

SG10K Pilot approved studies- updated as of 06 February 2024

| No. | Title of research | Lead Investigator | Institution | Date of approval |
|------------|--|--------------------------|--|-------------------------|
| 1 | Whole-genome reference panel of ~20,000 individuals | Sunjae Kim | Department of Biomedical Science, Seoul National University, Precision Medicine Institute at Macrogen, Korea | 7 Feb 2020 |
| 2 | Identification and functional annotation of disease-causal variants under positive selection | Mulin Jun Li | School of Basic Medical Sciences, Tianjin Medical University, China | 7 Feb 2020 |
| 3 | Identifying causal mutations for craniofacial microsomia | Yongbiao Zhang | BUAA-CCMU advanced innovation center for big-data based precision medicine, Beihang | 7 Feb 2020 |
| 4 | Reference panel of Chinese population based on whole genome sequencing data | Xin Jin | BGI-Shenzhen, University of Chinese Academy of Sciences, China | 7 Feb 2020 |
| 5 | Molecular basis and mechanisms underlying human spermatocyte developmental arrest (SDA) | Yuanwei Zhang | Life Science School, University of Science of Technology of China, China | 7 Feb 2020 |
| 6 | Genomic mutation profile of acute lymphoblastic leukemia | Jinyan Huang | Shanghai Institute of Hematology, Ruijin Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai, China | 7 Feb 2020 |
| 7 | Identification of drug pathways associated with potentially functional polymorphisms in the Singapore population | Caroline Lee Guat Lay | Department of Biochemistry, National University of Singapore, Singapore | 7 Feb 2020 |
| 8 | Diagnosis of neurodevelopmental disorders in India | Katta Mohan Girisha | Department of Medical Genetics,, Kasturba Medical College, Manipal, India | 7 Feb 2020 |
| 9 | Enhancing common variant imputation via RICOPILI | Stephan Ripke | Stanley Center for Psychiatric Research, The Broad Institute of Harvard and MIT; Department of Psychiatry and | 7 Feb 2020 |
| 10 | Identification of new susceptibility loci for complex traits in Han Chinese by imputing genotypes from Asian- | Hou-Feng Zheng | Westlake University, China | 7 Feb 2020 |

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| 11 | Incidental “causative” variants for inherited cardiac conditions in a Singaporean whole genome sequencing cohort. | Roger SY Foo | National University Heart Centre and Cardiovascular Research Institute, National University of Singapore, Singapore | 7 Feb 2020 |
| 12 | Genomic sequencing analysis on candidate genes for pemphigoid disease | Yonghu Sun | Shandong Provincial Institute of Dermatology and Venereology, China | 7 Feb 2020 |
| 13 | Determining the Carrier Frequency and Genetic Prevalence of Inherited Retinal Diseases | Hwei Wuen Chan | Department of Ophthalmology, National University of Singapore, Singapore | 16 Mar 2021 |
| 14 | Population genetics and rare developmental disorder risk in South Asians | Hilary Martin | Wellcome Sanger Institute, United Kingdom | 19 Mar 2020 |
| 15 | Somatic mutation in regulatory region of cancer genome | San Ming Wang | Faculty of Health Sciences, University of Macau, Avenida da Universidade Taipa, Macau, China | 2 Apr 2020 |
| 16 | Discovering Taiwanese-specific variants by comparing Taiwanese WGS with other Asian populations | Chien-Yu Chen | Department of Biomechatronics Engineering, National Taiwan University, Taiwan | 2 Apr 2020 |
| 17 | Population diversity in pharmacogenomic variants | Jun J. Yang | Department of Pharmaceutical Sciences, St Jude's Children's Research Hospital, Memphis, USA | 19 Apr 2021 |
| 18 | GWAS/WGS Analyses of Primary aldosteronism and related conditions in Malaysian Populations | Mun-Kit Choy | Division of Cardiovascular Sciences, The University of Manchester, United Kingdom | 17 May 2020 |
| 19 | Human GRIN gene mutations | Chian Ming Low | Pharmacology, National University of Singapore, Singapore | 3 Jul 2020 |
| 20 | Germline variants that are associated with cancer risk in Asian populations | Melvin Chua Lee Kiang | Division of Radiation Oncology, National Cancer Centre, | 18 Aug 2020 |

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| 21 | Comparison of genotype imputation of South East Asian data on various imputation software and reference panels | Yik Ying Teo | Saw Swee Hock School of Public Health, National University of Singapore, Singapore | 18 Aug 2020 |
| 22 | Variant landscape of RYR1 and CACNA1S gene based on whole genome sequencing of Singaporean population | Jian Jun Liu | Genome Institute of Singapore, Singapore | 15 Sep 2020 |
| 23 | Building databases of genomic variants for Vietnamese population | Duc-Hau Le | Department of Computational Biomedicine, Vingroup Big Data Institute, Vietnam | 16 Oct 2020 |
| 24 | Admixture history of Singapore Peranakan Chinese revealed by whole genome sequencing analysis | Roger Foo | Department of Medicine / National University Health System and Genome Institute of Singapore, Singapore | 16 Oct 2020 |
| 25 | MMR gene variation in ethnic Chinese population | San Ming Wang | Faculty of Health Sciences, University of Macau, Avenida da Universidade Taipa, Macau, China | 16 Oct 2020 |
| 26 | WeGene Imputation Reference Panel Project | Lizhong Wang | WeGene, Shenzhen, Guangdong, China | 15 Jan 2021 |
| 27 | Combining Short And Long-read Sequencing To Detect Noncoding And Structural Variants, And Haplotypes In Cone/ROD Dystrophy Patients | Rui Chen | Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, USA | 22 Feb 2021 |
| 28 | Improve genetic risk prediction on individuals of South and East Asian ancestries for common and actionable diseases. | Michael Simpson | Genomics plc, United Kingdom | 19 Apr 2021 |
| 29 | Data-driven analysis of carrier frequencies of autosomal recessive and X-linked diseases in the Asian population | Shen Gu | School of Biomedical Sciences, The Chinese University of Hong Kong, Hong Kong | 19 Apr 2021 |
| 30 | Genetic diversity of East Asian populations | Shuhua Xu | Key Laboratory of Computational Biology, Shanghai Institute of Nutrition and Health, University of Chinese | 19 Apr 2021 |

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| 32 | Exploring allelic architecture and sub population structure among South Asians | Salih Tuna | Global Gene Corporation Pte Ltd, Singapore | 16 Jul 2021 |
| 32 | A genome-wide association study for Parkinson's disease in the Asian population | Huifang Shang | Department of Neurology, West China Hospital, China | 16 Jul 2021 |
| 33 | Carrier frequencies for Hereditary Cancer and Inherited Cardiac Diseases in Singapore population | John Whay Kuang Chia | Curie Oncology Pte Ltd, Singapore | 16 Jul 2021 |
| 34 | Effective Imputation of Clinical Trials Data | Sarah Pendergrass | Genentech | 16 Sep 2021 |
| 35 | Tracing the evolution and migration history of the indigenous population from pennisular Malaysia | Boon Peng Hoh | UCSI University, Malaysia | 18 Feb 2022 |
| 36 | Genetic adaptations to environments across Eurasia | Anders Eriksson | University of Tartu, Estonia | 18 Apr 2022 |
| 37 | Whole-genome Sequencing Based Blood Group Antigen Automated Typing | JiaHai Shi | Synthetic Biology Translational Research Program, Yong Loo Lin School of Medicine, National University of Singapore | 21 Jun 2022 |
| 38 | High coverage whole genome sequencing (WGS) reveals insights into the historical migration of Malays population | Rozaimi Razali | Department of Biomedical Science, College of Health Sciences, Qatar University | 20 Dec 2022 |
| 39 | Investigation of genomic mechanism of diabetic kidney disease using whole genome sequencing data from multi-ethnic type 2 diabetic cohorts | Enrico Petretto | Duke-NUS Medical School, Singapore | 31 Mar 2023 |
| 40 | Timing of driver mutations for liver fluke associated cholangiocarcinoma in Asia | Thomas Crellen | The University Court of the University of Glasgow | 15 May 2023 |
| 41 | Investigating potential disease-related rare genetic variants in Parkinson's disease patients of South East Asian ancestry in Malaysia | Azlina Ahmad Annuar | University of Malaya, Malaysia | 15 Aug 2023 |

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| 42 | Elucidation of disease pathology by multi-omics analysis- homogeneity and heterogeneity of selection signatures in diverse populations | Yukinori Okada | Osaka University Graduate School of Medicine, Japan | 15 Aug 2023 |
| 43 | Identifying the functional consequences of genetic variation in Type 2 Diabetes Mellitus Susceptibility in Asia | John Grealley | Albert Einstein College of Medicine, USA | 4 Dec 2023 |
| 44 | Genetic investigation of common epilepsies in the East and Southeast Asian populations | Yen-Chen Anne Feng | Institute of Health Data Analytics and Statistics, College of Public Health, National Taiwan University, Taiwan | 4 Dec 2023 |
| 45 | Genome wide association studies of schizophrenia in South/Southeast Asian populations | Sathish Periyasamy | The University of Queensland, Australia | 26 Jan 2024 |