Quand et comment décider d'une biopsie chez l'enfant

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PNDS, Guidelines, EB-Burden

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« Plan Maladies Rares » in France since 2004

3 clinical expert centres for EB patients (CRMR)

**Paris** (MAGEC: coordination C Bodemer)
- Necker-EM (C Bodemer)
- Saint-Louis (EBourrat (C Blanchet Bardon))
- Avicenne

**Bordeaux-Toulouse**
- (coordination A Taieb)
- Toulouse (J Mazeereuw)

**Nice**
- (coordination JP Lacour)
Expert centres:
Responsible of care and clinical research

- Phenotype/genotype
- Clinical research
- Database and cohorts
- Translational therapies
- Therapeutic education
- School Integration
- Social Integration
- Epidemiology
- National protocol care
- ...
« Plan Maladies Rares » in France since 2004

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6 regional competence centres
National Protocol care
PNDS
National Protocol Care

Health Ministry: “the goal of PNDS is to explain diagnostic and therapeutic problems and the course of care for patients with a particular rare disease. It aims to optimize and harmonize the management and monitoring of rare disease throughout the country.

The PNDS can serve as reference to the regular doctor with the specialist, notably when determining the treatment protocol, in conjunction with the doctor council of Health Insurance and the patient, in the case of a demand of exemption from the patient’s contribution, in conformance with a disease except list.
EB National Protocol Care

The PNDS is thus essential

*to list all the needs of EB patients
-medical needs
-paramedical needs (ex. nursing time for dressing at home) and - no medical needs (ex arrangement of environment at home

*to make them recognize by the french health authorities

*to try to improve the care at home of each patient and thus the burden of patients and their families
EB patients: multidisciplinary medical management

Dermatology
Kinesitherapy
Functional re-education
Nutrition
Dietetician
Psychologist
ORL
Ophthalmologist
Dentists
Surgeons
Genetic counselling
Anatomopathologists
Social Workers ...

PNDS: multidisciplinary very hard work
EB National Protocol Care

- **Co-coordination:** Prs JP Lacour (Nice) et C Bodemer (Paris)

- **Pilot Group (PG)** with clinical EB experts of each CR
  E Bourrat, E Bourdon-Lanoy, C Bodemer (MAGEC, Paris)
  C Chiaverini et JP Lacour (Nice), J Mazereeuw (Toulouse)

- **Working groups (WG) with specialists involved in** the clinical (medical and paramedical) multidisciplinary team, for EB monitoring (Paris and Nice CR)

- **Expertise of DEBRA-France**
EB National Protocol Care

- Methods

Step 1: The review of all guidelines under the expertise of WG

Step 2: A systematic analysis of the literature data WG

Step 3: Redaction of a first document including 16 chapters covering all the fields (medical, paramedical, educational and socioeconomic fields) indispensable to the best monitoring of EB patients (document 1) PilotGroup

Step 4: Correction and criticism of the first document after a second reading by 4 experts (C Bodemer, E Bourrat, C Chiaverini, JP Lacour) (document 2)

Step 5: Synthesis and pagination of the document 2, with synthetic tables (document 3) ChB and JPL (in progress)

Step 6: Review of the document 3 by DEBRA-France and final document (document 4) (expected at last on november)
The PNDS will be essential

*to list *all the justified needs* of EB patents

-medicinal needs

-paramedical needs (ex. nursing time for dressing at home) and - no medical needs (ex arrangement of environment at home

*to make them recognize by the french health authorities

*to try *to improve* the care at home of each patient and thus the burden of patients and their families
Individual EB Burden
Societal Burden and Individual Burden

- **WHO**: notion of **Societal Burden** to quantify the health of a population and to determine the priorities of action in public health domain

- **Individual Burden**: disability (e.g. health-related Quality of Life [HRQoL], social integration, home-life, and use of medical resources (including consultations/medications) in the broadest sense of the term (psychological, social, economic and physical).

Essential to sensibilize the population and the authorities to the under-estimated disability of EB patients and their families
Individual EB-Burden score

Research Program: MAGEC-Necker

The Epidermolysis Bullosa Burden Scale (EB-BS): Development and Validation of a Tool

H Dufresne\textsuperscript{1,2}, S Hadj-Rabia\textsuperscript{1,2}, I Corset\textsuperscript{1,2}, S Chaumon\textsuperscript{2}, C Taieb\textsuperscript{3}, C Bodemer\textsuperscript{1,2}

Annual congress of Dermatology CARD 2014, under publication

**Aim of the study:** to permit the evaluation of an individual and familial burden score in EB patients (EB-BS), calculated on the basis of a specific burden questionnaire (EB-BQ) approved and designed to understand the tendency to changes in EB care and lifestyles.
Individual EB-Burden score
Research Program: MAGEC-Necker
The Epidermolysis Bullosa Burden Scale (EB-BS): Development and Validation of a Tool (EB-BQ)

**Method:** development of a self-administered EB Burden Questionnaire (EB-BQ) using standard methodology (3 phases: exploratory, development, and validation (psychometric analysis).

To ensure clinical and scientific rigor:

development by a multidisciplinary team: comprising experts in questionnaire design/development, experts in the management and care of patients with EB and QoL expert (CNIL authorization number 1690350).
Individual EB-Burden score

Research Program: MAGEC-Necker

The Epidermolysis Bullosa Burden Scale (EB-BS):
Development and Validation of a Tool (EB-BQ)

Method: 60 families were included and take part in the study

(Thank you very much!)

Results: A self administrated specific individual EB-BQ was validated leading to quantifiable items covering 4 dimensions:

― “Family life”, “Child's life”, “Disease and treatment”
― “Economic and social impact”.

The specific EB-BS is the sum of all individual items: 0-100
Individual EB-Burden score

Research Program: MAGEC-Necker

The Epidermolysis Bullosa Burden Scale (EB-BS): Development and Validation of a Tool (EB-BQ)

Interest of the EB-BQ

-Self questionnaire:

- Not limited by time or by costs (travel…)
- Possible to work on large scale
- Minimizes effects relating to interviewers personality
- Responses not biased due to embarrassment (shame)

ocuring in difficult family social and economic circumstances
Individual EB-Burden score

Research Program: MAGEC-Necker

The Epidermolysis Bullosa Burden Scale (EB-BS): Development and Validation of a Tool (EB-BQ)

Interest of the EB-BQ

- Self questionnaire
- EB-BS sensitive to be able to assess the impact of medical and non-medical management
- Permits a social and economic dimension and an indication of unmet needs for families at individual level, which would seem to increase societal burden.
Individual EB-Burden score

Research Program: MAGEC-Necker

The Epidermolysis Bullosa Burden Scale (EB-BS):
Development and Validation of a Tool (EB-BQ)

Interest of the EB-BQ

Translation and cross-cultural adaptation

The original French version of EB-BQ has been translated and undergone linguistic and cultural adaptation in English (US).
Individual EB-Burden score

Clinical Research program: FIMARAD-Radico

**FIMARAD**: French national network for all the rare skin diseases (Filière Maladies Rares Dermatologiques)

*Certification: February 2014  Coordinator: C Bodemer*

**RaDiCo** (rare diseases cohort) Program for the development of large cohorts, in the field of rare diseases.

Supported by the Ministry of the Research

Submission of the project: FIMARAD-BS 2013

Project selected by an international committee (July 2014)
Individual EB-Burden score

Clinical Research program: FIMARAD-Radico

Study design:
- Long term follow-up of large descriptive cohort involving the French reference centres and their network (FIMARAD) (with a shared database)

Objectives:
To evaluate the individual Burden of genodermatoses, in the aim of reducing it, through medical and non medical strategies

Methods: 2 indicators of burden: 1-individual BS calculated on the basis of BQ; 2-descriptive analysis of all resources
Individual EB-Burden score
Research Perspectives (for EB families)

-The change in EB-BS, but also in various items of the BQ, and in the resources used by each family, as observed through monitoring of the cohort, will be compared with the medical and non-medical strategies

-A cross-cultural evaluation of the individual burden score in different countries will be realized

-A descriptive analysis of the cost of EB for patients and their families

-The development of ancillary studies from a cohort clearly defined in terms of diagnosis and monitoring
A Teamwork
Motivation
Interaction
Brainstorming
Respect
Friendship