The 155th RIKEN BRC SEMINAR

Since 1917 BRC

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バイオリソースセンター1階　森脇和郎ホール

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The International Mouse Phenotyping Consortium (IMPC):
A comprehensive catalogue of gene function for the mammalian genome

The function of the majority of the genes in the human and mouse genomes remains dark. A major challenge for biomedical sciences is to build a comprehensive understanding of gene function that will support studies of rare and common disease and underpin advances in precision medicine. The International Mouse Phenotyping Consortium (IMPC) is building a catalogue of mammalian gene function by generating and phenotyping a knockout mouse line for every protein-coding gene. To date, 8,000 knockout mouse lines, many for poorly understood genes, have been generated and over 6,000 phenotyped in a coordinated effort involving more than a dozen global research centers and dedicated publicly-available online resources. Using a standardized adult phenotyping pipeline, centers test each mouse for more than 250 phenotypic parameters covering all major organ and disease systems. In addition, over 1,000 embryonic lethal mouse lines have been analyzed in a specialized embryonic development pipeline that uses high-resolution 3D imaging. All data is quality controlled and analyzed by a dedicated informatics consortium and all abnormal phenotypes automatically compared to clinical features of human disease populations to identify robust mouse models of disease. We present our discoveries into the enrichment of human Mendelian disease genes among the embryonic lethal strains, the pervasive and wide-ranging sexual dimorphism of phenotypic traits in both wild-type and mutant mice, and the over 300 new mouse models of human disease now available for further studies. The latest findings in relation to metabolism, hearing and aging will also be presented. The plethora of new genetic disease models as well as the basic and translational knowledge that has arisen from our analysis is being applied in collaboration with rare disease, biobank and other consortia to provide a more profound understanding of the function of human genetic variation.

当セミナーは、学生、研究者、技術者を対象としたものです。
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研究機関が発行する身分証をご持参になり、守衛所にて
入構証をお受け取りください。

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