

Research field:

Principal Investigator: Toshifumi Nomura

E-mail.address: nomura@md.tsukuba.ac.jp

URL: <https://dermatology-tsukuba.org>



Other Faculty Members

Associate Professor: Yoshiyuki Nakamura (Email address: ynakamura@md.tsukuba.ac.jp)

Major Scientific Interests of the Group

We aim to understand the pathomechanisms underlying genetic skin disorders, including ichthyoses and keratodermas.

Projects for Regular Students in Doctoral or Master's Programs

- 1) To unveil the pathomechanisms underlying Nagashima-type palmoplantar keratosis
- 2) To clarify how mutant *FLG* mRNAs are degraded in keratinocytes
- 3) To identify the mechanisms underlying the revertant mosaicism in ichthyosis with confetti

Study Programs for Short Stay Students (one week – one semester)

- 1) Not applicable

Selected Publications

- 1) Miyauchi T, Suzuki S, Takeda M, Peh JT, Aiba M, Natsuga K, Fujita Y, Takeichi T, Sakamoto T, Akiyama M, Shimizu H, Nomura T (corresponding author): Altered replication stress response due to CARD14 mutations promotes recombination-induced revertant mosaicism. *Am J Hum Genet* 108: 1026-1039, 2021.
- 2) Suzuki S, Nomura T (corresponding author), Miyauchi T, Takeda M, Fujita Y, Nishie W, Akiyama M, Ishida-Yamamoto A, Shimizu H: Somatic recombination underlies frequent revertant mosaicism in loricrin keratoderma. *Life Sci Alliance* 2: e201800284, 2019.
- 3) Ohguchi Y, Nomura T (equally contributed, corresponding author), Suzuki S, Takeda M, Miyauchi T, Mizuno O, Shinkuma S, Fujita Y, Nemoto O, Ono K, McLean WHI, Shimizu H: Gentamicin-induced readthrough and nonsense-mediated mRNA decay of *SERPINB7* nonsense transcripts. *J Invest Dermatol* 138: 836-843, 2018.