Canadian Dermatology Association

Association canadienne de dermatologie

Unmet needs in Pediatric Dermatology

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Disclosures

- No financial disclosures
- I am a Pediatrician-Dermatologist
- From an academic center
- From a privileged country

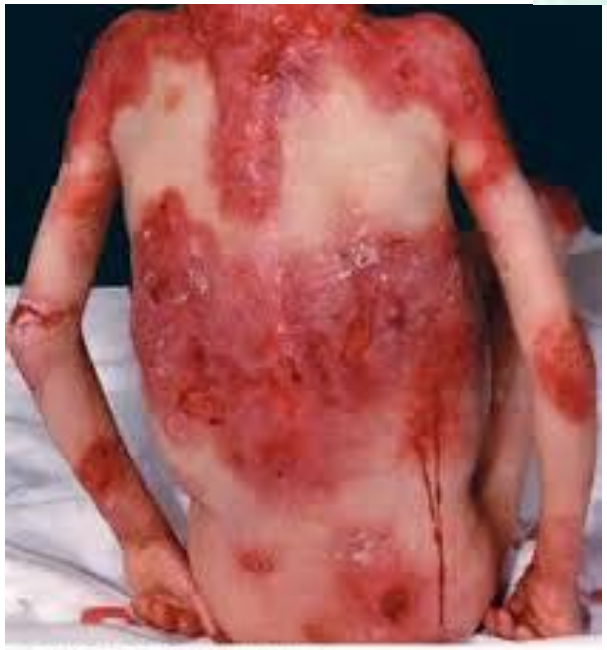
CHU Sainte-Justine
Mother and Child University Hospital Center

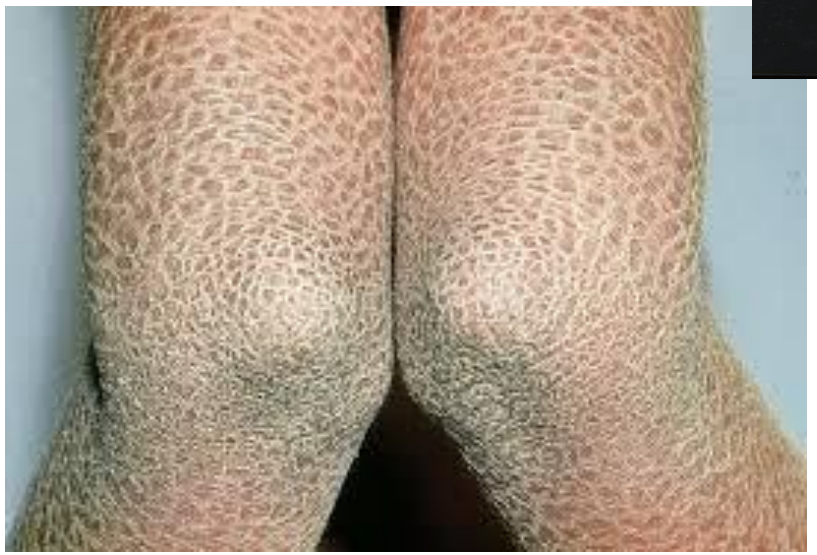
For the love of children

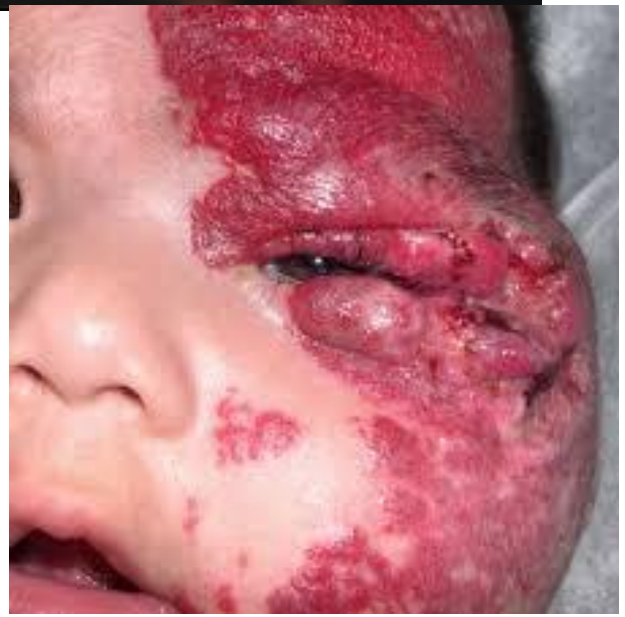
Université de Montréal

Rare or Orphan Diseases

- Definition varies
  - USA: <200,000 affected
  - EU : < 5/10,000 inhabitants
  - Japan : <50,000
- About 7000 rare diseases
- Many are genodermatoses, presenting during childhood (Epidermolysis bullosa, Ichthyoses…)

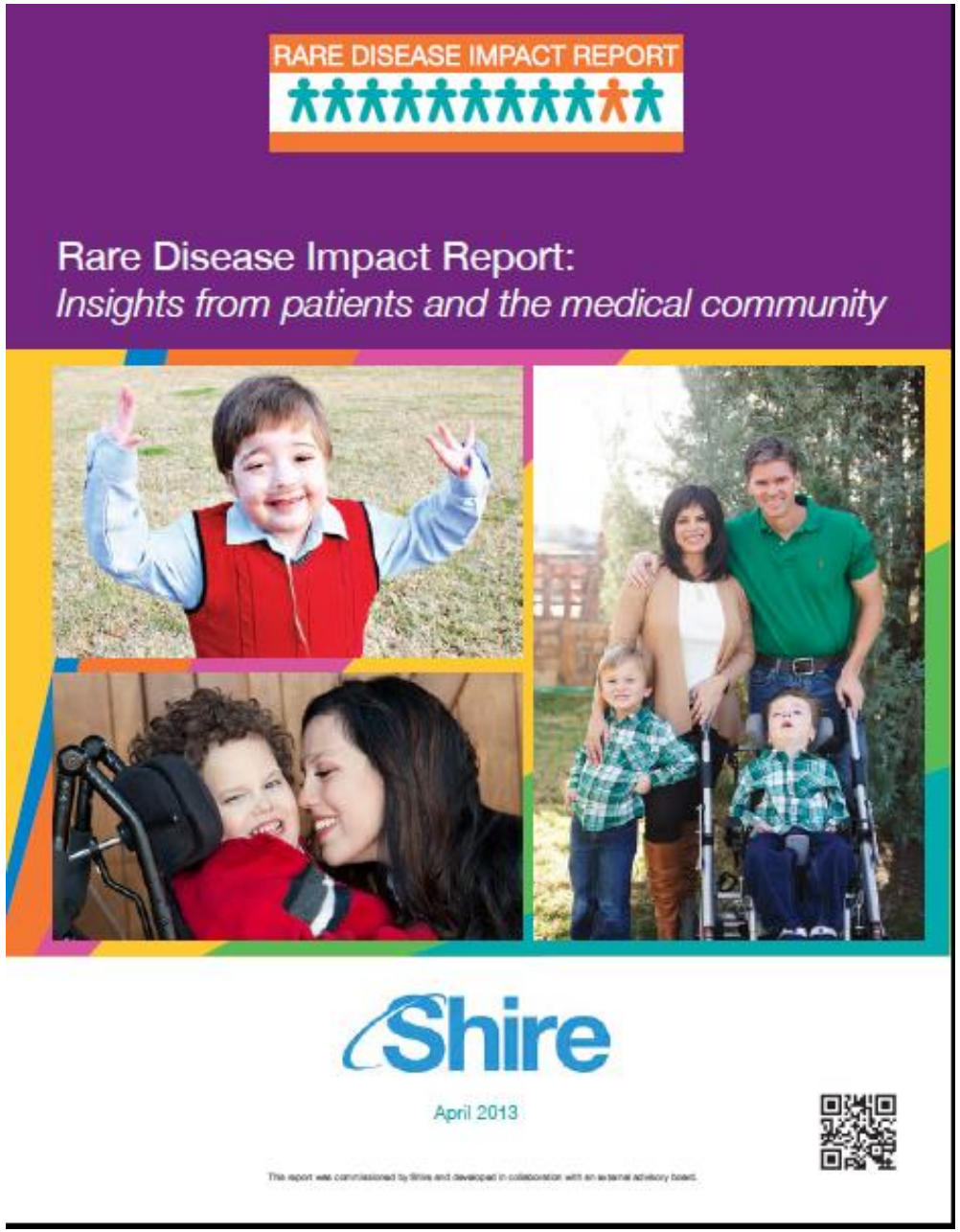






Challenges

- Small numbers of patients
- Complex histories and multisystem findings
- Longer clinical visits
- Average delay to diagnosis : 5 to 8 years
- Recent explosion in genetic knowledge
- Availability and cost of testing
- Lack of pediatric medication formulations/ official indications
- Transfer to adult care



A Leading Biotech Focused on Rare Diseases

THEME 1: There is a Lack of Resources and Information to Address Rare Diseases

Physicians (both primary care and specialists) have limited resources and information to properly diagnose/manage patients with rare diseases as compared to more common diseases seen.

PHYSICIANS

- Typical short office visit insufficient
- More office visits required for diagnosis
- Few opportunities to network with physicians treating rare diseases
- Difficulty in coordinating with other physicians involved

PATIENTS AND CAREGIVERS

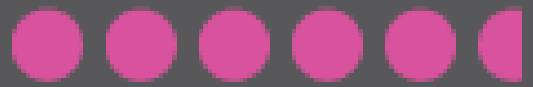
- Conflicting information about treatment options
- >60% needed to provide their healthcare professionals with info on their rare disease

Shire 2013 Rare disease impact report

Delays in Diagnosis

According to patients surveyed, it takes:

on average **7.6 years in the US**



on average **5.6 years in the UK**
for a patient with a rare disease to receive a proper diagnosis

According to patient/caregiver respondents, in order to get a proper diagnosis, a patient typically visits up to

8 physicians: 4 primary care and 4 specialists

and receives **2** to **3** misdiagnoses

Shire 2013 Rare disease impact report

THEME 2: The Economic Impact of Diagnosing and Managing Rare Diseases Is Significant

	US	UK
Had to use savings to pay for medical expenses	53%	31%
Not covered by insurance/National Health Service	55%	18%
Borrowed money from family/friends	37%	21%
Help from charity / public assistance	34%	18%
Negative impact on Credit score	32%	10%
Used retirement funds	23%	10%

Shire 2013 Rare disease impact report

Rare Diseases Take a Major Emotional Toll on Patients/Caregivers

	US	UK
Feelings of Depression	75%	69%
Anxiety/ Stress	86%	82%
Less interaction Friends / Family	70%	68%
Isolation	65%	57%
Worry about Future	90%	91%
Lack of available information	83%	81%

Shire 2013 Rare disease impact report

Strategies

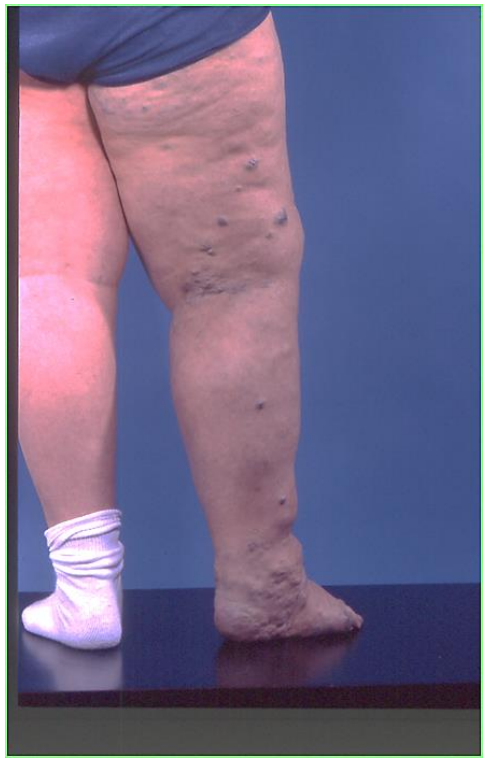
- Multidisciplinary clinics
- Collaborative multicentric and interdisciplinary research (PeDRA, HIG, French pediatric research group GRCSFD, ISSVA…)
- Patient-Family involvement, « Patient as Partner »
- Patient support groups
- Advocacy groups
- Collaboration between researchers and pharma to increase adapted therapeutic options for rare pediatric skin disorders

Areas for Future Research

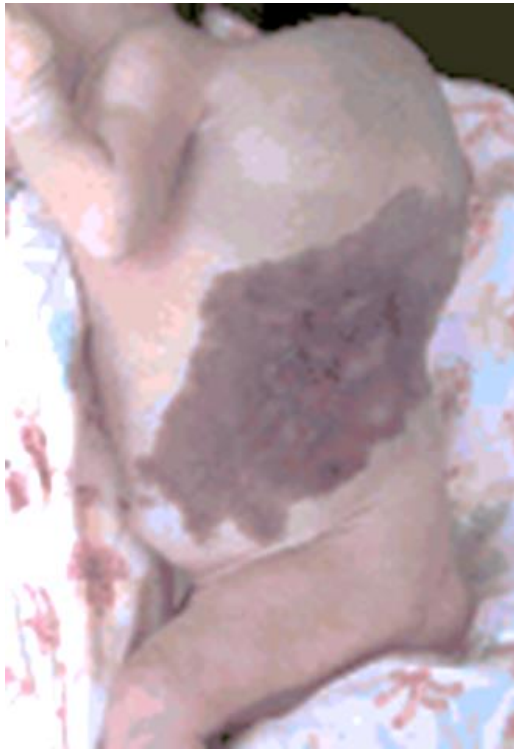
- Additional focus on cultural or regional differences of the impact of rare diseases
- Comparison of impact on patients with treatable vs non-treatable rare conditions
- Further explore primary care physician's perspective vs specialist's

Future of Genomics and Genetic Therapy

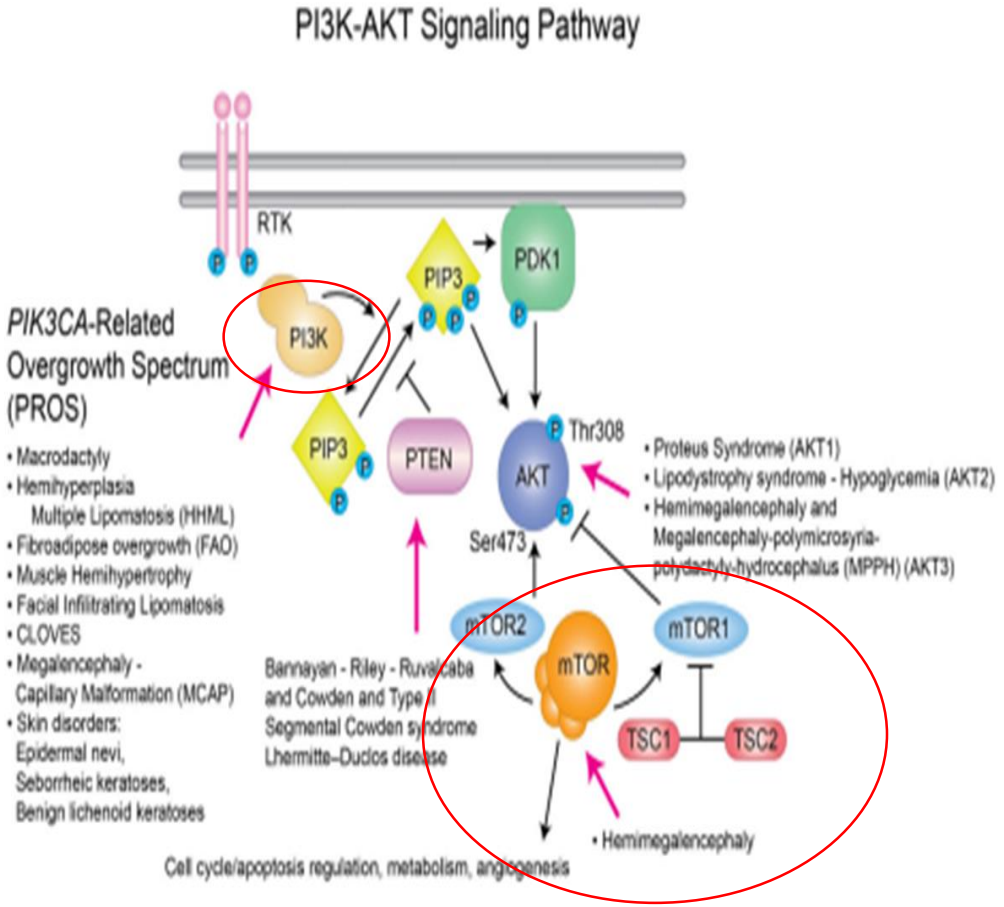
Complex Vascular Anomalies
The *PIK3CA story*



Klippel-Trenaunay syndrome



CLOVES syndrome



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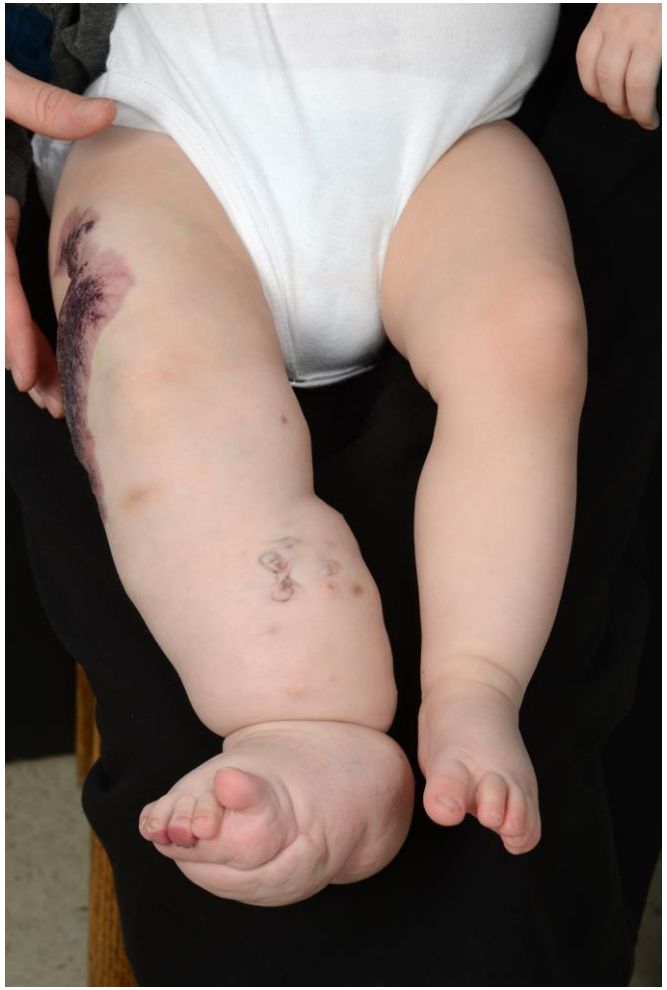
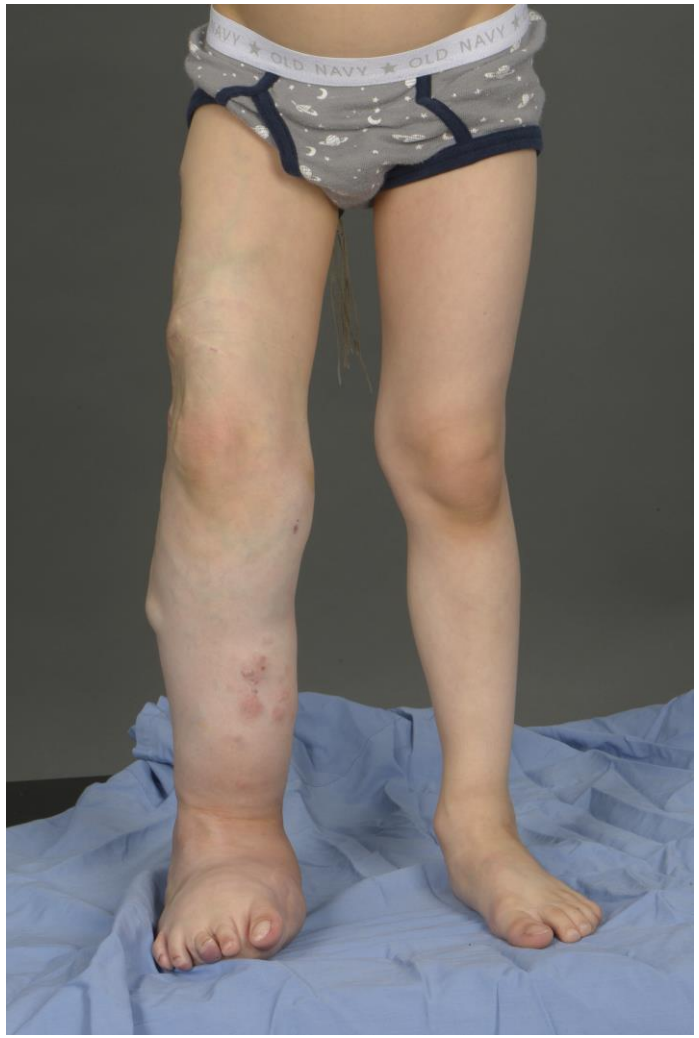
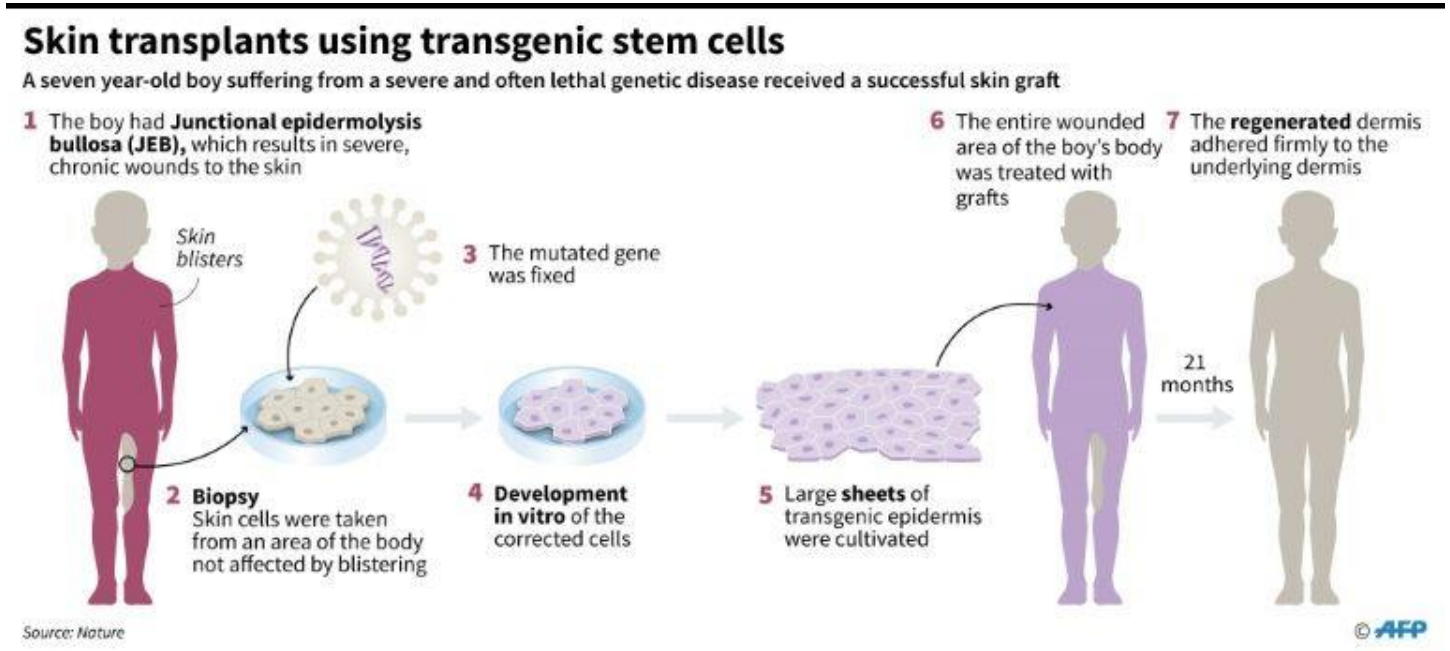
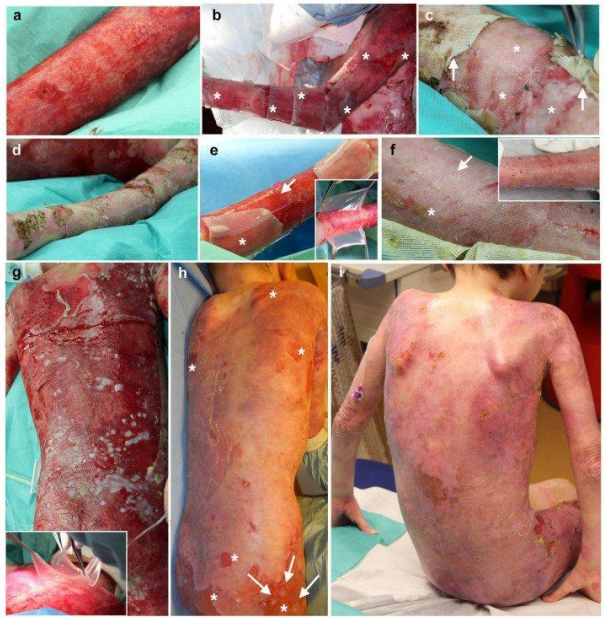




EPIDERMOLYSIS BULLOSA





Nature 2017;551:327-332

FUTURE is at our Door

- Collaborative studies of rare diseases lead to better understanding of their pathogenesis
- Targeted personalized medicine
- Development of specific drugs
- Need more Pediatric Dermatologists!