PRESS RELEASE –WHITE PAPER

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EXCELLENCE IN MOUSE PHENOGENOMICS

Date: 2020, Feb 4th

DEEP GENOME PROJECT: Mouse genetics is vital for understanding human disease and the future of healthcare

Only 3-5% of our DNA code is used for making proteins. Researchers are beginning to learn and understand what the other 95% is used for. But significant portions of the human genome, including much of the DNA that codes for proteins, are still 'dark' and not understood. These 'dark' genes are understudied, so their functions and links to disease remain unknown. This lack of knowledge limits developments in medicine and prevents us from producing new drugs that target disease-linked proteins and finding new genetic markers that could assist in early diagnoses, even before symptoms start to show.

An editorial published in ***Genome Biology*** and authored by 44 leading scientists, clinicians, and academics in 15 countries on 5 continents, is raising awareness of how limited our understanding is of the function of the majority of our genes and their products. These experts are working to catalogue all the genes in the mammalian genome, as well as their function and physical effects, to improve our understanding of disease and enable the discovery of the next generation of therapies.

The authors propose a **Deep Genome Project** to illuminate the 'dark' portions of the genome and provide a database that lists all mouse genes that are related to human genes, their functions and their role in disease. They propose four steps for the global mouse genetics research community to develop and deliver a better understanding of the genome, and to continue to improve the current resources that researchers and clinicians use for the study of disease, efficient diagnoses, development of treatments, and improved patient care.

The **first** step is to study the section of the mouse genome that produces proteins, which is only 3-5% of mouse DNA, describing which gene produces which protein and what happens when that protein stops working. The International Mouse Phenotyping Consortium (IMPC) is working to do this by turning off each gene and then studying the physical and chemical changes in a mouse. By 2021 over 9,000 mouse genes, around half the genome, will have been analysed and it is vital that this is completed in order to fully understand the impact of genetics on disease.

The **second** step is to target the noncoding section of the genome. This is the other 95% of the mouse genome. We know that DNA in the noncoding genome has important roles and can also have a significant effect on how genes function. This will help us understand how abnormalities in the noncoding genome can cause or contribute to disease.

The **third** step is to turn the genetic information and knowledge produced into clinical knowledge. Doctors, Specialists, Clinician-Scientists, and Researchers anywhere in the world will be able to use this information to study the role of genes in health and disease and find new targets for therapies.

The **fourth** step is to ensure fast and easy access to this information so that it can be integrated in the clinical decision-making process. By streamlining the production and analysis of these mouse models, clinicians could diagnose patients more easily and administer targeted therapies with a better chance of being effective early in a patient's course of disease.

The authors highlight that achieving and completing this Deep Genome Project will require continued funding, global collaboration and support in scientific research. To achieve it could transform biology, medicine, and global health.

Professor Steve Brown, Chair of the IMPC Steering Committee, and Professor Kent Lloyd, University of California at Davis, both senior authors on the paper, said: *"This call for a deep genome project echoes many of the strategic aims of the International Mouse Phenotyping Consortium (IMPC). The IMPC is already shining a powerful light on the function of the dark genome and its role in disease. We urgently build on this programme and undertake an even deeper and more extensive analysis of the mouse genome."*

Free access to the white paper

Lloyd, K.C.K., Adams, D.J., Baynam, G. et al. The Deep Genome Project. Genome Biol 21, 18 (2020).

Additional information:

Mouse models are of critical importance in studying and understanding the human genome. We share 97% of our DNA with mice and the mouse is a powerful tool using gene editing techniques for analysing the function of genes and their role in disease. The development of mouse models generated by research centres, such as those in the IMPC, is vital to inform our understanding of the impact of human genetic variation and its relationship to disease.

The **International Mouse Phenotyping Consortium, IMPC (www.mousephenotype.org)** comprises 20 of the leading mouse genetics centres worldwide. IMPC has been developing a complete functional catalogue of the mouse genome, linking each gene to disease, enabling a better understanding of how genetic variation in the human population causes disease and identifying new targets for therapeutic intervention.

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