Pediatric Dupuytren's disease: case report and review of the literature

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Dupuytren's disease (DD) is a rare entity in the pediatric population, especially in children younger than 10 years of age. We report the case of a 6-year-old boy with contracture and a left-hand nodule that was excised and histologically confirmed to be Dupuytren's. Although there are several causes of flexion contracture of the fingers in children, delayed diagnosis of DD can result in debilitating disease and therefore should be an early consideration when evaluating children with suspicious physical exam findings with or without symptomatic contracture, especially in the setting of a strong family history.

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Introduction

Classically considered a disease of adult onset, Dupuytren's disease (DD) in the pediatric population is rare, especially in children younger than 10 years of age. In this population, the initial assessment may be performed by pediatricians alone, and the differential diagnosis for flexure contracture of the hand is variable, with DD often a late consideration. We report the case of a 6-year-old boy with symptomatic contracture and a left-hand nodule histologically confirmed to be Dupuytren's. Although there may be several causes of palmar nodularity and flexion contracture in children, the aim of this study is to highlight the importance of early consideration of DD in the setting of contracture, especially when a familial association is elicited. Further, review of the current literature of the natural history and progression of DD is presented, and current management is discussed.

Case report

A 6-year-old boy was brought for evaluation by his grandmother after 3 months of increasing pain upon gripping objects, with no history of trauma. On examination, there was a firm, palpable nodule on the palmar aspect of the ring finger. While obtaining a detailed history, it was found that the family was of Nordic descent, and the grandmother was noted to have a Dupuytren's nodule on her hand in the same location (Fig. 1). The boy was subsequently taken to the operating room, where a Bruner incision was made on the palmar aspect of the ring finger along the ulnar border. A localized palmar fasciectomy was performed (Fig. 2). Histopathologic evaluation indicated fibromatosis consistent with Dupuytren's contracture (Fig. 3a–c). The patient had an uneventful postoperative course. At the 6-month follow-up, he had regained full function of the affected hand, with no signs of recurrence.

Discussion

DD is a fibroproliferative disorder of autosomal dominant inheritance. The disease causes a fixed flexion contracture of the hand; that is, the fingers are bent toward the palm and cannot be fully extended. In general, the disease is progressive and insidious, first manifested by painless thickening of the involved tendon progressing to hard nodule formation on the palmar fascia, which is ultimately replaced by a tendon-like fibrous cord resulting in contracture [1]. The fourth and fifth digits are most commonly affected. Risk factors for development of the disease include Northern European descent, family history, diabetes mellitus, and sex. However, after 80 years of age, the occurrence is equal in both sexes [2–4]. About 10% of patients with DD also have Peyronie disease (penile fibromatosis). Environmental and other epigenetic factors implicated in the epidemiology of this contracture remain an area of current investigation [5].

There are three overlapping phases in the development of DD, which include a proliferative stage, an involutional stage, and a residual stage [1]. Early DD is histologically hallmarked by hyperplasia and focal nodularity of the fascia. Enhanced, disorganized fibroblast proliferation with increased vascularity is pathognomonic for this proliferative stage. The involutional stage is characterized by the early formation of contracture as the number of myofibroblasts decreases and nodules become smaller, but more organized along the longitudinal axes of the hand. The residual stage shows joint flexion contracture with disappearance of nodules and the presence of thickened, fibrous cords. This classification, first described by Luck [1], is widely used in research, but is less useful in clinical practice among surgeons. In general, the physical exam may indicate a wide range of findings, depending on the timing of diagnosis.

Clinical assessment incorporates a thorough assessment of patient and family history, observed physical findings, measurement of contracture deformity, and a positive Hueston tabletop test [6] as part of the comprehensive diathesis. Whereas early DD may be manifested by painless cords, nodularity skin blanching, and atrophic pitting [7], advanced disease is characterized by prominent,

Fig. 1

The nodule present on the hand of both the patient (right) and his grandmother (left).

Fig. 2

Intraoperative photograph of the Bruner incision and excised lesion.

painful cords, contracture of the metacarpophalangeal joint and proximal interphalangeal joint joints with secondary deformity (Boutonniere and swan neck deformities), and the presence of Garrod's nodes (tender knuckle pads) [8]. Plantar fascia involvement (Ledderhose disease) is also an indicator of severe disease [8]. Routine diagnostic imaging is not indicated in the workup of this disease. However, when the diagnosis is less certain, MRI, computed tomography, and ultrasound have been used, with varying degrees of success [9]. Ultrasound may visualize hypertrophy of the palmar fascia and is particularly useful to localize nodules for intralesional injection therapy.

Staging for DD follows the grading system first established by Tubiana [10]. Each stage is determined by the total passive extension deficit or angle of contracture deformity present on physical exam (stages 1–4). The scoring system also takes into account the earliest presentation of DD, which contains neither nodule nor functional deficit (stage 0), and the presence of nodule alone, without contracture deformity (stage N). Modifications to this staging system have been proposed and staging has been revised to correlate the severity of symptoms with the angle of contracture and joint involvement [11]. The decision to perform a surgical intervention is informed by the degree of contracture (stage), failure of medical therapy, and, ultimately, functional status of the hand – progressive weakness, pain, and functional decline. An overview of the physical findings, staging, and treatment strategies is summarized in Fig. 4.

Although surgery remains the treatment of choice for DD in both children and adults, formal guidelines for surgical intervention are yet to be established. Treatment may include percutaneous fasciotomy (needle aponeurotomy) for early disease with minimal contracture, fasciectomy (limited, segmental or radical), and dermofasciectomy that removes the entire diseased fascia, cord, nodule, and overlying skin, with graft repair of the resultant defect [7,12,13]. Needle aponeurotomy has shown efficacy in patients with well-defined cords and is valuable as an alternative for patients in whom surgery is contraindicated, although recurrence rates can be as high as 50% [8,12]. Limited (selective) fasciectomy is performed under regional or general anesthesia, where diseased cord and fascia alone are excised [12]. Recently, the 5-year follow-up results from a randomized-controlled clinical trial comparing percutaneous needle fasciotomy and limited fasciectomy have been reported [14]. Recurrence rates were significantly higher in the limited fasciectomy group; however, time to recurrence was earlier in the needle fasciotomy cohort.

Nonsurgical options for the management of DD have been reviewed [15]. Reported modalities include observation (for stage N only), physical therapy, splinting, radiation, dimethylsulfoxide, vitamin E cream, ultrasound therapy, and enzymatic (collagenase) and corticosteroid injections [16–18]. Many experimental agents are being investigated, including 5-fluorouracil, botulinum toxin, immunomodulators, matrix metalloproteinase inhibitors, and hyperbaric oxygen therapy, for the effective treatment of DD [19–23]. To date, the safety and efficacy of nonsurgical therapies have not been validated in the pediatric population.

Despite its rarity, pediatric DD is not a new entity. In 1832, Baron Guillaume Dupuytren, a French anatomist and surgeon, first identified a 6-year-old boy with a permanent contracture of the ring and little fingers. This condition was subsequently known as 'Morbus Dupuytren', 'Dupuytren's disease,' or 'Palmar fibromatosis' [24]. The diagnosis was made without histological confirmation. The first histologically diagnosed pediatric case was in 1954 by Goetzee and Williams [25], who identified

Fig. 3
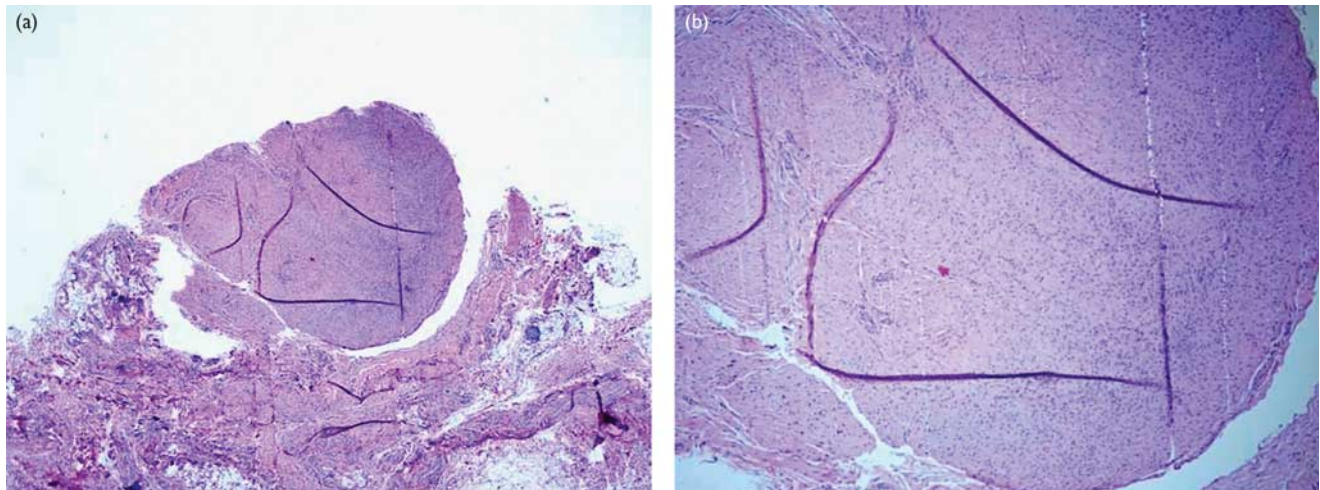
(a–c) Low-power (a, b) and high-power magnification (c) of hematoxylin and eosinophil-stained sections of the palmar nodule. Thick bundles of fibroblasts are separated by collagen fibers and slit-like, compressed blood vessels. The features are consistent with palmar fibromatosis.

Fig. 4
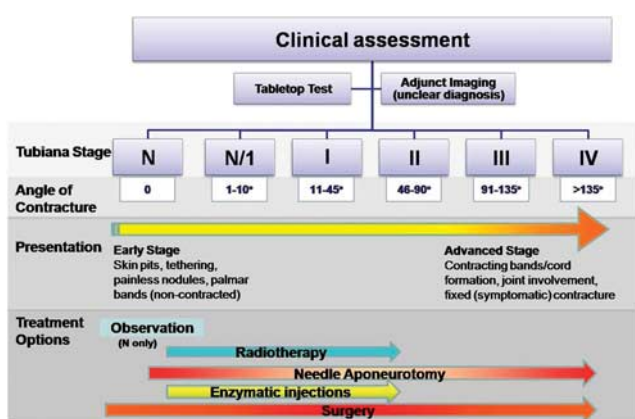
Schematic representation of the presentation, workup, staging, and management of Dupuytren's disease.

a 14-year-old boy with flexion contractures of the ring and little fingers. Since 1832, there have been fewer than 10 published cases of histologically proven DD in children younger than 10 years of age. The youngest patients diagnosed with confirmed DD were two 6-month-old infants, both treated successfully with surgical excision [26,27].

Fibroproliferative conditions other than DD are more commonly identified as the cause of pediatric contracture. The differential diagnosis includes infantile digital fibromatosis, camptodactyly, congenital ulnar drift, burns, infantile fibrosarcoma and calcifying aponeurotic fibromas, and more. Misdiagnosis has been reported (27), with debilitating consequences. Early consideration, in-depth assessment of history, and accurate diagnosis are essential; pathological evaluation of the specimen should always be performed with the goal of ruling out malignancy and confirming the diagnosis of DD among a broad differential. Diagnosis of the disease in childhood presents unique opportunities for long-term follow-up of disease recurrence and may provide further insight into the pathophysiology of this disease process.

Conflicts of interest
There are no conflicts of interest.

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