

Biological Agents and JAK Inhibitors for Dystrophic Epidermolysis Bullosa

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Tainan, Taiwan



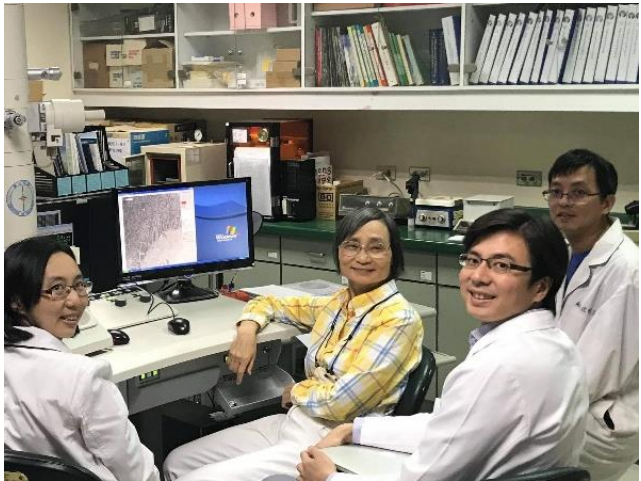
17 Oct 2024



EB History in Taiwan

1988

Electron
microscopy



Prof. Julia Yu-Yun Lee

2000

Genetic
study



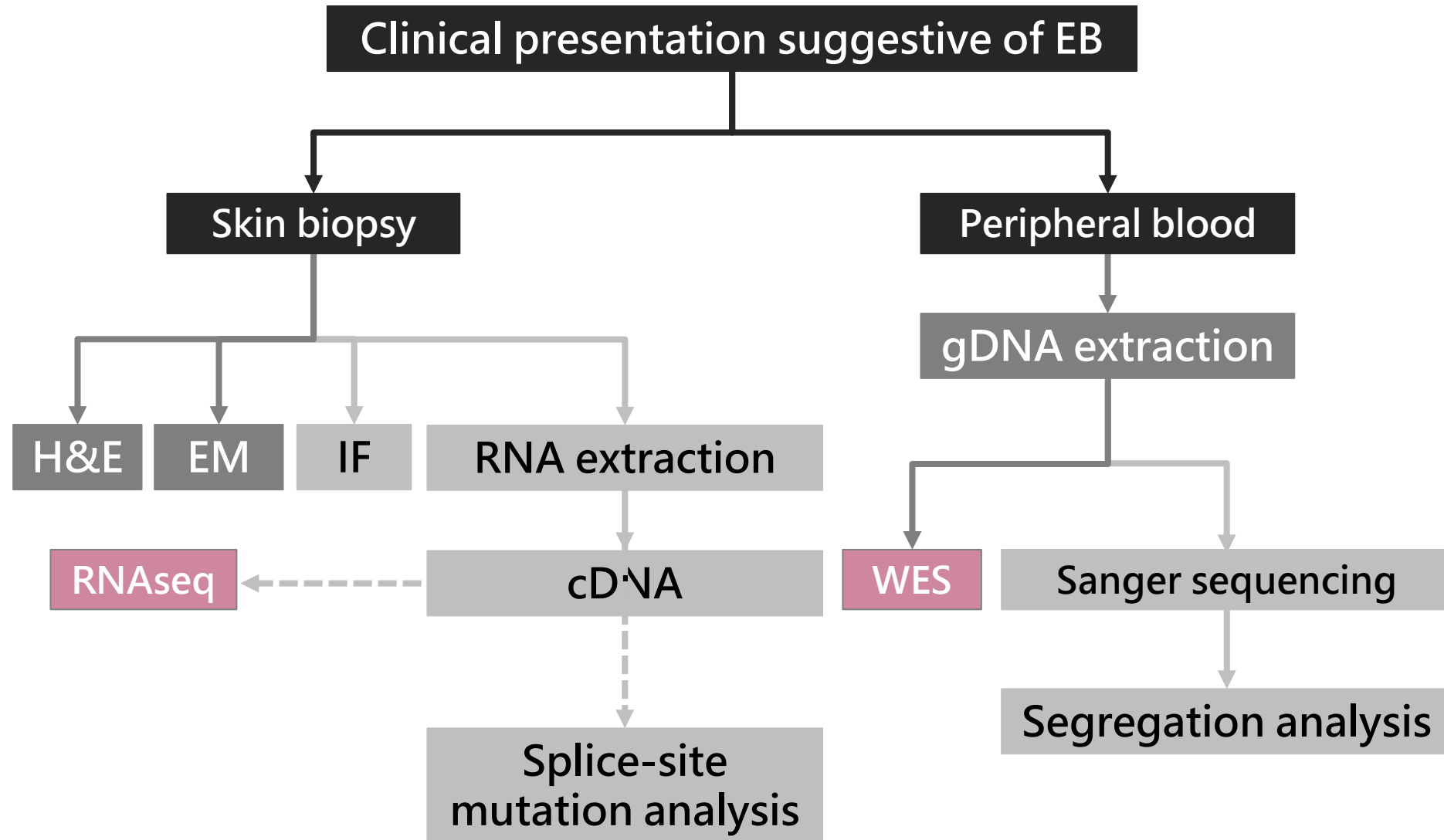
Dr. Sheau-Chiou Chao

2020

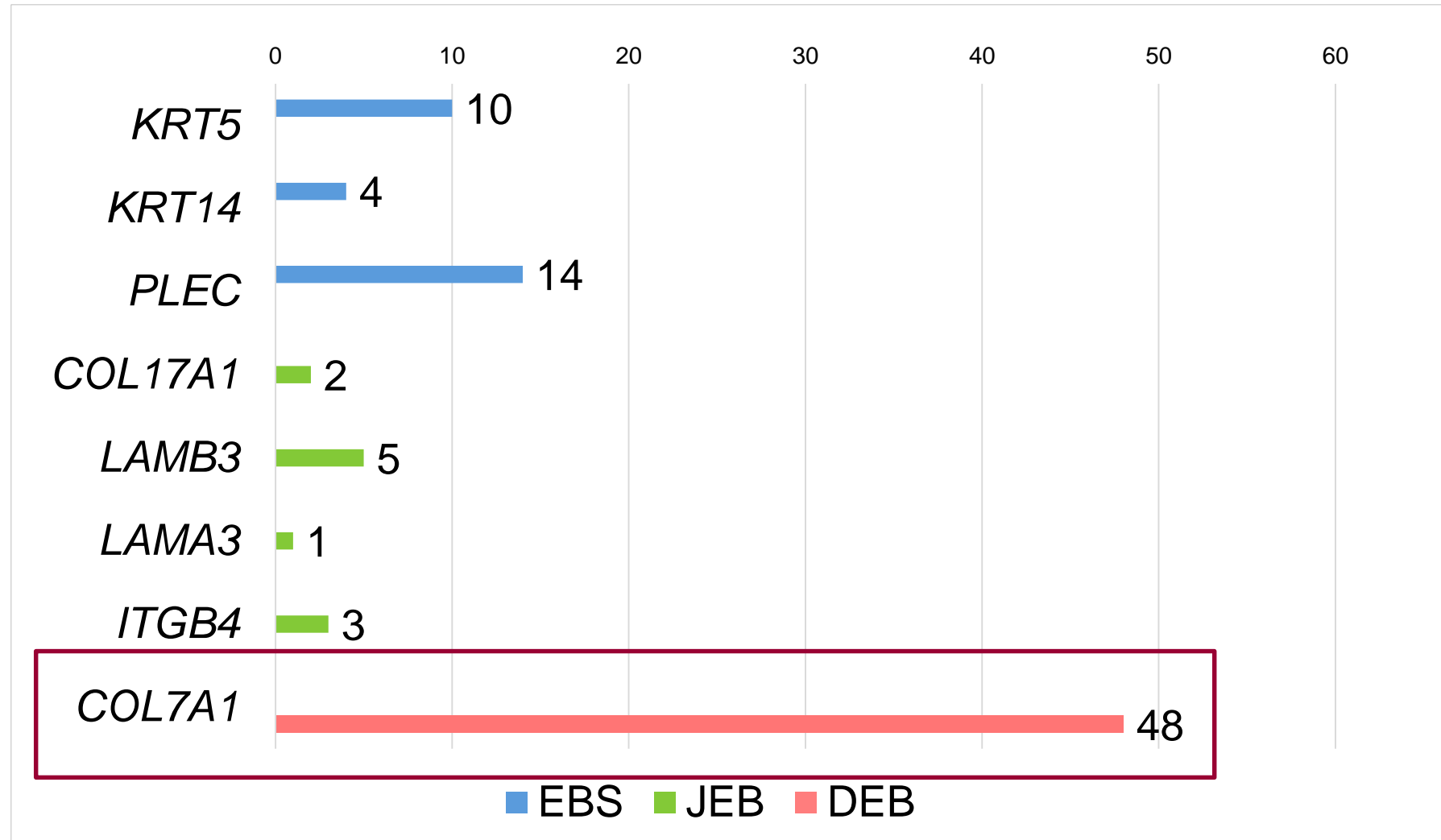
EB
clinics



NCKUH EB Genetic Study



Genetic study of EB patients in NCKU



Total 123 patients (till 2024.3)

EB pruriginosa



2

~ 30% of DEB

Severe itch

Poor tx response

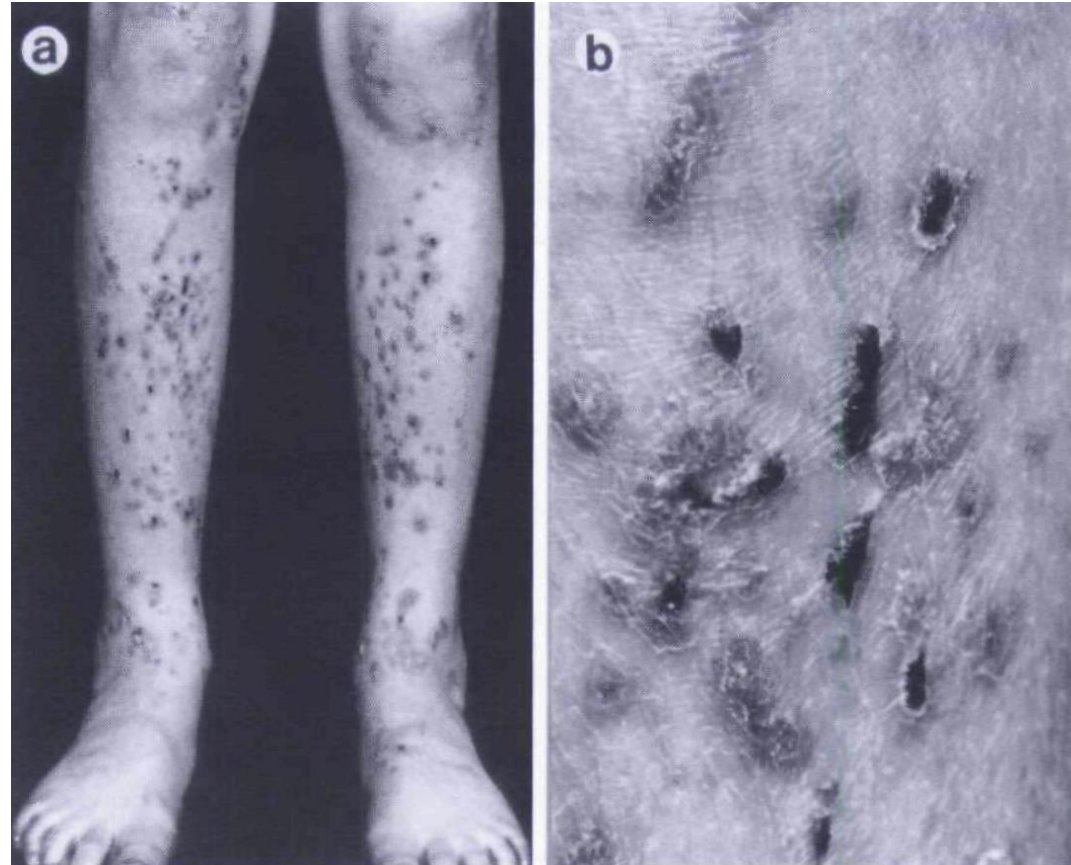
Epidermolysis bullosa pruriginosa: dystrophic epidermolysis bullosa with distinctive clinicopathological features



J.A.McGRATH, O.M.V.SCHOFIELD AND R.A.J.EADY

Department of Cell Pathology, St John's Institute of Dermatology, St Thomas' Hospital, London SE1 7EH, U.K.

Accepted for publication 22 October 1993



Dupilumab for EB pruriginosa

> [Br J Dermatol.](#) 2020 Jun;182(6):1495-1497. doi: 10.1111/bjd.18855. Epub 2020 Jan 29.

Treatment of epidermolysis bullosa pruriginosa-associated pruritus with dupilumab

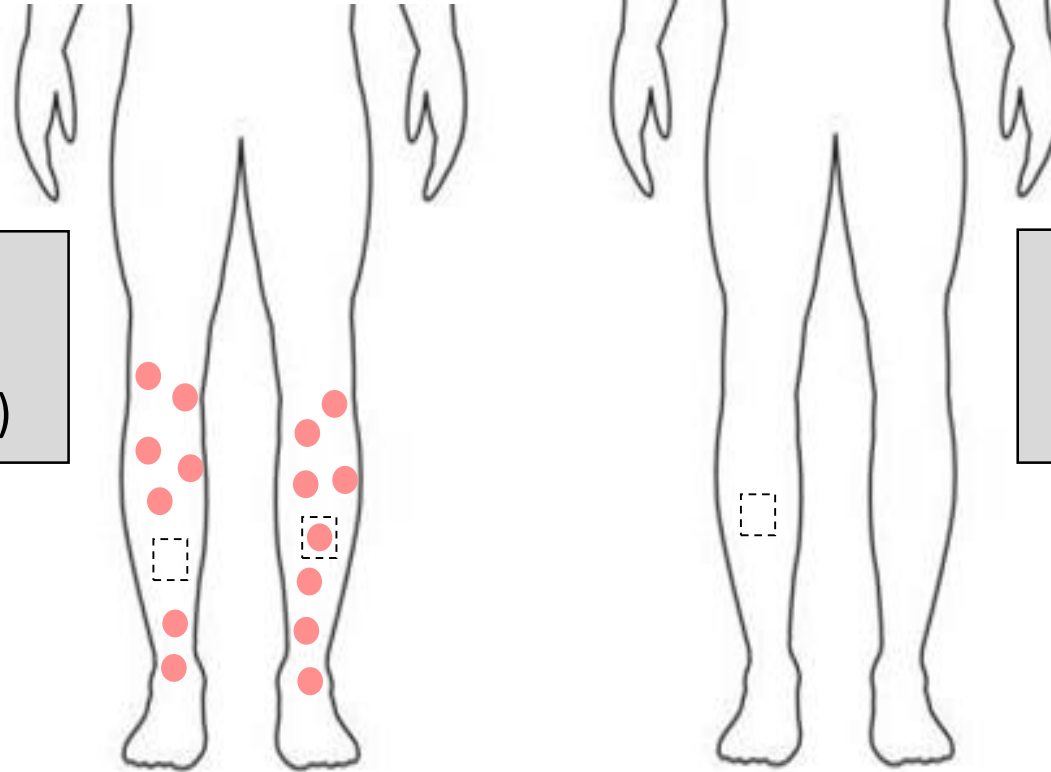


RNAseq study for EB pruriginosa

□ Skin biopsy

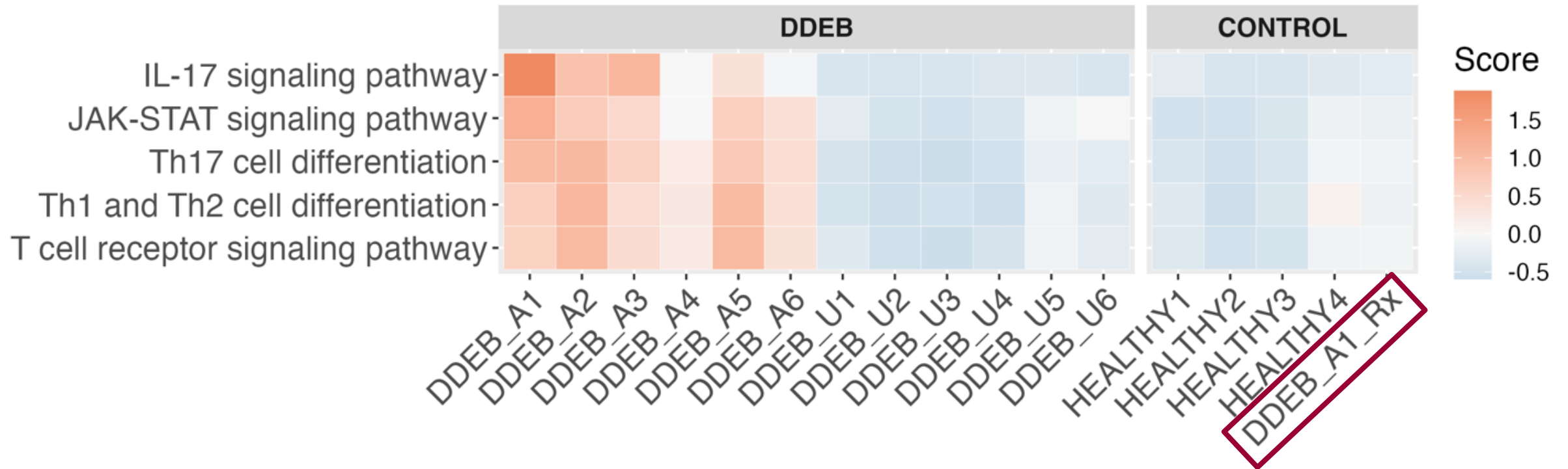
Bulk RNAseq

Affected (n=6)
Unaffected (n=6)
Post-Dupi Tx (n=1)



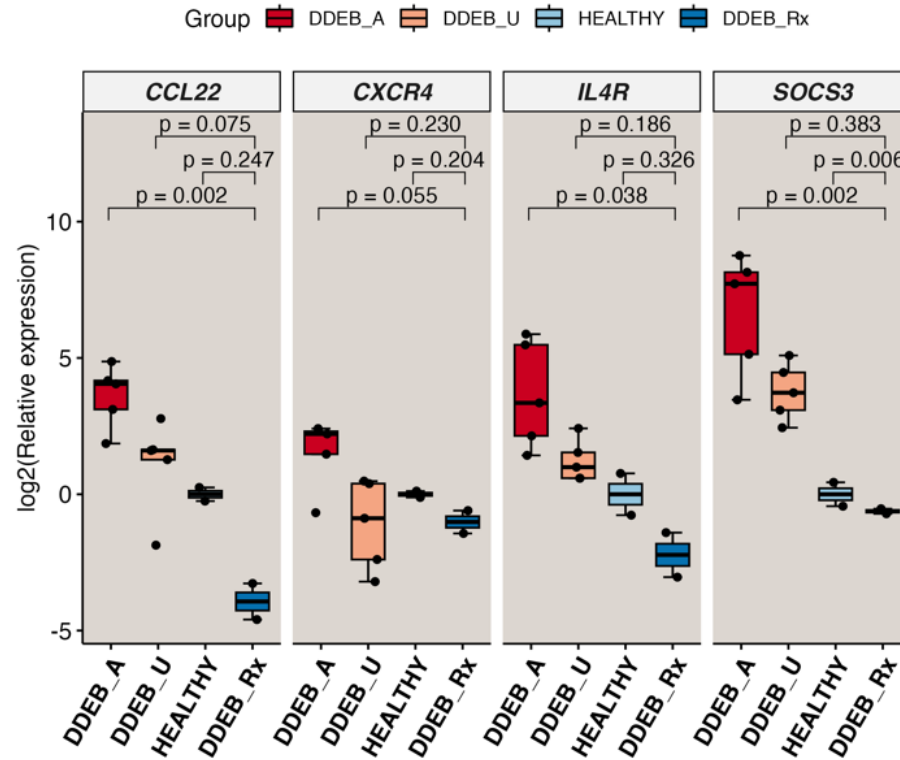
Healthy control (n=4)

Bulk RNAseq of EB pruriginosa

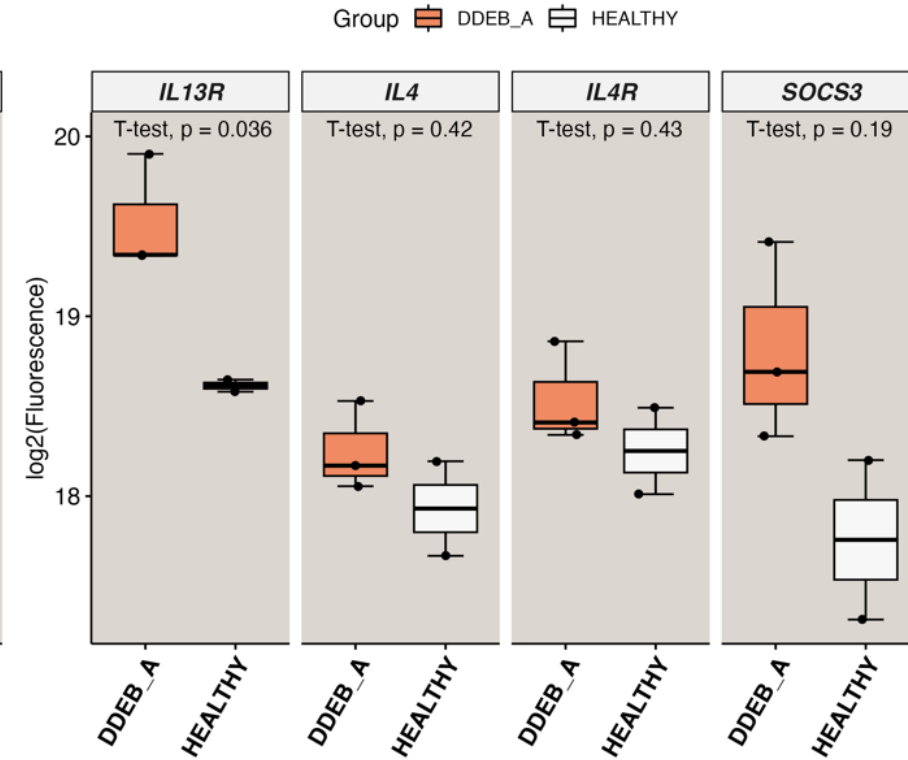


Th2-associated mRNA and protein studies of EB pruriginosa

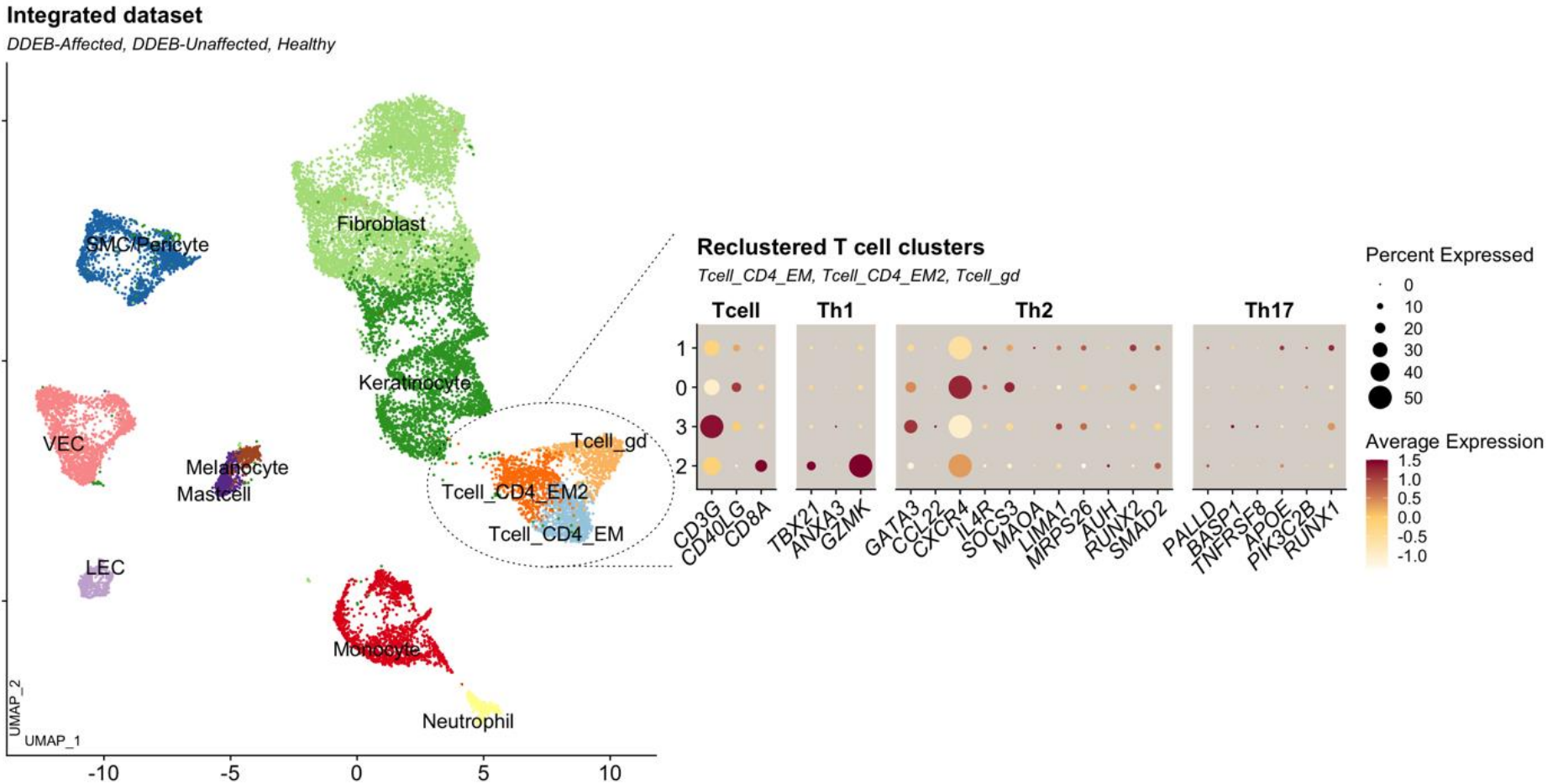
Th2 markers (mRNA)



Th2 markers (protein)



Single-cell RNAseq of EB pruriginosa



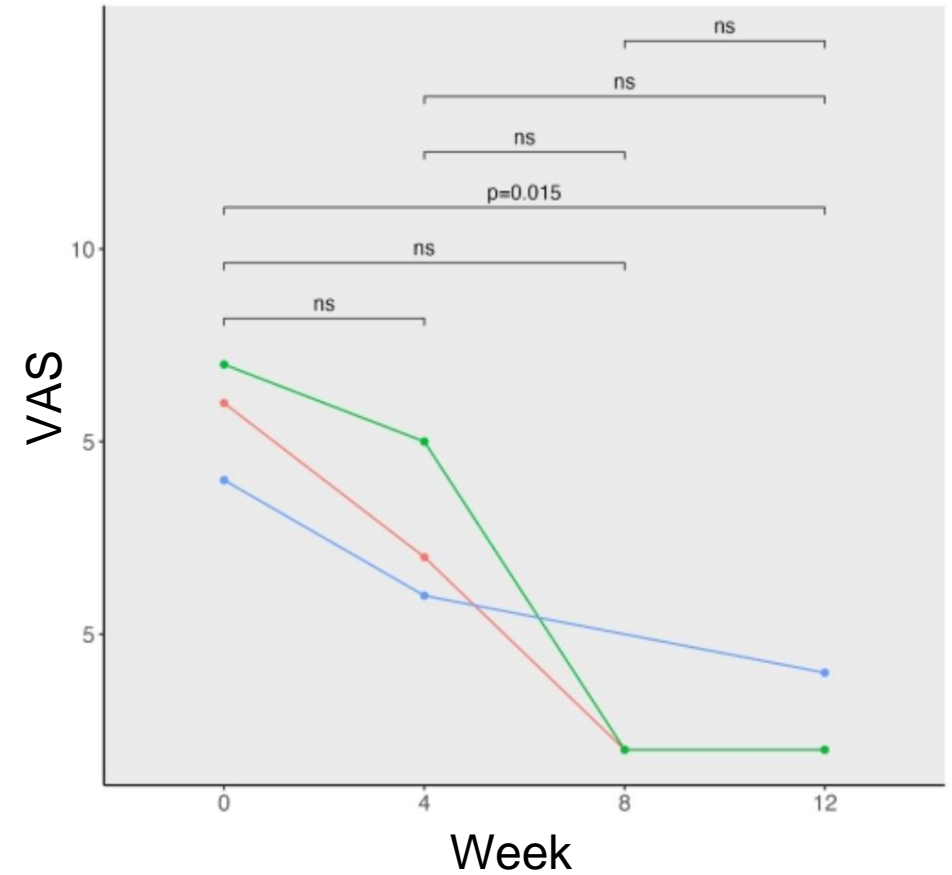
Affected (n=2)
Unaffected (n=1)
Healthy control (n=2)

Dupilumab for EB pruriginosa

Week 0



Week 12



JAKi for EB pruriginosa

RESEARCH LETTER

SKIN HEALTH AND DISEASE
Open Access

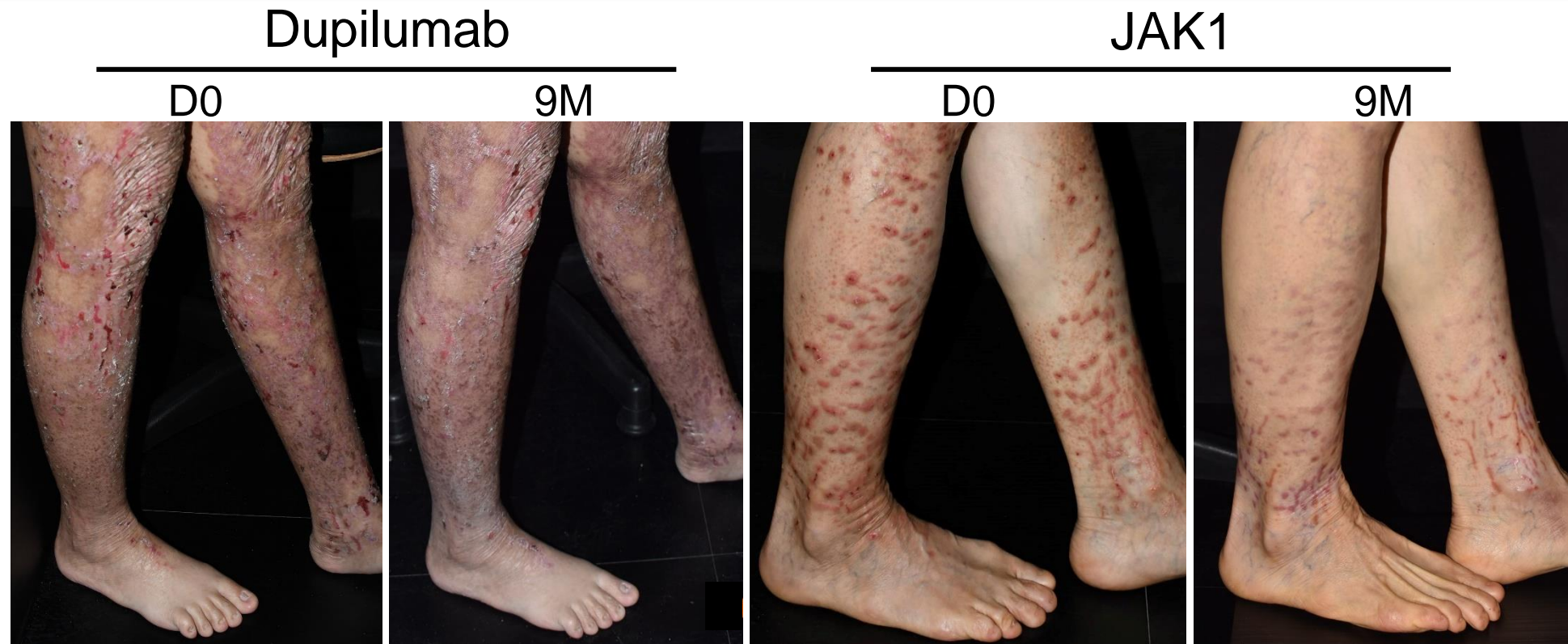
Real-world experience of using dupilumab and JAK inhibitors to manage pruritus in epidermolysis bullosa pruriginosa

Dupilumab
(N=3)

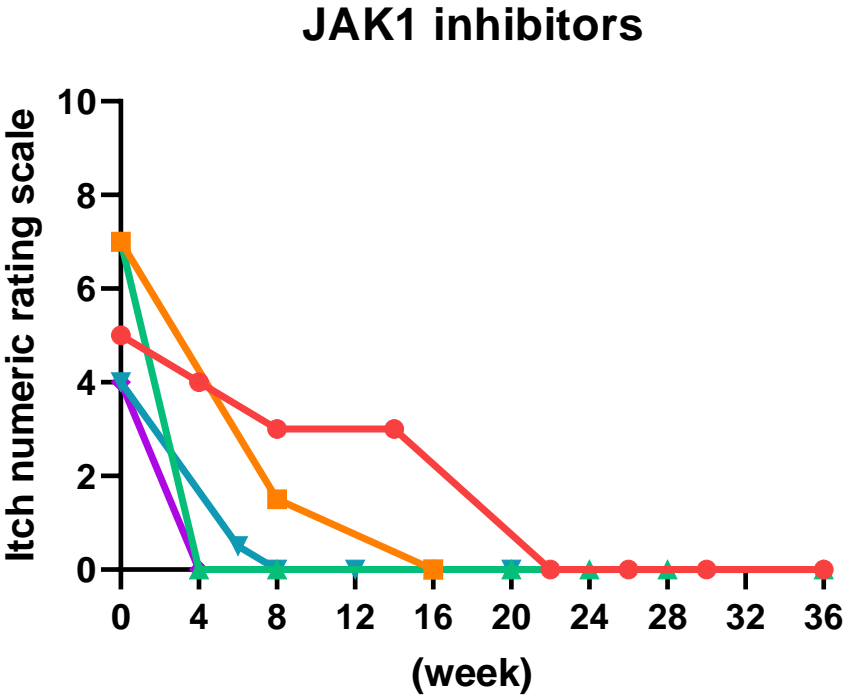
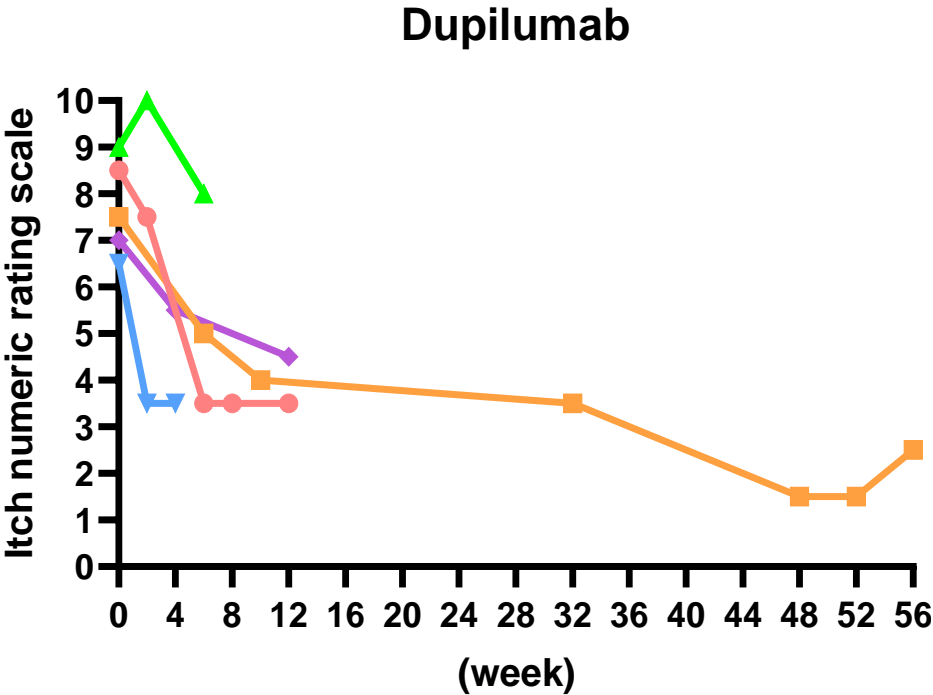
JAKi
(N=3)

Dupi --> JAKi
(N=3)

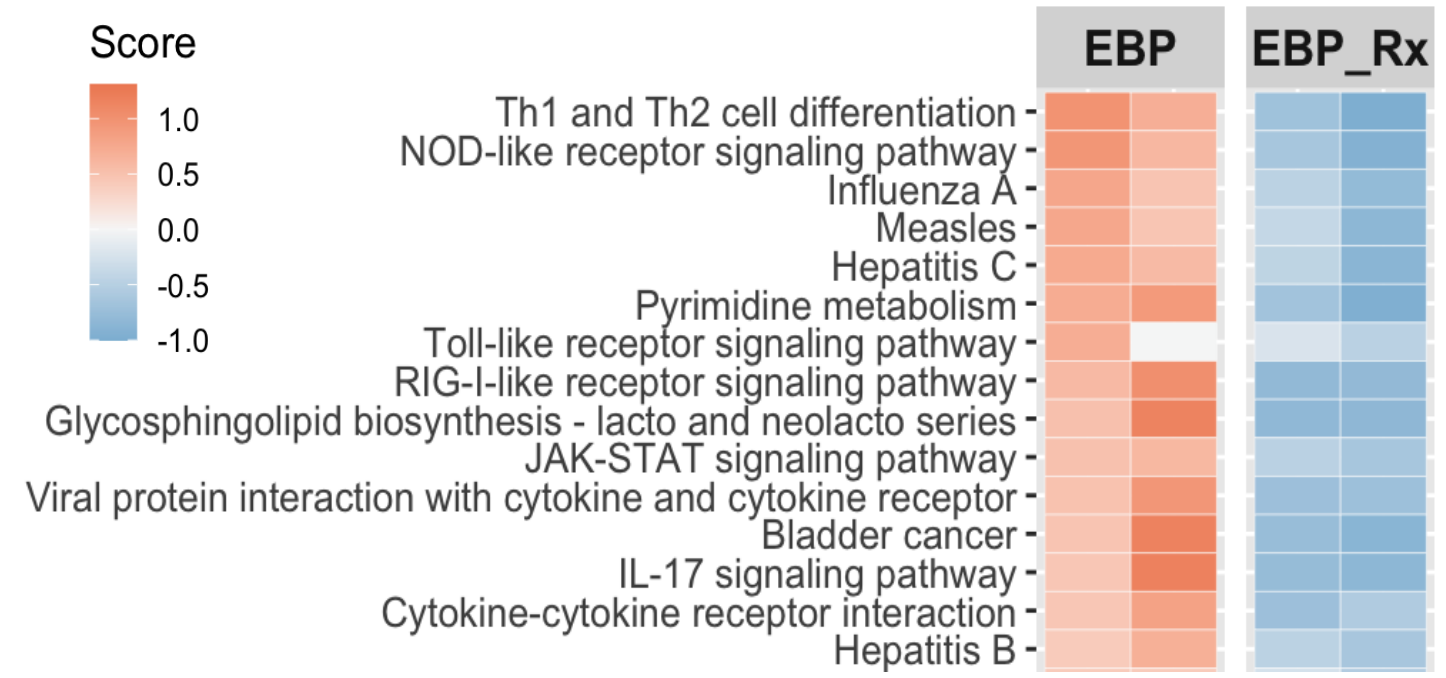
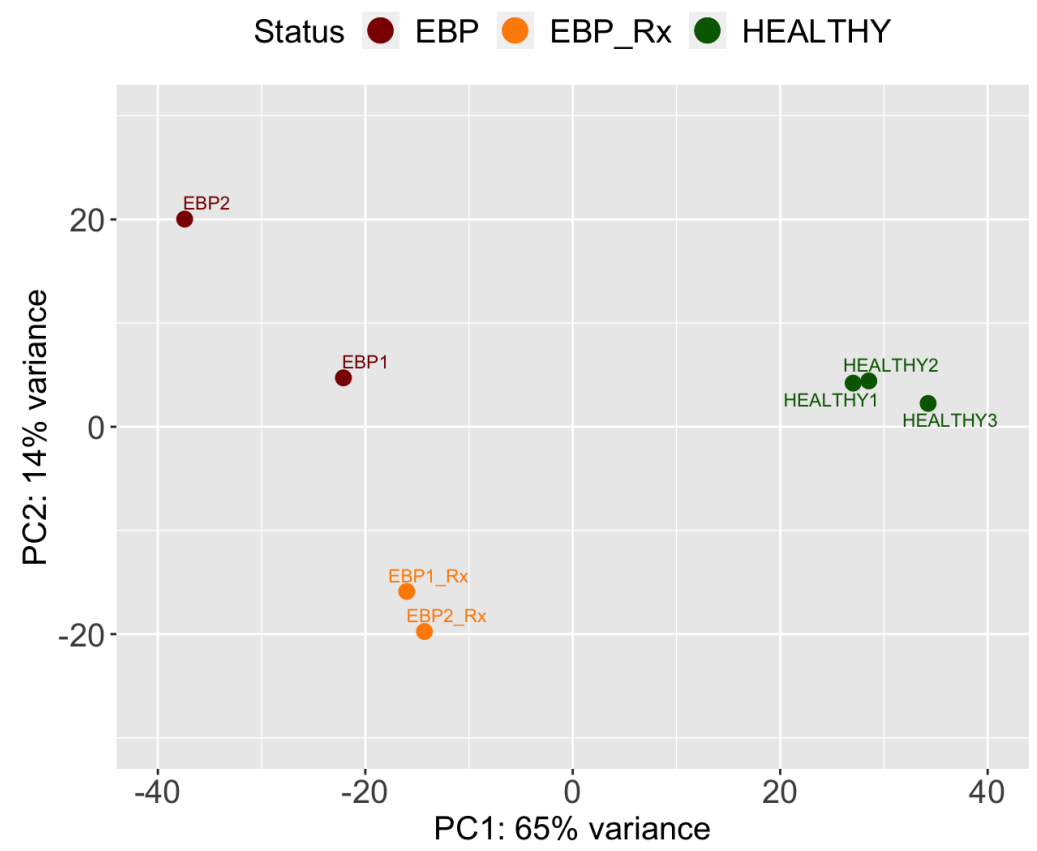
Real-world experience of using dupilumab and JAK inhibitors to manage pruritus in epidermolysis bullosa pruriginosa



Real-world experience of using dupilumab and JAK inhibitors to manage pruritus in epidermolysis bullosa pruriginosa



Transcriptomic changes after JAKi in EBP

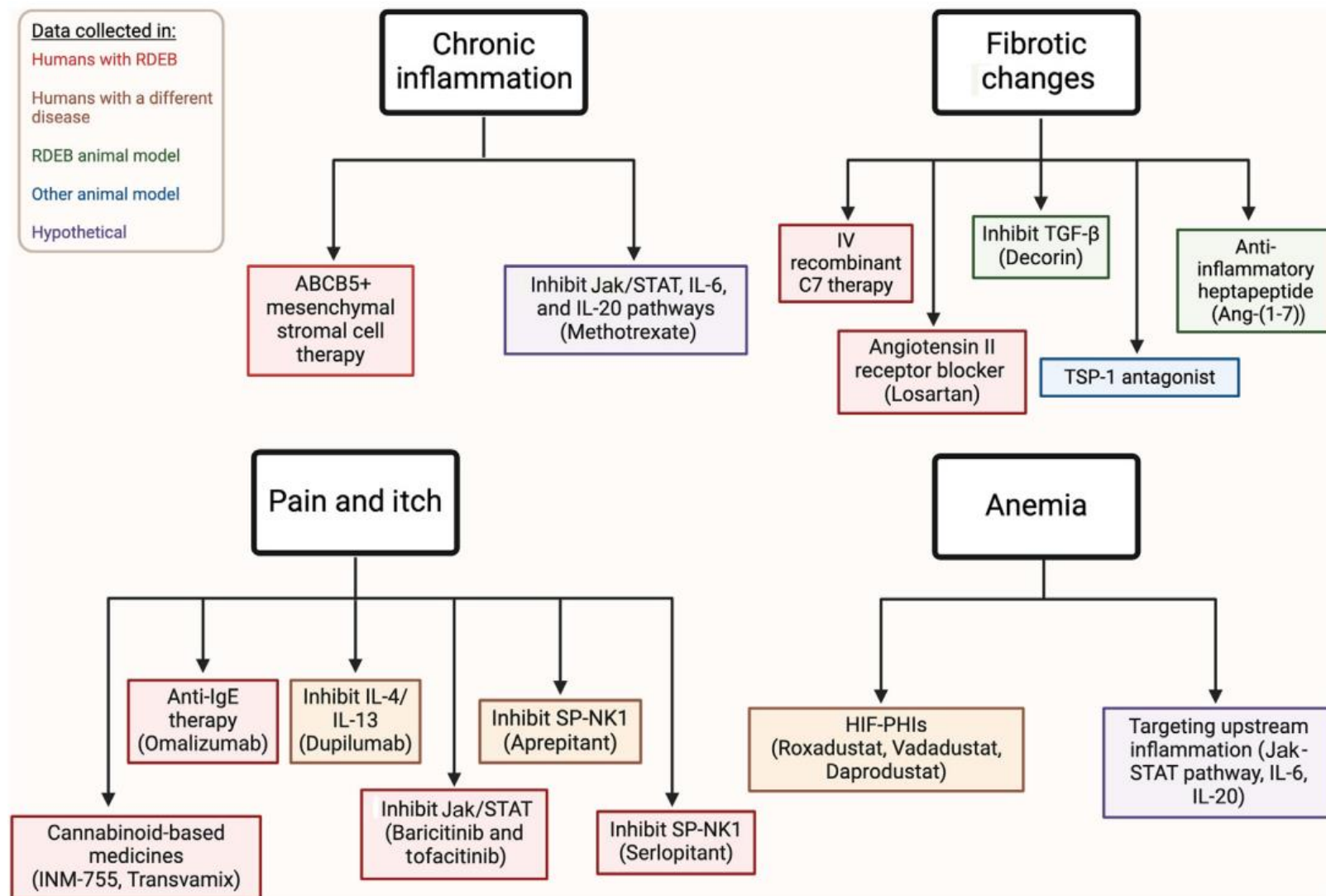


Recessive Dystrophic EB







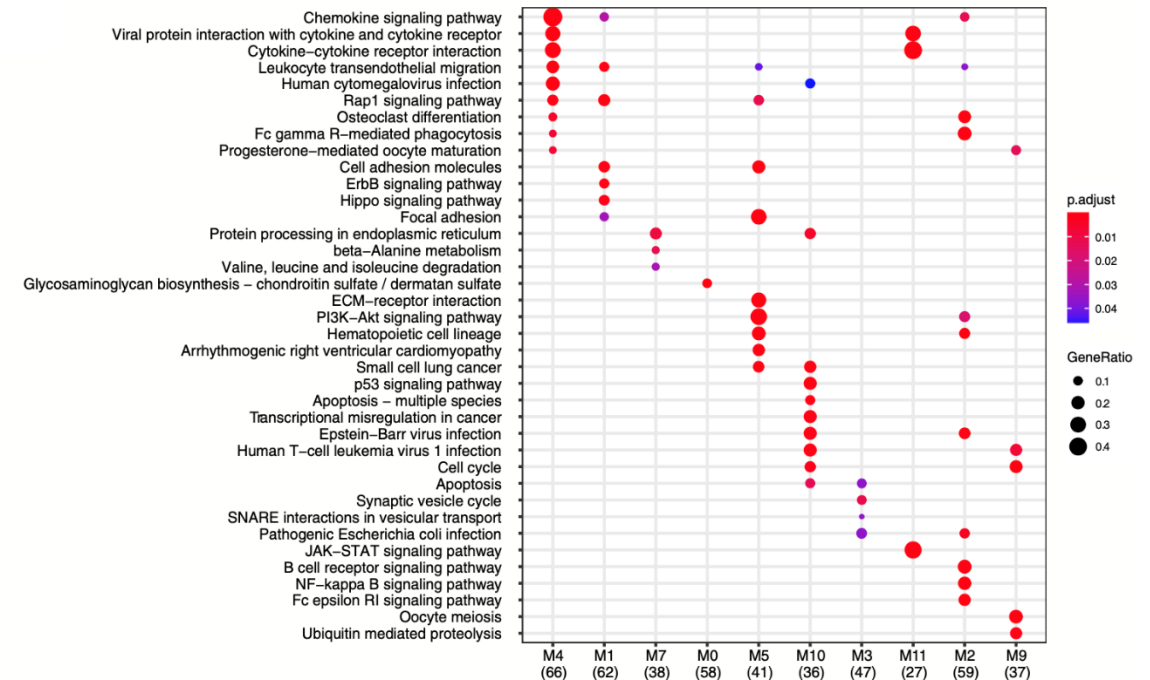
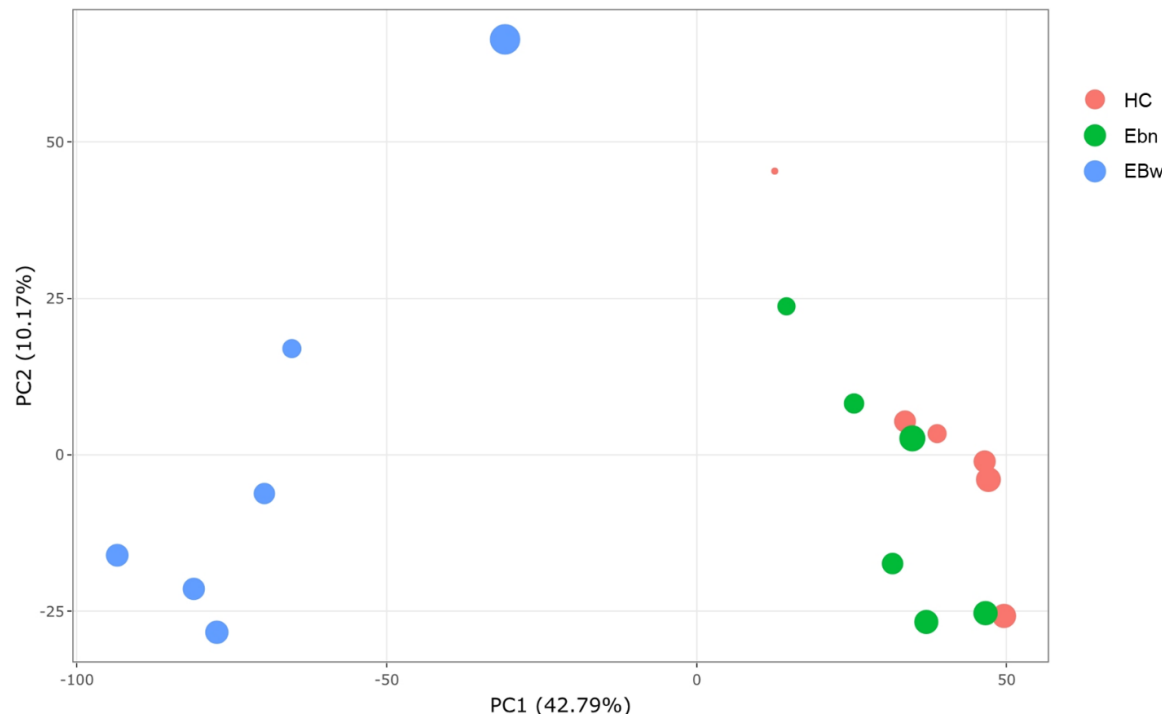
Data collected in:

- Humans with RDEB
- Humans with a different disease
- RDEB animal model
- Other animal model
- Hypothetical







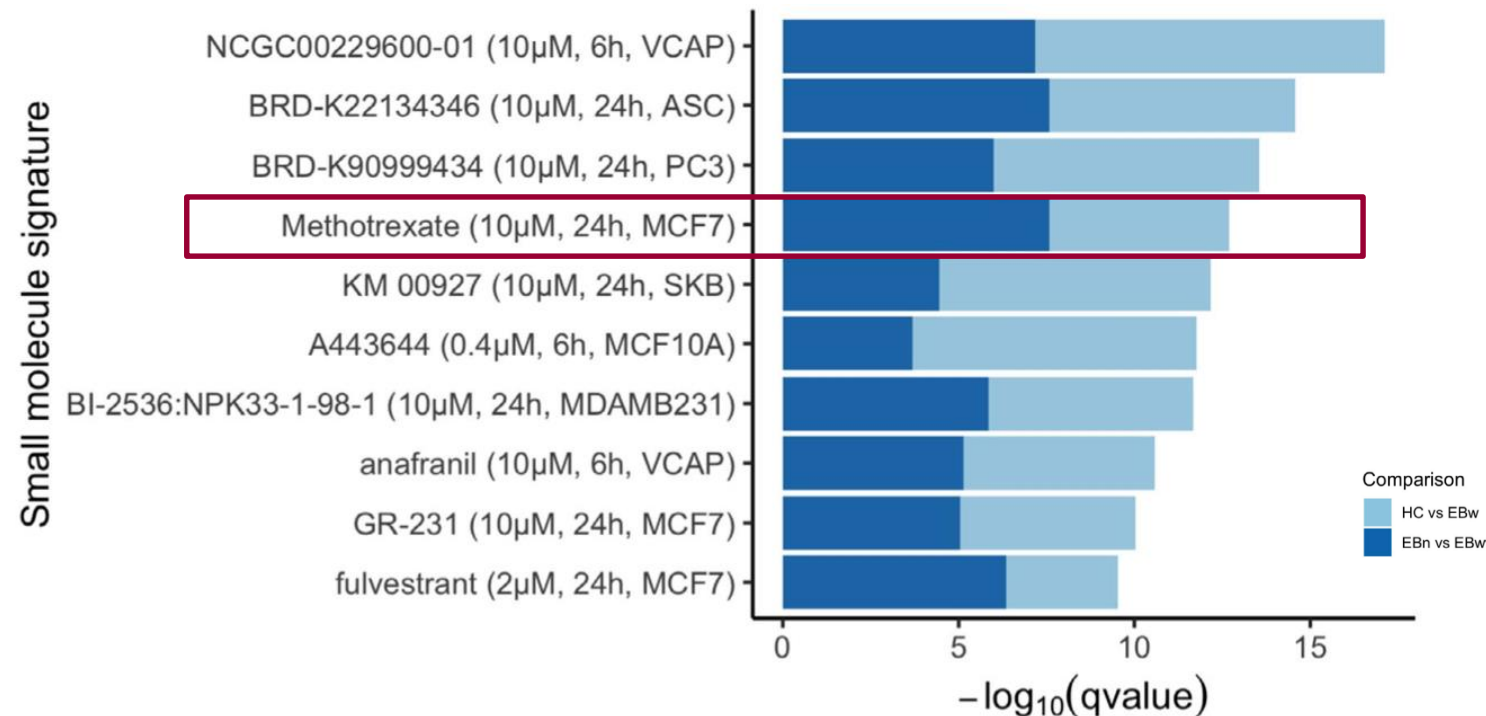
Transcriptomic profiling of recessive dystrophic epidermolysis bullosa wounded skin highlights drug repurposing opportunities to improve wound healing

Alexandros Onoufriadis¹  | Laura E. Proudfoot¹  | Chrysanthi Ainali²  |
Denis Torre³  | Maria Papanikolaou¹  | Tuntas Rayinda^{1,4}  | Ellie Rashidghamat¹  |
Retno Danarti⁴  | Jemima E. Mellerio¹  | Avi Ma'ayan³  | John A. McGrath¹ 

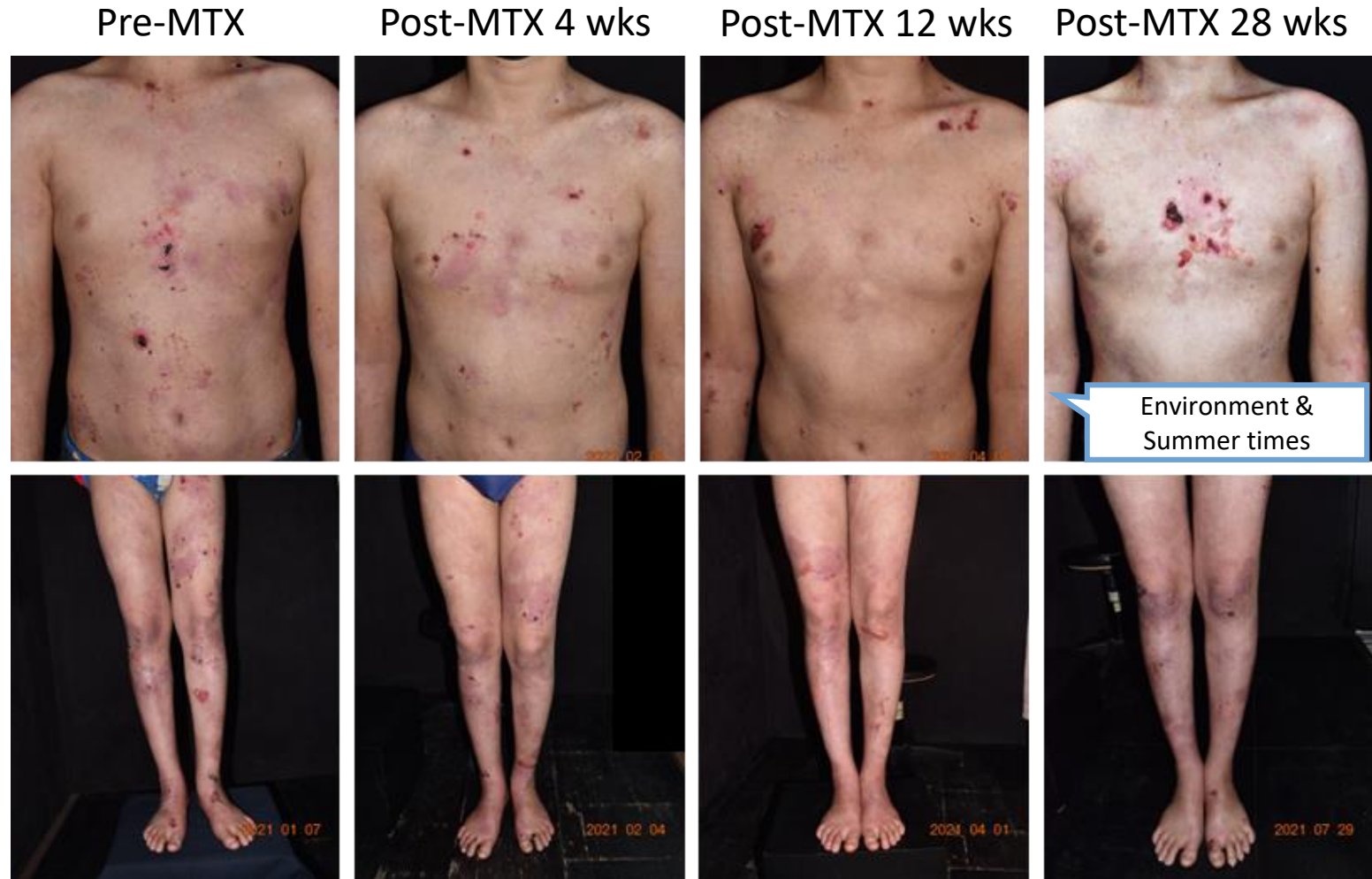
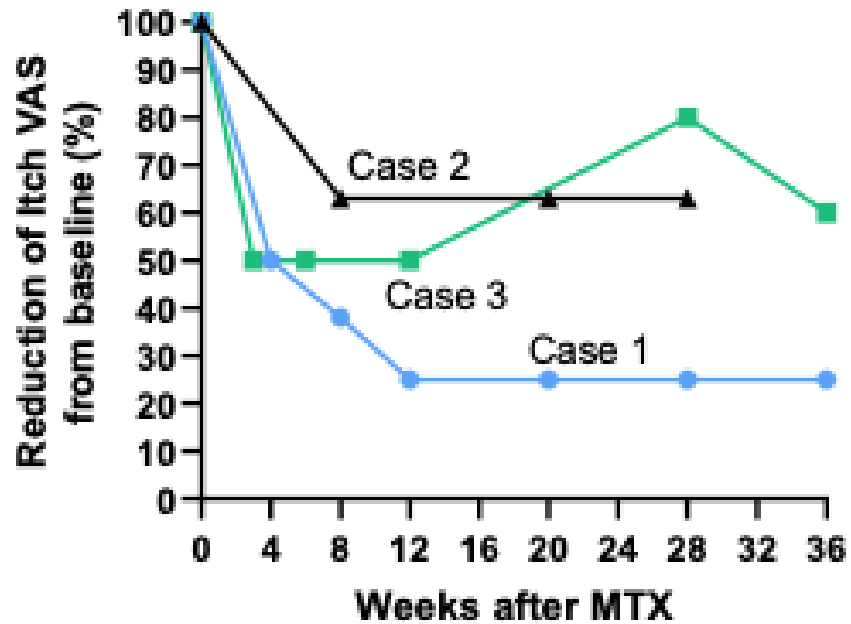


Transcriptomic profiling of recessive dystrophic epidermolysis bullosa wounded skin highlights drug repurposing opportunities to improve wound healing

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Methotrexate for RDEB



(Unpublished)

Take Home Message

Both dupilumab and JAKi alleviate itch and prurigo-like lesions in patients with EB pruriginosa, and our study suggested JAK inhibitors may be more effective than dupilumab. However, potential side effects should be considered when treating EB patients.

EB Team in NCKUH



Dermatologist



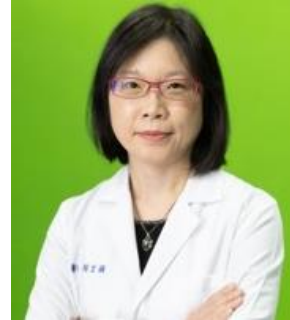
Dermatologist



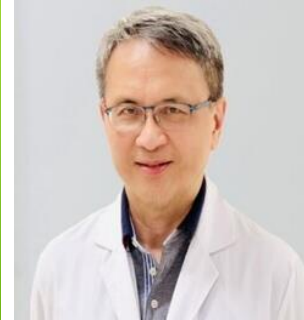
Dermatologist



Otolaryngologist



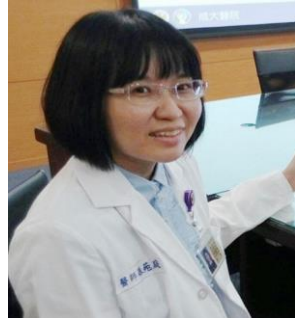
Pediatricist



Geneticist



Pediatric neurologist



Neurologist



General physician



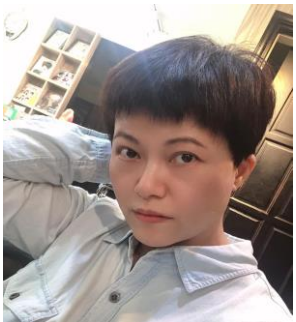
Pharmacist



Nutritionist



Ophthalmologist



Study Nurse



Study Nurse



Dentist



Nurse



Social worker



Genetic
Counselor

Acknowledgement

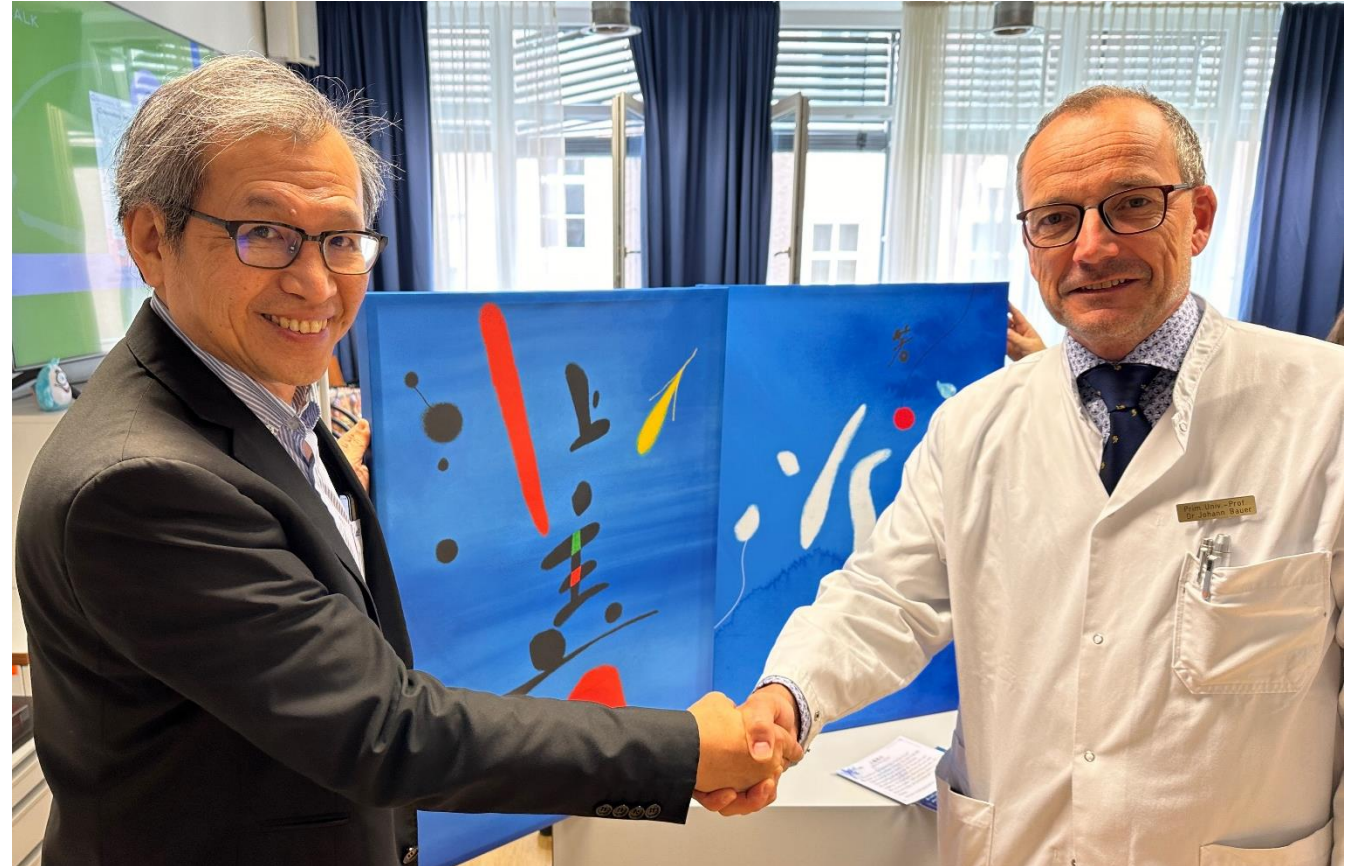




Keloid & GenoDermatoses Lab

Department of Dermatology, National Cheng Kung University Hospital, Tainan, Taiwan





15-16 Oct 2024

Thank you



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