

## **SERVICE STANDARDS FOR THE CARE OF PEOPLE WITH EPIDERMOLYSIS BULLOSA**

*These service standards were produced in the UK and have been adapted for international use. However, since national healthcare systems vary, adaptation to national circumstances is recommended.*

### **1 INTRODUCTION**

- 1.1 Epidermolysis bullosa, or 'EB', refers to a rare group of hereditary skin fragility disorders, in which the skin and other body linings have a greatly increased tendency to develop blisters after minor trauma. EB occurs worldwide and can affect all racial groups. It is inherited in a 'mendelian' manner, transmitted by single gene defects in autosomal dominant or recessive modes with a frequency calculated to be 17 EB births per million live births.
- 1.2 The clinical manifestations of EB are very diverse since a number of different forms of the condition exist with varying phenotypes and prognosis. The common feature of the condition is an exceptional vulnerability to blistering at the slightest trauma. This can involve many systems apart from the skin.
- 1.3 Currently there is no cure for any of the forms of EB. However, effective symptomatic and preventative treatment is available, and is best managed by dedicated, specialised EB centres.
- 1.4 Historically, a major strength of the EB service has been when there is a close collaboration between clinical services and the patient support group, DEBRA. The objective should be a unified service with strong involvement of patients, their families and their representative organisation.

### **2 PATIENT CENTRED CARE**

- 2.1 Patients and their families should be provided with comprehensive information about the diagnosis of their form of EB, management options, and, where appropriate, likely prognosis. They should be given sufficient time and support so that they can make informed decisions about their treatment. They should be reassured at the time of diagnosis that there is a supportive process in place which will be explained in detail at their first visit to the specialist centre. This process should start at their first visit to the specialist centre and continue throughout their care at the centre.
- 2.2 The information provided should be clear, understandable and appropriate for the patient's age, education and cultural background. Any verbal information should be documented for future reference and copied for the patient's personal use. Interpreters must be provided to allow staff to communicate verbally and in writing with patients who do not speak or understand the language used at the centre.

Arrangements must also be made to allow communication with patients with visual or hearing disabilities.

- 2.3 Patients, families (including carers and advocates) and general practitioners should have access to all members of the multidisciplinary team to discuss specific problems or concerns.
- 2.4 A specialist advisory service should be available to other hospital-based and primary care providers and to patients and families through appropriate means, including a telephone hotline. The patient organisation, DEBRA, may play a significant role in this service.
- 2.5 A range of psychological and social support services should be offered to meet the needs of patients and families and made available at the specialist centre and in the community. Links to facilitate this at local level should be developed.
- 2.6 Patients should be provided with information about relevant patient organisations.
- 2.7 The views of patients and families should be formally sought at regular intervals and the results communicated to the relevant service providers.
- 2.8 There should be formal arrangements for addressing complaints and other comments by patients, families, patient organisations and staff.
- 2.9 Staff in the multidisciplinary team should be trained in communication skills, including ways of breaking bad news.
- 2.10 Patients who are dissatisfied with their treatment plan must be informed of their right to a second opinion.
- 2.11 There should be written guidelines for shared care between the specialist centre, referring physician, voluntary agencies and general practitioner. Written guidelines may also be needed for sharing care with other specialist centres.

### **3 DIAGNOSIS AND ASSESSMENT**

- 3.1 Accurate diagnosis is essential for the immediate and future management of all EB patients. It may be very complex, or relatively straight-forward when undertaken in a specialised centre. It will take account of several factors including the medical and family history, the type and distribution of clinical lesions, including blisters and scars, the body systems affected, and the age at which the condition first presented.

3.2 Services must be consultant led and provided by specialists with experience in EB. The specialist team should be appropriately qualified and trained to manage patients of different age groups (infants, children, adolescents and adults).

### **3.3 Diagnostic Support**

3.3.1 There should be access to a range of relevant diagnostic procedures.

3.3.2 Where possible, the procedures should be organised around a 'one-stop' service to avoid unnecessary repeat visits by patients.

### **3.4 Laboratory Facilities**

3.4.1 A specialised laboratory service should be capable of carrying out all the tests relevant for the diagnosis of EB. This includes transmission electron microscopy and immunofluorescence microscopy of skin biopsy samples, and molecular biology techniques for the analysis of DNA or RNA from different sources, including blood samples.

3.4.2 There should be access to a range of hospital-based diagnostic services including histopathology, radiology, microbiology, virology, biochemistry, haematology and blood bank services.

3.4.3 Access to specialised services offering prenatal and pre-implantation genetic diagnosis should be available where appropriate.

3.4.4 Accreditation under national regulations should be mandatory for all clinical laboratories as should adherence to national standards of best practice..

3.4.5 The specialized laboratory should participate in reference functions for non-specialist laboratory providers and also provide resources and expertise for discussion of atypical and complicated cases.

3.4.6 Laboratories should participate in external quality assurance programs for each analytical service offered and provide open access to advice and recommendations concerning individual analytical procedures at non-specialist provider sources.

### **3.5 Other Diagnostic Facilities and Support Services**

3.5.1 There should be ready access to echocardiography.

3.5.2 DEXA scans, general radiology, CT and MRI scanning should be available as elective investigations.

## **4 CARE AND TREATMENT**

4.1 The care of patients with EB is complex. Management strategies will depend to some extent on the nature and severity of the condition, as well as the age of the patient. Although specific therapies have yet to emerge, stabilisation of the condition and prevention and management of complications can greatly improve the quality of life for the affected patients as well as the immediate family in the home setting.

## **4.2 Care setting**

4.2.1 In general, care of children within a children's hospital is preferable. In all cases national standards for the care of children in hospital must be met.

4.2.2 The centre must ensure that patients with EB can be admitted without delay to a suitable ward staffed by personnel familiar with care of patients with EB.

4.2.3 Staff on admitting wards and in Emergency departments must be aware of the centre's arrangements for care of patients with EB and ensure prompt referral to the EB team.

4.2.4 Each centre should have regular consultant or specialist led ward rounds of any EB in-patients and 24 hour access to specialist EB doctors or nurses for inpatients.

## **4.3 Team care**

4.3.1 Children and adults with EB require the care of large multidisciplinary teams. EB is a systemic disease. The numerous specialists involved need to be experienced and aware of the range and sometimes unique complications these patients can develop, depending on the type of EB. Patients may require access to the following specialists.

4.3.1.1 The team serving children with EB should include as full members

- Consultant dermatologist
- Consultant paediatrician
- Dermatology clinical nurse specialist
- Dietitian
- Physiotherapist
- Dentist
- Dental hygienist
- Pharmacist
- Psychologist
- Plastic surgeon with particular expertise in hand surgery
- Gastroenterologist
- General surgeon
- Ophthalmologist
- Clinical geneticist
- Social worker/ Social Care Manager

Endocrinologist  
Cardiologist  
Interventional radiologist  
Occupational therapist

4.3.1.2 The following specialist services should also be available locally for children

Radiology  
Occupational therapy  
Orthotics  
Palliative care  
Play therapy  
Pain clinic  
ENT  
Speech & language therapy  
Urology  
Podiatry  
Adolescent services and a framework for transitional care.

4.3.1.3 The team serving adults with EB should include as full members

Consultant dermatologist  
Dermatology clinical nurse specialist  
Nurses serving dedicated beds, skilled in nursing EB patients  
Dietitian  
Physiotherapist  
Dentist  
Dental hygienist  
Pharmacist  
Psychologist/psychiatrist  
Surgeon with particular expertise in hand surgery  
Plastic surgeon with particular expertise in removing tumours  
Gastroenterologist  
General surgeon  
Ophthalmologist  
Clinical geneticist  
Social worker  
Occupational therapist

4.3.1.4 The following specialist services, in addition to those listed for children (apart from play therapy), should be available locally for adults

Endocrinologist  
Oncologist (both medical and radiotherapy)  
Cardiologist

4.3.2 Arrangements for the transfer of adolescents from the paediatric services to adult care should be undertaken in a manner that takes account of their emotional management and chronological age.

- 4.3.3 The multidisciplinary team should include named specialist nurses involved in the administration of all aspects of care including assessment, continuing care and counselling. The nurses' duty rotas must be compliant with working time directives.
- 4.3.6 The multidisciplinary team should include a named physiotherapist, occupational therapist, dietitian and pharmacist. These members should facilitate best management of patients according to their national professional good practice guidelines.
- 4.3.7 Each centre should have a documented social support system to aid post-discharge care.

#### **4.4 Team process**

- 4.4.1 All members of the multidisciplinary team should attend quarterly audit meetings, in line with good clinical governance.
- 4.4.2 Minutes of multidisciplinary team meetings, including a register of attendance, should be recorded.

### **5 PALLIATIVE CARE**

- 5.1 Palliative care should be provided in or close to the patient's home with specific advice from the specialist centres.

### **6 MANAGEMENT AND ORGANISATION OF EB PROGRAMME**

- 6.1 There should be clear and accountable leadership of the service at each centre with a named lead clinician.
- 6.2 A named manager should support the lead clinician.
- 6.3 Job plans for the lead clinician and manager should include adequate time to perform their roles.
- 6.4 The lead clinician is responsible for coordinating the activities of the multidisciplinary team and ensuring good communication between team members.
- 6.5 The lead clinician is responsible for ensuring that staff are aware of the standards against which centres will be assessed and that mechanisms are in place to comply with those standards.
- 6.6 There should be evidence-based protocols and these should form part of a formal induction for all new staff joining the service.

- 6.7 Centres should participate in clinical governance activities within their host hospital that are relevant to the EB service.
- 6.8 There should be effective and sustainable workforce planning covering all professional disciplines included in the multidisciplinary team.
- 6.9 All staff should have regular appraisal and agreed professional development plans.
- 6.10 There should be regular business meetings to address issues specific to the EB service including financial reports, activity reports, education, audit, clinical governance and research.
- 6.11 Arrangements should be in place for timely and accurate collection of data.

## **7 COMMUNICATION WITH GENERAL PRACTITIONERS AND REFERRING PHYSICIANS**

- 7.1 General practitioners and referring physicians must receive adequate written feedback after patients are assessed.
- 7.2 When patients are discharged from hospital, general practitioners must be provided with an information pack detailing medication, potential side effects and drug interactions. Guidance must be included on procedures to adopt in case of a need for emergency admission.
- 7.3 After each visit to the follow up clinic, a letter detailing results of investigations and changes in medication must be sent to the general practitioner within seven working days. It is good practice to copy this letter to the patient with their consent.
- 7.4 A member of the EB team should be available at all times to answer calls from General Practitioners and others caring for the patient.

## **8 ADMINISTRATIVE SUPPORT**

- 8.1 Each centre should have adequate dedicated office accommodation for consultants, nurses, secretaries and data clerks.
- 8.2 Centres should have access to well organised secretarial support and facilities to allow mailing of discharge summaries within 5 working days and clinic letters within 7 working days.
- 8.3 Specific rooms should also be available for multidisciplinary team meetings and for counselling patients and their carers.
- 8.4 Centres should have sufficient IT facilities to allow rapid retrieval of patient details including results of laboratory and other diagnostic investigations.

8.5 All centres should have at least one dedicated data clerk to maintain an up-to-date database on all patients referred and assessed.

8.6 All centres should contribute up-to-date data to national EB registries.

## **9 EDUCATION AND TRAINING**

9.1 All members of the multidisciplinary team are expected to participate in continuing professional development by attending appropriate courses and meetings.

9.2 Consultant members of the multidisciplinary team should attend at least two specialist national or international meetings per year.

9.3 Trainees should be encouraged to participate in clinical research projects and to submit papers for presentation at local, national and international meetings.

9.4 Nursing staff should attend at least four relevant study days per year.

9.5 Specialised services are responsible for the provision of educational and audit programmes for all clinical staff, specialised nurses, clinical scientists, counsellors, undergraduate and postgraduate students as well as other personnel required to promote optimal patient care.

## **10 RESEARCH AND DEVELOPMENT**

10.1 Centres for the treatment of EB should belong to a national anonymised data registry to ensure that there is comprehensive evaluation of the burden of disease and the frequency of morbidity and of the specific disorders nationally.

10.2 Registries should record adverse events and treatment outcomes during the assessment of all patients diagnosed and receiving specialist care from the specialised service.

10.3 A record should be maintained of patients managed under a shared-care arrangement with a non-specialist unit.

10.4 Each centre should have a research strategy stating the goals of the research and proposed involvement of staff and other resources.

10.5 Centres should have sufficient staff, space and facilities to support the research strategy and a well-regulated funding system to support pilot/preliminary projects.

10.6 Centres should have a clinical information database with written protocols for data entry and quality assurance and a dedicated data clerk.

- 10.8 All clinical research studies must have ethical approval and patients must give fully informed consent before participating in research studies, in line with the requirements of good clinical practice.
- 10.9 The collection, storage and use of donated tissues and organs for research must conform to current national guidelines on the uses of biological materials for research.
- 10.10 All centres are expected to participate in national research projects and audits.
- 10.11 Centres are expected to present results of their research projects at national and international conferences and to publish them in peer reviewed publications.
- 10.12 Clinical practice should be influenced by regular review of research evidence derived from local, national and international research activity.

## **11 AUDIT**

- 11.1 There should be quarterly audit meetings, attended by all members of the multidisciplinary team , to confirm good practice and to identify and rectify deficiencies in patient care, inefficient processes and poor communications.
- 11.2 Minutes of audit meetings should be recorded.
- 11.3 Audit meetings should address:
  - 11.3.1 Clinical performance and outcome.
  - 11.3.2 Process-related indicators, e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow up etc.
  - 11.3.3 Stakeholder satisfaction including feedback from patients, their families, patient organisations, referring physicians and General Practitioners.
- 11.4 All centres must participate in any national audit commissioned.
- 11.5 Data recorded for the national audit should be accurate, complete and transmitted on time to the national audit database.
- 11.6 There should be a dedicated data clerk with responsibility for collection, storage and transmission of the audit data.
- 11.7 There should be a mechanism for checking the accuracy and completeness of the audit data.
- 11.8 Reports from the national audit should be distributed to all members of the

multidisciplinary team, and to the host hospital's management. Centres should review their results in comparison with other centres to identify areas for improvement.

- 11.9 Audit reports should be effectively anonymised to conceal patient identity and made available to patients, their General Practitioners and referring physicians as well as NSCAG and the relevant patient organisation.

***These service standards are based on those devised by the lead clinicians at St Thomas' Hospital, Great Ormond Street Hospital for Children, Birmingham Children's Hospital, Solihull Hospital and by DebRA.***