

Summary of needs not currently met for people with EB

INTEGRATION PROBLEMS ON A SOCIAL LEVEL, IN THE WORKPLACE AND IN SCHOOLS

From a social perspective it is the case, that sometimes the behaviour and the eyes of others hurt more than the open wounds. Every day families face uncomfortable stares and rejection. Parents of other children move their children away from children with EB because they fear it may be contagious. Children with EB are often denied access to public swimming pools for the very same reason. Using public transport can be very upsetting when people move seats to be further away from you.

Schools are often unable to implement support systems so that children with EB can receive the right education.

Lack of support in the work place can also mean it is very difficult for people with EB to integrate at work.

REFERENCE CENTERS

Our aim is that all patients with EB can be looked after in Specialist Reference Centres. These health care units have specialist medical staff to ensure that patients receive the correct care and advice. In order to make this happen, the current process for the official designation of the hospitals as reference centres needs to be completed and the Hospital Val d'Hebron in Barcelona (where hand surgery takes place) needs to be included within the plan.

BANDAGING MATERIALS AND OTHER ESSENTIAL PRODUCTS REQUIRED FOR THE TREATMENT OF EB:

Amongst the different essential treatments required for people with EB are the medical products (dressings, bandages, etc.), cosmetic products (specialist creams), prosthetic products and dietary supplements (liquid food).

Specialist bandaging material for the treatments, which are carried out daily or on alternate days need to be supplied in variable quantities and must be of a specific type. In most cases they cannot be substituted for other brands or generic options. Importantly they must be non adhesive, flexible, they must be long lasting (to avoid increasing the frequency of the treatments as these are very painful), they must help to reduce pain levels, be made from breathable materials that combat infection and bacteria, aid healing and help to minimize unpleasant odours, etc.

Nutritional supplements are often very important for people with EB. The open wounds and the body's response to healing, means that it is hard to get the necessary calorific intake and the right nutrients to aid this process. Many people with EB also suffer from gastro-intestinal complications like the narrowing of the oesophagus and dysphagia which make eating extremely difficult. This means that most people with EB require high calorie, high protein liquid foods that will provide the necessary nutrients to aid in the healing of wounds, to help fight infection, aid growth, sexual development and improve quality of life.

Prosthetic products like the splints for the retraction of the fingers are of vital importance both before the appearance of any complications and after this. They are also essential after any operations. Wheel chairs are imperative tools for the day to day of people with EB. There are times when

walking is too painful, but in some cases wheelchairs become a permanent extension of the person as deterioration progresses. These help them to maintain a level of independence in the home and at school or in the workplace.

Amongst other products, hydration and protective creams are so important to maintain the healthy areas of the skin. Specific creams are imperative to ensure hydration, elasticity and the flexibility of the skin in order to prevent the appearance of new wounds. Good hydration also helps to minimize skin irritation and with this break the cycle of irritation, scratching, open wounds, healing, and irritation.... Specialist non-irritant shampoos and soaps are essential as other products can be damaging to the skin.

On the 29th July 2015 the Ministry of health approved the accord that led to the agreement that the National Health Service in the 18 autonomous regions would cover the cost for treatment of people with EB.

In spite of this there are still many issues within the different autonomous regions and as a consequence there are still families who find it difficult to access treatments.

HOME VISITS

People with EB need specific care and treatment, which must be carried out by specialist nurses in the home of the patient. The reasons for this are:

The different types of wounds can be severe, chronic, infected, malignant, of different depth and in different places, they can ooze, be painful and exude smells...All these factors mean that patients require supervision from medical health care providers. The treatment of the wounds require specialist knowledge and understanding to recognize signs

of infection and to make sure the correct products are used for each type of wound. These wounds change and need to be monitored so that complications can be detected early and treatment prescribed quickly and accurately. Professional nurses must be involved in order to care for a person with EB.

Parents and principal carers do not normally have the experience and knowledge to deal with this level of care.

If parents or family members are acting as carers this can really affect their quality of life and can have a negative impact on relationships..

EB is a chronic disease so treatment can take between 1 and 4 hours a day making this extremely difficult to combine with family or working life.

Treatment should not take place in local surgeries because in the majority of cases they do not have bathing facilities and because of the high risk of infection. Treatment in the home environment is favourable and more comfortable for people with EB.

Despite the requirement to provide this service for people with EB this unfortunately does not happen in the majority of cases. In the instances where they do receive a level of home care it is often provided by inexperienced nursing staff. Local surgeries and hospitals are severely short staffed and under resourced and they often come unprepared and unqualified to treat patients with EB.

REHABILITATION

EB brings with it a number of severe complications from musculoskeletal issues (like the retraction of the fingers and articular problems, syndactyly, stiffness), nutritional complications like microstomia, tongue tie, difficulty swallowing, narrowing of the oesophagus, motor complications and speech difficulties.

Physiotherapists, occupational therapists and speech therapists are essential to monitor rehabilitation.

This is so important to help slow the progression of the disease. Because of the evolutionary nature of the condition it is of vital importance to start with treatments from birth.

However, rehabilitation in Spain is currently only offered after surgery or when deemed necessary and is not offered as a preventative measure which is imperative for chronic diseases like EB.

GENETIC TESTING

Genetic testing is important for people with EB so that the type of mutation can be isolated. This is fundamental to determine the prognosis for each individual and is imperative for family planning. Genetic testing also means that patients can take part in clinical trials.

Currently the team in charge of genetic testing in Spain is CIEMAT, they belong to CIBER, which is the rare diseases research organization at the Carlos III health institute. However, the testing is only available for research purposes and is subject to public or private finance for this use only. Genetic testing is therefore not available for people with EB through the national health services and as a consequence the DEBRA Butterfly Children's charity is currently financing

the salary for the geneticist responsible for the genetic testing of people with EB.

PRENATAL TESTING AND PRE-IMPLANTATION DIAGNOSIS

This testing which also gives families a choice of whether or not to have further children with or without the disease is not currently offered through the National Health Service.

ECONOMIC HELP FOR CARERS OF MINORS WITH CANCER OR OTHER RARE DISEASES

The royal decree of the 29th July 1148/2011 for the development of a financial care system for people affected by cancer or other serious illnesses includes a list of 109 disorders.

Despite the fact that Epidermolysis Bullosa is a chronic illness that seriously affects the quality of life of patients it is not included on this list.

ACCESS TO DISABILITY AND DEPENDENCY ALLOWANCES

Because of the lack of awareness and understanding surrounding the complications and the symptoms of the disease, families with EB are often denied access to the allowances that are so important to help their quality of life.

PSYCHOLOGICAL SUPPORT

Support and counselling from psychologists and psychiatrists is currently not available to people with EB

through the National Health Service despite the importance of this.

DENTISTRY

One of the many serious complications from EB is the oral health issues that affect swallowing and speech. Despite these issues the National Health Service does not cover dental healthcare for patients with EB.

TRAINING FOR HEALTH CARE PROFESSIONALS AND TEACHERS

In order to promote the correct care for people with EB, training must be put in place to help professionals provide the right care and support. This includes the production of specific guidelines and written documents to support them.