Concise report

Access to care for children and young people diagnosed with localized scleroderma or juvenile SSc in the UK

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Abstract

Objectives. To describe pathways of care and referral to paediatric rheumatology from onset of first symptom (noticed by the patient or their family) to diagnosis for children and young people diagnosed with localized scleroderma (LS) or juvenile SSc (jSSc).

Methods. Retrospective case note audit of patients under paediatric rheumatology care who presented during January 2005–January 2010. Data included disease subtype, sex, age at key points in the referral pathway and health care professional (HCP) contact. All patient and HCP data were pseudo-anonymized in accordance with good clinical practice.

Results. Data were from eight UK centres that saw 89 cases: 62 females, 26 males; 73 LS, 16 jSSc. Median time from first symptom to first HCP review was 4 (range 0–72) months (LS) and 1 (range 0–50) month (jSSc). Median time from first symptom to paediatric rheumatology review was 15 (range 1–103) months (LS) and 7 (range 0–50) months (jSSc). Median time from first HCP review to first paediatric rheumatology review was 11 (range 0–103) months (LS) and 2 (range 0–10) months. First HCP seen (74%) was usually a general practitioner. The referring HCP to paediatric rheumatology was usually a dermatologist (56%) for LS. Median time from first symptom to diagnosis was 13 (range 1–102) months (LS) and 8 (range 1–50) months (jSSc).

Conclusion. A prolonged interval occurs from first symptom to definitive diagnosis, which may adversely affect outcome. There is a need to raise awareness of this rare diagnosis and facilitate earlier recognition.

Key words: paediatric/juvenile rheumatology, scleroderma and related disorders, skin, education (patients), outcome measures, primary-care rheumatology, attitude of health professionals.

Introduction

Scleroderma in childhood has two distinct groupings: localized scleroderma (LS) and the much rarer juvenile SSc (jSSc). Both LS and jSSc are very rare paediatric diseases (estimated incidence per million of 3.4 and 0.27, respectively [1]). Diagnosis and classification [2, 3] of LS and jSSc are essentially clinical. There is no universal consensus of classification criteria for use in clinical practice or clinical trials. LS is differentiated from jSSc based upon clinical findings of almost exclusive cutaneous involvement. Recent reports have documented a higher rate of extracutaneous manifestations (approaching 25%) than previously thought in LS, leading to calls for earlier
multi-system investigation and more aggressive treatment in those with widespread disease [4].

The pathogenesis of scleroderma is complex and remains poorly understood. Excessive collagen deposition and autoimmune dysfunction seem to be key factors and small-vessel vasculopathy also appears to play a role in jSSc [5, 6]. Optimal treatment is controversial, largely due to a lack of high-quality clinical trials. Most paediatric rheumatologists advocate systemic immunosuppression in a significant proportion of cases to avoid organ damage (particularly in jSSc [7, 8]), progressive deformity, functional disability and disfigurement [9, 10]. CSs, MTX and etanercept are widely used and case reports document use of other biologic therapies.

In the UK there is no consensus regarding optimal management for children and young people with scleroderma. It is well recognized that optimal management of complex, rare conditions involves concentrating expertise within specialist centres and shared care with local centres as appropriate—this model is strongly advocated within the British Society for Paediatric and Adolescent Rheumatology (BSPAR) Standards of Care for children and young people with JIA [11]. Consequently many children and young people with scleroderma in the UK are managed by paediatric rheumatology multidisciplinary teams (MDTs) in collaboration with colleagues in dermatology or paediatrics. Pathways through which children and young people reach these teams are often complex; a recently published UK prospective cohort reporting incidence data received notifications from paediatric and adult dermatologists (44%), paediatric and adult rheumatologists (31%) and paediatricians (25%) [1].

The aim of this study was to describe care pathways and the involvement of health care professionals (HCPs) in these pathways. Anecdotally, children and young people with scleroderma may present to various HCPs working in primary care and hospital services, and experience a complex and often protracted pathway before the diagnosis is confirmed and appropriate treatment commenced. These care pathways have not previously been investigated; this study aims to provide an initial assessment and, it is anticipated, will inform the development of strategies to facilitate earlier recognition and triage to optimal care.

Methods

A retrospective case note audit was performed and included patients under paediatric rheumatology care at the time of the study and who presented between January 2005 and January 2010. Data were collected using pre-tested pro formas piloted in three centres. Data collected included the age when the first symptom was noticed, age when first seen by a HCP (and description of the HCP involved), age when first referred to paediatric rheumatology, age when first seen by paediatric rheumatology and when the diagnosis was first entered into the medical record (by any HCP). In each case, age was recorded in years and months. Further information was collected regarding LS and jSSc subtypes; pro forma A (LS) included the anatomical location, depth and size of skin lesion at presentation (if recorded in the medical record). Anecdotal evidence suggests significant variation in methods used to evaluate skin lesions (depth, size and inflammatory activity) in UK paediatric rheumatology centres. Collecting and analysing these data were beyond the scope of this project; however, further detailed work is planned to investigate how this information is collected in the UK. Pro forma B (jSSc) included information regarding evidence of organ system involvement (as documented in the medical record) prior to assessment at a paediatric rheumatology centre and at diagnosis. In current UK paediatric rheumatology practice, anecdotal evidence suggests clinicians do not consistently use a standardized protocol for assessing organ system involvement. We therefore chose to focus on expert clinician assessment rather than potentially exclude a significant proportion of cases considering the rarity of jSSc.

All data (patient, HCP and centre) were pseudonymized and collected in accordance with good clinical practice [12]. The study was deemed an audit of clinical care and exempted from ethical approval. Only UK training centres were invited to participate and cases were only included where diagnosis had been established by expert paediatric rheumatologist opinion. Paediatric rheumatology trainees who had attended the BSPAR annual trainee meeting (Bristol, UK, November 2009) collected the data under supervision of their responsible consultant paediatric rheumatologist.

Results

Eight centres provided data on 89 cases (73 LS, 16 jSSc). The majority were females (overall 62/89, 70%; within the LS group 49/73, 67%; and within the jSSc group 13/16, 81%). The median number of cases per centre was 9.5 (range 2–22). Participating centres consisted only of representative UK tertiary teaching paediatric rheumatology centres.

Classification for LS cases (at the time of study) was as follows: linear scleroderma, 49 cases (67%); plaque morphea, 8 cases (11%); general morphea, 6 cases (8%); Parry–Romberg, 1 case (1%); multiple subtypes, 3 cases (4%); unspecified, 2 cases (3%); and not recorded, 3 cases (4%). Lesion depth at diagnosis (as documented in the medical record) was as follows: dermis, 1 case (1%); subcutaneous, 10 cases (14%); fascia, 3 cases (4%); muscle, 6 cases (8%); bone, 0 cases (0%); joint, 5 cases (7%), not recorded, 42 cases (58%). Of the 73 LS cases, 6 reported other systemic involvement: 1 autoimmune hepatitis, 1 arthritis, 2 CNS involvement (1 headaches, 1 unspecified), 1 ocular involvement and 1 an associated leg-length discrepancy. Associations were based on specialist paediatric rheumatology opinion as recorded in the medical record. From collected data however, it was not possible to differentiate between comorbidities and true associations.

Table 1 summarizes HCP first seen and referring HCP and compares LS with jSSc cases. Table 2 summarizes the key stages in the care pathway and Table 3
summarizes intervals between the key stages in the care pathway. Table 2 also summarizes missing data that were proportionally represented across LS and jSSc cases.

In most cases (74%), the first HCP seen was a general practitioner (GP). The HCP referring to paediatric rheumatology was usually a dermatologist (56%) for LS, and varied for jSSc. The median number of HCPs seen prior to referral to paediatric rheumatology was 2 (range 1–5). The diagnosis was usually made at a similar time to first review by a paediatric rheumatologist; however, in some cases the diagnosis was made either significantly earlier or later than first review in paediatric rheumatology services (Table 3). Data were not collected regarding the physician making the diagnosis.

### Table 1: First seen and referring HCP for LS and jSSc

<table>
<thead>
<tr>
<th>HCP</th>
<th>Event in care pathway&lt;sup&gt;a&lt;/sup&gt;</th>
<th>HCP first seen</th>
<th>Referring HCP</th>
<th>Referring HCP</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>LS</td>
<td>jSSc</td>
<td>LS</td>
</tr>
<tr>
<td>General practitioner</td>
<td></td>
<td>45 (62)</td>
<td>10 (63)</td>
<td>5 (7)</td>
</tr>
<tr>
<td>Paediatrician</td>
<td></td>
<td>5 (7)</td>
<td>0</td>
<td>14 (19)</td>
</tr>
<tr>
<td>Dermatologist</td>
<td></td>
<td>9 (12)</td>
<td>0</td>
<td>41 (56)</td>
</tr>
<tr>
<td>Adult rheumatologist</td>
<td></td>
<td>0</td>
<td>1 (6)</td>
<td>5 (7)</td>
</tr>
<tr>
<td>Emergency department physician</td>
<td></td>
<td>2 (3)</td>
<td>1 (6)</td>
<td>1 (1)</td>
</tr>
<tr>
<td>Orthopaedic surgeon</td>
<td></td>
<td>0</td>
<td>0</td>
<td>2 (3)</td>
</tr>
<tr>
<td>Paediatric neurologist</td>
<td></td>
<td>0</td>
<td>0</td>
<td>3 (4)</td>
</tr>
<tr>
<td>Paediatric hepatologist</td>
<td></td>
<td>0</td>
<td>0</td>
<td>1 (1)</td>
</tr>
<tr>
<td>Paediatric cardiologist</td>
<td></td>
<td>0</td>
<td>0</td>
<td>1 (1)</td>
</tr>
<tr>
<td>Dentist</td>
<td></td>
<td>1 (1)</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Not recorded</td>
<td></td>
<td>11 (15)</td>
<td>4 (25)</td>
<td>0</td>
</tr>
</tbody>
</table>

Absolute numbers given with percentages in parentheses. No missing data; percentages rounded, therefore not totalling 100.

<sup>a</sup>As recorded in patient medical record (notes, referral letters, etc.).

### Table 2: Ages at key stages in care pathway for LS and jSSc

<table>
<thead>
<tr>
<th>Disease subtype</th>
<th>Age of onset of first symptom (noticed by family)</th>
<th>Age when seen by first HCP</th>
<th>Age when referred to paediatric rheumatologist</th>
<th>Age when first seen by paediatric rheumatologist</th>
<th>Age when diagnosis first entered into notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>LS</td>
<td>6.8 y (1 m–15 y)</td>
<td>7.7 y (11 m–14 y)</td>
<td>8.5 y (22 m–14.8 y)</td>
<td>8.8 y (24 m–16 y)</td>
<td>8.7 y (24 m–15.7 y)</td>
</tr>
<tr>
<td>jSSc</td>
<td>8.5 y (9 m–15 y)</td>
<td>9.6 y (11 m–17 y)</td>
<td>9.7 y (11 m–17 y)</td>
<td>9.7 y (11m–17.1 y)</td>
<td>9.6 y (11 m–17.7 y)</td>
</tr>
<tr>
<td>Missing data, no. of cases</td>
<td>1</td>
<td>24</td>
<td>3</td>
<td>3</td>
<td>10</td>
</tr>
</tbody>
</table>

Ages expressed as mean in years (y) and months (m). Ranges in parentheses.

### Table 3: Intervals between key stages in the care pathway for LS and jSSc

<table>
<thead>
<tr>
<th>Disease subtype</th>
<th>Time from first symptom to seeing first HCP</th>
<th>Time from first HCP review to first paediatric rheumatology</th>
<th>Time from referral to paediatric rheumatologist to first paediatric rheumatology review</th>
<th>Time from first paediatric rheumatology review to diagnosis</th>
<th>Time from first symptom to first paediatric rheumatology review</th>
<th>Time from first symptom to diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>LS</td>
<td>4 (0–72)</td>
<td>11 (0–103)</td>
<td>1 (0–6)</td>
<td>0 (–67 to 51)</td>
<td>15 (1–103)</td>
<td>13 (1–102)</td>
</tr>
<tr>
<td>jSSc</td>
<td>1 (0–50)</td>
<td>2 (0–10)</td>
<td>0.5 (0–6)</td>
<td>0 (–1 to 7)</td>
<td>7 (0–50)</td>
<td>8 (1–50)</td>
</tr>
</tbody>
</table>

Ages expressed as median in months. Ranges in parentheses.
Discussion

This is the largest series of patients with childhood-onset LS and jSSc to focus on access to paediatric rheumatology care. We demonstrate a protracted interval from observed onset of symptoms to diagnosis, and furthermore a prolonged interval to access to paediatric rheumatology MDTs. In a disease such as scleroderma, where inadequate treatment can lead to deformity and disability, such prolonged intervals are likely to adversely affect outcomes.

We acknowledge that this study has a number of limitations. First, we only recruited patients under paediatric rheumatology care, which is likely to reflect bias towards more severe disease and may represent a different clinical spectrum of scleroderma to that observed in dermatology care (where children with seemingly mild disease may never be referred to paediatric rheumatology services).

Secondly, there was no predetermined standard method for case ascertainment by participating centres. Some, but not all, centres had clinical databases. In some centres, cases were recalled from memory of the team members, introducing potential recall bias. Given that the total number of cases contributed by each participating centre was small, reflecting the rarity of the disease, we would suggest that recall of individual patients is likely to be accurate.

Thirdly, this was a retrospective analysis of case notes, which likely introduced further bias in terms of the accuracy of the written documentation. Notably, there were missing data observed in several case notes as described.

Fourthly, there is no universally accepted diagnostic test for scleroderma, making verification of case ascertainment challenging; furthermore, there is no consensus on diagnostic criteria for LS and the ACR criteria for jSSc have been shown to be insensitive [13]. The 2007 proposed diagnostic criteria [3] for jSSc were not available at the start of the audit and, while useful, are not universally accepted in clinical practice or clinical trials. We therefore took a pragmatic approach to accept the clinical diagnosis documented by a consultant paediatric rheumatologist or dermatologist. The first documentation of the diagnosis was made in most cases by a paediatric rheumatologist at the initial assessment, although in LS cases more diagnoses were made by a dermatologist, reflecting differing pathways of care.

Given these biases and limitations, it is noteworthy that the age at which symptoms were first noticed (by the child, young person or their family) and the time between disease onset and first review by a paediatric rheumatologist was similar to other published retrospective and prospective cohorts [1, 14, 15], suggesting that this patient group is representative.

Consensus among paediatric rheumatologists is that jSSc and most LS lesions require systemic immunosuppression and MDT input; in LS this is especially important if lesions are recent, multiple or involving face or limb, as such lesions can be disfiguring and potentially disabling. Many such cases were observed in this cohort. It is therefore likely that patients with more severe disease are likely to benefit most from early systemic immunosuppressive therapy aimed at minimizing irreversible damage and functional loss. Therefore the observed protracted interval to confirmation of diagnosis and starting treatment is important and may adversely affect long-term outcome.

This study demonstrates that receiving an early diagnosis and obtaining access to specialist care are important challenges to improving outcomes for children with these rare conditions. It is noteworthy that some of the protracted interval arises from delay from first symptom to first HCP visit and referral to paediatric rheumatology. This suggests a need to raise public awareness as well as awareness among primary care physicians and paediatricians to consider scleroderma earlier and refer patients for specialist opinion.

Conclusion

This study suggests pathways to care are complex and involve many HCPs. We have observed a protracted interval from time of first symptom onset to diagnosis and referral to paediatric rheumatology. Access to care is therefore a major challenge when considering improving outcomes for children and young people receiving a diagnosis of scleroderma. Further work is required to raise awareness of this rare diagnosis, facilitate earlier recognition and obtain a referral to specialist services.

Rheumatology key messages

- Children with LS face complex pathways to care that involve many HCPs.
- A protracted interval exists between first symptom and referral to paediatric rheumatology services.
- Accessing care is a major challenge when considering improving outcomes for children with LS.

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