Motivation

The 2025 Lombardy Prize, under the theme "Innovative Methods of Early Diagnosis or Preventive Medicine," was jointly awarded to **Professor Mark Caulfield** and **Professor Douglas Easton** for their fundamental research in genomic medicine, which led to the identification of genes associated with increased susceptibility to various diseases, particularly cancer and cardiovascular disease. The awardees have pioneered the translation of genomic technologies impacting on early diagnosis, disease prevention and precision medicine.

Professor Sir Mark Caulfield (London, UK) has made outstanding progress in identifying genes that increase susceptibility to cardiovascular diseases. He inspired the scientific strategy and led the creation of the 100,000 Genomes Project for whole-genome sequencing, with over 90,000 participants. The 100,000 Genomes Project's findings include the discovery of over 2,000 gene regions that influence blood pressure and cardiovascular disease and can be used to predict the risk of adverse cardiovascular outcomes. Upon clinical trials, some findings have changed clinical practice, including international guidelines for the primary prevention of hypertension and heart attack. The 100,000 Genomes Project has successfully advanced its ambitious goal of bringing the most advanced genomic technologies to the patient's bedside. Professor Caulfield's philosophy was to democratize access to clinical data and whole genome sequencing, creating a data center that would allow a coalition of over 3,400 international scientific researchers from 33 countries to democratically access the 100,000 Genomes datasets. His commitment and vision have had a transformative impact, with the integration of the Genomic Medicine Service into the UK's National Health Service: the UK boasts the first free system of genomic analysis for diagnosis and precision medicine. The “Genomic Analysis in Children Group” and the “Generation Study” initiatives have led to a broad newborn screening program through genome sequencing for the early identification and diagnosis of potential avoidable harm in children, with the aim of reducing morbidity and mortality.

Professor Douglas Easton (Cambridge, UK) is a genetic epidemiologist who has made fundamental contributions to the understanding of genetic susceptibility to cancer, with a particular focus on breast, ovarian and prostate cancer. He has been a pioneer in the genetic epidemiology of cancer-predisposing genes (including BRCA1 and BRCA2) and in the development of statistical methods.
He has coordinated international consortia that have conducted large-scale genetic studies to identify and characterize gene variants associated with cancer susceptibility risk; these multi-omics data are also useful for identifying biomarkers for cancer onset (early diagnosis) and potential drug targets. He and his collaborators have developed important and widely used cancer risk prediction models, such as BOADICEA (Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm), a risk model for familial breast and ovarian cancer, two cancers with high mortality in women. The BOADICEA algorithm has received regulatory approval in the EU and UK and is currently used worldwide for genetic screening and risk management counselling. His studies on cancer predisposition genes have advanced our understanding of genetic cancer susceptibility, informing early diagnosis strategies and enhancing the effectiveness of preventive medicine.

Professor Caulfield and Professor Easton are world leaders in the discovery and population-level translation of genomic medicine in cardiovascular disease and various cancers. They have pioneered and implemented genetic risk models that enable truly personalized preventive screening to reduce disease incidence and mortality, improve quality of life and reduce global healthcare costs.

Short for communication

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