

A12/S(HSS)/a

**2013/14 NHS STANDARD CONTRACT
FOR EPIDERMOLYSIS BULLOSA SERVICE (ALL AGES)**

PARTICULARS, SCHEDULE 2 – THE SERVICES, A - SERVICE SPECIFICATION

Service Specification No.	A12/S(HSS)/a
Service	Epidermolysis bullosa service (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Epidermolysis bullosa (EB) encompasses a group of rare inherited disorders that cause life-long blistering and ulceration of the skin and mucus membranes. Blistering is often apparent at or soon after birth, but the severity of the condition varies greatly depending on the specific type of EB. Some forms have problems limited only to the skin, whereas other forms involve many other organs.

EB is incurable at present and severe types of disease shorten life expectancy. In the most severe type of EB, Herlitz-junctional EB, infants rarely survive beyond the first year or two of life. In another severe form, severe generalized recessive dystrophic EB, patients have life-long chronic blistering, scarring, contractures and malnutrition, and will usually die in the third to fifth decades due to disseminated skin cancer. However, whilst the disease is incurable, a huge amount can be achieved to improve the quality of life, ameliorating the effects of blistering and ulceration of the skin, along with preventing and treating the multiple complications that arise.

EB is a rare disease; it is estimated to affect 1 in 17,000 live births and there are around 5,000 people living with EB in the UK. Whilst individuals and families with the milder forms of EB may not seek medical help or a specialist opinion, those with more severe forms will require life-long, intensive medical input from many different specialists and disciplines. As an extremely rare disease, very few dermatologists or other specialists will have had much exposure to EB and will not have the specialist knowledge to deal with patients in a holistic way. Also, by virtue of being rare, there are very few published guidelines or evidence-based clinical trials in EB. However, it is well accepted in the EB community worldwide that EB patients are best cared for in a multi-disciplinary team (MDT) setting. Key papers about managing EB are listed

below

2. Scope

2.1 Aims and objectives of service

Aims and General Overview

The national Epidermolysis Bullosa (EB) service provided by Birmingham Children's Hospital NHS Foundation Trust, Great Ormond Street Hospital for Children NHS Foundation Trust (GOSH), Guys & St Thomas' Hospital NHS Foundation Trust Heart of England NHS Foundation Trust (GSTT) aims to provide diagnosis and assessment of infants, children, adolescents and adults with suspected or known EB, along with treatment and long-term support.

EB is a heterogeneous group of inherited diseases characterised by blistering of the skin and mucosae following minor mechanical trauma. Different types of EB vary in their severity and associated non-cutaneous features. Whereas the milder forms result in painful blistering limited to localised areas of skin, more severe forms are associated with death in infancy, or life-long chronic skin loss, scarring, pain, systemic involvement and early death from metastatic skin cancer.

There is great variability in severity between the different forms. In the most severe forms, death is usual within the first few months of life. A precise genetic diagnosis can be made in over 90% of cases.

Currently, no specific treatment is available for any form of EB. Therefore, the principal goals of management are:

- to make a precise diagnosis as early as possible;
- to provide accurate information relating to prognosis and the natural history of a patient's disease;
- to relieve symptoms;
- to provide the best possible quality of life for patients and their families;
- to prevent complications of the disease;
- to provide genetic counselling to affected individuals and families.

Patients

The following conditions are regarded as falling within the spectrum of EB:

- all forms of EB simplex, including plakophilin and desmoplakin deficiency
- all forms of dystrophic EB
- all forms of junctional EB, including laryngoonychocutaneous (LOC) (Shabbir) syndrome
- Kindler syndrome
- Epidermolytic ichthyosis (in babies and children)
- other severe genetic skin fragility conditions

The section below will be refined and revised in 2013/14 to reflect the work being undertaken to better clarify the allocation of patients to one of two categories - "mild" or "severe" - according to the severity of their disease and their medical needs.

'Mild':

all patients requiring input from the basic EB team only (dermatologist and/or paediatrician, CNS, podiatrist)

'Severe':

all patients requiring input from the basic EB team and additional EB team specialists.

Severity should be reassessed after school entry and at transition, but can be reassessed at any point if clinical need indicates this is necessary. Patients with 'mild' disease will generally need to be seen in one of the designated centres at any early stage, for precise clinical and, in some instances, skin biopsy or molecular diagnosis.

'Mild' patients

Having determined that a patient has 'mild' disease, it may be possible for care to be provided locally as part of a shared care arrangement with the national service.

'Severe' patients

Patients with 'severe' disease will need to be assessed during the early neonatal period in their local neonatal unit, by medical and/or nursing staff from one of the designated centres. Clinical diagnosis is exceedingly difficult at this age, therefore, unless the diagnosis within the family is known; a biopsy will be taken for immunohistochemical and/or electron microscopy diagnosis and sent to the designated diagnostic laboratory.

A decision will be made whether it is preferable to transfer the child immediately to one of the designated centres, or whether initial care would most safely and comfortably be provided without transfer (prematurely transporting a child may be harmful). If the child is not transferred, arrangements should be made, where appropriate, for follow-up in one of the designated centres.

Objectives and expected outcomes:

- to offer accurate and timely diagnosis of EB, including, where necessary, genetic counselling and information regarding future recurrence risks in the family;
- to provide, where possible, accurate prognostic information to the patient and family;
- to provide symptomatic care e.g. pain control;

- to monitor for and, where possible, to prevent complications of the disease;
- to raise awareness and to offer surveillance for cutaneous squamous cell carcinoma, where appropriate, and to offer comprehensive oncology and surgical care within the context of a skin cancer multi disciplinary team;
- to support parents of affected children, and EB patients themselves in having children, through liaison with obstetric teams and midwives, as well as referring for accurate genetic counselling;
- to actively engage and support adolescents in transitioning from the paediatric to adult EB centres;
- to assess the quality of life of patients at transition;
- to provide an excellent EB diagnostic service with rapid turnaround times for skin biopsy specimens and molecular screening;
- to provide prenatal diagnosis through first trimester DNA-based testing, foetal skin biopsy and pre-implantation genetic diagnosis (PGD). Funding for PGD must be sought from the patients' local commissioners before treatment is commenced.
- to be seen as the leading clinical service and a source of expert advice for the diagnosis and management of EB within the NHS;
- to provide support, advice, and guidance to the wider NHS on the management of patients with EB within their local care provider structure;
- to provide high quality information for patients, families and carers in an appropriate and accessible format;
- to develop the experience, knowledge and skills of the multi-disciplinary team (MDT) to ensure high quality sustainable provision of the national service;
- to develop and participate in active research programmes to improve the clinical care of EB patients, working towards more effective therapies and, eventually, a cure. Whilst this element is not nationally funded there is an expectation that all parts of the national service will actively participate in research.
- EBQoL or PedsQL should be undertaken periodically in adults and children above 2 years, respectively. EBQoL should be done in all patients at transition.
- length of life and age at death should be recorded across the different EB sub-types and can be used as a longer term outcome measure for the service.

2.2 Service description/care pathway

Service description

Care of EB patients should be delivered in a variety of ways: outpatient clinics; multi-disciplinary day case reviews; day case reviews; inpatient admissions (elective and emergency); outreach Clinical Nurse Specialised (CNS) visits. Patients should have easy access to the EB team and be informed of how emergency access and support can be obtained.

Paediatric EB service

The EB service can be divided into outpatients, day care, inpatients, outreach and telephone consultation as well as teaching and training.

Outpatients

Children should be seen in clinic by a consultant dermatologist or paediatrician, and EB clinical nurse specialist, at least once a year.

Inpatient, planned admission

Children with severe types of EB require multiple reviews on average every 3 to 6 months. Since patients come from all over England this may require a night or two in hospital. During their stay, children will have multiple consultations with the EB MDT team.

Key core disciplines for paediatric EB patients

- Consultant dermatologist
- Consultant paediatrician
- EB clinical nurse specialist
- Dietician
- Physiotherapist
- Occupational therapist
- Ophthalmologist
- Dentist
- Psychologist
- Interventional radiologist
- Palliative care/pain clinician
- Plastic/hand surgeon
- Podiatrist
- Service co-ordinator
- Administrative support

Other MDT members of the wider EB team to be determined as deemed necessary for the EB patient population. These wider disciplines should be accessed via normal NHS referral mechanisms close to the patient's local care providers, and, where appropriate consist of specialists and therapists including dental hygienist, gastroenterologist, urologist, speech and language therapist, ear nose and throat (ENT) surgeon, cardiologist, endocrinologist and orthotics.

Initial assessment: paediatrics

Neonates with 'severe' disease will be assessed in their local neonatal unit, by an EB CNS in communication with the EB paediatrician or dermatologist. Clinical diagnosis is exceedingly difficult at this age; therefore a biopsy will usually be taken by the nurse and sent for immunohistochemical +/- electron microscopic diagnosis to the designated EB diagnostic laboratory. A decision should be made whether it is preferable to transfer the child immediately to one of the nationally designated centres, or whether initial care would most safely and comfortably be provided without transfer (prematurely transporting a child may be harmful). If the child is not transferred, arrangements should be made for follow-up in one of the designated

centres when appropriate, but with outreach CNS support until then. Many babies with Herlitz JEB will receive all care locally and from the outreach team without being reviewed face-to-face in either paediatric specialist centre.

For older babies and children who are fit to travel, initial assessment will be as outpatient or day case at one of the nationally designated centres.

Adult EB service

Key core disciplines for adult EB patients

- Consultant dermatologist
- EB CNS
- Dietician
- Physiotherapist
- Occupational therapist
- Ophthalmologist
- Dentist
- Psychologist/psychotherapist
- Interventional radiologist
- Palliative care/pain clinician
- Plastic/hand surgeon
- Podiatrist
- Service co-ordinator
- Administrative support

Other MDT members of the wider EB team to be determined as deemed necessary for the EB patient population. These wider disciplines should be accessed via normal NHS referral mechanisms close to the patient's local care providers, and where appropriate consist of specialists and therapists including, dental hygienist, gastroenterologist, urologist, speech and language therapist, ENT surgeon, cardiologist, endocrinologist and orthotics.

Outpatients

Adult EB patients should be reviewed at least annually with a wider MDT as required. Generally, those with more severe forms of EB, especially those with a previous history of skin cancer, may be reviewed every 3-6 months. The adult outpatient EB clinics must be consultant dermatologist led and an EB CNS must also be present. The clinics should have facilities for taking skin biopsies and blood samples for the EB laboratory.

Day cases

Some EB patients may require minor treatments e.g. blood or iron infusion, biopsy of suspicious skin lesions, as a day case attendance.

Inpatients

The service should have access to inpatient facilities for the admission of patients electively for a variety of procedures e.g. oesophageal dilatation, excision of squamous cell carcinoma. Emergency admission may be required if a patient is medically unwell as a consequence of their EB e.g. septic, dehydrated, renal problems. The EB team will make a decision as to whether admission locally or to the specialist centre is most appropriate based on clinical need.

Management in between annual reviews

Between annual review visits to the national centre, patients should be seen as required by local paediatricians, dermatologists and by their nursing teams. It is the responsibility of the national centre to ensure clear management plans are communicated to local care providers. In some cases patients may need to be transferred to the designated centres in the event of acute illness or complications requiring specialised care. The designated centres should ensure communication indicates when this would be deemed necessary.

Outreach (adults and paediatrics)

The EB outreach service is fundamental to providing the best possible care for babies, children, adolescents and adults with EB. Outreach is delivered as direct care and advice supporting local care providers. It is also an integral part of the service but must be delivered within an appropriate geographical area. The service must demonstrate the team resource is being used efficiently and effectively.

Paediatric patients

When a neonate is born with skin fragility or blistering, one of the two paediatric EB centres is contacted. Clinical images and history are reviewed by the relevant EB team. Once a diagnosis of EB is confirmed or felt to be highly likely, advice should be given immediately along with couriering specialist dressings and written information, if necessary. It is expected, ideally in the first 24-48 hours, that one of the EB CNSs will travel to the infant and provide immediate training to parents and nursing staff on how to manage the blistering and reduce the risk of further skin damage. CNSs should perform a diagnostic skin biopsy along with blood sampling from the child and family for mutation analysis.

The service should continue this outreach service following discharge from hospital, tailored to the individual's and family's need in order to teach families and the local paediatric community team in caring for the child at home, with an emergency and end of life care plan where appropriate. This will avoid the need for the family having to travel long distances to the EB centre with a very sick child who may suffer undue skin damage or airway difficulties if made to travel. The lead consultant may also visit the child and family if necessary to establish links with local services.

Adult patients

The service via the EB CNSs should provide outreach support where necessary. It

may be necessary to visit patients ahead of multi-disciplinary clinic appointments to undertake full skin examination and photography (which are sometimes not feasible in hospital) and identify suspicious areas of skin that will require examination and biopsy in hospital. The CNS may also visit teenagers from the paediatric EB services ahead of transfer to the adult service to aid smooth transition and gain a better understanding of the individual's requirements.

It is also important that links with local care providers are maintained ensuring skills and knowledge about EB is passed on so patients are well supported by their local care providers.

For other types of EB, the outreach service provides support in optimising skin care management as well as training of the local team.

Genetic counselling

This is not a core part of the service and therefore EB teams can provide general advice with some detailed input from the EB consultant. The teams are required to signpost to appropriate local genetic services in line with the shared care model. Genetic services exist which can support patients and patients should be referred to them for further support as appropriate. This must be completed with funding agreed from the patient's local commissioners.

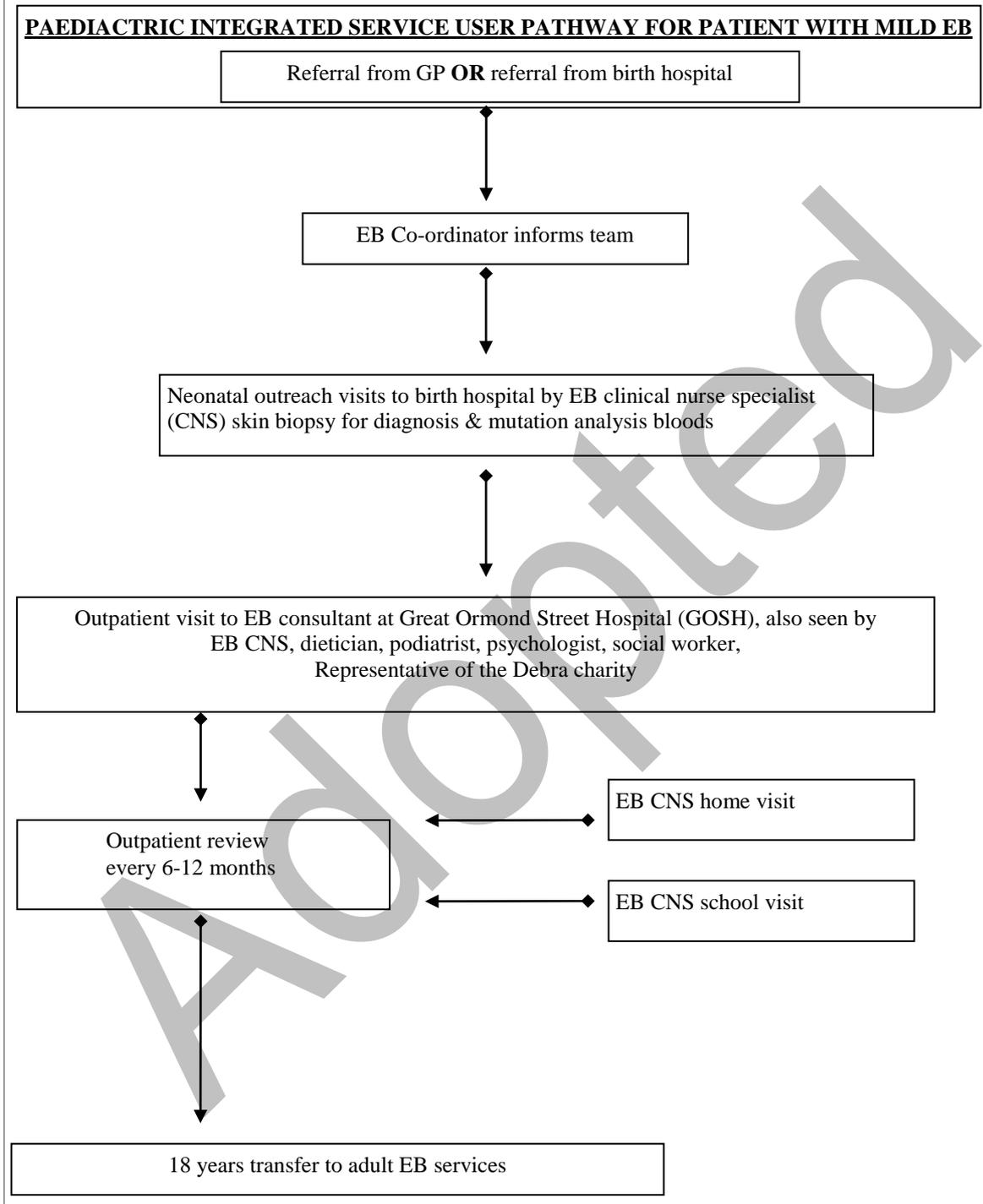
Surgical procedures

A number of surgical interventions/ procedures are undertaken as part of this service. These include the following :

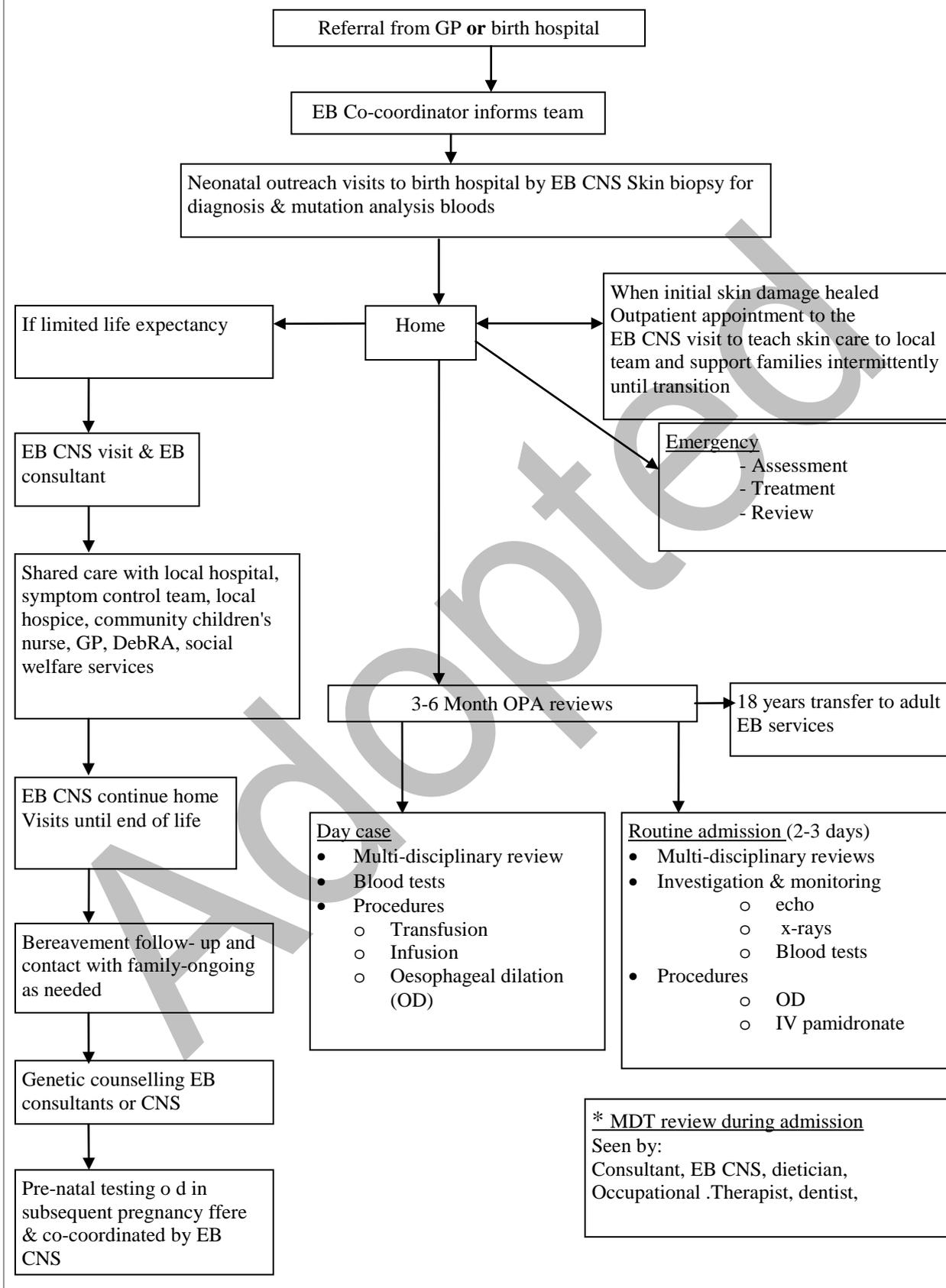
- Oesophageal dilatation
- Insertion of gastrostomy tube
- Surgery to manage contractures e.g. of hands
- Excision of skin cancers/amputation/regional lymph node dissection
- Insertion of central venous access
- Tracheostomy

The surgical procedures not included are those which are unrelated to the underlying diagnosis, or that need to be carried out in the specialist centre due to issues surrounding handling, surgery or anaesthesia due to the underlying diagnosis of EB e.g. tonsillectomy, orthopaedic procedures, circumcision. All 'severe' patients should have surgical procedures carried out at the specialist centre, unless agreed otherwise by the specialist centre. For any procedures undertaken that are unrelated to the underlying diagnosis of EB the trust must obtain prior agreement from the local commissioner before proceeding. In some 'mild' cases, however, EB patients may be referred to local care providers for these procedures with advice on how to support the patient during the procedure.

3.1 Service model and care pathways



INTEGRATED SERVICE USER PATHWAY FOR PAEDIATRIC PATIENTS WITH SEVERE EB



Days/hours of operation

Monday to Friday 9am - 5pm.

Out of hours, a telephone on-call dermatology service should be available and there should be clear arrangements for emergency on-call available via the provider's main switchboard.

A dermatology consultant is required to be on call out of hours at all times for any emergencies.

On call arrangements

Patients and carers should be given information about how to access the dermatologist on-call during emergency situations and out of hours. It is not necessary for the EB nurses to be on call.

Laboratory facilities

The EB service across all 4 centres is serviced by the Robin Eady National Diagnostic Laboratory at Guys & St Thomas' Hospital NHS Foundation Trust which is one of the foremost and well-respected international centres for EB diagnosis from skin biopsies, molecular testing and prenatal diagnostics. The lab offers skin biopsy EB diagnosis (immunohistochemistry, electron microscopy, molecular testing based on skin cDNA), genetic testing of the 14 known EB genes, and prenatal testing for severe forms of EB (DNA-based, foetal skin biopsy and pre-implantation genetic analysis (alongside the Assisted Conception Unit at GSTS)). Some genetic testing of 2 EBS genes and other skin fragility genes e.g. TGM5, KRT1/2/6/10/16/17, desmoplakin, is also currently undertaken at Ninewells Hospital Dundee.

The EB CNSs can co-ordinate prenatal testing, but funding for this must be agreed with the patient's local commissioners beforehand.

All providers must demonstrate that patients, carers and care providers are given support and advice on how to manage the condition once an initial diagnosis and management plan have been given, with advice on how to gain access from the national team when required.

Patient engagement and involvement

National EB service providers will work with the NHS England to ensure that sufficient consideration is given to communications with all stakeholders. The EB patient support groups and charities should be very closely involved in service design. Service changes and improvements should be tailored around identified needs.

Risk Management

The provider will ensure that professionals within the trust adhere to the standards of their regulatory bodies.

Care delivered by the national EB service must be of a nature and quality to meet the care standards, specification and agreement for the service. It is the trust's responsibility to notify the commissioner on an exceptional basis should there be any breaches of the care standards. Where there are breaches, any consequences will be deemed as being the trust's responsibility.

Patients must be managed in line with the specification and care standards. Any deviation from these, which has not been approved by the NHS England, is at the trust's risk both clinically and financially. It is the trust's responsibility to inform the commissioners of any such non-approved deviations on an exceptional basis.

Where a patient's presentation challenges the assumptions that underpin the specification, service standards and contractual arrangements, it is the trust's responsibility to inform the commissioners on an exceptional basis, prior to any treatment (except for emergency treatment) so that the implications of the patient's requirements can be considered. This does not affect situations where the Individual Funding Application process applies.

National EB service

The national EB service is commissioned NHS England and requires the designated centres to provide optimum care and support to patients with EB within the funds that have been agreed and allocated. This funding includes all consumables including dressings required in outpatients and the occasional inpatient stay settings. Dressings that are required once patients have been returned to their local care providers are prescribed by the GP and local care providers. Patients are not required to bring in prescribed dressings from home for an inpatient admission.

EB nursing

An element of the funds for the EB service is allocated to EB nursing. The following EB nursing principles should apply across all national EB centres.

National EB service revised nursing principles

The nursing team employed and based at the 4 national centres cover England only. Patients in Scotland have a dedicated EB nursing team.

Expectation from the nursing team:

- The nursing teams **MUST** be employed by the provider for core and non-core EB nursing within the nationally designated centres;
 - This ensures appropriate professional, clinical and corporate governance.
 - This would prohibit nursing staff from holding dual contracts with the patient support group.

- Nurses will only work with patients registered and active at their nationally designated centre (hospital & outreach);
 - All patients must be registered at a national EB centre (ideally this will be the closest centre but a choice of centre MUST be offered to all patients);
 - A patient is considered as active if seen a minimum of once a year in a face to face consultation with the EB team (this can be an EB nurse (some patients may fall outside of the 12 month review timeline but cannot exceed 14 months)); (N.B a telephone call does not constitute a consultation.)
- Patients should receive outreach from the EB centres closest to them to ensure effective use of resources;
 - It is recognised that there are some historical links with patients and these need to be managed effectively within the resources allocated – all patients MUST be offered choice.)
- NHS England resources must be used to address the core nursing priorities identified below (table 1);
 - Non-core activities should be supported from DebRA, which may or may not involve investment in the provider trust.
- Providers will manage within the resources provided;
 - Providers must record and report all activity classified as core nursing priorities and non-core functions as per contract and service level agreement (SLA).
 - Service reviews and reports must give assurance to the NHS England that core nursing priorities are being delivered. Non-core activities to be reported to Dystrophic Epidermolysis Bullosa Research Association (DeBRA).

Table 1: The table below lists the core nursing priorities and non-core activity which have been agreed by DeBRA, the patient support group.

Core nursing priorities	Definition
Outpatient care	Support in outpatient clinics at centre relating to EB outpatients direct care or care advice.
Inpatient	Support for inpatient clinics at centre relating to EB inpatients direct care or care advice. EB nurses to support ward & clinical staff to deliver care (advise, train and co-work)
Outreach within appropriate geographical area	Outreach delivered as direct care advice supporting local care providers. It also an integral part of the service but must be delivered within an appropriate geographical area. The service must demonstrate the nursing resource is being used effectively.
Telephone advice	Telephone advice to patients and health professionals for patients registered at

	the national centre.
Transition support	Nurses must provide advice, support and guidance throughout transition (both adult & paediatric)
Pre-natal diagnosis advice	This is not a core part of the service and however EB nurses to give general advice and then co-ordinate pre –natal screening and signpost to appropriate local services in line with the shared care model. This must be completed with funding agreed from the patient's local commissioners.
Genetic counselling	This is not a core part of the service and therefore EB nurses to give general advice and then refer to EB consultant or signpost to appropriate local genetic services in line with the shared care model. Genetic services exist which can support patients and patients should be referred to them for further support. This must be completed with funding agreed from the patient's local commissioners.
Palliative care	This is also not a key aspect of the specification. EB nurses to signpost patients/ families to available resources. Support/ educate / liaise with local services/ providers to deliver EB aspect of palliative care.
Bereavement support	EB nurses will only undertake one follow up post death of EB patient. Additional support to be provided by DeBRA.
Management relating to core work	This is applicable to the lead nurse only who will be responsible for the management of the wider EB nursing team. Audit, writing letters, activity recording, email, service development & liaison with patients will be undertaken by all EB nurses.
Teaching healthcare professionals	This relates to teaching healthcare professionals/carers/ families on managing EB for specific patients registered at the national centre, and local clinical guideline development. Joint partnership with DeBRA to improve wider education combining core and non- core activities.
Professional training & development	Mandatory training. Professional development relating to the role in line

	with job description and NHS study leave policy & agreement.
Non-core functions	Definition
Bereavement support	Undertaken after initial core visit as described in core activity of EB & DeBRA organisation
Charitable functions /activities	Raise awareness in the public arena, at schools, places of work, general audience. e.g. dermatology conferences, charitable events pharmaceutical meetings etc.
Research	The NHS England does not fund research activity and therefore this is a non core item. To be agreed in SLA with DeBRA
Product development/ evaluation	The NHS England does not fund research activity and therefore this is a non-core item. To be agreed in SLA with DeBRA
Publications	Non-core – publications, articles, research projects, product development write ups, development of guidelines for products & research and national & international best practice guidelines.
Teaching wider (as in core)	Conferences Teaching in wider circles – public arena.
Management relating to non core activities	Service monitoring & audit for non-core activities. This relates to the lead nurse responsible for the management of the wider EB team.

Sub-Contractual Arrangements with DebRA

The NHS England must be made aware of any sub-contractual arrangements between the provider and DebRA, if it has a material impact on the delivery of the EB service.

The NHS England is not involved in negotiating or mediating any contractual arrangement between the provider and DebRA.

The EB nursing principles will be included as part of the service specification which will form part of the contract.

These principles will be reviewed alongside the review of the service specification.

EB nurses in designated centres must ensure activity is recorded and reported as required.

Discharge criteria

Some patients may be discharged from the service when:

- they are able to self-manage with support from local NHS providers. In these circumstances the service will liaise with local services to provide advice on management and transfer of clinical information. Access to the national service for support and advice during difficult episodes will be clearly communicated to the patient and local care providers. OR
- they are a competent adult patient who chooses to disengage with the service

Other discharge conditions are :

- repeated non-attendance at more than two consecutive clinic appointments at the EB centre's discretion – notification in writing and by telephone to patient, GP and referrer. Patients not seen annually must be discharged back to the GP unless there is a medical or social reason for their having delayed appointments to beyond one year.
- patients can transfer their care to another national centre at any point in their care. For repeated non attendance by paediatric patients, providers should instigate the local safeguarding policy to ensure patients are receiving support and care as appropriate. The EB should work with the GP and/or health visitor to ascertain whether non-attendance is indicative of a child protection issue such as non-compliance by a parent/guardian. Paediatric patients should only be discharged when there is a clear indication that care is no longer required, or is being given elsewhere and discussions have been completed with parents/ legal guardians and the patients GP.

Transition to adult service

Transition arrangements should be in place across both adult and paediatric providers. An audit or survey of the experiences of patients who have transitioned should be undertaken. Transition of adolescent patients should begin around the age of 12 -14 years. Patients should transition to the adult service by the age of 18 years. All patients should undertake an EB Quality of Life questionnaire assessing the impact of different aspects of disease on quality of life at the time of transition.

2.3 Population covered

This service is commissioned to provide access to patients from England and Scotland. If people with EB from Northern Ireland and Wales attend designated centres in England, separate commissioning arrangements should be in place and agreed. Centres are still required to report all activity undertaken in the service.

2.4 Any acceptance and exclusion criteria

Accessibility/acceptability

The service is accessible to all patients with suspected EB regardless of sex, race or gender. Providers require staff to attend mandatory training on equality and diversity and the facilities provided to offer appropriate disabled access for patients, family and carers. When required the providers will use translators and ensure information is available in appropriate languages.

The provider has a duty to co-operate with the commissioner in undertaking Equality Impact Assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation.

Referral criteria

Referrals for clinical services will be accepted from general practitioners, dermatologists, paediatricians, neonatologists or any other clinician who believes their patient may have a form of EB or other genetic skin fragility disorder. Referrals may also come directly from the EB CNSs who may have seen EB patients in the community. Referrals are triaged by a consultant dermatologist or paediatrician.

No tests need to have been carried out before referral; indeed, skin biopsy should be positively discouraged by local teams and instead performed by the EB centre if indicated. If there is clinical doubt about the diagnosis from the secondary care referrer, it may be helpful to send photographs via email for the EB centre to review before referral.

Referral to the lab for diagnosis

Prior to sending samples, it is essential to contact the EB lab. Prenatal diagnosis does not form part of the designation, so it is important to contact the lab well in advance to check availability for these tests and ensure funding streams are identified. This should be agreed with the patients' referring primary commissioners.

Once the lab agrees to accept the sample:

For diagnostic skin biopsies, instructions regarding suitable specimens will be sent out, together with specialised fixative and transport solutions.

The EB lab is designated to support diagnosis for patients resident in England. In many cases, particularly in neonates, a skin biopsy is necessary so that subsequent molecular testing can be directed toward the appropriate gene(s). In older children and adults, however, clinical review may be adequate to diagnose the form of EB clinically, in which case molecular testing of the appropriate gene can be done without recourse to skin biopsy. Generally, a clinical diagnosis of a specific EB subtype sufficiently firm to proceed to molecular testing should come from one of the 4 EB centres. Occasionally, a clinician from outside these centres may be sufficiently experienced in EB to be confident in making a clinical diagnosis of EB subtype such

that molecular testing can be undertaken. In other instances, review of emailed photographs from a referring clinician may help decide whether skin biopsy is necessary or whether it is possible to proceed straight to molecular testing.

For EB cases originating in Ireland or overseas, the first contact should be made directly with the EB lab.

Cases covered by the NHS England service for EB are directly funded. No charge is made to the referring NHS trust – this does not include pre-implantation genetic diagnosis (PGD) where prior agreement for funding must be received from the patient's PCT. There is a special NHS agreement for Scotland. A charge is made for all other cases, including private patients - please contact the EB lab for details.

Routes

It is envisaged that patients will be referred from multiple sources but primarily from dermatologists and from paediatricians, including neonatologists.

Referrals will be accepted from any clinician with clinical suspicion that their patient may have EB in consultation with the EB lab and/or a clinical EB centre.

Exclusion criteria

Patients referred who are found not to suffer from EB will be discharged back to the referring team, for example, infants with staphylococcal scalded skin syndrome or incontinentia pigmenti, immunobullous diseases, etc.

Acquired EB, a very rare auto-immune condition arising in later life, is excluded from this service.

Response time, detail and prioritisation

The providers of the EB service will aim to see:

- new babies within 48 hours
- non-urgent new patients within 18 weeks unless delayed appointment requested by patient or family
- urgent new patients within 2 weeks

2.5 Interdependencies with other services

Shared care

Some 'mild' and 'severe' patients' medical care can be jointly provided with their local teams. Individualised shared care plans for patients should be developed and agreed with the EB team at all national centres. It is the responsibility of the nationally designated EB providers to provide education within the NHS to raise and maintain awareness of EB and its management. This will also support sustaining the service in the wider NHS.

Continuing care

Continuing Care will in some cases be provided by local primary and secondary care teams, though most will require longer-term follow-up at the national centre which will normally take the form, primarily, of visits to the EB clinic.

The national providers will form a relationship with local health and social care providers to help optimise any care for EB provided locally for the patient. This may include liaison with consultants, GPs, community nurses or social workers etc.

DebRA

The patient support group should not fund any part of the core nationally designated service. Support from DebRA should be delivered in the form of the non-core activities e.g. funding nurses to attend conferences and training as outlined in the nursing principles.

Sub-contractors

A sub-contractual arrangement exists between Guys & St Thomas' NHS Foundation Trust and GSTS Laboratory that provides lab support to all 4 providers.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

The providers of the national EB service must ensure they are fully integrated into their trust's corporate and clinical governance arrangements and must comply fully with Care Quality Commission (CQC) requirements in terms of quality and governance. The hub centres are responsible for overseeing the governance arrangement of any spoke clinic provided under sub-contractual arrangements.

Each centre will ensure that:

- regular meetings take place with patient representatives;
- all practitioners participate in continuous professional development and networking;
- patient outcome data is recorded and audited across the service.

The commissioners and service will conduct a formal Joint Service Review at least every six months. All centres must participate in service related national audit - audit meetings should address:

- clinical performance and outcome
- process-related indicators, e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow up etc.
- stakeholder satisfaction levels, including feedback from patients, their families, referring clinician and General Practitioners.

4. Key Service Outcomes

Quality Performance Indicator	Threshold	Method of measurement	Consequence of breach	Report Due
QoL questionnaire to be undertaken at transition				

5. Location of Provider Premises

Birmingham Children's Hospital NHS Foundation Trust
Great Ormond Street Hospital for Children NHS Foundation Trust
Guys & St Thomas' Hospital NHS Foundation Trust
Heart of England NHS Foundation Trust

Adopted