CASE REPORT

Disabling pansclerotic morphoea of childhood

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Accepted 23 January 2018

SUMMARY

Disabling pansclerotic morphoea (DPM) of childhood is a severe and often fatal variant of deep morphoea. It usually starts in childhood and rarely seen in adults. The course of the disease is progressive with lifelong morbidity in the form of joint contractures and immobility. The causes of mortality include complications such as sepsis, gangrene and cardiopulmonary involvement. Herein, we discuss the case of a 15-year-old girl with limb deformity and finger contractures, that is, bone involvement. The diagnosis of DPM of childhood was fortuitously made after the correction of limb deformity, when the patient was seen in the dermatology department for evaluation of skin discolouration on the thighs.

BACKGROUND

Disabling pansclerotic morphoea (DPM) of childhood is a rare and debilitating variant of localised scleroderma or morphoea. It is characterised by deep cutaneous sclerosis and can extend up to the fascia, muscle and even the underlying bone. It can result in poor quality of life due to contractures and musculoskeletal atrophy, and can have a fatal course due to complications such as sepsis, gangrene and bronchopneumonia.¹ It is three to four times more common in girls than boys.² Herein, we present a case of DPM, which was incidentally diagnosed when a patient was seen in the dermatology office for hyperpigmentation of the thighs.

CASE PRESENTATION

A 15-year-old Asian-Indian girl was seen for evaluation of brown discolouration of both the thighs. The discolouration started at the age of 11 years, was gradual in onset and without any associated symptoms. She initially noticed discolouration of posterolateral aspect of the left leg behind the lateral malleolus. She gradually developed difficulty in walking, shortening of lateral four toes, foot and limb deformity causing her to walk in equinus position. She never sought medical attention and attributed it to an old leg injury. She was seen by orthopaedics for limb deformity, and 2 months ago, she underwent left tendo-Achilles lengthening, open reduction and internal fixation. She was then sent to the rheumatology clinic for finger contractures and was diagnosed with juvenile inflammatory arthritis. She was then referred to the dermatology clinic for brownish patches on the thighs. She initially developed it at the age of 7 years associated with tightening of the skin on the left leg. It was followed by limb deformity, painful

finger contractures by age 9 and appearance of similar patches on both thighs by age 11.

Examination revealed a 12×10 cm brownish plaque on the lower gluteal fold and upper part of posterior right thigh (figure 1A); a groove seen above and behind the lateral malleolus of the left leg, associated with muscle atrophy and shortened foot (figure 1B,C); light brownish indurated plaque on the left thigh with absence of hair and surrounding erythema, skin was hide-bound with inability to pinch (figure 1D) and finger contractures due to sclerosis (figure 2). Similar skin lesions were noted on the neck and scalp but there was no alopecia.

INVESTIGATIONS

Skin biopsy obtained from the left thigh had sparse superficial and deep perivascular and periappendageal lymphohistiocytic infiltrate. There was marked thickening of collagen bundles in the lower reticular dermis. The thickened bundles are closely packed to give hyalinised appearance (figure 3A,B).

TREATMENT

We diagnosed her with DPM and started on methotrexate 15 mg weekly and prednisone ~1 mg/kg/day along with phototherapy. We intend to continue methotrexate for 1 year and steroids for at least 3 months.

DISCUSSION

Morphoea encompasses a wide spectrum of conditions, ranging from superficial, circumscribed sclerotic plaques to severe, generalised and pansclerotic forms. Other clinical variants include: plaque-type morphoea—comprising superficial, guttate and nodular variants; generalised morphoea; linear scleroderma and Parry-Romberg syndrome (progressive hemifacial atrophy); and deep morphoea—including morphoea profunda, eosinophilic fasciitis and DPM of childhood.³ Histologically, these disorders may show similar inflammatory and sclerotic findings, the major difference being the depth of these changes. Even their clinical and epidemiological characteristics can overlap.^{4,5}

Morphoea has been traditionally differentiated from systemic sclerosis based on the clinical findings of almost exclusive cutaneous involvement (with some involvement of the underlying musculature) and absence of internal organ involvement. However, there is accumulating evidence that a significant proportion of these patients develop extracutaneous manifestations during the course of the disease. For example, in a multicentre study

To cite: Jamalpur I, Mogili HR, Koratala A. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-222132

Figure 1 (A) Indurated brown-coloured plaque on the posterior aspect of the right thigh. (B) Groove above and behind the lateral malleolus of the left leg. (C) Shortening of the lateral four toes and left foot. (D) Brownish hide-bound skin on the left thigh.

of 750 children, 168 had at least one extracutaneous manifestation, of which arthritis was the most common (~47%), followed by neurological (eg, seizures), vascular (eg, vasculitic rash), ocular (eg, uveitis) and gastrointestinal manifestations (eg, reflux disease) seen in 17%, 9.3%, 8.3% and 6.2%, respectively.⁶ Fortunately, our patient did not have any of these manifestations but we have to be vigilant during the course of follow-up.

The treatment for DPM is unsatisfactory and none of the existing therapies has shown to arrest the progression of disease activity. Physical therapy remains the mainstay of non-pharmacological management and may aid in maintaining functional ability, muscle strength and prevention of flexion contractures. In addition, systemic therapy is recommended in DPM, as there is a significant risk for disability. Combination therapy with methotrexate, steroids and phototherapy with ultraviolet A (UVA) has shown to be effective than monotherapy. UVA irradiation is thought to reduce procollagen synthesis and induce the expression of collagenase-1, an enzyme that cleaves collagen bundles.⁷ Other drugs that have been tried with limited benefit include antimalarials, colchicine, cyclosporine, tacrolimus,

Figure 2 Finger contractures.

Figure 3 (A, B) Skin biopsy demonstrating marked lymphohistiocytic infiltrate (arrows) and thickened collagen bundles giving a hyalinised appearance in the lower reticular dermis.

penicillamine, intravenous immunoglobulins, interferon-gamma and recombinant human relaxin.³ Newer drugs such as sildenafil, a phosphodiesterase type 5 inhibitor and bosentan, an endothelin receptor antagonist, have shown some promise in ulcerative forms of DPM.⁸ Surgical intervention is usually considered if the disease is progressive despite aggressive medical and physical therapy.⁹ In our case, there was a very gradual progression towards ankle deformity and DPM was actually diagnosed after the correction of limb deformity.

Patients with morphoea, regardless of the subtype experience delay in diagnosis, which likely impacts outcome. Therapeutic decision-making is largely determined by the specialty of the physician rather than characteristics of the disease and many treatments with little or no proven efficacy are used, while others with proven efficacy are underused. For example, in a cohort of 224 patients, Johnson *et al* found that 63% were given the diagnosis more than 6 months after onset. Interestingly, dermatologists predominantly prescribed topical treatments or phototherapy, even to patients with linear and generalised morphoea, while rheumatologists frequently prescribed systemic immunosuppressive agents and physical therapy.¹⁰ Though DPM is a rare disease, any child with unexplained finger contractures and limb deformities in the early years of life should undergo detailed cutaneous examination for skin pigmentation with sclerotic changes and possible skin biopsy to confirm the diagnosis.

Learning points

- ▶ Disabling pansclerotic morphoea is the least common but highly morbid variant of localised scleroderma or morphoea.
- ▶ Children with unexplained finger contractures and limb deformities should undergo detailed cutaneous examination for skin pigmentation with sclerotic changes and possible deep biopsy to exclude this disease.
- ▶ Timely referral to appropriate specialists aids in making correct diagnosis and potentially alleviates procedures that could harm the patient.
- ▶ Systemic therapy with methotrexate, steroids and phototherapy along with physical therapy forms the mainstay of treatment. Surgical reconstruction may be required in resistant cases.

Contributors IJ and HRM drafted the initial version of the manuscript and procured the images. AK revised the manuscript for critically important intellectual content and prepared the final version.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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