

## 主な研究課題・発表代表論文

皮膚科学講座 Dermatology

研究領域 上皮情報解析医科学

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## 主な研究課題

- ・膿疱性乾癬の病態解明
- ・遺伝性皮膚・毛髪疾患の発症機構の解明
- ・形成外科領域の新規術式の開発に向けた研究

## 発表代表論文

- 1) Onitsuka M., et al.: A homozygous loss-of-function variant in the MPO gene is associated with generalized pustular psoriasis. *J Dermatol.*, 50 : 664-671, 2023.
- 2) Yagi S., et al.: Different degree of loss-of-function among four missense mutations in the EDAR gene responsible for autosomal recessive hypohidrotic ectodermal dysplasia may be associated with the phenotypic severity. *J Dermatol.*, 50 : 349-356, 2023.
- 3) Takasu H., et al.: Lower Eyelid Reconstruction Using a Myotarsocutaneous Flap while Considering the Superior and Inferior Palpebral Sulci. *Plast. Reconstr. Surg. Glob. Open*, 10 : e4147, 2022.
- 4) Asano N., et al.: Characterization of EDARADD gene mutations responsible for hypohidrotic ectodermal dysplasia. *J. Dermatol.*, 48 : 1533-1541, 2021.
- 5) Yasuno S., et al.: Evidence for a dominant-negative effect of a missense mutation in the SERPING1 gene responsible for hereditary angioedema type I. *J. Dermatol.*, 48 : 1243-1249, 2021.
- 6) Murata M., et al.: Two cases of severe congenital hypotrichosis caused by compound heterozygous mutations in the LSS gene. *J. Dermatol.*, 48 : 392-396, 2021.
- 7) Okita T., et al.: Functional studies for a dominant mutation in the EDAR gene responsible for hypohidrotic ectodermal dysplasia. *J. Dermatol.*, 46 : 710-715, 2019.
- 8) Yasuno S., et al. : Sporadic case of Darier disease caused by a novel splice-site mutation in the ATP2A2 gene. *Clin. Exp. Dermatol.*, 44 : e10-e12, 2019.
- 9) Okita T., et al. : Two Japanese families with hypohidrotic ectodermal dysplasia: Phenotypic differences between affected individuals. *J. Dermatol.*, 46 : e99-e101, 2019.
- 10) Asano N., et al.: Identification of a novel splice site mutation in the LIPH gene in a Japanese family with autosomal recessive woolly hair. *J. Dermatol.*, 46 : e19-e20, 2019.