博 士 論 文 発 表 会 (公 聴 会) の お 知 ら せ

下記の通り博士論文発表会（公聴会）を開催しますので、お知らせいたします。

日時：7 月 31 日 13 時 00 分から

場 所：理学研究科棟 B301

タイトル：Phenotype-genotype relationships in Xenopus sox9 crispants provide insights into campomelic

dysplasia and vertebrate jaw evolution

（ツメガエルの sox9 変異体群を用いたヒト屈曲肢異形成症と脊椎動物の顎進化の研究）

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本セミナーは統合生命科学研究科セミナーとしてプログラム共同セミナーの対象です。

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要旨

Since CRISPR-based genome editing technology works effectively in the diploid frog Xenopus tropicalis, a

growing number of studies have successfully modeled human genetic diseases in this species. However,

most of their targets were limited to non-syndromic diseases that exhibit abnormalities in a small fraction of

tissues or organs in the body. This is likely because of the complexity of interpreting the phenotypic

variations resulting from somatic mosaic mutations generated in the founder animals (crispants). In this

study, we attempted to model the syndromic disease, campomelic dysplasia (CD), by generating sox9

crispants in X. tropicalis. The resulting crispants failed to form neural crest cells at neurula stages and

exhibited various combinations of jaw, gill, ear, heart, and gut defects at tadpole stages, recapitulating part of

the syndromic phenotype of CD patients. Genotyping of the crispants with a variety of allelic series of

mutations suggested that the heart and gut defects depend primarily on frame-shift mutations expected to

be null, whereas the jaw, gill, and ear defects could be induced not only by such mutations but also by

in-frame deletion mutations expected to delete part of the jawed vertebrate-specific domain from the

encoded Sox9 protein. These results demonstrate that Xenopus crispants are useful for investigating the

phenotype-genotype relationships behind syndromic diseases and examining the tissue-specific role of each

functional domain within a single protein, providing novel insights into vertebrate jaw evolution.

※学外者の方で博士論文発表会に参加を希望される場合は、事前に、理学系支援室

(ri-gaku-sien@office.hiroshima-u.ac.jp)まで連絡してください。事前に連絡が無い場合は、参加できません。