医学セミナー・第 16 回分子遺伝疫学セミナー
“Seminars in Medical Sciences” Lecture

ゲノム医科学リサーチユニット　難治性免疫疾患・アレルギーリサーチユニット

Whole-genome mutational landscape and characterization of noncoding and structural mutations in liver cancer

Speaker: **Dr. Akihiro Fujimoto** (Associate Professor, Department of Drug Discovery Medicine, Graduate School of Medicine, Kyoto University)
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Date:　January 11, 2018 (Thurs)
Time:　17:00-18:15
Venue: Innovation Building, 8F Auditorium （健康医科学イノベーション棟8階講堂）

This seminar is one of the seminars for the subject “Seminar in Medical Sciences” in Doctoral Programs in Biomedical Sciences and Clinical Sciences. The seminar will be given in English, but questions in Japanese are also welcome.

Liver cancer, which is most often associated with virus infection, is prevalent worldwide, and its underlying etiology and genomic structure are heterogeneous. Here we provide a whole-genome landscape of somatic alterations in 300 liver cancers from Japanese individuals. Our comprehensive analysis identified point mutations, structural variations (STVs), and virus integrations, in noncoding and coding regions. We discovered recurrently mutated coding and noncoding regions, such as long intergenic noncoding RNA genes (*NEAT1* and *MALAT1*), promoters, and regulatory regions. STV analysis found a significant association with replication timing and identified known (*CDKN2A*, *CCND1*, *APC*, and *TERT*) and new (*ASH1L*, *NCOR1*, and *MACROD2*) cancer-related genes that were recurrently affected by STVs, leading to altered expression. These results emphasize the value of whole-genome sequencing analysis in discovering cancer driver mutations and understanding comprehensive molecular profiles of liver cancer, especially with regard to STVs and noncoding mutations.

References

1. Fujimoto A, Furuta M, Totoki Y, Tsunoda T, Kato M, et al. (2016) Whole genome mutational landscape and characterization of non-coding and structural mutations in liver cancer. **Nat Genet** 48: 500-509
2. Fujimoto A, Totoki Y, et al. (2012) Whole-genome sequencing of liver cancers identifies etiological influences on mutation patterns and recurrent mutations in chromatin regulators. **Nat Genet** 44: 760-76
3. Fujimoto A, et al. (2010) Whole-genome sequencing and comprehensive variant analysis of a Japanese individual using massively parallel sequencing. **Nat Genet** 42: 931–936

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