Unusual presentation of more common disease/injury

Aggressive Dupuytren's diathesis in a young woman

Ivan Couto-Gonzalez, Beatriz Brea-Garcia, Antonio Taboada-Suárez, Eduardo González-Álvarez

Department of Plastic and Reconstructive Surgery, Complejo Hospitalario Universitario de Santiago de Compostela, Santiago de Compostela, Spain

Correspondence to Ivan Couto-Gonzalez, ivan_couto@hotmail.com

Summary

Dupuytren's disease is a palmar fascia pathology characterised by flexion contracture of the involved fingers in late stages. It has been described a higher prevalence in northern populations, especially in men in the fifth or sixth decade of life.¹ Alcohol consumption, liver disease, diabetes mellitus, epilepsy and some of its treatments, tobacco smokers or hand workers are only a example of the different aetiologies proposed. There are a group of patients presenting with more aggressive clinical manifestation named Dupuytren's diathesis. This subtype could frequently present a positive family history, ectopic fibromatosis and bilateral hand affectionation.

The authors present a 24-year-old woman referred to the Plastic Surgery Department because severe fingers contracture with an associated aggressive plantar fibromatosis. She referred familial history in her first degree relatives (mother and three of seven brothers and sisters). The first clinical manifestations in her hand began when she was 7 years old.

BACKGROUND

Dupuytren's disease (DD) is a frequent pathology developed in patients over the fifth decade of life. Ectopic fibromatosis are possible but not very common.

In this case we describe a quite unusual case presented in a young woman with a severe clinical manifestation in both hands and an associated strong fibromatosis process on her feet. Family history, bilateral palmar disease and ectopic fibromatosis support risk factors for diathesis according to Hueston but these are infrequent presented in a so young woman.

CASE PRESENTATION

A 24-year-old woman was referred to the Plastic and Reconstructive Surgery Department with DD with advanced flexion contracture in the fifth ray of both hands as well as palmar lesions. The patient was an immigrant from the northern region of Portugal. The patient was teetotal and a non-smoker with no significant past medical history. Asked about family DD history, the patient confirmed affectionation of her mother and her grandmother and three men and the own patient of her seven brothers and sisters. On physical examination a severe flexion contracture in the fifth ray of

Figure-1

Figure 1 Skin pits and hypertrophic scar-like lesions were observed over the distal palmar crease of the left hand.

Figure 2 Fibromatous mass located in the right heel.

Figure 3 Syndactyly process between fifth and fourth toes in the right foot.

both hands were noted (Tubiana's stage IV). Skin changes including pits and hypertrophic scar-like lesions were observed over the distal palmar crease of the left hand (figure 1). No previous trauma was registered in this area. Asked about the presence of any ectopic fibromatosis the patient showed the severe fibromatous deposits located on both feet (figures 2–4). She admitted that the process in

hands and feet started when she was 7 years old, first in her hands like 'small scars' in the distal palmar fascia area and then, similar lesion occurred at plantar fascia and heels. Retraction on digits was developed a few years after the first signs of the disease, along her childhood. The fifth rays in both hands were mainly affected with an associated deficit for usual manual labours. The fibromatous process

Figure-4

Figure 4 Fibromatous mass located in the medial aspect of the left foot.

caused a syndactyly process between fifth and fourth toes in the right foot and a bulky connective tissue deposit developed in the heel. A similar deposit was described in the medial aspect of her right foot.

INVESTIGATIONS

Blood analysis, chest radiography and ECG realised in the preoperative study were normal.

Postoperative histological analysis of the affected palmar fascia tissues revealed myofibroblast cells surrounded by a dense connective tissue matrix. Mitosis (MIB-1 measured) was minimal and immunohistochemical markers vimentin and HF-35 actin were positive.

Organised collagen fibres were the only finding when digital cords were analysed. There were not found myofibroblast and MIB-1, vimentin and HF-35 actin were negative.

TREATMENT

We decided surgical relapse of the flexion contracture in the ulnar aspect of the left hand. A partial dermofasciectomy was practiced, removing affected tissues in the fifth, fourth and third ray. The defect was covered with a full thickness skin graft harvested from the volar aspect of the left elbow. Capsulotomy was necessary to achieve an acceptable joint extension in the proximal interphalangeal (PIP) joint of the fifth finger, but full extension could not be obtained and a residual contracture remained after surgery.

OUTCOME AND FOLLOW-UP

The patient made a good postoperative review and was discharged 2 days after surgical intervention. Skin graft healed without any complication and normal activity was resumed 4 weeks after surgery.

Currently the patient is waiting for surgical intervention in the right hand but refuses any surgical treatment in her feet.

DISCUSSION

DD is a fibroproliferative disorder that results in the development of abnormal tissue in the palmar fascia extending to any digit.² Its course is progressive and in advanced stages a flexion contracture in the involved finger is noted because the pathological retraction cords. DD is a classical pathology in hand surgery and the first mentioned reported was in the 17th century.³ The aetiology of the process remains unknown nowadays but several hypotheses have been proposed along the 20th century trying to find the factors which predispose an individual to DD. Alcohol consumption and associated liver disease,^{4 5} epilepsy,⁶ diabetes,⁷ smoking,⁸ manual labour with repetitive trauma over the affected palmar fascia⁹ or genetic and hereditary factors,^{10 11} autoimmune disorders^{12 13} have been associated to DD but there is little conclusive evidence regarding these factors.¹⁴

There is a probed familial predisposition to DD and it has been described as one of the most common hereditary connective tissue disorders in Caucasians.¹⁵ Several inheritance patterns including autosomal recessive have been proposed as the likely mode of inheritance.¹ Mutation in transforming growth factor β receptor-1 (TGF- β 1) is a probed genetic alteration associated to an increased incidence of DD.¹⁶ Zf9 transcription factor gene mutation has been also related to enlarged levels of TGF- β 1 expression in tissues, inducing DD by enhancing the fibrogenetic effect of TGF- β 1 gene.¹⁷ DD seems to be a complex oligogenic rather than a monogenic condition that segregates into multiple model of inheritance¹ where environmental factors may have a great impact.

The initial presentation in DD usually is a nodule formation, mostly located in the distal palmar fascia, nearly the proximal or distal creases. Nodules are usually accompanied with funnel shaped lesions of the skin relief named 'skin pits'. With disease evolution, there is a nodule regression as the cords formation occurs. The cords can remain in the palm or even extend to the fingers. Pretendinous cords formation in the palm cause flexion contractures of the metacarpophalangeal joints. Digital cords cause PIP flexion contractures.¹⁴

There have been descriptions of fibromatosis located in different places to palmar or palmodigital fascia but occurring simultaneously with DD. These deposits are known as ectopic fibromatoses.¹⁸ The most frequent ectopic fibromatoses are knuckle pads (located on the dorsal aspect of PIP joints), plantar fibromatosis or Lederhose's disease^{19 20} and penile fibromatosis or De La Peyronie's disease.^{21 22}

Hueston described *diathesis* to explain a morbid habit that would predispose an individual with DD to an aggressive course and greater tendency for recurrence after surgery. Factors described by Hueston were bilateral palmar disease, family history of DD, ectopic lesions (DD found outside the palmar surface) and ethnicity.²³ Other associated factors, and recently reported with increased severity, include male gender and a young age at onset.²⁴

Aggressive treatment is recommended for the patients with high risk of recurrence, especially in young patients with rapid and progressive disease. Primary dermofasciectomy repaired with a skin graft is the most recommended option.²⁵ It is based on the idea, that recurrence will not occur beneath an skin graft.²⁶

Learning points

- ▶ DD mainly affects Caucasian males since the fifth decade of life.¹ In a small number of patients it could be diagnosed in young females, even in the childhood.²⁷
- ▶ Bilateral hand affectation, family history or ectopic fibromatosis are probed to be signs of aggressiveness with a faster development of the lesions and higher tendency to recidive (Dupuytren's diathesis).²³
- ▶ DD seems to be a complex oligogenic condition that segregates into multiple model of inheritance¹ where environmental factors would play an important role.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. **Burge P.** Genetics of Dupuytren's disease. *Hand Clin* 1999;**15**:63–72.
2. **Bayat A, McGrouther DA.** Management of Dupuytren's disease-clear advice for an alusive condition. *Ann R Coll Surg Engl* 2006;**88**:3–8.
3. **Dahmen G, Kerckhoff F.** Langzeitbeobachtungen operativ und konservativ behandelter Dupuytren'scher Kontrakturen. *Arch Orthop Unfallchir* 1967;**61**:187–202.
4. **Noble J, Arafat M, Royle SG, et al.** The association between alcohol, hepatic pathology and Dupuytren's disease. *J Hand Surg Br* 1992;**17**:71–4.
5. **Attali P, Ink O, Pelletier G, et al.** Dupuytren's contracture, alcohol consumption and chronic liver disease. *Arch Int Med* 1987;**147**:1065–7.
6. **Early PF.** Population studies in Dupuytren's contracture. *J Bone Joint Surg* 1962;**44**:602–13.
7. **Rosenbloom AL, Silverstein JH.** Connective tissue and joint disease in diabetes mellitus. *Endocrinol Metab North Am* 1996;**25**:473–83.
8. **An HS, Southworth SR, Jackson WT, et al.** Cigarette smoking and Dupuytren's contracture of the hand. *J Hand Surg Am* 1988;**13**:873–4.
9. **Liss GM, Stock SR.** Can Dupuytren's contracture be work-related? Review of evidence. *Am J Ind Med* 1996;**29**:521–32.
10. **McFarlane RM.** Dupuytren's disease. *J Hand Ther* 1997;**10**:8–13.
11. **Dal Cin P, De Smet L, Sciort R, et al.** Trisomy 7 and trisomy 8 in dividing and non-dividing tumor cells in Dupuytren's disease. *Cancer Genet Cytogenet* 1999;**108**:137–40.
12. **Burch PRJ.** Dupuytren's contracture: an auto-immune disease? *J Bone Joint Surg* 1966;**48**:312–19.
13. **Menzel EJ, Piza H, Zieliski C, et al.** Collagen types and anticollagen-antibodies in Dupuytren's disease. *Hand* 1979;**11**:243–8.
14. **Hughes TB, Mechrefe A, Littler JW, et al.** Dupuytren's Disease. *J Hand Surg Am* 2003;**3**:27–40.
15. **Hunter JAP, Ogdon C, Norris MG.** Dupuytren's contracture. I: Chemical pathology. *Br J Plast Surg* 1975;**28**:10.
16. **Bayat A, Stanley JK, Watson JS, et al.** Genetic susceptibility to Dupuytren's disease: transforming growth factor beta receptor (TGFBR) gene polymorphisms and Dupuytren's disease. *Br J Plast Surg* 2003;**56**:328–33.
17. **Bayat A, Watson AS, Stanley JK, et al.** Genetic susceptibility to Dupuytren's disease: association of Zf9 transcription factor gene. *Plast Reconstr Surg* 2003;**111**:2133–9.
18. **Brenner P, Rayan GM.** *Dupuytren's disease a concept of surgical treatment.* New York, USA: Springer-Verlag Wien, 2003:10.
19. **Brouet JP.** Etude de 1000 dossiers de maladie de Dupuytren. In: Tubiana R, Hueston JT, eds. *La maladie de Dupuytren.* Paris: Expansion Scientifique Française, 1986:98–105.
20. **Rao GS, Luthra PK.** Dupuytren's disease of the foot in children: a report of three cases. *Br J Plast Surg* 1988;**41**:313–15.
21. **Jordan GH.** Surgery for Peyronie's disease. In: Libertino JA, ed. *Reconstructive urologic surgery.* St Louis Baltimore Carlsbad: Mosby, 1998:539–51.
22. **Brenner P, Krause-Bergmann A, Ha Van V.** Die Dupuytren'sche Kontraktur in Norddeutschland-Epidemiologische Erfassungsstudie und Hand von 500 Fällen. *Unfallchirurg* 2001;**104**: 303–11.
23. **Hueston JT.** *Dupuytren's contracture.* Edinburgh: E&S Livingstone, 1963:51–120.
24. **Hindocha S, Stanley JK, Watson S, et al.** Dupuytren's diathesis revisited: evaluation of prognostic indicators for risk of disease recurrence. *J Hand Surg Am* 2006;**31**:1626–34.
25. **Tubiana R.** Surgical indications. In: Tubiana R, Leclercq C, Hurst LC, Badalamente MA, Mackin EJ, eds. *Dupuytren's disease.* London: Martin Dunitz, 2000:218–22.
26. **Gordon S.** Dupuytren's contracture: recurrence and extension following surgical treatment. *Br J Plast Surg* 1957;**9**:286–8.
27. **Foucher G, Lequeux C, Medina J, et al.** A congenital hand deformity: Dupuytren's disease. *J Hand Surg Am* 2001;**26**:515–17.

This pdf has been created automatically from the final edited text and images.

Copyright 2010 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.

BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Couto-Gonzalez I, Brea-Garcia B, Taboada-Suárez A, González-Álvarez E. Aggressive Dupuytren's diathesis in a young woman. *BMJ Case Reports* 2010;10.1136/bcr.12.2009.2592, date of publication

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow