

# 大学院特別講義

(医歯学先端研究特論) (生命理工学先端研究特論)

(医歯理工学先端研究特論)

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記

**演題: Fuz mutant mice reveal shared mechanisms between ciliopathies and FGF syndromes**

**講師: Dr. Karen Liu**

**(Craniofacial Development & Stem Cell Biology)**

**日時: 平成 25 年 5 月 10 日 (金)**

**16 時 00 分 ~ 18 時 00 分**

**場所: 共用セミナー室 11 (M&D タワー6 階)**

**講演要旨:** Ciliopathies are a broad class of human disorders, with craniofacial dysmorphology as a common feature. Among these is high arched palate, a condition that affects speech and quality of life. Using the ciliopathic Fuz mutant mouse, we find that high arched palate does not, as commonly suggested, arise from midface hypoplasia. Rather, increased neural crest expands the maxillary primordia. In Fuz mutants, this phenotype stems from dysregulated Gli processing, which in turn results in excessive craniofacial Fgf8 gene expression. Accordingly, genetic reduction of Fgf8 ameliorates the maxillary phenotypes. Similar phenotypes result from mutation of oral-facial-digital 1 (Ofd1), suggesting that aberrant transcription of Fgf8 is a common feature of ciliopathies. High arched palate is also a prevalent feature of fibroblast growth factor (FGF) hyperactivation syndromes. Thus, our findings elucidate the etiology for a common craniofacial anomaly and identify links between two classes of human disease: FGF-hyperactivation syndromes and ciliopathies.

**【連絡先】 分子発生学分野 井関祥子 (内線 5579)**