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### Authors

Clement, Barak C  
Forster, Christopher  
Logemann, Nicholas

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# Focal linear elastosis in a patient with joint hypermobility syndrome

Barak C Clement DO, Christopher Forster MD, Nicholas Logemann DO, FAAD

Affiliations: Walter Reed National Military Medical Center, Bethesda, Maryland

Corresponding Author: Barak C. Clement, 85 Cherry Circle, Groton CT, 06340, Email: [Barak.c.clement2.mil@mail.mil](mailto:Barak.c.clement2.mil@mail.mil)

## Abstract

Focal linear elastosis (FLE) is a benign skin finding characterized by hypertrophic linear plaques with abnormal elastic fibers on histology. We present a unique case in which focal linear elastosis occurred in the setting of joint hypermobility syndrome (JHS). Our patient, a 20-year-old man with a medical history significant for symptoms consistent with JHS, had been followed by the rheumatology clinic for many months. He was referred to the dermatology department for further evaluation of asymptomatic longitudinal bands on his back that had been present for many years. He denied trauma but endorsed a history of 'stretch marks.' On examination there were numerous horizontally oriented, firm, linear, yellow to flesh colored bands, all non-tender to palpation. Punch biopsies were performed of involved and uninvolved skin. Histopathology of normal skin revealed no significant abnormalities whereas involved skin demonstrated broadened collagen bundles in the deep dermis. The elastic fiber stain, Verhoeff-Van Gieson, revealed a gross increase in the number of elastic fibers, fragmented fibers, fibers with "paintbrush" or widened-ends, fibers of varying thickness, and clumped fibers. This combination of histopathologic and clinical features was consistent with FLE.

**Keywords:** focal linear elastosis, joint hypermobility syndrome, Ehlers-Danlos

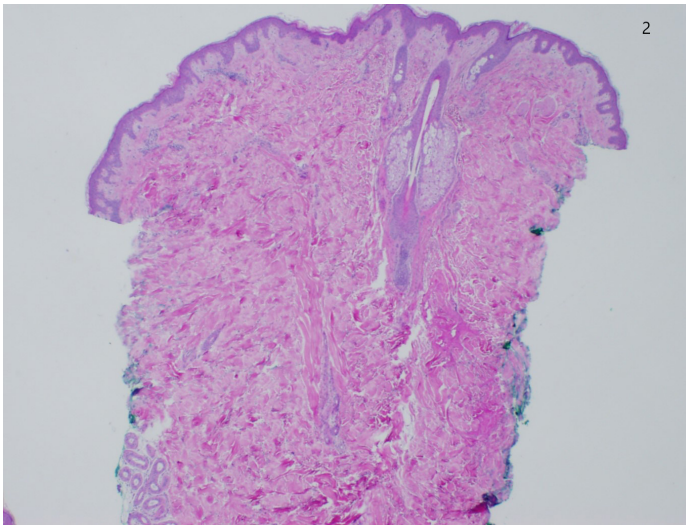
## Introduction

Focal linear elastosis (FLE), a term first coined in 1989

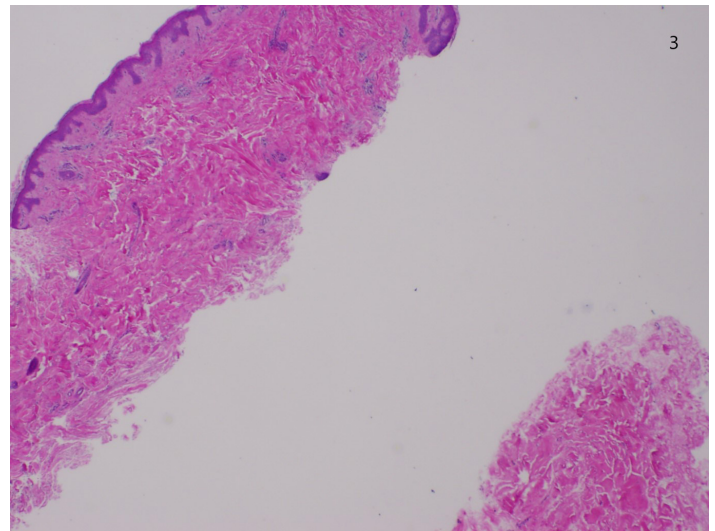
by Berket et al. [1], is a rare cutaneous abnormality described in, to the best of our knowledge, less than 35 patients since originally being defined. To date, FLE has not been reported in patients with other connective tissue disease processes that include diseases of impaired collagen or elastic fibers. FLE clinically manifests as multiple hypertrophic linear plaques usually yellowish in color, most often located in the lumbosacral region. On histologic examination, the hallmark finding is increased, as well as abnormal, elastic fibers. The pathogenesis of FLE is not currently well understood but it is thought by some to be a variant of striae distensae [2] or a keloid of elastic fibers [3, 4]. Herein we report FLE occurring in a patient with (JHS, formerly Ehler-Danlos Syndrome [EDS] Type III). The clinical presentation and histopathological characteristics of FLE and JHS are described and we postulate possible relationships



**Figure 1.** Demonstrates the gross physical findings of horizontally oriented yellow to flesh colored linear plaques located on the mid-lower back.



**Figure 2.** Demonstrates the normal, intact punch biopsy of uninvolved skin. H&E, 10%.



**Figure 3.** Demonstrates the uncharacteristic separation of the dermis that occurred during the punch biopsy of lesioned skin. H&E, 15%.

between FLE and JHS.

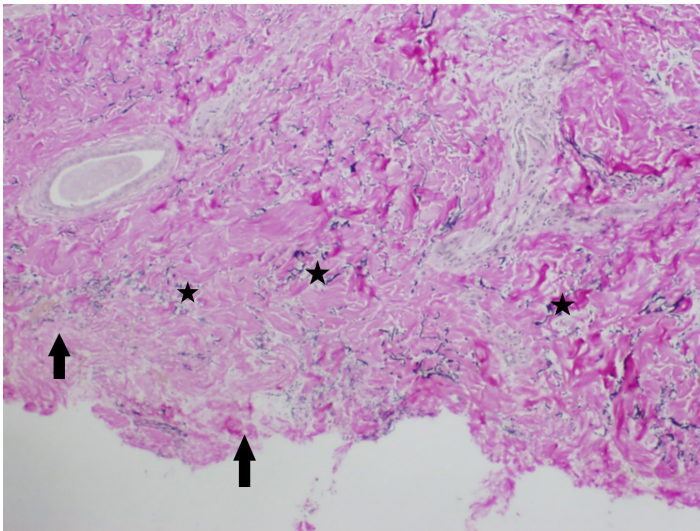
### Case Synopsis

Our patient is a 20-year-old man referred to dermatology clinic by the rheumatology department for further evaluation of firm, linear bands on his back that had been asymptomatic but present for many years. The patient had a constellation of symptoms, which included headaches, joint pains, and joint laxity. The rheumatology consultants determined the patient's condition was consistent with JHS [5, 6]. During their exam they noted raised linear lesions on his mid back and referred him to the dermatology department for further evaluation. At his appointment he denied ever having rapid or large weight loss or gain, being overweight in the past, or suffering trauma to the area, although he did report that his peers remarked that the lines on his back resembled 'lashings' (**Figure 1**). On physical exam, numerous, non-tender, strikingly linear, firm, yellow/flesh colored bands oriented horizontally in the lumbosacral region were noted. Biopsies were performed of involved and nearby uninvolved skin. Of note, when performing the punch biopsy of a linear plaque (involved skin), the skin was noted to be 'tougher' than anticipated, offering uncustomary resistance to the punch tool during the procedure. Furthermore, on attempting to procure the specimen with pick-ups from the biopsy site, the superficial portion of the biopsy was noted to separate loosely from the underlying tougher dermis (**Figure 2**) and in order to obtain tissues from the deeper dermis and

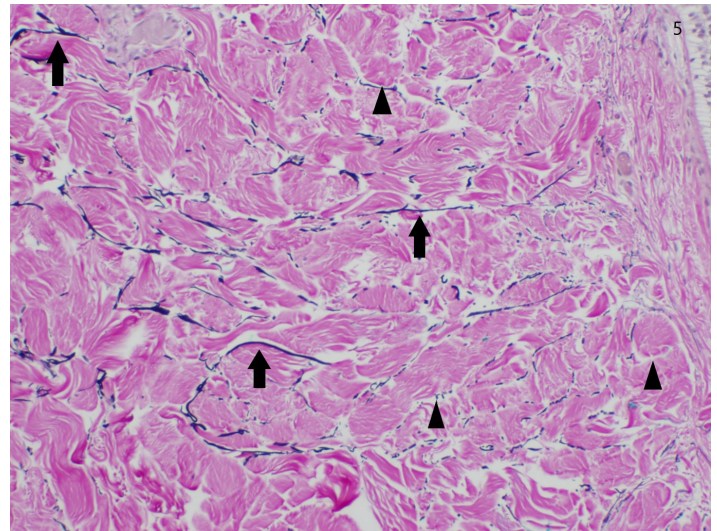
subcutis, sharp dissection was required. In addition to these findings, the specimen obtained was noted to be 'sticky' and we had difficulty transferring it from the pickups to the specimen cup. When it was finally in the specimen container it clung adherently and tenaciously to the perimeter until vigorous agitation dislodged it into the formalin pool. The biopsy procedure for the normal comparison skin was unremarkable with the entirety of the punch being easily procured (**Figure 2**).

### Case Discussion

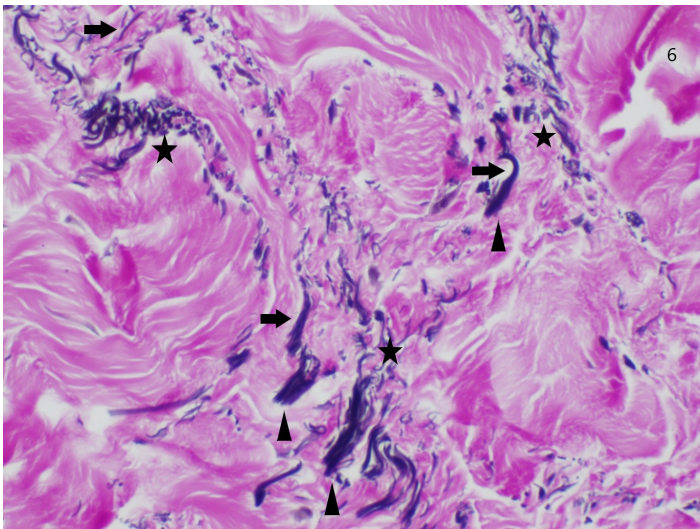
FLE has previously been described as a benign and independent finding unrelated to or associated with any systemic condition. To date, only a few dozen cases of FLE have been reported and although it initially appeared to be a condition with predilection for elderly Caucasian men, it has since been reported in individuals of African, Japanese, Korean, and Turkish heritage without partiality. Furthermore it has been found in patients between the ages of 7 all the way to 89 and in various areas of the body including the posterior thigh, lumbosacral region, neck, and even the face in one individual [7]. Males and females are both affected, though overall FLE does seem to have a penchant for males at a ratio of about 5:1 [7]. Histologically, normal to hypertrophic collagen is observed separating an increased number of elastic fibers [7]. These findings are most pronounced in the subpapillary and reticular dermis [8]. Elastic fibers tend to clump together and show evidence of fragmentation [4]. When treated with a



**Figure 4.** Stained with Verhoeff-Van Gieson. There is a lack of elastic fibers (arrows) in the area adjacent to the aforementioned "split" when compared to the more superficial or deep tissue's elastic fiber (star) quantity. 25%.



**Figure 5.** Slide of uninvolved skin treated with Verhoeff-Van Gieson stain at high magnification. Note the uniform elastic fibers (arrows) and evenly distributed collagen bundles (arrowheads), 40%.



**Figure 6.** Slide of involved skin treated with Verhoeff-Van Gieson stain at high magnification. Note the varying thicknesses of elastic fibers (arrows), clumping and aggregation pattern (stars) and the frayed or "paintbrush" ends (arrowhead) of elastic fibers at high magnification, 100%.

Verhoeff-Van Gieson stain, samples are notable for thin, wavy, elongated and fragmented elastic fibers that separate the dermal collagen bundles with the ends of elastic fibers showing apparent fraying resembling a "paintbrush" [7]. These findings, along with the increase in number and varying thickness of elastic fibers, are the hallmark of this dermatosis [8]. Our patient's biopsy was consistent with these findings demonstrating broad hypertrophic wavy collagen bundles separated by elastic fibers of varying thickness (**Figure 6**). Throughout the biopsy specimen elastic fiber aggregation was observed

and there were malformed fibers demonstrating the classic "paintbrush" ends associated with FLE (**Figure 6**). In addition, and curiously, when obtaining the biopsy, there was a separation in the dermis (**Figure 2**). On histology, a marked diminution of elastic fibers was noted in the area of separation (**Figure 5**).

JHS is the most common condition among the hereditary connective tissue disorders, which include EDS, osteogenesis imperfecta, Marfan syndrome, and Stickler syndrome [9]. Most authorities consider it the same as EDS-hypermobility type, formerly EDS type III [10, 11]. Exact prevalence of JHS is unknown [12, 13], but with recently implemented new criteria used for diagnosing JHS, increased numbers have been reported in some musculoskeletal disease clinics [14, 15]. JHS can affect individual joints or the whole body, with onset in the 1st and 2nd decades. It is more common in women, Asians, and West Africans, and is reported to have a heritability of 70% [16]. Although JHS follows a dominant inheritance pattern, the penetrance is variable. Most patients with JHS do not have an abnormality of collagen or genes encoding for it, or similar molecules [13] though there is a small percentage that have a deficiency of tenascin X, a large extracellular glycoprotein [17, 18]. The clinical split observed in procurement of the punch biopsy specimen as described above could potentially be related to defects in elastic fibers as visualized on histology (Figures 3, 4). Some authors have speculated that the elastic fiber defects could relate to improper

expression of tenascin [19]. Since the presence of a commonly occurring biological marker has not been identified, the diagnosis of JHS is made clinically based on symptoms using the Brighton criteria and the Beighton hypermobility score. Brighton uses major and minor category inclusion criteria in which 2 major criteria, 1 major and 2 minor criteria, or 4 minor criteria are necessary for diagnosis. Some of the more common extra-dermal symptoms of JHS include joint pain, varicosities, hernias, joint subluxations/dislocations, and eye abnormalities like drooping eyelids or myopia [15]. Patients with JHS, like those with more severe manifestations of EDS, also have significant skin findings. Soft, hyperextensible skin prone to easy bruising, scarring, and striae are commonly associated with this condition [5, 9, 20]. Microscopic skin changes, when present, include flower-like collagen fibrils in the papillary and/or reticular dermis as well as variable collagen fibril diameters and abnormal spacing and orientation of the fibrils [21].

Although the changes observed in our patient with FLE and those associated with the skin of patients with JHS are not identical, propensity for forming striae in patients with JHS is intriguing. FLE pathogenesis is ill defined as yet but many have theorized a relationship between striae and FLE formation [4, 22, 23]. Whereas more research is necessary to definitively link the two and more specifically to determine whether TGF- $\beta$  and the keloid repair processes are involved [2], the fact remains that striae accompanied by FLE have been found coexisting in a number of patients [2, 4, 22-24]. Given the suspected connection between striae distensae and FLE and the tendency for patients with JHS to develop horizontal striae in the lumbosacral region [9, 25], we suggest this may have been the cause for developing FLE in our patient. Furthermore, we propose that FLE may be associated with the striae, which patients with JHS are prone to in general. To this end, we agree with Ramlogan et al. that FLE is underreported [22] and suggest that, in the presence of other symptoms consistent with JHS, the finding of FLE may be a potential clue to the clinical diagnosis of JHS or other hereditary connective tissue disorders. We also believe that whereas more studies are necessary to determine the pathogenesis of FLE and the definitive association between it, striae, and JHS, it would behoove clinicians of patients

presenting with symptoms consistent with JHS to look for FLE during their physical exam as it could be a harbinger of an underlying connective tissue abnormality.

## Conclusion

In conclusion, we report a patient with FLE in the context of JHS, an association hitherto unreported. In general, whereas the pathogenesis of FLE is not entirely clear, it is our estimation that FLE may be an under-recognized entity, especially in patients with underlying connective tissue disorders. Finally, if FLE is considered in the clinical differential diagnosis, a 'sticky punch' may be a potential clinical clue to the diagnosis of the disease.

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