CATALOGUE OF GLOBAL GENOMIC MEDICINE IMPLEMENTATION INTIATIVES

Policy / Implementation Frameworks

Title	Human Heredity and Health in Africa (H3Africa) Whitepaper
Type of initiative	Policy / Implementation Framework
Geographic Region	Africa
Date	2011
Description	
Policy Issues considered	▪ Collaborative Centers ▪ Research Projects ▪ Bioinformatics Networks ▪ Biorepositories ▪ Societal Implications research
Website	H3Africa Whitepaper

Title	National Health Genomics Policy Framework
Type of initiative	Policy / Implementation Framework
Geographic Region	Australia
Date	2017 – 2020
Description	
Policy Issues considered	▪ Person-Centred Approach ▪ Genomics literate workforce ▪ Sustainable and strategic investment in cost-effective genomics ▪ Maximising quality, safety and clinical utility of genomics in health care ▪ Responsible collection, storage, use and management of genomic data Along with enablers: ▪ Collaborative governance and leadership ▪ Stakeholder engagement ▪ National and international partnerships
Website	Australian National Health Genomics Framework Draft Implementation Plan

Title	NSW HEALTH – Genomics Strategy
Type of initiative	Policy / Implementation Framework
Geographic Region	Australia - New South Wales
Date	2017
Description	
Policy Issues considered	▪ Leadership and governance ▪ Focussed application: clinical need, validity and utility ▪ Service delivery: commissioning and utilising genomic technology ▪ Genomic data and infrastructure: handling “big data” ▪ Preparing the workforce for genomics ▪ Community engagement: maintaining public trust and confidence
Website	NSW Health Genomics Strategy

Title	Genetic and Genomic Healthcare for Victoria 2021
Type of initiative	Policy / Implementation Framework
Geographic Region	Australia - Victoria
Date	2017
Description	Including genomic information into routine healthcare requires additional work such as: ▪ strengthening the healthcare system so that Victorians, regardless of their age, location or background, benefit from safe, fast and fair inclusion of genomic information into routine healthcare ▪ building trust so that Victorians are confident they are being provided with the best possible care and that their and their family's genomic information will be handled and used in accordance with their wishes ▪ raising awareness about the use of genomic information in healthcare, its benefits and limitations ▪ growing knowledge so that Victoria is a leader in using genomic information in routine healthcare
Policy Issues considered	Four priorities identified for action in the first 12 to 24 months are: ▪ developing and implementing a state-wide genetic and genomic services plan to ensure more equitable access to appropriate and sustainable services ▪ establishing a genomic health clinical network to improve the safe and fair adoption of genomic healthcare practice by the health workforce ▪ undertaking community consultations to address some of the key ethical, legal and social issues associated with including genomic information into routine healthcare to inform Victorian Government policy and funding decisions ▪ reducing superbugs and improving detection of infectious disease outbreaks through strengthening of microbial genomics activities in Victoria to improve the health of Victorians
Website	Genetic and Genomic Healthcare for Victoria 2021

Title	Genomics and public health in Belgium
Type of initiative	Policy / Implementation Framework
Geographic Region	Belgium
Date	2010
Description	Information translated from French
Policy Issues considered	▪ Structure of translational research in Belgium ▪ Translational research and its funding ▪ Promoting Collaboration in Translational Research ▪ Review and evaluation of clinical utility and validity of genetic testing ▪ Regulation of genetic testing ▪ Encouraging innovation in the biotechnology and healthcare sectors ▪ Integrating genomics into everyday practice
Website	Genomics and Public Health in Belgium

Title	Genome Canada Strategic Plan 2012-17
Type of initiative	Policy / Implementation Framework
Geographic Region	Canada
Date	2012-2017
Description	Plan is broader than medicine and health but objectives apply to health as well as other sectors
Policy Issues considered	▪ Respond to societal needs by generating discoveries and accelerating their translation into applications ▪ Attract greater investment in genomics research from a broad range of stakeholders, in particular the private sector ▪ Enhance the impact of genomics by transforming knowledge of the ethical, environmental, economic, legal and social challenges and opportunities into sound policies and practices ▪ Enhance recognition of the value of genomics by increasing stakeholder appreciation of genome science, its applications and its implications
Website	Genome Canada Strategic Plan

Title	Genome BC - A Genomics Strategy for British Columbia's Health Sector
Type of initiative	Policy / Implementation Framework
Geographic Region	Canada - British Columbia
Date	2015
Description	
Policy Issues considered	▪ Scientific understanding and technical capabilities ▪ Alignment and prioritization ▪ Health information technology (IT) infrastructure, tools, and governance ▪ Clinical and laboratory infrastructure and capabilities ▪ Regulatory guidelines ▪ Harmonized ethical consent ▪ Privacy and anti-discrimination policies and legislation ▪ Reimbursement guidelines ▪ Healthcare professional capacity, awareness, training and adoption
Website	Genome BC Health Sector Strategy

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Title	Strategic Development of Genomic Medicine in Hong Kong (the Strategy)
Type of Initiative	Policy / Implementation Framework
Geographic Region	Hong Kong – S.A.R, China
Date	2020
Description	A blueprint to drive the local development of genomic medicine in order to harness its huge potential in precise diagnoses, personalised treatment and surveillance of diseases.
Policy Issues considered	The recommendations of the strategy include: ▪ launching the Hong Kong Genome Project ▪ enhancing clinical services in genetics and genomics ▪ nurturing talents in genomic medicine ▪ enhancing public engagement in genomic medicine ▪ enhancing the laboratory network with effective referral mechanism and centralisation of advanced genetic and genomic tests ▪ facilitating the establishment of a biobank network for genomic research ▪ enhancing the regulation on use of genetic data for insurance and employment purposes ▪ promoting the proper use of genetic and genomic tests
Website	Strategic Development of Genomic Medicine in Hong Kong Hong Kong Food and Health Bureau

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Title	National Strategy for Personalised Medicine
Type of Initiative	Policy / Implementation Framework
Geographic Region	Denmark
Date	2017-2020
Description	Lays the tracks for the use of Personalised Medicine in the Danish healthcare system – and for the research that is to realise the potential of Personalised Medicine.
Policy Issues considered	▪ Transparent governance structure with nationwide involvement ▪ Clear legal framework addressing ethical principles and data privacy and security ▪ Patients and citizens must be involved ▪ A technological infrastructure with secure, efficient and equal access ▪ Genomics research must be international and deeply integrated in the healthcare system ▪ Tools and competencies to use genetic data ▪ Denmark must have an attractive development environment in relation to personalised medicine
Website	National Strategy for Personalised Medicine

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Title	Shaping Europe's Vision for Personalised Medicine Strategic Research and Innovation Agenda (SRIA)
Type of initiative	Policy / Implementation Framework
Geographic Region	Europe
Date	2015
Description	Implementation of personalised medicine as opposed to genomics specifically but similar policy issues (27 organisations from 14 countries across Europe)
Policy Issues considered	▪ Developing Awareness and Empowerment ▪ Integrating Big Data and ICT Solutions ▪ Translating Basic to Clinical Research and Beyond ▪ Bringing Innovation to the Market ▪ Shaping Sustainable Healthcare
Website	Shaping Europe's Vision for Personalised Medicine

Title	Improving Health through the use of Genomic Data
Type of initiative	Policy / Implementation Framework
Geographic Region	Finland
Date	2015
Description	
Policy Issues considered	▪ Ethical principles and legislation govern the responsible use of genomic data ▪ Genomics research is closely integrated into healthcare ▪ Healthcare professionals have the knowledge and skills to use genomic data ▪ Finland has information systems enabling the effective use of genomic data ▪ Genomic data are widely used in healthcare based on individual and population needs ▪ Individuals are able to make use of genomic data in their own lives ▪ Finland is an internationally attractive research and business environment in the field of genomics
Website	Finland's Genome Strategy

Title	The International Cancer Genome Consortium for Medicine (ICGCmed) Whitepaper
Type of initiative	Policy / Implementation Framework
Geographic Region	Global
Date	2016
Description	Implement the ambitious project of analyzing the genomes of more than 200,000 patients by the end of 2025 and linking this data to high-quality clinical information including treatment information and outcomes
Policy Issues considered	▪ Scientific and technical knowledge and standards ▪ Data Management and coordination ▪ Clinical Data coordination ▪ Ethics and Policy ▪ Communications and Outreach
Website	International Cancer Genome Consortium

Title	National Biotechnology Development Strategy
Type of initiative	Policy / Implementation Framework
Geographic Region	India
Date	2015 - 2020
Description	Plan is broader than medicine and health and genomics but there are a number of genomic medicine specific actions
Policy Issues considered	▪ Human genome research and development ▪ Availability of genome engineering technologies ▪ Bioinformatics support
Website	India National Biotechnology Development Strategy

Title	The policy of public health genomics in Italy
Type of initiative	Policy / Implementation Framework
Geographic Region	Italy
Date	2013
Description	Information translated from Italian ▪ Definition of areas of intervention of public health genomics (PHG) ▪ Collection of evidence on current PHG experience in Italy ▪ Definition of the best instruments for the promotion of genome-based knowledge among health professionals and citizens ▪ Development of evidence-based recommendations for the appropriate use of genetic testing of complex diseases ▪ Identification of the critical points for the translation of genomics into clinical practice
Policy Issues considered	▪ Define the policy and system setup ▪ Exercise Influence on Interlocutors ▪ Ensuring the implementation of policies ▪ Establish and maintain collaboration and partnerships ▪ Ensuring accountability ▪ Knowledge-Based Management
Website	Italian Public Health Genomics Policy

Title	Genomic Medicine in Korea: Plan and Infrastructure Genome Technology to Business Translation Program
Type of initiative	Policy / Implementation Framework
Geographic Region	South Korea
Date	2014-2021
Description	Plan is broader than medicine and health. It has a non-health component and infrastructure components.
Policy Issues considered	▪ Translational genomics for personalized medicine ▪ Support for translational medicine ▪ Production & utilization of Korean genomics research resources ▪ Ethics & legal & social implications (ELSI) ▪ Joint projects
Website	Genomic Medicine in Korea

Title	Thailand's National Biotechnology Policy Framework (2012-2021)
Type of initiative	Policy / Implementation Framework
Geographic Region	Thailand
Date	2012-2021
Description	Plan is broader than medicine and health and genomics
Policy Issues considered	▪ Advance critical platform technologies such as genome, nutrigenomics, pharmacogenomics, proteomics and drug discovery
Website	Thailand National Biotechnology Policy Framework

Title	Genomics Thailand
Type of Initiative	Policy / Implementation Framework
Geographic Region	Thailand
Date	2019-2023
Description	A National Strategic Initiative on Precision Medicine Strategy 1 Research Strategy 2 Service Strategy 3 Bioinformatics Information Management Strategy 4 Ethics, Law and Society Strategy 5 Human Development Strategy 6 New manufacturing economy
Policy Issues considered	▪ Research – prioritisation of research area, clinical geneticist and genetic counsellor training ▪ Informatics – high performance computing, bioinformatician training, bioinformatic pipelines, genome data analysis service ▪ Service – genetic technology assessment, quality assurance program for genetic tests, BRCA1/2, Hereditary Colon Cancer and pharmacogenomic tests, guidelines for clinical geneticists, establishment of a Society for Human Genetics, Genetics test and treatment coverage package ▪ Social and Ethical issues – Genetic non-discrimination Act, Ethical, Legal and Social implications program
Website	Genomics Thailand

Title	Building on our inheritance - Genomic technology in healthcare - A report by the Human Genomics Strategy Group
Type of initiative	Policy / Implementation Framework
Geographic Region	United Kingdom
Date	2012
Description	
Policy Issues considered	▪ Translating genomic innovation to establish clinical validity and utility ▪ Service delivery: commissioning and utilising genomic technology ▪ Biomedical informatics: underpinning genomics ▪ Preparing the workforce ▪ Developing the legal and ethical framework ▪ Engaging the public and building awareness
Website	Building on our inheritance

Title	National Human Genome Research Institute Strategic Plan - Charting a course for genomic medicine from base pairs to bedside
Type of initiative	Policy / Implementation Framework
Geographic Region	United States of America
Date	2011
Description	
Policy Issues considered	▪ Imperatives for genomic medicine - making genomics-based diagnostics routine, defining the genetic components of disease, comprehensive characterization of cancer genomes, practical systems for clinical genomic informatics, the role of the human microbiome in health and disease ▪ Bioinformatics and computational biology - data analysis, data integration, visualization, computational tools and infrastructure, training. ▪ Education and training - strengthening primary and secondary education, conducting public outreach, building healthcare providers' genomic competencies, preparing the next generation of genomics researchers ▪ Genomics and society - psychosocial and ethical issues in genomics research, psychosocial and ethical issues in genomic medicine, legal and public policy issues, broader societal issues
Website	Charting a Course for Genomic Medicine

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Demonstration Projects / alliances / initiatives

Title	Population Genome Program
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Abu Dhabi
Date	2019
Description	This program aims to be the first of its kind worldwide to provide Emirati individuals with their own high-quality genome as a baseline and incorporate genomic data into healthcare management. Purpose: ▪ to provide an Arab reference genome to assist in the health of the population ▪ to equip physicians and other healthcare practitioners with high quality information and knowledge ▪ to enable advanced diagnosis and treatment options ▪ to deliver personalized and prevention programs tailored to an individual's unique genetic makeup. The Genome Program will have two stages: a. Research Stage: That focuses on understanding the genetic variation of the population; b. Strategy Stage: That aims to deliver improved health outcomes on the basis of research insights.
Policy Issues considered	▪
Website	Population Genome Program
Cohort Size	The project will be executed in phases. An initial phase will be to sequence 100,000 samples.
Cohort Description	Local population on voluntary basis
Type of Genomic Data	Short and long read sequencing technology
Disease Area	

Title	H3Africa Initiative
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Africa
Date	2011
Description	A network of NIH and Wellcome Trust funded research sites located cross Africa. The overarching objectives of the Consortium are to foster pan-continental collaboration to nurture research and develop and support African scientists
Policy Issues considered	▪ Collaborative Centers ▪ Research Projects ▪ Bioinformatics Networks ▪ Biorepositories ▪ Societal Implications research
Website	H3Africa
Cohort Size	Some studies in H3Africa may involve 10,000 or more individuals
Cohort Description	Initially, H3Africa will support two pilot studies: interactions between disease-causing micro-organisms and human hosts and non-communicable disease. H3Africa will seek to understand the interaction between genetic susceptibilities and environmental changes — such as diet — that may be leading to the increased incidence of these diseases
Type of Genomic Data	Genomic tools will likely include genotyping chips used in genome-wide association studies, WES and eventually, WGS
Disease Area	▪ Communicable diseases (Tuberculosis, Human African Trypanosomiasis) ▪ Non-communicable diseases (Sickle Cell Disease, monogenic disorders, Stroke, Cardiovascular disease, Hyper tension, Type 2 Diabetes Mellitus and Obesity) ▪ Pharmacogenomics ▪ New and innovative ideas

Title	Centre for Arab Genomic Studies (CAGS)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Arab States
Date	2003
Description	Arab Council of CAGS was formed in 2005 and since 2014 has had representations from Bahrain, Egypt, Jordan, Kuwait, Lebanon, Oman, Qatar, Saudi Arabia, Sudan, and Tunisia
Policy Issues considered	▪ Catalogue of Transmission Genetics in Arabs (CTGA) database ▪ Communication and education through publications ▪ Research projects on genetic diseases in Arab populations such as rare skeletal abnormalities ▪ Workforce training through the Pan Arab Human Genetics Conferences ▪ Ethical. Legal and social issues
Website	Centre for Arab Genomic Studies

Title	Precision Medicine Initiative
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Argentina
Date	2017
Description	Aim was to establish the scientific know-how and protocols required to implement “omic” technologies in the clinical practice, by performing a proof of concept of the process in a small number of patients. Three grants were awarded: • Clinical Genomics of Pediatric Diseases • Creation of a National Biobank • Cancer Genomics. Projects involve both healthcare institutions harboring NGS equipment and research groups from academia This project was developed following the creation of the Argentinean Genomic and Bioinformatic Platforms in 2012. This 4-year project, with the help and financing of the National Ministry of Science and Technology (MINCyT), aimed to develop and transfer genomic and bioinformatic knowledge as well as facilitate those services to public and private parties. During this period the first three whole genomes were entirely sequenced. Following this in 2016, another project was launched to sequence 100 exomes of patients with rare genetic disorders throughout the country, “100 Exomes Campaign”.
Policy Issues considered	▪ Development of targeted sequencing panels ▪ Biobanking
Website	Argentina Precision Medicine Initiative Earlier project 100 exomes Project
Cohort Size	1,000 (Precision Medicine Initiatives)
Cohort Description	
Type of Genomic Data	
Disease Areas	Pediatric and cancer

Title	Australian Genomics Health Alliance
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Australia
Date	2016 – 2020
Description	AGHA funding of AUD$25 million (NHMRC Grant)
Policy Issues considered	▪ A national diagnostic and translational research network ▪ A national approach to data federation and analysis ▪ Economic analysis and policy implications for the health system ▪ Genomic workforce, education and ethics
Website	Australian Genomics
Cohort Size	Approx. 4000 in 5 years
Cohort Description	Patients from across Australia are being recruited into Flagship projects, and the data from their clinical care will drive national diagnostic networks and provide evidence on the benefits of genomic medicine
Type of Genomic Data	Targeted gene panels, WES, WGS, RNA sequencing, Mitochondrial DNA sequencing (different clinical flagship projects have different approaches)
Disease Areas	▪ Rare Diseases ▪ Cancer

Title	Sydney Genomics Collaborative
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Australia - New South Wales
Date	2014
Description	
Policy Issues considered	
Website	Sydney Genomics Collaborative
Cohort Size	Approx. 8,295
Cohort Description	▪ Medical Genome Reference Bank - approximately 4,000 whole genome sequences from healthy, aged people to be used for control purposes in disease-specific genomic research ▪ NSW Genomics Collaborative Grants – funding to undertake whole-genome sequencing to improve understanding of the genetic causes of disease ▪ Genomic Cancer Medicine Program – a research program dedicated to applying genomics to the understanding, early detection, prevention and management of cancer including the Cancer Molecular Screening and Therapeutics (MoST) Program and Genetic Cancer Risk in the Young Study
Type of Genomic Data	WGS
Disease Areas	▪ Schizophrenia ▪ Metastatic melanoma, ▪ mitochondrial disease ▪ congenital heart disease ▪ mendelian ▪ epilepsy ▪ rare disease ▪ blinding retinal dystrophy ▪ genetic disorders of bone ▪ inherited cardiomyopathies

Title	Queensland Genomics Health Alliance (QGHA)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Australia - Queensland
Date	2016
Description	
Policy Issues considered	▪ Workforce Development ▪ Evaluation of Clinical Genomics ▪ Genomic Testing Innovation ▪ Genomic Information Management ▪ Ethics, Legal and Social Implications of Genomics
Website	Queensland Genomics
Cohort Size	
Cohort Description	Patients will be recruited across a range of clinical demonstration projects
Type of Genomic Data	various
Disease Areas	Round 1 disease areas are: ▪ Melanoma ▪ Lung cancer ▪ Hospital acquired infections ▪ Maturity onset diabetes of the young (MODY)

Title	SA Genomics Health Alliance
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Australia – South Australia
Date	2017
Description	
Policy Issues considered	▪ Develop an interactive genomics network with a visible state and national presence ▪ Ensure genomics services provided in South Australia are state of the art ▪ Integrate genomics into existing and future research and innovation activities ▪ Educate patients, healthcare professionals and researchers about genomics ▪ Reduce the barriers to accessing genomics technologies and support their implementation in healthcare
Website	SA Genomics Health Alliance

Title	Melbourne Genomics Health Alliance (MGHA)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Australia - Victoria
Date	2014-2019
Description	
Policy Issues considered	▪ assessing the impact of genomic sequencing for patients in 16 different disease areas ▪ building genomic knowledge and experience among healthcare professionals ▪ establishing Alliance-wide systems to support genomic sequencing in practice for the benefit of patients, doctors, laboratory scientists and medical researchers
Website	Melbourne Genomics
Cohort Size	3854 across demonstration and current clinical projects
Cohort Description	Current phase 2016-2019 - 11 disease areas spanning adult and paediatric medicine and bacterial analysis. Patients invited to participate will have genomic sequencing at the same time as their usual care
Type of Genomic Data	various
Disease Areas	▪ Hereditary neuropathies (also called CMT) ▪ Focal epilepsy ▪ Hereditary colorectal cancer ▪ Acute myeloid leukaemia (AML) and bone marrow transplants ▪ Immunology ▪ Dilated cardiomyopathy ▪ Congenital deafness ▪ Childhood syndromes ▪ Complex care in children ▪ Advanced non-Hodgkin lymphoma ▪ Advanced solid cancers ▪ Bone marrow failure ▪ Controlling superbugs ▪ Complex neurological and neurodegenerative diseases ▪ Genetic kidney disease ▪ Perinatal autopsy

Title	Brazilian Initiative on Precision Medicine (BIPMed)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Brazil
Date	2015
Description	
Policy Issues considered	▪ Focus on genomic databases and data sharing through beacons
Website	Brazilian Initiative on Precision Medicine
Cohort Size	Multiple disease specific cohorts
Cohort Description	BIPMed-WES-db provides information obtained from Whole Exome Sequencing experiments and includes 106 subjects BIPMed-Array-db contemplates 264 individuals and the data were obtained from microarray-based experiments (Affymetrix GenomeWide SNP 6.0 array) BIPMed-EE-db provides information about specific mutations found in patients with different types of epileptic encephalopathies BIPMed Craniofacial Anomalies: Brazilan Molecular Database on Craniofacial Anomalies is the genotype of 253 variants in 41 genes, with 358 Healthy individuals with no history of oral clefting in the past three generations, and 157 patients with Non-syndromic Cleft Lip and Palate BIPMed-BRCA: Brazilian node of the BRCA Challenge of the Global Alliance for Genomics and Health (GA4GH) shows variants found in the Brazilian population
Type of Genomic Data	various
Disease Areas	Epileptic encephalopathies, Craniofacial Anomalies, Breast cancer and related cancers

Title	Human Genome and Stem-Cell Research Center (HUG-CEL)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Brazil
Date	2013
Description	The Human Genome and Stem-Cell Research Center (HUG-CEL) at the University of São Paulo (USP) expands the scope of the original Human Genome Research Center (HGRC/RIDC I) that was established in 2000. The original center concentrated initially on Mendelian disorders, mainly neuromuscular, craniofacial, and mental disabilities. These activities were expanded in 2005 through the introduction of stem cell research in an effort to understand gene expression and differentiation in complex genetic disorders such as autism and amyotrophic lateral sclerosis, and to evaluate stem cell-based disease therapy The current center has been expanded to include research into the genetics and genomic instability associated with aging and degenerative diseases, epigenetic mechanisms involved in disease manifestation, and phenotypic variability between individuals with identical Mendelian disease mutations. In addition, the 'Over 80' project will compare genome variation and brain functioning (MRI) of healthy Brazilians over age 80 with a group of people over age 60 without prior selection based on good health
Policy Issues considered	▪ research on stem cell and human genetic disease ▪ diagnostic services that includes clinical and laboratory evaluation of affected persons and counselling for their families ▪ educational and teaching programs that aim to bring the latest breakthroughs in Genetics and stem cell research to high school students and teachers and present relevant information to health workers and science journalists
Website	HUG-CEL

Title	DNA do Brasil
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Brazil
Date	2019
Description	Project intends to sequence the complete genomes of approximately 15,000 Brazilians that are already part of the Elsa Brasil study (a Longitudinal Study of Adult Health which aims to investigate over 20 years the factors behind chronic diseases).
Policy Issues considered	▪
Website	DNA do Brasil
Cohort Size	15,000
Cohort Description	Public servants from 35 to 74 years old, in six Brazilian cities - São Paulo (USP)), Belo Horizonte (UFMG), Porto Alegre (UFRGS), Salvador (UFBA), Rio de Janeiro (Fiocruz) and Vitória (UFES).
Type of Genomic Data	
Disease Areas	

Title	Genome Canada
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Canada
Date	2000-current
Description	Genome Canada is a not-for-profit organization, funded by the Canadian Government to act as a catalyst for developing and applying genomics and genomic-based technologies to create economic and social benefits for Canadians Genome Canada designs and administers a suite of programs with funding in three core investment areas: ▪ Large-Scale Science - supports projects using genomic approaches to address challenges in Canada's economic sectors, as well as strategic initiative programs that address national and international opportunities. These programs also include genomics-related research undertaken from the perspective of the social sciences and humanities – known as GE3LS research (i.e. genomics and its ethical, environmental, economic, legal and social aspects) ▪ Leading-Edge Technologies - support for Genomics Technology Platforms to ensure that Canadian researchers have access to the latest technologies, as well as for programs in technology development, bioinformatics and computational biology ▪ Translation - programs that bring genomics-derived solutions to needs in industry, the public sector and society at large
Policy Issues considered	
Website	Genome Canada
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	CanDIG - Canadian Distributed Infrastructure for Genomics
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Canada
Date	
Description	Fully distributed platform that allows national-scale, privacy-maintaining analyses of locally-controlled data sets
Policy Issues considered	
Website	CanDIG
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	China Precision Medicine Initiative
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	China
Date	2016
Description	Broader than genomics. US$9.2 billion, 15-year project as part of China's 13th Five-Year Plan (2016-2020)
Policy Issues considered	
Website	China Precision Medicine
Cohort Size	Initial phase - Beijing Institute of Genomics under the Chinese Academy of Science will spend four years collecting genetic information of 4,000 volunteers
Cohort Description	
Type of Genomic Data	WGS (2000 volunteers)
Disease Area	

Title	Hong Kong Genome Project (HKGP)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Hong Kong – S.A.R, China
Date	2019
Description	To perform 20 000 cases (or 40 000 to 50 000 whole genome sequencing) in two phases for a period of six years; with a) the pilot phase (2 000 cases or about 5 000 genomes) to cover patients with undiagnosed disorders, and cancers with clinical clues linked to possible hereditary components and b) the main phase (18 000 cases or 45 000 genomes) to expand the coverage to other diseases and research cohorts which could benefit from whole genome sequencing. The 2019-20 Budget reserved $1.2 billion (Hong Kong dollars - 155 million U.S. dollars) to take forward the HKGP in six years. The Government will set up the Hong Kong Genome Institute (HKGI), a company limited by guarantee wholly owned by the Government, to coordinate the implementation of the HKGP in partnership with Food and Health Bureau (FHB), the Department of Health, the Hospital Authority, universities, private hospitals, and the research and development sector.
Policy Issues considered	The project will promote the clinical application of genomic medicine by acquiring genomic data from the local population, establishing testing facilities, and nurturing talent.
Website	Hong Kong Genome Project
Cohort Size	20,000
Cohort Description	
Type of Genomic Data	WGS
Disease Areas	Initially undiagnosed disorders and hereditary cancers

Title	Genome Denmark
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Denmark
Date	
Description	A national platform for sequencing and bioinformatics, which includes universities, hospitals and private firms. The platform is established through two large demonstration projects and investments in technological equipment The project is focussed on anti-cancer vaccines and the establishment of a Danish reference genome
Policy Issues considered	
Website	Genome Denmark
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	Dubai Genomics
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Dubai
Date	2018
Description	Plan to sequence the DNA of the entire population, in a bid to improve the health of the 3 million residents.
Policy Issues considered	Phase I focuses to build the necessary infrastructure for genomic medicine and starts large-scale whole-genome sequencing. Phase II aimed at creating novel artificial intelligence capability to help with complex sequence analysis layered with longitudinal data with the focus on the accurate prediction of risks associated with genetic-related illnesses. Phase III focuses on the science of precision medicine by collaborating with interested pharmaceutical companies and academia to identify and design the drugs of the future.
Website	Dubai Genomics
Cohort Size	Population of 3 million
Cohort Description	Entire population – unclear if voluntary or can opt out
Type of Genomic Data	
Disease Areas	

Title	Estonian Genome Center of the University of Tartu (EGCUT) Development Plan for 2015-2021
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Estonia
Date	2015-2021
Description	Estonian Biobank (EGCUT) is a population-based longitudinal biobank established in 2000 and representing about 5% of Estonia's adult population (i.e. a total of 52,000 participants). The whole cohort of the EGCUT is now fully genotyped and 2,500 individuals have been whole-genome sequenced. In 2018 the Estonian government allocated €5 million for a new initiative to recruit and genotype 100,000 new biobank participants as part of its National Personalized Medicine program. The project is implemented according to Estonia's Human Genes Research Act using the same broad consent form used for the first 50,000 participants in the Estonian Biobank. The project aims to collect the DNA of 100,000 individuals and generate personalised genetic reports for the participants using the Illumina Global Screening Array. The project will link genetic data with the Estonian National Health Information System, to enable physicians to take people's personal genetic information into account when assessing health risks. Sample collection began in April 2018.
Policy Issues considered	▪ Management and development of the genome bank - goal is to create "gene cards" with DNA microchips for all gene donors and sequence at least 5,000 full genomes by 2020 ▪ Providing feedback to gene donors - carry out a continuing education program in order to prepare family practitioners and other appropriately qualified doctors for offering first-line genetic counselling ▪ Linking research with the development activities – international collaborations, participation in international biobanking networks and delivery of Estonian Health Insurance Fund tests ▪ Studies - developing post graduate study materials ▪ Strengthening the material base - maintaining and upgrading research infrastructure including sequencing equipment, information management and decision support software ▪ Ensuring the functionality of the organisation – maintain ISO standards, integrate data sources such as national registries and EHRs and start providing feedback to donors

	▪ Serving society - apply the results from the genetic studies for the improvement of overall public health ▪ Outreach - informing the public of new technologies and achievements in medical genetics via national and international training programs, conferences, information days and the internet ▪ Linking genetic data with the Estonian National Health Information System
Website	Estonian Genome Center Estonian Government give 100,000 Estonians information about their genetic risks
Cohort Size	Original project - 5,000 full genomes by 2020 2018 Project – 100,000 Illumina Global Screening Array
Cohort Description	
Type of Genomic Data	WGS and Illumina Global Screening Array
Disease Area	

Title	Ubiquitous Pharmacogenomics (U-PGx)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Europe
Date	2016
Description	Horizon 2020 funded - €14,9 million, duration of 60 months (kick off January 2016) The Netherlands, Spain, UK, Italy, Austria, Greece, Slovenia
Policy Issues considered	▪ Enabling pre-emptive testing – Shared guidelines, genotyping technology, training and education, IT solutions and clinical decision support systems ▪ Implementation of pre-emptive PGx testing (PREPARE study) – clinical implementation and evaluation (Netherlands, Spain, Austria, Italy Greece Slovenia and UK) and HTA / cost effectiveness analysis ▪ Implement pre-emptive genotyping of a - PREemptive Pharmacogenomic testing for prevention of Adverse drug REactions (PREPARE) ▪ Next step in the future – systems pharmacology and gene-drug-drug interactions and NGS ▪ ELSI, dissemination and communication – dissemination and communication, and legal and societal implications
Website	Ubiquitous Pharmacogenomics
Cohort Size	8,100 in PREPARE study - panel of more than 40 variants in 13 pharmaco-genes in a prospective, international, block-randomized, controlled study
Cohort Description	Within 3 years, starting January 1, 2017, 8,100 patients will be pre-emptively tested for more than 40 clinically relevant PGx markers across 13 important pharmacogenes. For 4,050 patients, their results will be used by their healthcare providers to guide the dose and drug selection and the other half of patients assigned to the control group will receive standard of care but will be provided with their results after the study ends. Data on therapy outcome and other parameters collected during the study period will be analyzed in 2020
Type of Genomic Data	PGx markers and panels of PGx markers
Therapeutic Drug groups	▪ Antiarrhythmic ▪ Analgesic ▪ Anticancer ▪ Anticoagulation ▪ Antidepressant ▪ Antiepileptic ▪ Antihypertensive ▪ Anti-infective ▪ Antipsychotic

	▪ Cholesterol-lowering ▪ Immune-suppressive ▪ Other – Atomoxetine and 2nd prescription oestrogen containing drug

Title	FINNGEN
Type of Initiative	Demonstration project / alliance / initiative
Geographic Region	Finland
Date	2017 - 2023
Description	The FinnGen study is based on a public-private partnership between Finnish universities, biobanks, hospital districts, and several international pharmaceutical companies, to drive research, implementation, and economic development in the field of personalized medicine. The project is expected to continue for ten years, with a budget of €59M. Funding comes from seven international pharmaceutical companies and Business Finland. The active phase during which biobank samples are collected and genotyped is planned to continue for six years. The FinnGen Project is divided into two phases, FinnGen 1 (years 1-3) and FinnGen 2 (years 4-6)
Policy Issues considered	The FinnGen study will utilise samples collected by a nationwide network of Finnish biobanks. The study is based on combining genome information with digital health care data from national health registries. The FinnGen study plans to utilise 500 000 unique samples collected by a nationwide network of Finnish biobanks. The project aims to produce close to complete genome variant data of 500 000 biobank participants using GWAS genotyping. The GWAS data are combined with phenotype data produced from several national health registries.
Website	FINNGEN

Title	French Plan for Genomic Medicine 2025
Type of initiative	Demonstration project / alliance / initiative
Geographic Region	France
Date	2016
Description	Focus on cancer, rare diseases, complex diseases
Policy Issues considered	▪ Establish instruments for a genomic care pathway ▪ Ensure operational development and expansion of the system in a safe technical, ethical framework ▪ Establish monitoring and steering tools to make the adjustments required throughout implementation of the Plan while ensuring public involvement
Website	French Plan for Genomic Medicine

Title	Project 10K
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Israel
Date	2019
Description	Project 10K is a long-term observational study to gather information on lifestyle and illness in an Israeli population. It monitors the population of subjects over time in order to find a relationship or relationship between exposure to certain factors and the onset of illness. The study uses advanced techniques to personally characterize a unique group of 10,000 people in Israel.
Policy Issues considered	Project 10K is a clinical study to find novel diagnostic, prognostic, and therapeutic biomarkers for diseases based on applying state of the art machine learning methods to deep phenotypic and multi-omics measurements of 10,000 human volunteers over a 10-year period.
Website	Project 10K
Cohort Size	10,000
Cohort Description	people aged 40-70
Type of Genomic Data	low-pass sequencing to gather accurate genome-wide genetic data and oral microbiome data
Disease Areas	

Title	Implementation of Genomic Medicine Project
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Japan
Date	2014
Description	FY2014 budget : JP¥ 5,5B Goals to be achieved by 2015 ▪ Construct biobank network ▪ Establish Central Genome Center (CGC) and Medical Genome Center (MGC) ▪ Construct Japanese reference genome sequence (Japanese RefSeq) ▪ Build comprehensive genomic variation database linked to clinical phenotypes Goals to be achieved by 2020–30 ▪ Improvements for the medical and health care of lifestyle diseases ▪ Establish predictive diagnostics for cancer incidence and severe Adverse Drug Reactions ▪ Start clinical genomic research for depression and dementia ▪ Clarify the pathogenesis of neurological diseases, etc
Policy Issues considered	▪ Biobank Network - BioBank Japan, National Center Biobank Network, Tohoku Medical Megabank Project ▪ Large scale genomic research - Central Genome Center ▪ Clinical application of genomic information - Medical Genome Center
Website	Japan Implementation of Genomic Medicine Project
Cohort Size	BioBank Japan - Disease-oriented, 200,000 patients National Center Biobank Network - disease-oriented biobanks through 6 national centers Tohoku Medical Megabank Project - Population-based biobank, long-term survey of disaster residents 150,000 local residents (local-resident cohort (80,000 people) and three-generation cohort (70,000 people))
Cohort Description	
Type of Genomic Data	
Disease Areas	▪ autism ▪ cancer – childhood, lung, breast, prostate ▪ epilepsy ▪ depression ▪ stroke ▪ diabetes ▪ congenital heart disease ▪ dementia ▪ diabetic nephropathy ▪ drug side effects

Title	National Institute of Genomic Medicine (INMEGEN)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Mexico
Date	2004-2009
Description	
Policy Issues considered	▪ Build an innovative organizational design: INMEGEN system ▪ Establish the initial infrastructure ▪ Make strategic alliances for the Nationwide Development of Genomic Medicine ▪ Perform high-quality scientific research in genomic medicine ▪ Apply world-class genomic technology to common health problems ▪ Reach excellence in teaching and training programs ▪ Support scientific research and academic programs ▪ Comply and investigate on ethical, social, and legal issues ▪ Translate scientific knowledge into products and services
Website	National Institute of Genomic Medicine Mexico
Cohort Size	
Cohort Description	
Type of Genomic Data	various

Disease and Research Areas	▪ Construction of a 1,500,000 SNPs Haplotype Map of the Mexican population
	▪ Genomic structure of Mestizo and Amerindian populations
	▪ DNA copy number variation in Mestizo and Amerindian populations
	▪ Common complex diseases:
	  ○ Diabetes Mellitus
	  ○ Cardiovascular diseases
	  ○ Obesity
	  ○ Autoimmune diseases (SLE, RA, others)
	  ○ Asthma
	  ○ Age-related macular degeneration
	▪ Cancer Genomics and Proteomics:
	  ○ Breast
	  ○ Lung
	  ○ Gastric
	  ○ Prostate
	  ○ Thyroid
	  ○ Melanoma
	▪ Pharmacogenomics
	▪ Nutrigenomics

Title	Genomics Aotearoa (GA)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	New Zealand
Date	2017
Description	The Government is investing $35 million over 7 years in this new cross-institutional advanced genomics research platform in order to produce high quality research capability for the whole science sector.GA will cement national collaborations between genomics and bioinformatics researchers and users, across theme areas of health, environment and primary production
Policy Issues considered	▪ delivery of a national genomics data repository and bioinformatics analytical platform ▪ strengthen international linkages ▪ development of an indigenous genomics platform which embeds Māori management of indigenous genomics research practice and data ▪ undertake a suite of nationally significant and enabling research activities - in the health area this includes understanding the variation in the genomes of the New Zealand population, which will be used by health providers to improve diagnosis and prognosis of disease and genetic conditions
Website	Genomics Aotearoa

Title	Philippine Genome Center (PGC)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Philippines
Date	2009
Description	A multidisciplinary institution that combines basic and applied research for the development of health and therapeutic products, and improved agriculture and aquatic resources. The mission is a deeper understanding and judicious application of advanced knowledge and emerging technologies in genomics and bioinformatics in health and medicine, agriculture, biodiversity, forensics and ethnicity, industry and the environment for the benefit of Filipinos and the rest of humanity.
Policy Issues considered	The goals are: ▪ implement and promote research program-driven agenda on identified priority areas of national need and of competitive advantage in order to achieve a leading position in the country, region, and in the world ▪ train future scientists, researchers and experts in genomics and bioinformatics ▪ promote a link between academic research, government and private industries for the development of genome-based applications ▪ provide access to state-of-the-art tools for genomic research and bioinformatics in order to strengthen the academic and research infrastructure of the country
Website	Philippine Genome Center
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	Infectious diseases such Dengue and Influenza Pharmacogenomics Type 2 Diabetes Hypertension, Dyslipidemia and Coronary Artery Disease Colorectal Cancer

Title	POLARIS (Personalized OMIC Lattice for Advanced Research and Improving Stratification)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Singapore
Date	2013
Description	POLARIS was established by A*STAR in 2013 to pilot the application of clinical genomics in the treatment and diagnosis of medical diseases in Singapore and the region
Policy Issues considered	▪ Working with Ministry of Health and other national bodies to ensure that the deployment of new technologies is accompanied by a robust framework for informed patient consent
Website	POLARIS Singapore
Cohort Size	
Cohort Description	
Type of Genomic Data	Panels, WES
Disease Areas	▪ Eye Disease ▪ Cancer panels and stratification ▪ Tuberculosis ▪ Rare diseases (undiagnosed disease in children)

Title	CIBERER (Center for Network Research on Rare Diseases)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Spain
Date	2006
Description	CIBERER aims to develop high quality cooperative and innovative research into rare diseases, fostering the transfer of results to clinical practice. The specific objectives are mainly based on the development of new treatments and improving access to diagnosis of rare diseases. Aims: ▪ Establish and provide access to data, standardised relevant information in rare diseases ▪ Carry out molecular and clinical characterisation of rare diseases ▪ Further translational, preclinical and clinical research into rare diseases ▪ Rationalise the ethical procedures and rules in rare diseases
Policy Issues considered	The Genetic Medicine Programme objectives are: ▪ to lead the development of innovations in genomic platforms ▪ to provide support for preclinical research on rare epilepsies and related diseases, including Lafora disease ▪ to promote physiopathological study for its therapeutic application and diagnosis in rare and complement-mediated vascular pathologies
Website	CIBERER
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	Rare diseases ▪ Lafora disease and other rare genetic epilepsies ▪ Neuromuscular diseases: muscular dystrophies, spinal muscular atrophy ▪ Vascular and immune system diseases; disorders affecting the vascular endothelium causing pathologies such as HHT and complement deficiencies.

Title	Genomic Medicine Sweden (GMS)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Sweden
Date	August 2017 - July 2018
Description	1 991 250 kronor will be invested to provide detailed information on how precision medicine can be implemented in a coordinated manner within Swedish healthcare. Specific Objectives: 1) Investigate and Propose Organization for GMS 2) Define Implementation Strategies and Design Pilot Studies for Unusual Hereditary Diseases and Cancer 3) Investigate two key challenges: Health Economics and Informatics 4) Ensure continued commitment and resources for the next phase of the Project Implementation of Genomic Medicine in Healthcare Long-term goal, sequencing of approximately 25,000 patients annually. National resource for academia and industry for the development of pharmaceuticals, diagnostics and software.
Policy Issues considered	Health Economics Informatics
Website	Genomic Medicine Sweden and Karolinska Institutet
Cohort Size	
Cohort Description	The primary focus will be patients with rare inherited diseases and cancer, but sequencing will also be performed in other areas such as in complex diseases and the microbiome.
Type of Genomic Data	
Disease Areas	

Title	Swiss Personalised Health Network (SPHN)
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	Switzerland
Date	2016
Description	A national initiative designed to promote the development of personalized medicine and personalized health in Switzerland During the period 2017–2020, priority is given to the development of a nationally coordinated data infrastructure ensuring data interoperability of local and regional information systems with special emphasis on clinical data management systems enabling effective exchange of patient data (e.g. disease phenotypes)
Policy Issues considered	• Information Management and data sharing • ELSI • Coordination and governance
Website	Swiss Personalised Health Network
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	Genomics England
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	United Kingdom - England
Date	2013 -
Description	approx. £300 million originally over 4 years from 2014-2017 but subsequently extended to 2018 to sequence 100,000 genomes. Includes public, private and philanthropic contributions Extended further in September 2018 with commitment from Secretary of State for Health and Social Care for: ▪ expansion of the 100,000 Genomes Project to see 1 million whole genomes sequenced by the NHS and UK Biobank in five years ▪ from 2019, the NHS will offer whole genome analysis for all seriously ill children with a suspected genetic disorder, including those with cancer and the NHS will also offer the same for all adults suffering from certain rare diseases or hard to treat cancers ▪ an aspiration to sequence 5 million genomes in the UK, within an unprecedented five-year period
Policy Issues considered	▪ Sequencing challenge ▪ Data challenge ▪ Security challenge ▪ Delivery benefits to patients ▪ Ethical issues – consent, privacy, confidentiality, ownership/commercialisation benefits ▪ Industry development
Website	Genomics England
Cohort Size	1,000,000 – 5,000,000
Cohort Description	NHS patients who have certain rare diseases, plus their families, or those who have cancers that fit inclusion criteria
Type of Genomic Data	WGS
Disease Areas	▪ Rare Diseases ▪ Cancer

Title	All of Us
Type of initiative	Demonstration Project / alliance / initiative
Geographic Region	United States of America
Date	2016
Description	US$300 million (as at FY 2017 budget)
Policy Issues considered	Biobanking Communications and engagement Data and Research support Participate Technology systems
Website	All of Us
Cohort Size	1,000,000+
Cohort Description	Anyone over the age of 18 who is living in the United States will be able to join the All of Us Research Program, either directly through the JoinAllofUs.org website or through participating health care provider organizations. Genomes, health records and physiological measurements
Type of Genomic Data	Pilot phase – whole genome genotyping and WGS
Disease Area	▪ Recruitment is not specific to a patient group or disease area. It is open to all

Title	Project Baby Bear
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	United States of America - California
Date	2018, 2-year duration
Description	A $2 million state-funded initiative will work with infants on Medi-Cal from four different cities: Fresno, Oakland, Sacramento and San Diego. The goal is to derive genetic diagnoses for at least 100 children admitted to neonatal intensive care units (NICU) in their communities with severe but unexplained symptoms.
Policy Issues considered	Rapid whole genome sequencing of Medical-enrolled infants for diagnosis of genetic diseases and genomic medicine guidance in California Children's Services neonatal and pediatric intensive care units
Website	Rady Children's Institute for Genomic Medicine
Cohort Size	~125
Cohort Description	Seriously ill infants in CCS-accredited NICUs and PICUs
Type of Genomic Data	Rapid whole genome sequencing
Disease Area	Childhood onset genetic diseases

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Population –specific genetic variation studies

Title	GenomeAsia 100k
Type of initiative	Population –specific genetic variation study
Geographic Region	Asia
Date	
Description	India, Singapore, Korea and USA. Looking to partner with Pakistan, Bangladesh, Sri Lanka, Malaysia, Indonesia, Myanmar, Nepal. Bhutan, Thailand, Vietnam, Laos and Japan Sequence and analyze 100,000 Asian individual's genomes to help accelerate Asian population specific medical advances and precision medicine
Policy Issues considered	
Website	GenomeAsia
Cohort Size	100,000
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	GenomeIndia Initiative & Genome India Project
Type of Initiative	Population –specific genetic variation study
Geographic Region	India
Date	2017
Description	Initiative to sequence thousands of Indian individuals belonging to different geographic locations and diverse population groups in order to unravel details of the Indian genome. Building a catalogue of genetic variations will pave the way for conducting better research on genetic disorders and will also enable physicians to provide accurate diagnosis of genetic disorders at a much cheaper price in the near future. In 2019 the Department of Biotechnology (DBT) announced the first phase of an initiative called the Genome India project which will catalogue the genomic data of 10,000 Indians by involving 22 partner organisations including public health institutions. Data will be collected over three years and linked to biobanks and biorepository.
Policy Issues considered	
Website	GenomeIndia Initiative Genome India Project
Cohort Size	Over 2000 individuals (GenomeIndia Initiative) Up to 10,000 individuals (Genome India Project)
Cohort Description	
Type of Genomic Data	WGS
Disease Areas	

Title	Iranome
Type of Initiative	Population –specific genetic variation study
Geographic Region	Iran
Date	2017
Description	Established the Iranome database (www.iranome.com) by performing whole exome sequencing on 800 individuals from eight major ethnic groups in Iran. The groups included 100 healthy individuals from each of the following ethnic groups: Arabs, Azeris, Balochs, Kurds, Lurs, Persians, Persian Gulf Islanders and Turkmen. They represent over 80 million Iranians.
Policy Issues considered	
Website	Iranome
Cohort Size	800
Cohort Description	Health individuals from major ethnic groups
Type of Genomic Data	WES
Disease Areas	

Title	Genome of the Netherlands (GoNL)
Type of initiative	Population –specific genetic variation study
Geographic Region	Netherlands
Date	
Description	DNA of 750 Dutch people-250 trio's of two parents and an adult child-plus a global genetic profile of large numbers of Dutch will disclose a wealth of new information, new insights, and possible applications
Policy Issues considered	
Website	Genome of the Netherlands
Cohort Size	750
Cohort Description	250 trio's of two parents and an adult child
Type of Genomic Data	
Disease Areas	

Title	The Nordic Alliance for Clinical Genomics (NACG)
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Nordic countries - – Norway, Denmark, Iceland, Sweden, Finland
Date	2018
Description	NACG aims to: ▪ Facilitate the responsible sharing of genomic data, bioinformatics tools, sequencing methods and best practices for interpretation of genomic data. ▪ Enhance quality of genomic data and processes, and explore methodologies to provide assurance. ▪ Understand legal barriers to the implementation of personalised medicine and to engage with key stakeholders that influence these barriers ▪ Develop demonstration projects that challenge perceived legal barriers that limit responsible and ethical sharing of genomic and health data. ▪ Build bridges between research and clinical communities, technologies and practices to foster innovation
Policy Issues considered	
Website	The Nordic Alliance for Clinical Genomics (NACG)
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	BigMed
Type of Initiative	Demonstration Project / alliance / initiative
Geographic Region	Norway
Date	2018
Description	The aim is to identify and address the bottlenecks for clinical implementation of precision medicine and lay the foundation for an ICT platform to facilitate the development of data driven decision support.
Policy Issues considered	The work in BigMed is organized through four clinical areas: rare diseases, colorectal cancer, sudden cardiac death and frost bites. The suggested solutions will be based on the clinical needs identified in each disease area and will focus on the following themes: ▪ ICT infrastructure and data provisioning ▪ legal and ethical considerations ▪ bioinformatic pipelines and data sharing
Website	BigMed
Cohort Size	
Cohort Description	
Type of Genomic Data	
Disease Areas	Rare diseases, colorectal cancer, sudden cardiac death and frost bites

Title	Qatar Genome
Type of Initiative	Population–specific genetic variation study Complete Integration
Geographic Region	Middle East
Date	2015
Description	The Qatar Genome Programme (QGP) is a population-based project generating large databases combining whole genome sequencing and other omics data with the comprehensive phenotypic data collected at Qatar Biobank.
Policy Issues considered	▪ Ethical and Social implications ▪ Data and Information
Website	Qatar Genome

Title	Saudi Human Genome Program (SGP)
Type of initiative	Population-specific genetic variation study
Geographic Region	Saudi Arabia
Date	2013
Description	~SAR300 Million budget over 3 years A 3-year project to find the genes responsible for the genetic diseases that impact the Kingdom, by sequencing the genomes of 20,000 subjects, representing both the general population, and all the major diseases
Policy Issues considered	
Website	Saudi Human Genome Program
Cohort Size	20,000
Cohort Description	General population and all major diseases
Type of Genomic Data	WGS

Disease Areas	The project will focus on all rare severe (early onset) diseases in the early phases, and then on the highest impact common diseases:
	▪ Rare Disease:
	  ○ Anemia
	  ○ Blindness
	  ○ Deafness
	  ○ Skeletal Deformation
	  ○ Mental Retardation
	▪ Common Disease:
	  ○ Cardiovascular
	  ○ Diabetes

Title	Southern African Human Genome Programme (SAHGP)
Type of initiative	Population –specific genetic variation study
Geographic Region	South Africa
Date	2011 -
Description	A national and regional initiative that aims to unlock the unique genetic character of southern African populations Its vision is to improve quality of life by understanding human genetic diversity, to be achieved via the following objectives: 1. developing capacity for genomic research in southern Africa 2. establishing a sustainable resource for genomic research (including a regional sample repository and database) 3. translating the information and knowledge into improvements in human health The programme aims to make a significant contribution to understanding DNA variation among southern Africans and how this affects the health of the people of the region
Policy Issues considered	
Website	Southern African Human Genome Programme (SAHGP)
Cohort Size	Pilot study - 24 individuals
Cohort Description	Pilot study - 8 Coloured and 16 black south-eastern Bantu-speakers)
Type of Genomic Data	deep whole-genome sequencing
Disease Areas	

Title	SweGen
Type of initiative	Population –specific genetic variation study
Geographic Region	Sweden
Date	
Description	A dataset that contains whole-genome variant frequencies for 1000 Swedish individuals generated within the SweGen project
Policy Issues considered	
Website	SweGen
Cohort Size	1000
Cohort Description	
Type of Genomic Data	
Disease Areas	

Title	Turkish Genome Project
Type of Initiative	Population-specific genetic variation study
Geographic Region	Turkey
Date	2017
Description	The Turkish Genome Project is planned in two phases: the first targeting 100,000 genomes in the next 3 years, and then reaching to 1,000,000 genomes before 2023. A major component of the project will be on complex phenotypes with high heritability index such as obesity, diabetes, cardiovascular, neuropsychiatric, endocrine, and rheumatologic diseases. A parallel-executed component will focus on rare diseases, an area that Turkey has contributed significantly over the years. And yet another major component of the project is on cancer with particular emphasis on familial forms of these diseases.
Policy Issues considered	
Website	Turkish Genome Project
Cohort Size	100,000 (phase one), 1,000,000 (phase two)
Cohort Description	Healthy individuals plus rare and complex disease patients
Type of Genomic Data	WGS
Disease Areas	Rare diseases, complex diseases and familial cancer

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Complete Integration / Implementation Roadmaps

Title	Australian Genomics Health Futures Mission
Type of Initiative	Complete Integration / Implementation Roadmap
Geographic Region	Australia
Date	2018
Description	The Government will invest $500 million over 10 years in an Australian Genomics Mission to help save or transform the lives of more than 200,000 Australians through research into better testing, diagnosis and treatment. The first project will be 'Mackenzie's Mission' – a new $20 million trial in pre-conception screening for rare and debilitating genetic birth disorders, such as Spinal Muscular Atrophy, providing vital information and hope for hundreds of parents who face difficult choices in starting a family.
Policy Issues considered	Key features of the Australian Genomics Mission include: ▪ new and expanded clinical flagship studies to tackle rare diseases, rare cancers and complex conditions; ▪ new clinical trials and technology applications allowing Australian patients to benefit from the latest medical research; ▪ increased academic and researcher collaboration and new career pathways; ▪ co-investment with philanthropy and business to support new industries; ▪ community dialogue to better understand the value of genomics and gain appreciation of the privacy, legal, social and familial impact; and ▪ analytical power backed by national standards and protocols that ensure secure data holdings, access, analysis and sharing to benefit Australians.
Website	Australia Genomics Health Futures Mission

Title	Genome Canada National Initiative for the Clinical Implementation of Precision Health
Type of Initiative	Complete Integration / Implementation Roadmap
Geographic Region	Canada
Date	2018
Description	Genome Canada launched a national initiative for the clinical implementation of precision health, focusing on a rare disease pilot program as a foundational step. This program promises every rare disease patient in Canada the opportunity for a diagnosis and hope for treatment. By establishing shared and effective policies, processes, techniques and technologies, it will also form the first step towards a national roll-out of precision health for all Canadians.
Policy Issues considered	Broadly, the national strategy/pilot initiative features three main components: ▪ a national rare disease cohort is envisioned to be established through the collection and sequencing of 30,000 samples from rare disease patients and their families. ▪ a national platform is being set up to provide mechanisms and best practices for the collection and sharing of data, including privacy policies, informed consent and other ethical and legal frameworks. ▪ clinical implementation will advance through working with provincial and regional centres and partners to establish clinical sites and achieve regulatory approval and accreditation.
Website	Genome Canada National Initiative for the Clinical Implementation of Precision Health

Title	Generation Genome – Annual Report of the Chief Medical Officer
Type of initiative	Complete Integration / Implementation Roadmap
Geographic Region	United Kingdom - England
Date	2016
Description	The report discusses the current state of genomic service provision in the NHS in England. It explores the potential of genomics to improve health and prevent ill-health
Policy Issues considered	▪ Systems and services including governance ▪ Research ▪ Data, Standards and Regulation ▪ Engaging staff and patients ▪ Other
Website	Generation Genome

Title	GENOME UK - The future of healthcare
Type of Initiative	Complete Integration / Implementation Roadmap
Geographic Region	United Kingdom
Date	2020
Description	Over the next ten years the ambition is to create the most advanced genomic healthcare system in the world, underpinned by the latest scientific advances, to deliver better health outcomes at lower cost. This will be done by: ▪ working together across the four nations and reducing boundaries between clinical care and research ▪ supporting earlier detection and faster diagnoses ▪ use genomics to target interventions to specific groups of patients ▪ support patients in understanding what genomics means for their health ▪ bring the full might of the capabilities in this field to bear against new global pandemics and threats to public health
Policy Issues considered	▪ Diagnosis and personalised medicine, Prevention & Research ▪ Engagement and dialogue with the public, patients and the healthcare workforce ▪ Workforce development ▪ Supporting industrial growth in the UK ▪ Maintaining trust through ethical frameworks, data security and appropriate regulation ▪ Delivering nationally coordinated approaches to data and analytics
Website	Genome UK

Title	Qatar Genome
Type of Initiative	Population–specific genetic variation study Complete Integration
Geographic Region	Middle East
Date	2015
Description	The Qatar Genome Programme (QGP) is a population-based project generating large databases combining whole genome sequencing and other omics data with the comprehensive phenotypic data collected at Qatar Biobank.
Policy Issues considered	▪ Ethical and Social implications ▪ Data and Information
Website	Qatar Genome