



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

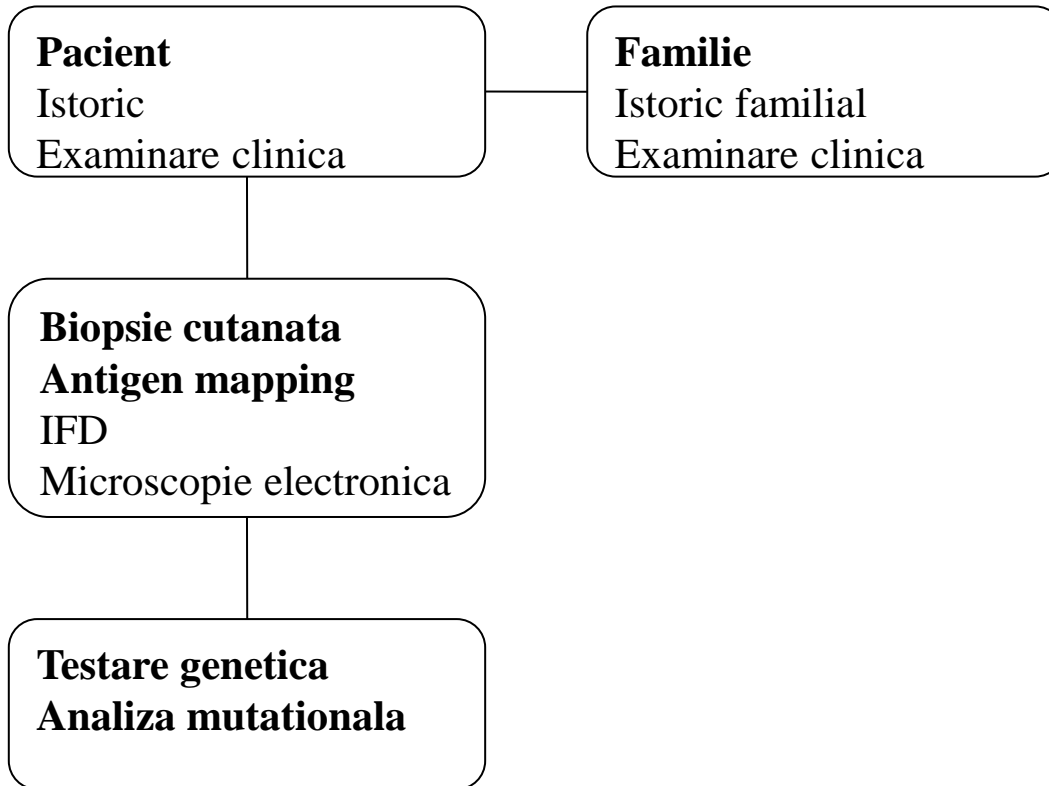
- Un grup de afectiuni congenitale caracterizat de fragilitate a tegumentelor si mucoaselor, ce se manifesta prin aparitia de bule si eroziuni, fie spontan, fie dupa traumatisme minore.
- **Clasificare-4 grupuri majore:**
 - 1. EB simplex (EBS)**- K5, K14, PLEC
 - 2. EB distrofica (EBD)**- COL7A1
 - 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





Management EB-PN

1. Ingrijirea leziunilor buloase si erozive
 - Materiale sanitare, medicamente, pansamente
2. Controlul infectiilor
 - Educarea pacientilor/familiilor
3. Prevenirea complicatiilor
4. Ameliorarea simptomelor (prurit si durere)

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 LOTTIⁱ; Irene TAGLIENTE^j; Annalisa
CIASULLI^k; Maria Rosaria MARCHILI^l; Giuseppe SABATINO^m; Erika BURCIUⁿ;
Rodica COSGAREA^o; Klaus FRITZ^p; George-Sorin TIPLICA^a

Echipa

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



Colaborare

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

ERN-Skin



The screenshot shows the ERN-Skin website. At the top left is a logo with a yellow star and a red and blue circle. The main header contains the text "DERMATOLOGY & RARE SKIN DISORDERS" and "THE NETWORK ON RARE SKIN DISEASES FOR PROFESSIONALS AND PATIENTS". Navigation links include "Art Contest 2018", "2017 ERN-SKIN Course", "2017 GENOD Course", "Our Network", "ERN-Skin", "E-Training", "Rare Skin Disorders", "Events", "Newsletters", and "Contact". A search bar is located on the right side of the header.

The main content area features a navigation menu with "ERN-Skin in brief", "What is a ERN?", "Missions", "Diseases & Transversal Approaches", and "Members". Below this is a sub-header "A pilot ERN for Rare and Undiagnosed Skin Disorders" with a "Links" button.

The central section is titled "ERN-SKIN" and includes the text "Developing a European Reference Network for Rare and Undiagnosed Skin Diseases". It features a navigation menu similar to the one above. The main text states: "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."

Below the text is a map of Europe titled "ERN-SKIN" and "56 EXPERT CENTRES FROM 18 COUNTRIES". The map shows various countries with red and blue circular markers indicating expert centres. Countries labeled include Sweden, Finland, Denmark, Lithuania, Poland, Czech Rep., Austria, Hungary, Romania, Croatia, Italy, Germany, France, Belgium, Netherlands, Ireland, Spain, and United Kingdom.

On the right side of the page, there is a "RESOURCES DIRECTORY ON RARE SKIN DISEASES" section, a "News" section with a link to "9 August 2017 - The 2017 Course on Genodermatoses", a "Follow us" section with social media links for Facebook (1.1m likes), LinkedIn (59 followers), and Twitter (@FRDermatology, 5663 followers), and a "Newsletter" section with a subscription form.

ERN-Skin

34	Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico	Italy	Sophie Guez
35	Fondazione Luigi Maria Monti-Istituto Dermatologico 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	Derma-to-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



E-Rare ERA-Net for Research Programmes on Rare Diseases

European Research Area

FUNDING & CALLS INFRASTRUCTURES SUPPORTED PROJECTS **ABOUT E-RARE** NEWS & MEDIA PUBLICATIONS EVENTS RELATED RD INITIATIVES

The Project

The successful linking of research funding organizations in E-Rare-1 and the subsequent exemplary joint funding activities have attested the need of, and the acknowledgment from, the research community for transnational funding of collaborative, multidisciplinary and ambitious projects on rare diseases. It has leveraged funding for rare disease research in countries without specific programmes for rare diseases and thus enabled the participation of researchers in these countries to transnational projects.

The current E-Rare-3 project will extend and strengthen the transnational cooperation on rare disease research funding organizations in the 5-year period of 2015-2019 by building on the experience and results of the previous ERA-Net programmes E-Rare-1 and E-Rare-2. It aims to provide an international model platform for implementing Joint Transnational Calls. The consortium comprises 25 institutions from 17 European, Associated and non-European countries. Its International dimension will be directly translated into close collaboration with IRDIRC and other relevant European and international Initiatives.

Extranet **The Project** Objectives Partners Organization Work Plan

10 years

Rare diseases are life threatening or chronically debilitating conditions from which not more than five affected persons per 10,000 citizens in the European Union (EU) suffer. It is estimated that 6000-8000 different rare diseases exist, affecting between 6% and 8% of the population in the course of their lives. This means that the total number of people affected by rare diseases in the EU is between 26 and 30 million. Most rare diseases are genetic diseases, the others being rare cancers, autoimmune diseases, congenital malformations, toxic and

Looking for collaborations Contact Interactive FAQ

If you are looking for collaborations E-Rare provides you with several search and communication tools.

Join

Log In

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!

