

Epidermolysis bullosa research publications

NCKU institutional related (till July, 2024)

1. Marieta Papanikolaou, Leigh Nattkemper, Natashia Benzian-Olsson, Lu Liu, Alyson Guy, Han Lu, Tendai Kadiyirire, Ping-Chen Hou, Wilson Aala, Sonia Serrano, Rashida Pramanik, Nina Walters, Konstantina Dimitrakopoulou, Su Lwin, Emily Kalfas, Jose Satoc, Roman Laddach, Domenico Cozzetto, Bjorn Thomas, Evangelia Kesidou, Ellie Rashidghamat, Guy Orchard, Edel A O'Toole, Chao-Kai Hsu, Mansoor Saqi, Martin Steinhoff, Alexandros Onoufriadis, Gil Yosipovitch, Hannah Gould, Jemima E Mellerio, John A McGrath (2024, Jul). Th2 response drives itch in dystrophic epidermolysis bullosa pruriginosa: a case control study. *Journal of the American Academy of Dermatology*, 91(1):130-133..
2. Wilson Jr F Aala, Ping-Chen Hou, Yi-Kai Hong, Yu-Chen Lin, Yu-Rong Lee, Wei-Ting Tu, Marieta Papanikolaou, Natashia Benzian-Olsson, Alexandros Onoufriadis, Hans I-Chen Harn, Daw-Yang Hwang, Siao Muk Cheng, Kurt Lu, Peng-Chieh Chen, John A McGrath, Chao-Kai Hsu (2024, Mar). Dominant dystrophic epidermolysis bullosa is associated with glycolytically active GATA3+ Th2 cells which may contribute to pruritus in lesional skin. *British Journal of Dermatology*, ljae110. doi: 10.1093/bjd/ljae110
3. Yi-Chia Tsai, Wei-Ting Tu, Chun-Lin Su, Yu-Wen Cheng, Pei-Ling Chi, Chao Kai Hsu, Yang-Yi Chen (2024, Feb). Self-improving dystrophic epidermolysis bullosa with a novel heterozygous missense variant in the COL7A1 gene in a Taiwanese family. *Wound Repair and Regeneration*, doi: 10.1111/wrr.13159.
4. Meng-Ling Li, Yi-Kai Hong, Yu-Chen Lin, Ken Natsuga, Hideyuki Ujiie, Kentaro Izumi, Hiroaki Iwata, Chao-Kai Hsu (2023, Dec). Transcriptomic response of peripheral blood mononuclear cells to dupilumab in a 65-year-old patient with bullous pemphigoid. *Clinical and Experimental Dermatology*, 49(1):73-74.
5. Brice Patangan Serquiña, Niña Aala Gabaton, Yu-Chen Lin, Han-Tang Wang, Chao-Kai Hsu, Bryan Edgar Guevara (2023, Jul). Adult-Onset Epidermolysis Bullosa Pruriginosa in a 52-Year-Old Filipino Female: A Case Report. *Dermatology Practical & Conceptual*, 13(3): e2023158.
6. Ping-Chen Hou, Nathalie Del Agua, Su M Lwin, Chao-Kai Hsu, John A McGrath (2023, Jun). Innovations in the Treatment of Dystrophic Epidermolysis Bullosa (DEB): Current Landscape and Prospects. *Therapeutics and Clinical Risk Management*, 19:455-473.
7. Wei-Ting Tu, Wei-Ting Tu, Peng-Chieh Chen, Wan-Rung Chen, Jing-Yu Wang, Yi-Ting Huang, Yi-Huei Wu, Chun-Lin Su, Yen-An Tang, Hiroaki Iwata, Ken Natsuga, Sheau-Chiou Chao, H Sunny Sun, Ming-Jer Tang, Julia Yu-Yun Lee, John A. McGrath, Chao-Kai Hsu (2022, Dec). Mutational analysis of epidermolysis bullosa in Taiwan by whole-exome sequencing complemented by RNA sequencing: a series of 77 patients. *Orphanet Journal of Rare Diseases*, 17(1):451.
8. Ya-Fen Chen, Hsin-Chin Lu, Ping-Chen Hou, Yu-Ching Lin, Wilson Jr Aala, Alexandros Onoufriadis, John A McGrath, Ying-Lan Chen, Chao-Kai Hsu (2022, Aug). Plasma metabolomic profiling reflects the malnourished and chronic inflammatory state in recessive dystrophic epidermolysis bullosa. *Journal of Dermatological Science*, 107(2):82-88.
9. Yu-Chen Lin, Wei-Ting Tu, Ping-Chen Hou, Hsin-Yu Huang, Peng-Chieh Chen, Chung-Hsing Chang, Julia Yu-Yun Lee, John A McGrath, Chao-Kai Hsu (2022, Aug). Autosomal dominant epidermolysis bullosa simplex exacerbated by

hyperkeratotic scabies. *Journal of Dermatology*, 49(8):283-284.

10. Chung-Hao Hsu, Wei-Ting Tu, Peng-Chieh Chen, Julia Yu-Yun Lee, Chao-Kai Hsu, Tsu-Man Chiu (2022, May). Novel compound heterozygous ITGB4 mutations underlie lethal junctional epidermolysis bullosa with pyloric atresia and aplasia cutis congenita. *Journal of Dermatology*, 49(5):154-156.
11. Tomoko Miyake, Ken Natsuga, Takatsune Umayahara, Seiko Naito, Junko Yoshimoto, Akemi Senoo, Han-Tang Wang, Chao-Kai Hsu, Osamu Yamasaki, Shin Morizane (2022, Jan). Case of inherited epidermolysis bullosa simplex with KLHL24 gene mutation in Japan. *Journal of Dermatology*, 49(1):24-25.
12. Ken Natsuga*, Satoru Shinkuma*, Chao-Kai Hsu*, Yasuyuki Fujita, Akira Ishiko, Katsuto Tamai, John A McGrath. (2021, Dec). Current topics in Epidermolysis bullosa: Pathophysiology and therapeutic challenges.. *Journal of Dermatological Science*, 104(3):164-17.
13. Ping-Chen Hou, Han-Tang Wang, Stasha Abhee, Wei-Ting Tu, John A McGrath, Chao-Kai Hsu (2021, Nov). Investigational Treatments for Epidermolysis Bullosa. *American Journal of Clinical Dermatology*, 22(6), 801-817.
14. Ping-Chen Hou, Ken Natsuga, Wei-Ting Tu, Hsin-Yu Huang, Brandon Chen, Liang-Yu Chen, Wan-Rung Chen, Yi-Kai Hong, Yen-An Tang, Julia Yu-Yun Lee, Peng-Chieh Chen, H Sunny Sun, John A McGrath, Chao-Kai Hsu (2021, Aug). Complexity of Transcriptional and Translational Interference of Laminin-332 Subunits in Junctional Epidermolysis Bullosa with LAMB3 Mutations. *Acta Dermato-Venereologica*, 101(8): adv00522.
15. Jia-Horung Hung, Ping-Chen Hou, Fu-Chin Huang, Chao-Kai Hsu (2021, Apr). Topical gentamicin ointment induces LAMB3 nonsense mutation readthrough and improves corneal erosions in a patient with junctional epidermolysis bullosa. *Clinical & Experimental Ophthalmology*, 49(3):309-312.
16. Ping-Chen Hou, Wei-Ting Tu, Peng-Chieh Chen, Bryan Edgar K Guevara, Yu-Fen Yen, Hsin-Yu Huang, Julia Yu-Yun Lee, Ming-Jer Tang, Pao-Lin Kuo, John A McGrath, Chao-Kai Hsu (2021, Apr). A de novo COL17A1 splice-site mutation causing a 7-bp deletion in a Taiwanese patient with junctional epidermolysis bullosa. *European Journal of Dermatology*, 31(2):267-269.
17. Tak-Wah Wong, Chao-Chun Yang, Chao-Kai Hsu, Cheng-Han Liu, Julia Yu-Yun Lee (2021, Feb). Transplantation of autologous single hair units heals chronic wounds in autosomal recessive dystrophic epidermolysis bullosa: A proof-of concept study. *Journal of Tissue Viability*, 30(1):36-41.

Other hereditary skin diseases research publication

NCKU institutional related (till July, 2024)

1. Arti Nanda*, Yi-Han Chang*, Hui-Ching Cheng*, I-Tzu Lai, Atlal Al-Lafi, John A McGrath, Chao-Kai Hsu (2024, Mar). Autosomal recessive plantar keratoderma with ragged periungual hyperkeratosis caused by a homozygous missense variant in KRT16 . *British Journal of Dermatology*, 190(4):588-590.
2. Yu-Chen Lin, Yi-Kai Hong, Wilson Jr F Aala, Kiyotaka Hitomi, Masashi Akiyama, John A McGrath, Chao-Kai Hsu (2024, Mar). Tofacitinib ameliorates skin inflammation in a case of severe autosomal recessive congenital ichthyosis . *Clinical and Experimental Dermatology*, 11ae080.
3. Ping-Chen Hou, Yi-Kai Hong, Yu-Chen Lin, Min-Chia Yang, Chung-Ting Hsu, John A McGrath, Chao-Kai Hsu (2024, May). Transcriptomic response of peripheral blood mononuclear cells to secukinumab in epidermolytic ichthyosis. *Clinical and Experimental Dermatology*, 49(6):626-628.
4. Roy Luister C Acos, Yi-Han Chang, Yu-Chen Lin, Dianne Katherine R Salazar Paras, Yu Fujimura, Hajime Nakano, Eijiro Akasaka, Ken Natsuga, Bryan Edgar K Guevara, John A McGrath, Chao-Kai Hsu (2024, Feb). Variable clinical consequences of mosaicism for c.1167dupC in IKBKG in male and female patients with incontinentia pigmenti and related phenotypes. *Clinical and Experimental Dermatology*, 49(3):298-301.
5. Chang-Yu Hsieh, Yu-Chen Lin, Hui-Ching Cheng, Chih-Yu Chang, Wei-Ting Tu, Chao-Kai Hsu, Tsen-Fang Tsai (2024, May). Identification of a novel frameshift mutation in cathepsin C gene in a patient with coexisting Papillon-Lefevre syndrome and rheumatoid arthritis. *Journal of the European Academy of Dermatology and Venereology*, 38(5): e410-e412.
6. Bryan Edgar K Guevara, Jay Mohamad Ryan M Aquino, Rosa Beatriz S Diaz, Andrea Isabel T Contreras, Yu-Chen Jeng, Hsin-Yu Huang, Chao-Kai Hsu, Bai Shariffah B Pandita-Reyes (2023, Aug). Infantile generalized pustular psoriasis with excellent response to secukinumab: a case report. *Experimental dermatology*, 32(8):1296-1298.
7. Yu-Chen Lin, Yu-Chen Jeng, Wilson Jr F Aala, Yi-Kai Hong, Pin-Hsuan Chen, Ya-Ru Chuang, Chao-Chun Yang, Chao-Kai Hsu (2023, Aug). Transcriptomic responses of peripheral blood mononuclear cells to cyclosporin and etanercept in a female infant with juvenile generalized pustular psoriasis. *Experimental Dermatology*, 32(8):1299-1305.
8. Yu-Chen Jeng, Yu-Chen Lin, Chao-Kai Hsu, Julia Yu-Yun Lee, Chaw-Ning Lee (2023, Aug). Generalized pustular psoriasis caused by bupropion with rapid recurrence after self-rechallenge. *Experimental Dermatology*, 32(8):1306-1307.
9. Chia-Yu Chu, Chih-Hung Lee, Hua-En Lee, Yung-Tsu Cho, Chao-Kai Hsu, Tom C Chan, Song-Chou Hsieh, Kai-Che Wei (2023, Jun). Taiwanese dermatological association (TDA) consensus for the management of pemphigus. *Journal of the Formosan Medical Association*, 122(7):540-548.
10. Yi-Han Chang, Hsing-San Yang, Hsin-Yu Huang, Julia Yu-Yun Lee, Chao-Kai Hsu (2023, Apr). A possible role for second-hit postzygotic GJB2 mutation in prokeratotic eccrine ostial and dermal duct nevus. *Journal of Dermatology*, 50(4):556-560.
11. Ken Natsuga, Yoshikazu Furuta, Shota Takashima, Takuma Nohara, Hsin-Yu Huang, Satoru Shinkuma, Hideki Nakamura, Yousuke Katsuda, Hideaki Higashi,

- Chao-Kai Hsu, Satoshi Fukushima, Hideyuki Ujiie (2022, Jul). Cas9-guided haplotyping of three truncation variants in autosomal recessive disease. *Human Mutation*, 43(7):877-881.
12. Min-Chia Yang, Yu-Chen Lin, Chien-Hao Huang, Tsang-Ming Ko, Meng-Che Tsai, Pao-Lin Kuo, Julia Yu-Yun Lee, John A. McGrath, Chao-Kai Hsu (2022, Jul). Incontinentia pigmenti in a male infant and a proposed diagnostic algorithm. *Clinical and Experimental Dermatology*, 47(7):1366-1368.
 13. Chao-Kai Hsu, Nan-Yu Cheng, Chao-Chun Yang, Yun-Yo Yen, Sheng-Hao Tseng (2022, May). Investigating the clinical implication of corneometer and mexameter readings towards objective, efficient evaluation of psoriasis vulgaris severity. *Scientific Reports*, 12(1):7469.
 14. Ping-Chen Hou, Yu-Rong Lee, Bryan Edgar K Guevara, Peng-Chieh Chen, John A McGrath, Chao-Kai Hsu (2022, Mar). Transcriptomic response of peripheral blood mononuclear cells to secukinumab in an 8-year-old boy with juvenile generalized pustular psoriasis. *Journal of the European Academy of Dermatology and Venereology*, 36(5):378-381.
 15. Alexandros Onoufriadis, Umar Niazi, Konstantina Dimitrakopoulou, Jeremias Reich, Chrysanthi Ainali, Maria Papanikolaou, Evangelia Kesidou, Chao-Kai Hsu, Mansoor Saqi, John A McGrath, Kristian Reich (2022, Feb). Transcriptomic Analysis of Blaschko-Linear Psoriasis Reveals Shared and Distinct Features with Psoriasis Vulgaris. *Journal of Investigative Dermatology*, 142(2):489-493.
 16. Chao-Chun Yang, Yun-Yo Yen, Chao-Kai Hsu, Nan-Yu Cheng, Shih-Yu Tzeng, Shih-Jay Chou, Jun-Ming Chang, Sheng-Hao Tseng (2021, Apr). Investigation of water bonding status of normal and psoriatic skin in vivo using diffuse reflectance spectroscopy. *Scientific Reports*, 11(1):8901.