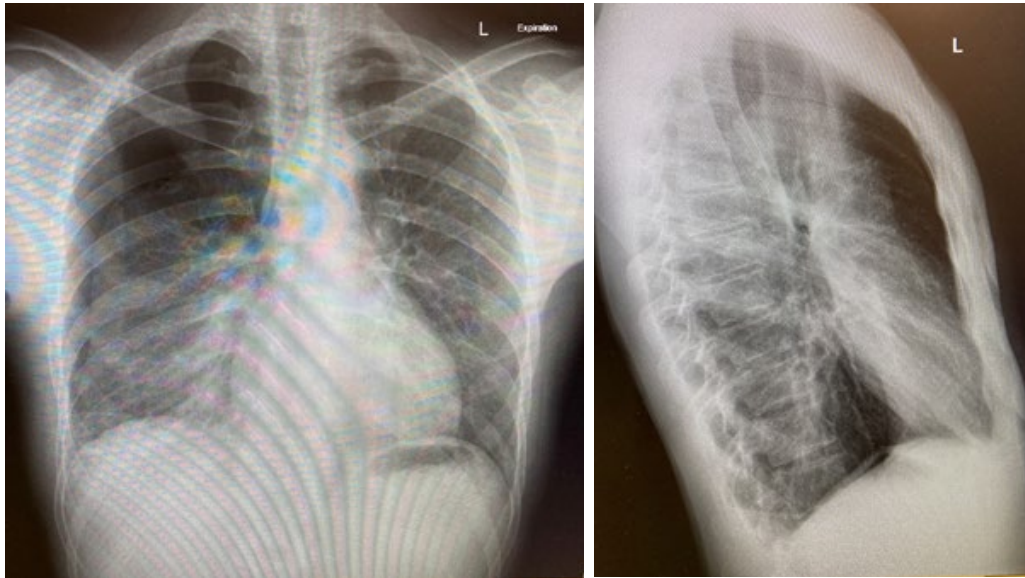


**Figure 1:** Hypermobility of the fingers and thumb, widened scarring and polaris keratosis.

### **Case Report 3: Spontaneous Pneumothorax in a Patient with A Marfanoid Body Habitus**

A Caucasian male in his late thirties presented to the Emergency Department with a four-day history of shortness of breath and continuous right sided chest pain. He had never experienced such symptoms previously. He was mainly concerned about worsening shortness of breath. His past medical history included a traumatic amputation of his left forearm and had been prescribed medical marijuana for chronic phantom limb pain. Otherwise, he had no other significant medical history. He had no significant family history of cardiac, or respiratory events. On examination his vital signs were within normal parameters. He was noted to have a marfanoid appearance with a tall narrow body habitus, arachnodactyly, high arched palate, mild pectus excavatum and

blue sclera. Due to his traumatic limb amputation, it was not possible to assess arachnodactyly using the little finger to thumb around the patient's opposing wrist or assess his arm span to height ratio. The patient had standard blood tests, telemetry monitoring an ECG, analgesia and antiemetics. A Chest Xray revealed a large right sided pneumothorax without mediastinal shift. (See Figure 2). A small-bore pigtail cannula was inserted in the Emergency Department and the patient was referred to the surgical team for management. The patient recovered well and was discharged 48 hours after initial presentation. Given the patient's examination findings he was referred for an urgent outpatient echocardiogram with cardiology follow up and general practice follow up to exclude Marfan's Syndrome and other Thoracic Aortic Aneurysm and Dissection (TAAD) syndromes.



**Figure 2:** AP and lateral views on plain chest x ray demonstrating a large pneumothorax in the right apex



**Figure 3:** AP view on plain chest x ray demonstrating resolution of the pneumothorax post pig-tail catheter insertion.

#### **Case 4: Severe Primary Dysmenorrhea in a Patient with Peripheral Joint Hypermobility**

A Caucasian female in her teenage years presented to the emergency department with her mother with complaints of severe menstrual cramps.

Her medical history included chronic fatigue and long covid since April of 2022. She reported severe dysmenorrhea and menorrhagia. She had a history of spontaneous bilateral shoulder dislocations and multiple musculoskeletal sprains and injuries. Other past medical history included childhood asthma, chronic headaches, lightheaded and dizzy spells.

Her only regular medication was 10mg amitriptyline nocte. She had no known allergies.

On examination the patient's vital signs were normal. No abnormalities were noted on respiratory or cardiology examination. The patient's abdomen was soft with tenderness in the suprapubic area.

The patient was noted to have a Beighton score of 4/9 localised to the hands. She did not have blue sclera. She had soft, hyperextensible skin to 2cm on the dorsal skin surface. The patient had a positive Gorlin sign and was able to perform the reverse



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prayer sign (namaskar) sign.

The patient's pelvic pain resolved post administration of 10mg of oxycodone and a further 5mg oxycodone over a 4-hour period in the emergency department. A transabdominal ultrasound did not reveal any emergent gynaecological phenomena.

She was referred to gynaecology and rheumatology for further investigations for HSD and endometriosis.

### **Case 5: Dysphagia in a Patient with Generalised Joint Hypermobility**

A Caucasian patient in her early thirties presented to the Emergency Department with concerns of acute on chronic dysphagia. Her symptoms started 9 months earlier when she had sustained a bite over her anterior neck from a dingo attack. The wound had become infected initially, however after it had healed the patient experienced ongoing symptoms of anterior neck instability and a sensation of pressure in her throat. In the last 2 weeks she reported struggling to lie flat at night due to the severity of her symptoms

The patient's past medical history was suspicious for HSD/EDS and included multiple spontaneous joint dislocations and sprains, chronic widespread myalgia, chronic anxiety and 'brain fog'. The patient had been diagnosed with Postural Orthostatic Tachycardia syndrome (POTS) previously, however, was not taking any regular medications.

She reported an allergy to penicillin.

On examination the patient had normal vital signs and appeared systemically well. There were no abnormalities on visual inspection of the patient's neck. She was able to talk in full sentences with no disturbances of voice. Examination of her oropharynx was unremarkable. There were no abnormal movements noted on inspection of swallowing and no signs to suggest the presence of a goitre, or other mass.

The patient was noted to have thin, pale and soft velvety skin. Hyperextensible skin was noted to be 2cm on the dorsum of her hands with very hyperextensible skin was noted on her arms and neck. Her Beighton score was 8/9. Blue sclera was noted. Papyraceous scarring was noted.

An X ray of the patient's neck was unremarkable and did not reveal any structural lesions.

The patient was considered safe to discharge from an Emergency Department perspective.

The patient was referred to her GP for further investigations of dysphagia and to rheumatology for work up for possible hypermobile EDS given the nature of the past medical history and extent of physical findings on examination.

### **Discussion**

Patients with signs of HCTDs, or diagnosed HCTDs present

several challenges for doctors working in the emergency medicine. The presenting complaints of the patients in the five case studies will now be discussed in relation to HCTDs.

Patients with suspected HCTDs must undergo referrals to rheumatology and genetic services. Whilst some presentations might seem clinically distinct and related to a specific condition such as Marfan's Syndrome, there is significant overlap between the clinical appearances of HCTDs and therefore thorough screening and expert assessment is required to prevent missed, or incorrect diagnoses.

Patients who present with central chest pain with known Thoracic Aortic Aneurysm and Dissection (TAAD) syndromes such as Loeys-Dietz, Marfan's Syndrome and familial TAAD require work up to exclude acute aortic Syndromes including ruptured aneurysms and aortic dissection. Aortic dissection can also occur not only in vascular EDS, but in other less common types of EDS including classical (cEDS), kyphoscoliotic (kEDS), and dermatosparaxis Ehlers-Danlos syndrome (dEDS) [12].

The patient in Case 1 already had a diagnosis of Loeys-Dietz Syndrome (LDS) and therefore the possibility of an aortic dissection in the context of chest pain was easier to identify, however the patient reported most emergency department doctors in previous presentations were not aware of the connection between LDS and aneurysm and dissection. This highlights the need for further education on conditions that can cause TAAD.

Patients with known HCTDs including TAAD might present to the Emergency Department multiple times due to chest pain and concerns about aortic dissection. This presents a problem with repeat CT imaging and accumulated high doses of radiation.

D dimer has a sensitivity of 96% and specificity of 70% for detecting AD [13]. If the D dimer is less than 500 ng/ml the sensitivity increases [13]. Some researchers have reported D-dimer has different serum levels in different types of aortic dissection, with significantly higher levels present in Stanford A type dissection 6 hours post admission [14].

The Aortic Dissection Detection Risk Score (ADD RS) is a scoring method that consists of 3 components, see box 1. A score of 1 or more combined with a positive D dimer improves diagnostic accuracy in detecting AD [15, 16]. Patients with HCTDs will automatically receive a score of 1 on the ADD RS. A score of 2 or more along with a positive D dimer would indicate the need for imaging but does not resolve the issue of accumulated doses of radiation in these patients.

Nazerian Ascione et al. [17] evaluated consideration of additional risk factors such as fluoroquinolone use at time of presentation, immunosuppressive, or glucocorticoid drugs, or systemic autoimmune diseases, but did not find these assisted with diagnosing acute AD.

If clinical signs, and presentation is highly suspicious for a dissection, imaging might be unavoidable, however alternative modalities could potentially be selected including point of care ultrasound (POCUS), or Magnetic Resonance Angiography (MRA). Although MRA a very sensitive and specific imaging modality for detection of aortic dissection, it is not readily available in rural emergency department contexts.

POCUS has been reported to have a sensitivity of 67-80% and specificity of 99-100%, but only if an intimal flap is identified [18]. This will appear as a hyperechoic dissection flap within the walls of the aorta. POCUS is useful in type A dissection but is can also be used to identify type B dissection. Transoesophageal echocardiography has a sensitivity and specificity of 95% [19], but might not be available in an emergency department context.

High-risk condition such as Marfan syndrome, family history of aortic disease, known aortic valve disease, known thoracic aortic aneurysm, or previous aortic manipulation, including cardiac surgery (1 point)  
Pain in the chest, back, or abdomen described as abrupt, of severe intensity, or a ripping/tearing sensation (1 point) Physical examination findings of perfusion deficit, including pulse deficit, systolic blood pressure difference, or focal neurologic deficit, or with aortic diastolic murmur and hypotension/shock (1 point)  
>1 Consider proceeding directly to CTA or other conclusive imaging  
≤1 Proceed to D-dimer testing. If dimer <500 ng/mL, consider stopping workup of dissection, if ≥500 ng/ml, consider CTA

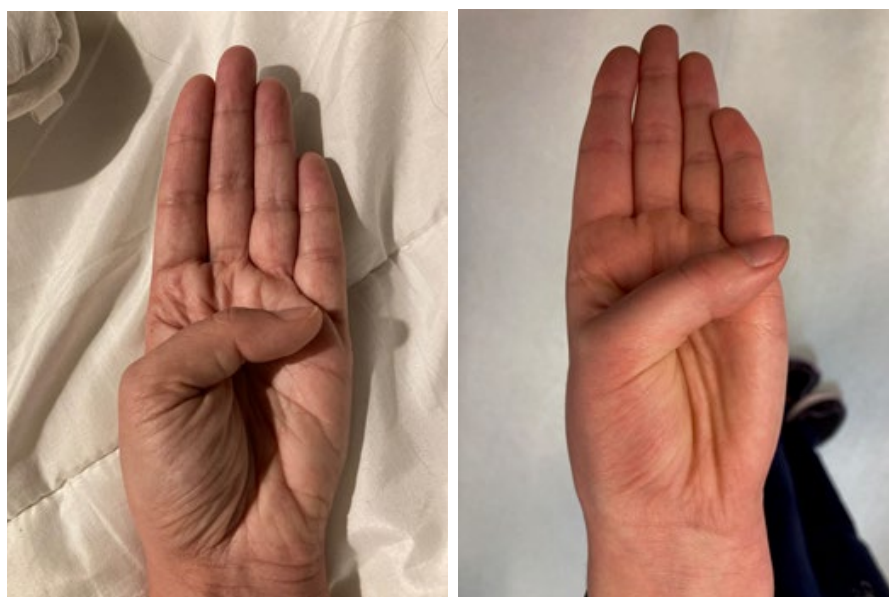
**Box 1:** Aortic Dissection Detection Risk Score + D Dimer for Type A Aortic Dissection [16].

A retrospective review performed by Rashed et al. [20] noted aortic dilatation with greater frequency in hEDS (20.7%) compared with only 7.7% of patients with HSD [20]. In hEDS male patients 50% had moderate to severe dilatation [20]. 1.9% of patients with HSD/EDS had extra-aortic arterial involvement such as spontaneous coronary artery dissection, or SCAD plus celiac artery pseudoaneurysm [20].

For patients who present with chest pain and an undiagnosed HCTD, but might have clinical features suggestive of one, such as a marfanoid body habitus, generalised joint hypermobility and hyperextensible skin, the authors recommend screening for the presence of an aortic root aneurysm in light of the fact this increases the risk of dissection. 60% of patients with a known TAAD who also developed dissection had aneurysms of less than

5.5cm maximal diameter [21]. Additionally, it has been identified that patients with TAAD who have an aortic root aneurysm of diameter of 4.0cm - 4.4 cm conferred an 89-fold increased risk of aortic dissection relative to a control diameter of ≤3.4cm, whilst a diameter ≥4.5cm confer an increased risk of 6000-fold compared to controls of ≤3.4cm [21].

The thumb palm test has been shown to have a specificity of 98.5% for detection of an aneurysm [22]. This paper received criticism by Diao et al. [23] for overestimating the true prevalence of aneurysms in the general population, however Elefthariades et al. [24] stand by their original research findings and recommend the thumb to palm test [24]. The authors recommend its use in patients who have other signs suggestive of connective tissue disease weakness who present with chest pain as discussed.



**Figure 4:** Negative and Positive Thumb-Palm Sign Respectively.

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The patient in Case 2 was an apparently well young male who presented post a syncopal event. Syncope in HCTDs is not uncommon and can represent a catastrophic event such as a ruptured aneurysm, dissection, ruptured viscera, or can result from a more benign condition such as autonomic dysfunction, or dysautonomia which has a close association with HSD and EDS [25,26]. Postural Orthostatic Tachycardia Syndrome (POTS) exists in 31%-94% of patients with EDS [25,27] and around 5% of MFS patients [28] and might occur in other HCTDs. The patient was noted to have widened scarring and polaris keratosis. Polaris keratosis has been reported in literature previously in association with HSD and hypermobile EDS [29]. Patients who present with recurrent benign syncopal events and a high Beighton Score particularly those who experience palpitations and orthostatic tachycardia might have dysautonomia and should be screened for possible HCTDs.

The patient in Case 3 presented with a spontaneous pneumothorax and was noted to have Marfanoid features on examination. Spontaneous pneumothorax is associated with Vascular Ehlers Danlos Syndrome, Marfan's Syndrome, Homocystinuria, Cutis Laxa, Loeys-Dietz Syndrome and other genetic disorders [30]. Mutations in the FLCN gene have been associated with sporadic and familial cases of spontaneous pneumothorax [30]. Literature reports spontaneous pneumothorax occurring in 4-11% of patients with Marfan's Syndrome with a median age of 40 [31,32]. Occasionally it can be the presenting feature of the condition. Any patient presenting with a spontaneous pneumothorax with a marfanoid body habitus should be screened for MFS, but also other HCTDs including LDS as there can be significant phenotypical overlap between all the HCTDs [33].

The Beighton Score has been called into question as the sole tool for identification of GJH [34,35]. It has significant limitations, if it is the only tool used for screening for a HCTD GJH particularly in the context of EDS [34]. Particularly with certain types of HCTDs such as Vascular Ehlers Danlos Syndrome where peripheral joint hypermobility is typical, relying on GJH alone to make a diagnosis is problematic. Other scoring tools exist including the Contompasis Score [36], the Rotes-Querol Score [37], the Hospital Del Mar Scor [38] the Lower Limb Hypermobility Assessment Tool [39] and Upper Limb Assessment Tool [40], however they might prove impractical in emergency medical settings.

The patient in Case 4 is a typical example of how a low Beighton Score can result in healthcare professionals potentially missing other signs of HCTDs. This patient had a Beighton score of 4. Sing et al. [41] recommend a cut off score of  $\geq 5$  in this age group, however this patient presented with other signs and a clinical history suggestive of a HCTD including a positive Gorlin's sign which is positive in 50% of patients with EDS [42], blue sclera, hyperextensible skin and a history of spontaneous bilateral shoulder dislocation. The patient in Case 1 is also an example of how the Beighton Score can be misleading in assessment of GJH as other comorbid conditions such as RA can decrease joint ROM and therefore present as hypomobility in joints that might have been previously hypermobile. This should be taken into consideration

when assessing such patients, especially if a diagnosis of an HCTD does not already exist.

This demonstrates why it is important to assess the patient holistically when suspecting a HCTD and not rely solely on GJH assessment. using only the Beighton Score as a method for screening for HCTDs.

In Case 4, the presenting gynaecological complaint is frequently reported in HSD and EDS. One study reported 50% of patients with HSD and EDS experienced dysmenorrhoea and 50% reported menorrhagia [43] whilst another study reported 72% of patients experienced dysmenorrhoea and 76% menorrhagia [44].

There is research underway investigating the relationship between GJH and long covid with one study reporting COVID-19 can induce fibromyalgia-like symptoms in patients with HSDs and other research exploring whether patients with long Covid are more likely to have GJH [45-47]. The patient reported significant limitations of ADLs and lack of school attendance as a result of post-covid fatigue. Fatigue can also be caused by autonomic dysfunction in such patients [48]. There are case reports of post covid-19 related POTS [49, 50] and other viral infections are also known triggers in development of POTS [51]. In patients with all the above features of case 4 a high index of clinical suspicion of HCTDs must exist.

The patient in Case 5 presented with a sensation of pressure in her throat and chronic dysphagia which was causing her difficulties in lying flat at night. High dysphagia, altered voice, choking, and deep neck pains are common ENT problems reported in patients with HSD and EDS [52, 53]. Dysphagia reportedly occurs in 37-62% of patients with EDS [53]. In such patient's mechanical dysfunction causes food residue to remain in the throat post swallowing resulting in delayed motility and discomfort [52, 53]. The patient's high Beighton Score and other relevant examination findings, a medical history including POTS raise the clinical suspicion for a HCTD such as hypermobile EDS and resulted in a direct referral to rheumatology for further evaluation. Many patients with undiagnosed HCTD will present to emergency departments with minor complaints and an opportunity to make a diagnosis is frequently missed due to a lack of understanding of these conditions. In the event a percentage of these patients might have an increased risk of serious complications relating to connective tissue weakness, the authors feel there is a requirement for a screening tool to assist doctors in recognising HCTDs.

Patients who potentially represent the more severely affected end of the spectrum of HCTDs such as Vascular Ehlers Danlos Syndrome (VEDS), Arterial Tortuosity Syndrome (ATS) and patients who have one of the Thoracic Aortic Aneurysm and Dissection Syndromes (TAAD) such as Marfan's Syndrome, Loeys-Dietz Syndrome etc must be referred for appropriate workup and diagnosis post ED presentation, to prevent delays, or missed diagnoses and poor patient outcomes.

Screening patients for such conditions in emergency department presentations need not be very time consuming, nor onerous and can be ascertained as part of history taking and examination. The primary author has developed a tool that could be used to screen patients for HCTDs, however validation of this clinical tool is required.

The authors propose the following tool for use in an emergency

department context.

### **Proposed Screening Tool for Identification of HCTDs in the Emergency Department\*:**

\*(For patients who present with aortic dissection, rupture of hollow viscera, these patients should automatically be referred for screening of HCTDs by treating teams, however for patients with less serious conditions this algorithm can be used)

#### **Criteria A: 1, or more of the following conditions in emergency department presentation, or past medical history:**

- Postural Orthostatic Tachycardia Syndrome/Inappropriate sinus tachycardia syndrome/Postural hypotension
- Unexplained recurrent syncope
- Spontaneous pneumothorax
- Unusual surgical complications
- Spontaneous atraumatic joint dislocations and subluxations
- Significant musculoskeletal injuries with minimal, or no trauma
- Unprovoked VTE
- Involvement with ballet, martial arts, gymnastics and other sports requiring joint hypermobility
- Fracture with minimal trauma
- Severe primary dysmenorrhoea
- Mitral valve prolapse
- Spontaneous CSF leak
- Nerve compression disorders
- Chiari malformation
- Hernias
- Family history of sudden death under the age of 55
- Chronic widespread musculoskeletal pain

**Plus**

#### **Criteria B: Examination Findings:**

A Positive Beighton score\* ( $\geq 6$  aged 3–7 years,  $\geq 5$  aged 8–39 years,  $\geq 4$  aged 40–59 years,  $\geq 3$  aged 60–69 years and  $\geq 2$  aged 70+ years. For males the suggested cut-offs are  $\geq 5$  aged 3–7 years,  $\geq 4$  aged 8–39 years,  $\geq 2$  aged 40–59 years and  $\geq 1$  aged 60+ years. [41])

**Or**

a score of  $\geq 2$  of the 5 part Questionnaire (5pQ) [55]

**Or**

At least 3 of the following clinical features:

- Hyperextensible skin  $\geq 1.5$ cm on dorsal surface of hand
- Marfanoid body habitus
- Blue sclera
- Atypical scars (keloid, atrophic, or papyraceous)
- Positive Gorlin sign (tongue to nose) [42]
- Positive reverse prayer sign (hands behind back with hands in prayer position) [56–58]
- Bifid uvula
- Thin translucent skin with prominent veins
- Soft, doughy, or velvety skin
- Craniofacial abnormalities
- Other skeletal abnormalities (ie severe scoliosis)
- Dysmorphic hands, or feet



Patients who score at least 1 point from past medical history plus a positive Beighton Score, or at least 2 points on the 5pQ or at least 3 of the clinical features listed, then these patients should be referred for investigation of a HCTD.

- Patients with diagnosed and undiagnosed HCTDs frequently attend emergency departments with a wide variety of presentations
- Existing knowledge regarding HCTDs contributing to ED presentations is limited and this represents a knowledge gap in the literature that demands further research.
- Delays in diagnosis of HCTDs are common, however, with training and application of appropriate screening this can be avoided
- Emergency department doctors are often the first point of contact for patients with problems related to undiagnosed HCTDs. Therefore, screening patients can assist with referrals for diagnosis and avoid missed, or delayed diagnoses
- Emergency physicians require additional education on the significance of these conditions in emergency medicine.
- A validated and reliable screening tool to assist emergency doctors in recognising possible HCTDs requires development.
- The authors propose one such screening tool for HCTDs, however further research in this area is required to validate this tool.

**Box 2: Key Learning Points.**

### Conclusion

Patients with diagnosed and undiagnosed HCTDs frequently present to emergency departments, however the relationship between presenting complaints and underlying HCTDs is not always recognised by doctors. There are currently no formal screening tools to identify such patients in an emergency medicine context. Doctors working in emergency medicine require education to assist with recognition of the clinical signs and symptoms that might represent a HCTD underlying the patient's presentation to the emergency department. Reliance on only generalised joint hypermobility is problematic and might potentially result in missed diagnoses. Patients should be assessed based on their past medical history and other examination findings that might be suggestive of connective weakness and these are included in the proposed score developed by the primary author. This tool requires further validation.

### Conflict of Interests

The authors have no conflict of interests to declare. No funding was received for the authoring, or publication of this paper.

### Patient Consent

The authors obtained patient consent for use of photographs and information and approval from the hospital CEO for the authoring and publication of this paper

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