Introduction to Mammalian Genome Special Issue: Mammalian Genetic Resources

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We are delighted to introduce a new special issue of Mammalian Genome—**Mammalian Genetic Resources**. Resources in the field of mammalian genetics are key to fundamental discoveries and advances in biological and biomedical sciences, which themselves underpin transformative developments in genomic and precision medicine. The emergence and awareness of mammalian genetic resources, from archives to data, is an important aspect of Mammalian Genome's mission to publish outstanding science in mammalian genetics which impacts upon *Precision and Functional Genomics*.

This is the first of an annual series. Each year we plan updates in a number of areas, while at the same time introducing new material, covering novel emerging resources with wide impact as well as updating critical developments in existing resources. We look forward to the annual special issue becoming a favoured source of information for researchers in genetics and genomics.

This issue incorporates review articles and commentaries on new developments along three major themes:

- *Data and data resources*—including updates on major data resources and novel analyses of existing datasets
- *Guidelines, protocol standards and nomenclature*
- *Genetic resources*—encompassing reports from archives, to large-scale functional genomics programmes, to tissue banks

In this year's issue, we are publishing a total of 23 articles, including one research contribution that bears upon the future of mouse genetic tools.

Data and data resources

Genomic and genetic datasets from mammalian model organisms are increasingly comprehensive and complex, often encompassing high-dimensional data. A number of articles address developments in data science across mammalian species. Ringwald et al. summarise the latest developments from Mouse Genome Informatics (MGI) at the Jackson laboratory, a vital global data source. Long et al. discuss recent progress in the annotation of the mouse genome, including the impact of techniques such as single-cell sequencing and the assessment of 3D genome architecture. In a related article, Takada et al. present the MoG + dataset and developments in cataloguing genomic variants across ten inbred strains from three key mouse subspecies used in biomedical research. The Multiple Genome Viewer reported by Richardson et al. is a valuable tool for visualisation and comparison of multiple annotated genomes, including subspecies of mice as well as other mammals. The latest Cre drivers are integral to mouse genetic studies, and Perry et al. review the CrePortal resource which provides expression and activity data for recombinase alleles. Kladunski et al. review the Rat Genome Database (RGD), the key online resource for genetic and physiologic data, and its integration with human, mouse and other human disease model

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datasets. Timmermans and Libert present MousePost and RatPost, which catalogue and provide searchable datasets of coding variants in the mouse and rat. Szpirer reports on and discusses an updated inventory of rat genes identified as underlying diseases or playing a role in critical biological processes that are altered in disease. Finally, Rogers discusses the many data resources, including the Macaque Genotype and Phenotype Database, for Rhesus macaques (*Macaca mulatta*), which are one of the most extensively studied non-human primates.

Guidelines, protocol standards and nomenclature

Guidelines, protocol standards and nomenclature are important pillars for ensuring reproducibility and robustness in biomedical research. We include three reviews that address critical areas in ensuring high standards of model organism research. Birling et al. discuss the need to ensure continuity of quality as biological materials are transferred between laboratories and the various aspects of laboratory animal quality that should be confirmed when sharing rodent research models. On a parallel note, Perry and Smith emphasise the importance of unique, concise and informative nomenclature, which is also a vital plank in mouse genetics research and the assurance of reagent quality and identity. Robust and reproducible phenotyping data are paramount in ensuring the translatability of model organism research and Ehlich et al. from the INFRAFRONTIER consortium set out a comprehensive list of quality principles that should be applied when undertaking comprehensive, systemic phenotyping.

Genetic and genomics resources

The expansion and applicability of genetic and genomics resources across diverse model organisms continues unabated—a reflection of their utility in addressing the many challenges that are faced by genomic and precision medicine. In the mouse, the diversity of genetic resources and the continued expansion of functional genomics programmes are an exemplar of the global enthusiasm for model organisms as they contribute to progress across the translational spectrum from studies of gene function to preclinical studies and the development of novel therapeutics. The International Mouse Phenotyping Consortium (IMPC) has been in the vanguard of producing a comprehensive catalogue of mammalian gene function through generation and characterisation of a mutant mouse null resource for all mouse–human orthologues. Peterson and Murray report that the community is well over half the way in this endeavour and they find

that less than 3400 mouse–human orthologues remain to be studied to complete the overall goal of complete functional annotation of the protein-coding portion of a mammalian genome. In a related article, Muñoz-Fuentes et al. assess the impact of the broad-based phenotyping pipeline employed in IMPC and the phenotype data produced as a resource for uncovering pleiotropy and co-morbidities and multi-morbidities in disease. Ju et al. report on the latest progress from the National Resource Centre for Mutant Mice in China and the development of knockout strains, floxed strains and a new panel of Cre drivers. Of course, null mutants represent only one allelic variant for uncovering gene–phenotype relationships, and reflecting on this, Tosh et al. review the availability of mouse models of aneuploidies for understanding human chromosome disorders, focussing particularly on efforts to develop and characterise mouse models of Down syndrome. Ju et al. have also contributed a research article on the generation of full-length balancer chromosomes using Cre/loxp recombination. While balancer chromosomes carrying partial inversion of a chromosome have been reported before, large-fragment balancers are able to maintain mutations distributed across entire chromosomes.

The archiving and distribution of these myriad genetic resources require significant national and regional repositories which have the scale and expertise to serve the wider biomedical sciences community. In this special issue, we include reports from three major global repositories for mouse resources. At the RIKEN BioResource Research Centre, Japan, Mizuno-Iijima et al. report on the mouse strains available through the National BioResource Project core facility. The RIKEN facility is part of the wider Asian Mouse Mutagenesis Resource Association (AMMRA) and Chin et al. report on the wider activities of AMMRA, not only in the provision of resources, but also in areas such as mutagenesis and phenotyping, and promoting mouse genetics research across Asia and Australia. Amos-Landgraf et al. report on the Mutant Mouse Resource and Research Center (MMRRC) Program, the pre-eminent public mutant mouse repository and distribution archive in the US, a regionally distributed effort comprising several vivaria and an Informatics Coordination and Service Centre.

Finally, we include three articles on genetic resources for model organisms that play an increasingly important and critical role in discovery and translational research. Genomics and genetics studies in dogs continue to have a profound impact on translational research and Buckley et al. review best practices for the imputation of genotypes in low-pass sequencing from dogs. Turning to pigs, Boettcher et al. review the landscape of swine models for translational oncology research and their role in preclinical therapeutic and device testing. Considering again the important role of non-human primates in biomedical research, Witham and

Wells discuss the utility and development of non-human primate tissue banks.

We hope that you enjoy reading and reflecting on these articles, which provide a timely perspective to current developments in mammalian genetic resources. We will be very glad to hear from you as we develop the second special issue in this series, and to consider reviews and commentaries that explore new areas, as well as articles that provide important updates on areas that have already been covered. We look forward to hearing from you.

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