ICPerMed INTERNATIONAL CONSORTIUM

ICPerMed Workshop
Advancing Personalised Medicine Through Technology Development
November 14-15, 2023

Centre for Research in Genodermatosis and Epidermolysis Bullosa: finding answers to rare skin diseases

CONICET

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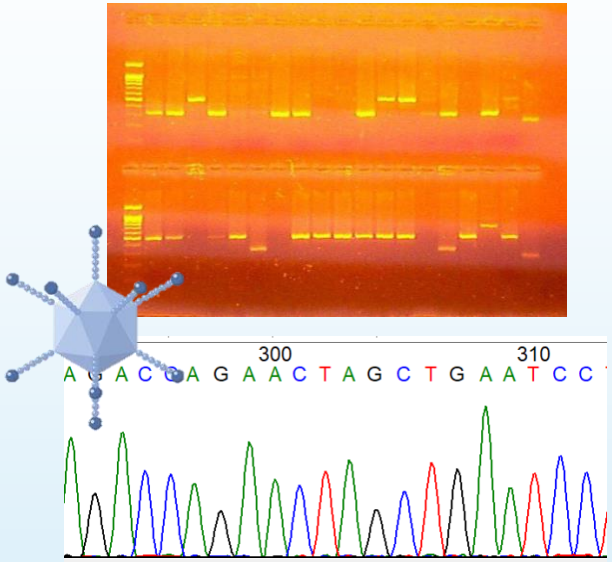
Genodermatosis

- Inherited skin disorders (>700)
- Most genodermatoses are rare diseases
- High clinical and genetic diversity (>500 genes)
- Symptomatic treatment
- Syndromic or non-syndromic

Our Journey - 2009



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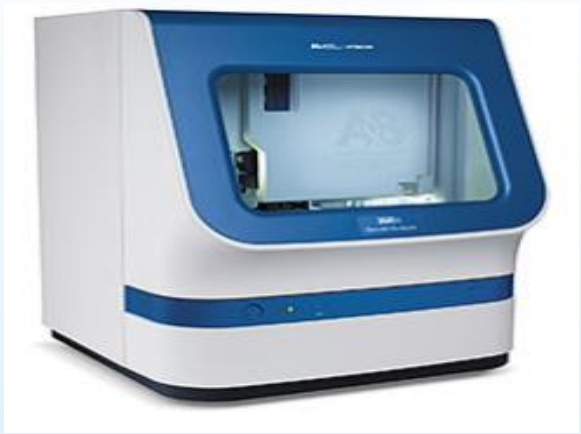

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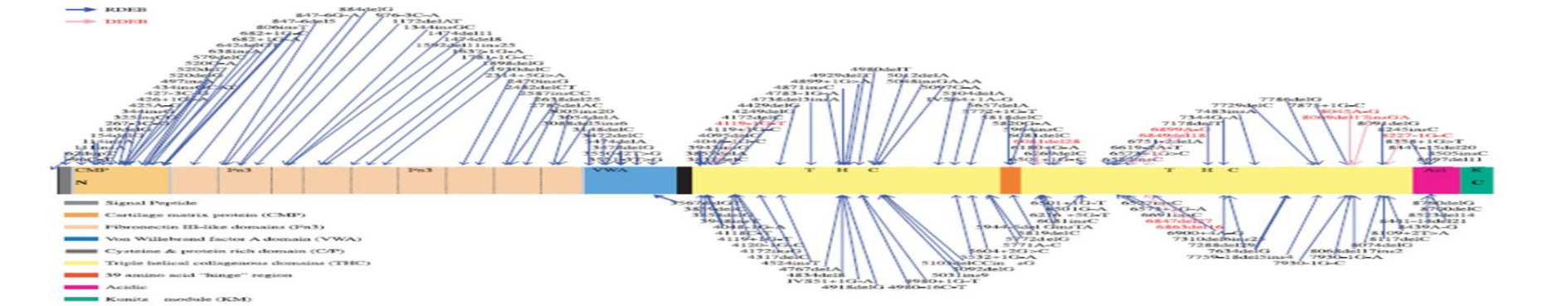
Dr. Graciela Manzur

- Dystrophic Epidermolysis Bullosa
- 2-12/1.000.000

Dystrophic Epidermolysis Bullosa
COL7A1 Sanger Sequencing (118 exons)



Dang et al Exp Dermatol. 2008 Jul;17(7):553-68

CHALLENGE: variants not reported in databases

ACTION: describe the genetic background in our region

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Our Journey - 2011

CHALLENGE: diagnosis of other genodermatosis

ACTION:

Research Project

- access to molecular diagnosis
- database mutational profiles of genodermatosis in our population

Our Journey - 2014

Foundation of the Centre for Research in Genodermatoses and Epidermolysis Bullosa, School of Medicine, University of Buenos Aires at the R. Gutiérrez Children's Hospital

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Our Journey - 2015

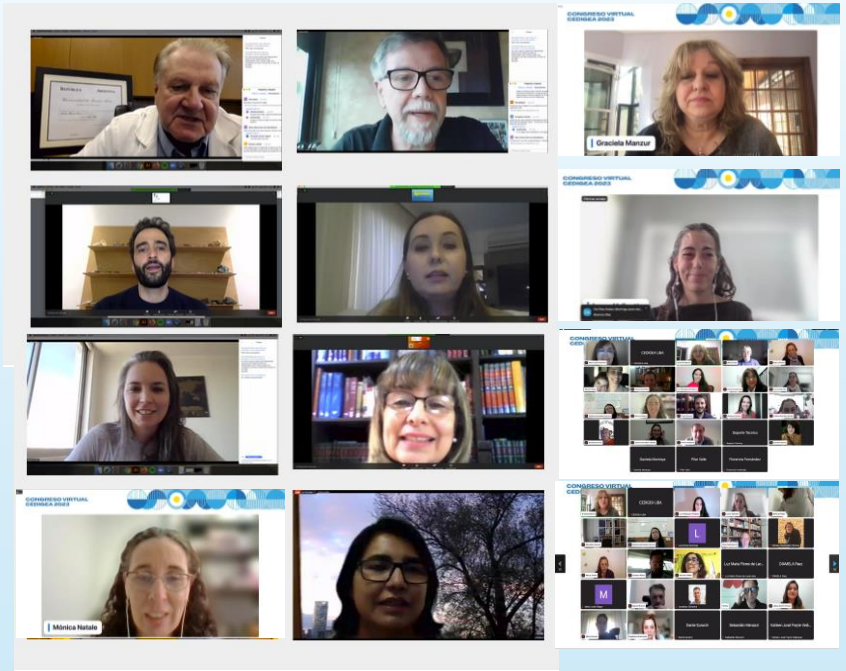
CHALLENGE: prevent long distance travel

ACTION: Genodermatosis Network and meetings

Our Journey - 2020

CHALLENGE: shorten the diagnostic odyssey

ACTION: virtual conferences



2,500 participants,
40 countries,
50 national and
15 international speakers

Our Journey - 2020

CHALLENGE: adult patients

ACTION: CEDIGEA at Hospital de Clínicas, UBA

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Dystrophic Epidermolysis Bullosa

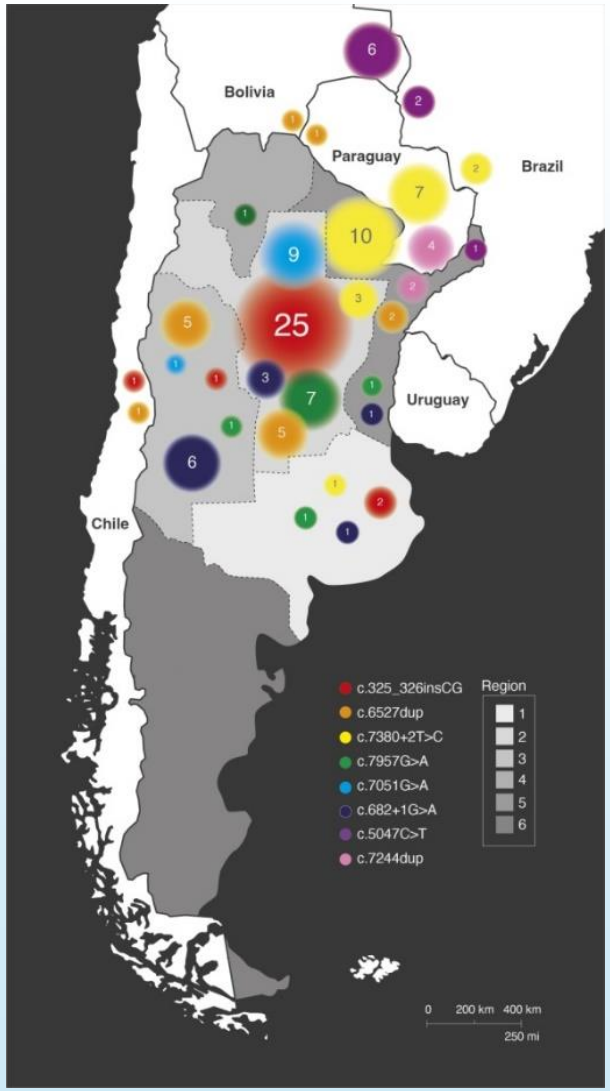
Received: 26 April 2022 | Revised: 22 June 2022 | Accepted: 1 August 2022
DOI: 10.1002/ajmg.a.62957

ORIGINAL ARTICLE

AMERICAN JOURNAL OF medical genetics PART A WILEY

Analysis of *COL7A1* pathogenic variants in a large cohort of dystrophic epidermolysis bullosa patients from Argentina reveals a new genotype–phenotype correlation

Mónica Inés Natale[1] | Graciela Beatriz Manzur[1,2,3] | Silvina Beatriz Lusso[1] | Eliana Cella[4] | María Elsa Giovo[5] | Romina Andrada[6] | Juana Goitia[7] | María Florencia Fernández[8] | Patricia Silvia Della Giovanna[9] | María José Guillamondegui[10] | Mariángeles Domínguez[11] | Olga Gutiérrez[12] | Agustín Izquierdo[13] | Heliana Hernández Herrera[1,3] | Luz Graciela Velázquez Perdomo[1,3] | Alicia Susana Mistchenko[1] | Laura Elena Valinotto[1,14]



- 181 patients
- 95 variants, 36 novel
- New phenotype-genotype association

Epidermolysis Bullosa Kindler

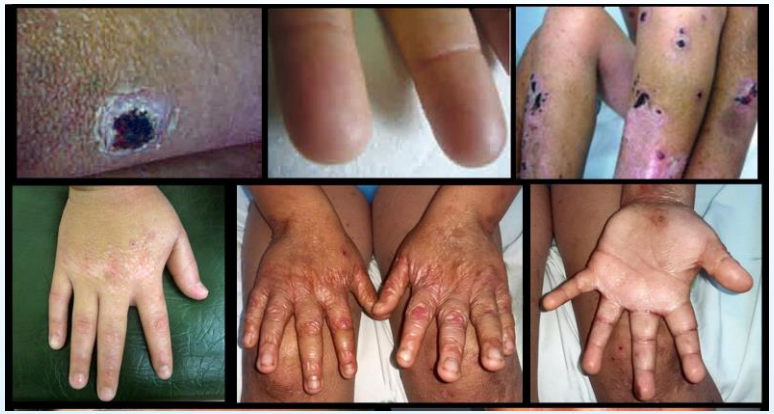
DOI: 10.1111/pde.14076

CASE REPORT

Pediatric Dermatology WILEY

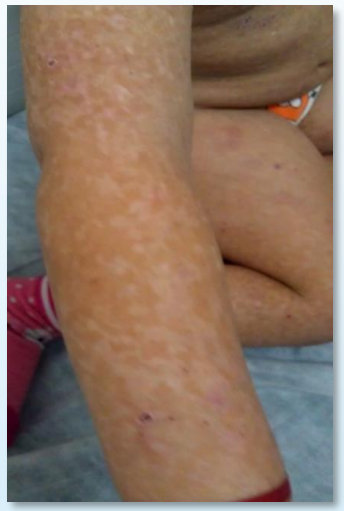
A novel pathogenic *FERMT1* variant in four families with Kindler syndrome in Argentina

Laura Elena Valinotto PhD[1,2] | Mónica Inés Natale MSc[1] | Silvina Beatriz Lusso MSc[1] | Eliana Cella MD[3] | Olga Gutiérrez MD[4] | Fernando Sebastiani MD[5] | Graciela Beatriz Manzur MD[1,6]



- 250 reported cases in the world
- *FERMT1, novel* c.450delG variant, homozygous

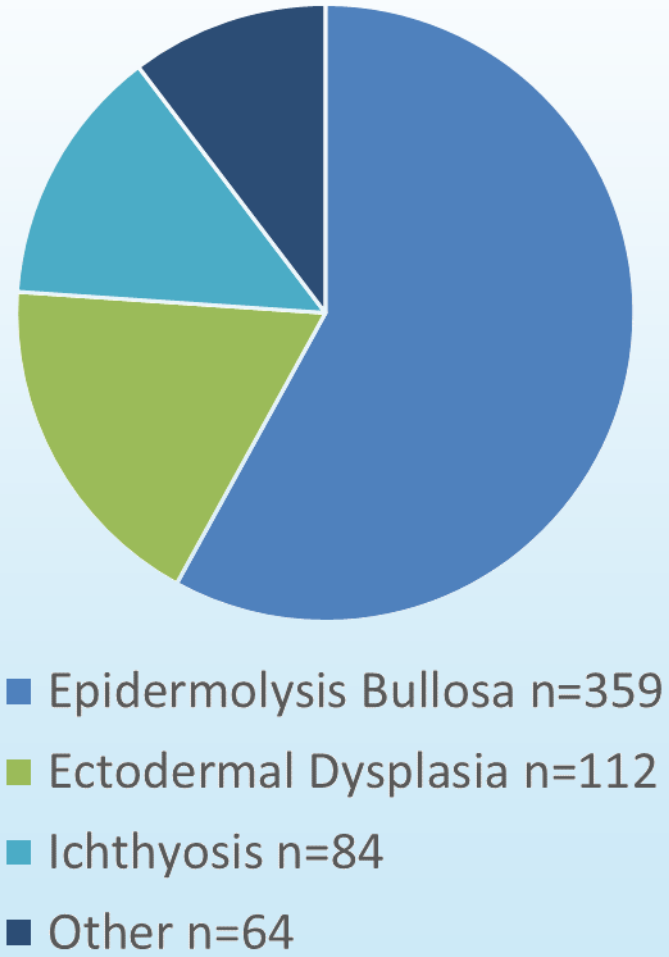
Epidermolysis Bullosa Simplex with mottled pigmentation COMING SOON....



- EBS 1-2/50.000
- EBS-MP <100 reported cases
- *KRT5* p.Pro25Leu 90-95%
- *KRT14* p.Met119Thr 2-5%
- 40 PATIENTS FROM 12 UNRELATED FAMILIES
- DEBRA UK

debra
THE BUTTERFLY SKIN CHARITY
Help stop the pain of EB

2009-2023



- 908 Patients, 619 molecular diagnosis (68%)

- 43 different genodermatosis, 53 genes
- 20% of the patients have novel variants
- Genodermatosis network in Argentina, Paraguay and Bolivia.

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Director: Graciela Manzur
DD: Alicia Mistchenko

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Thank you for your kind attention

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