Experienta Centrului de Referinta Colentina. 5 ani de activitate

National Conference RDD 2018

1 Martie 2018

Alina Suru, Carmen Maria Sălăvăstru, George-Sorin Ţiplica

Puncte de discutie

- Program National Boli Rare
- Centru Referinta
- Cercetare

Programul National de Boli Rare

2013 Clinica Dermatologie II & CNAS: Programul Național pentru Boli Rare în cadrul Spitalul Clinic Colentina.

- Pacientii pediatrici: Compartimentul Clinic de Dermatologie Copii
- Pacientii adulți: Clinica II Dermatologie.

Epidermoliza buloasa

- Un grup de afectiuni congenitale caracterizat de fragilitate a tegumentelor si mucoaselor, ce se manifesta prin aparitia de bule si eroziuni, fie spontan, fie dupa traumatisme minore.

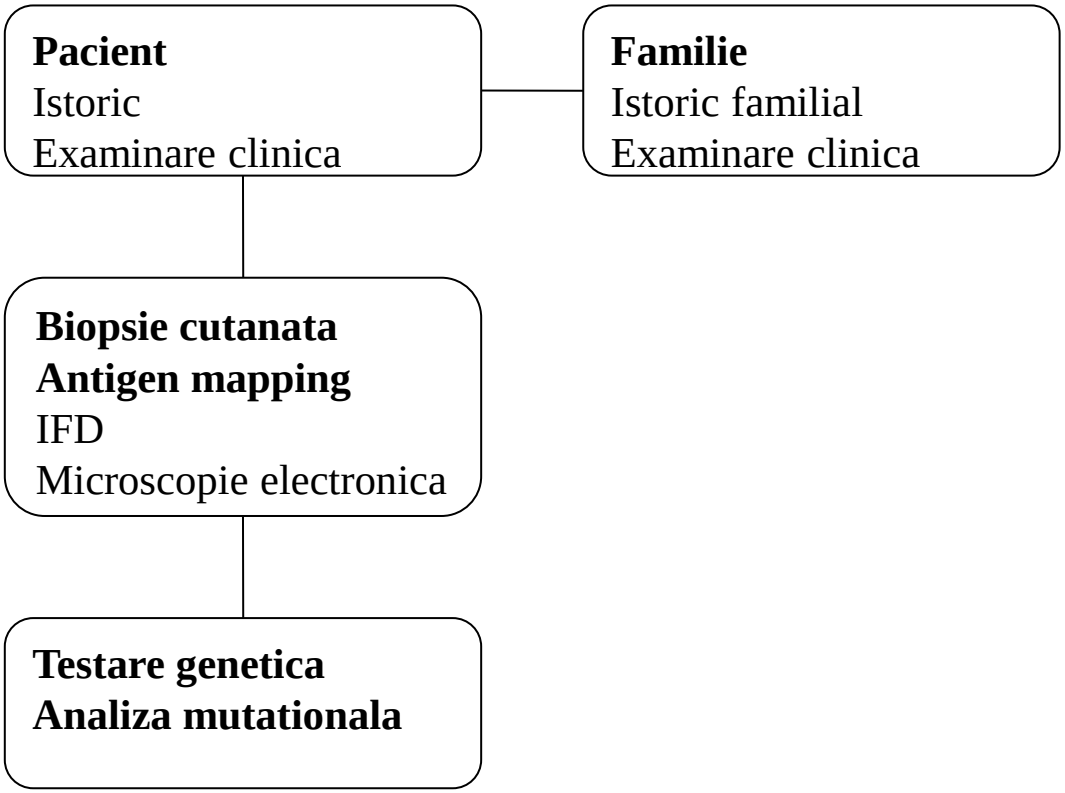
- **Clasificare-4 grupuri majore**:

1. **EB simplex (EBS)**- K5, K14, PLEC
2. **EB distrofica (EBD**)- COL7A1
3. **EB jonctionala (EBJ)-**LAMB3, COL7A1, LAMC2, LAMA3
4. **Sindrom Kindler-**FERMT1, gena pt kindlin 1

Programul National de Boli Rare

- 28 de pacienți EB, 25 monitorizati
- 18 dintre pacienți EB distrofica
- 5 EB simplex
- 1 pacient cu EB dobândită
- 1 pacient EB jonctionala (născut în decembrie 2017-al doilea din această categorie născut în doi ani)

Diagnostic


Pacient
Istoric
Examinare clinica
Familie
Istoric familial
Examinare clinica
Biopsie cutanata
Antigen mapping
IFD
Microscopie electronica
Testare genetica
Analiza mutationala

Management EB-PN

1. Ingrijirea leziunilor buloase si erozive
2. Controlul infectiilor
3. Prevenirea complicatiilor
4. Ameliorarea simptomelor (prurit si durere)

- Materiale sanitare, medicamente, pansamente
- Educarea pacientilor/familiilor

MÆDICA - a Journal of Clinical Medicine

EDITORIALS

MAEDICA – a Journal of Clinical Medicine
2013; 8(2): 200-205

Recommended Strategies for Epidermolysis Bullosa Management in Romania

Carmen Maria SALAVASTRU[a]; Eli SPRECHER[b]; Mihaela PANDURU[c]; Johann BAUER[d]; Caius Silviu SOLOVAN[e]; Virgil PATRASCU[f]; Horia Silviu MORARIU[g]; Anca TUDORACHE[h]; Torello LOTTI[i]; Irene TAGLIENTE[j]; Annalisa CIASULLI[k]; Maria Rosaria MARCHILI[l]; Giuseppe SABATINO[m]; Erika BURCIU[n]; Rodica COSGAREA[o]; Klaus FRITZ[p]; George-Sorin TIPLICA[a]

Echipa

- Prof. Dr. Carmen Maria Sălăvăstru
- Prof. Dr. George-Sorin Ţiplica
- As. sef. Erika Aleonte
- Doctoranzi: Alina Suru, Alexandra Butacu, Ionela Manole
- Medici rezidenti: Adelina Şendrea, Monica Popescu

Colaborare

- Institutul “Victor Babes”: Prof. Gherghiceanu (IFD, microscopie electronica)
- IOMC: pediatrie, neonatologie, genetica (dr. Plaiasu)

ERN-Skin


GENODERMATOSES & RARE SKIN DISORDERS
THE NETWORK ON RARE SKIN DISEASES FOR PROFESSIONALS AND PATIENTS
Art Contest 2018
2017 ERN-SKIN Course
2017 GENOD Course
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Rare Skin Disorders
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What is a ERN?
Missions
Diseases & Transversal approaches
Members
A pilot ERN for Rare and Undiagnosed Skin Disorders
Links
ERN-SKIN
Developing a European Reference Network for Rare and Undiagnosed Skin Diseases
Search
On June an application for the creation of a European Reference Network (ERN) in the field of dermatology was submitted to the European Commission (EC).
The ERN-Skin, it is 56 centres of expertise in the field of rare and complex skin diseases from 18 countries gathered in one network to bring highly-specialised, high-quality and accessible healthcare closer to the patients.
ERN-SKIN
56 EXPERT CENTRES
FROM 18 COUNTRIES
Sweden
Finland
United Kingdom
Denmark
Lithuania
Netherlands
Ireland
Poland
Czech Rep
Belgium
Austria
Hungary
France
Romania
Croatia
Spain
Italy
Germany
RESOURCES DIRECTORY ON RARE SKIN DISEASES
News
9 August 2017 – The 2017 Course on Genodermatoses
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ERN-Skin

34	Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico	Italy	Sophie Guez
35	Fondazione Luigi Maria Monti-Istituto Dermopatico dell'Immacolata (IDI) IRCCS	Italy	Biago Didona
36	Ospedale Pediatrico Bambino Gesù, IRCCS	Italy	May El-Hachem
37	Azienda Ospedaliera di Padova (AOP)	Italy	Anna Belloni Fortina
38	Azienda Ospedaliera Sant'Orsola Malpighi	Italy	Annalisa Patrizi
39	Hospital of Lithuanian University of Health Sciences Kauno Klinikos	Lithuania	Vesta Kucinskiene
40	AMC	Netherlands	Raoul Hennekam
41	Erasmus University medical Centre	Netherlands	Suzanne Pasmans
42	Maastricht University Medical Centre	Netherlands	Peter Steijlen
43	Academisch Ziekenhuis Groningen (UMCG) - Centre for Blistering Diseases	Netherlands	Marcel Jonkman
44	Heliodor Swiecicki Hospital, Poznan - Dept. Of Dermatology	Poland	Marian Dmochowski
45	Dermato-Pediatry Dpt., Bucharest	Romania	Carmen Maria Salavastru
46	Hospital Sant Joan de Déu and Hospital Clinic Group	Spain	Maria Asuncion Vicente Villa
47	Uppsala Genodermatoses Centre	Sweden	Marie Virtanen
48	Barts Health NHS Trust	UK	Edel O'Toole
49	Birmingham Children's Hospital NHS FT	UK	Celia Moss
50	Great Ormond Street Hospital Foundation Trust	UK	Veronica Kinsler
51	Guy's and St Thomas' NHS Foundation Trust - Dept Of Dermatology	UK	Jemima Mellerio
52	Guy's and St Thomas' NHS Foundation Trust - Dept of oral Dermatology	UK	Jane Setterfield

Cercetare

- Colaborare cercetatori de top din domeniul EB

Cercetare

- Studii clinice EB

-A Phase 3, Multi-center, Randomized, Double-Blind, Placebo Controlled Study of the Efficacy and Safety of SD-101 Cream in Patients with Epidermolysis Bullosa

-Double-blind, Randomised, Vehicle-controlled, Phase III, Efficacy and Safety Study with 24-month Open-label Follow-up of Oleogel-S10 in Patients with Inherited Epidermolysis Bullosa

Va multumesc!