解剖学・発生学（高橋 智）
Anatomy and Embryology (TAKAHASHI Satoru)

TAKAHASHI Satoru, M.D.
Professor/Director
Laboratory Animal Resource Center,
Transborder Medical Research Center
University of Tsukuba

E-mail address: satoruta@md.tsukuba.ac.jp
URL: http://www.md.tsukuba.ac.jp/basic-med/anatomy/embryology/

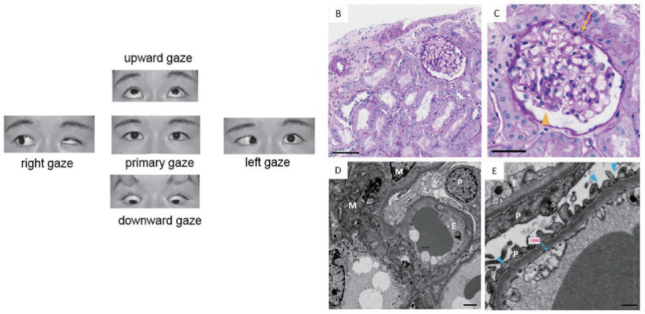
細胞分化におけるLarge Maf転写因子群の機能解析

Large Maf転写因子群は、遺伝子の発現を調節する転写調節因子ですが、ヒトおよびマウスではMafA、MafB、c-Maf およびNrl の4種類が知られています。これらの因子は、目、内耳などの感覚器、副甲状腺、膵臓などの内分泌器、骨、軟骨、筋肉などの運動器、腎臓など泌尿器系に発現しており、それらの臓器の細胞分化と機能維持に重要な働きをしていることが明らかとなっています。またこれらの遺伝子異常により、ヒト疾患が発症することも報告されています。ゲノム編集を用いた遺伝子改変マウスを用いて、Large Maf転写因子群の細胞分化と機能維持、疾患発症における機能解析を行なっています。

Functional Analysis of Large Maf Transcription Factors in Cell Differentiation

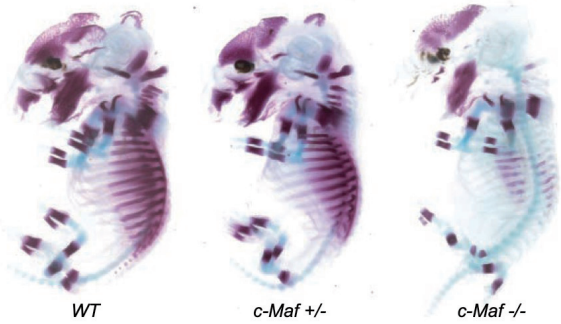
Large Maf transcription factors are transcriptional regulators that control the expression of target genes. Four factors, MafA, MafB, c-Maf and Nrl are known in both human and mouse. These factors are expressed in sensory organs such as the eyes, inner ear, endocrine organs such as parathyroid, pancreas, exercisers such as bones, cartilage and muscles, urinary system such as kidney, and are used for cell differentiation and function maintenance of these organs. It is also reported that human diseases develop due to these genetic abnormalities. Using genetically modified mice by genome editing, we are performing functional analysis of Large Maf transcription factors in cell differentiation, function maintenance and disease onset.

Duane's Retraction Syndrome and FSGS (Focal Segmental Glomerulosclerosis) caused by point mutation in DNA binding domain of MafB in Japan



(Sato Y. et al. Kidney International. 2018)

c-Maf deficient mouse displays bone formation defects



(Nishikawa K et al, J Clin Invest, 2010)