Genomic Medicine in Korea:

Plan & Infrastructure

Center for Genome Science
Korea National Institute of Health
Ministry of Health and Welfare, Korea

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Overview of Multi-ministry genome project

<table>
<thead>
<tr>
<th>Official Name</th>
<th>Genome Technology to Business Translation Program</th>
</tr>
</thead>
<tbody>
<tr>
<td>Duration</td>
<td>2014 ~ 2021(4+4, 2 steps, $500Mill/8 years)</td>
</tr>
<tr>
<td>Final Goals</td>
<td>Implementation of Genomic Medicine</td>
</tr>
<tr>
<td></td>
<td>Exploration of genomics-based biological resources</td>
</tr>
<tr>
<td>Objectives</td>
<td>Development for novel diagnosis &amp; treatment for personalized medicine (Human)</td>
</tr>
<tr>
<td></td>
<td>Exploitation of high value-added natural resources using genomics (non-Human)</td>
</tr>
<tr>
<td></td>
<td>Construction of infrastructure for activation of genome information industry (infra)</td>
</tr>
<tr>
<td></td>
<td>Establishing genomic research infrastructure for fundamental genomics technology (infra)</td>
</tr>
</tbody>
</table>
Multi-ministry genome project

Human
- Research & support for translational genomics
- Production & utilization of Korean genomic information
- Construction of human genomics information center

Agriculture
- Agricultural genomics for industrialization
- Agriculture genomics analysis
- Construction for agricultural genomics information

Marine life
- Exploration of marine life resources using genomics
- Support for marine life genomics

Basic research
- Genomics core technology development
- Construction of infrastructure for genome analysis

Industrialization
- Software development for industrialization of genome information
- Development of business model for genomics

Joint research project

MHW: Ministry of Health and Welfare
MA: Ministry of Agriculture, Food, and Rural Affairs
MOF: Ministry of Oceans and Fisheries
MOTIF: Ministry of Science, ICT, and Future Planning
MOTI: Ministry of Trade, Industry and Energy
Cooperative links between ministries

Genome Research to Business System

Basic research
- MOTIP
  - Joint research foundation for genome analysis
  - Project for developing future core technology
  - Enhancement of future genomics research infrastructure

Application research
- MHW
  - Genomics for personalized medicine
  - Support for translational genomics
  - Production of Korean resources for genomics
- MAFR
  - Microbes genomics for industrialization
  - Agricultural plant genome analysis
  - Enhancement of agricultural information
- MCF
  - Exploration of marine life using genomics
  - Support for marine life genomics

Industrial application
- MITE
  - Constructing open computing facility
  - Optimized application to industry
  - Infrastructure for business system & industrialization

Joint research project
## Vision & Mission for Genomic Medicine at MHW

### Vision
- Promotion of public health and personalized medicine

### Mission
- Development of prevention, diagnosis and treatment using genomics
- Acquisition of resources & information for disease genomics

### Projects
- Translational genomics for personalized medicine
- Support project for translational medicine
- Production & utilization project of Korean genomics research resources
- Ethics & legal & social implications (ELSI)
- Joint projects
Projects

Ministry of Health & Welfare

- **Translational genomics for personalized medicine**
  - Research strategy for disease genomics
  - Clinical genomics research for personalized medicine

- **Support project for translational genomics**
  - Center for Pharmacogenomics
  - Integrated center for genomics technology

- **Production of Korean genomics resources**
  - Production of genomics information based on cohort
  - Production & standardization of major diseases genomics data
  - Production & standardization of cancer genomics data

- **ELSI**
  - Disease mechanism
  - Host-Microbe interaction
  - Korean reference genome
  - International collaboration

Joint projects

Integrated system for genomic information
Conceptual diagram of project organization

- **Center for Genomics Information**
  - Translational genomics
  - Strategy for disease genomics
  - Genomics for disease mechanism
  - Management of genome resources

**With genomic information**

- Prediction of disease progress & prognosis
- Reducing complication & mortality
- Selection of responsive group
- Early diagnosis & treatment
- Development of preventive method
- Prediction of disease onset

- **Center for Pharmacogenomics**
- **Center for Integrated Genomics Technology**
- **Center for ELSI**
Ministries of Health and Welfare Projects in 2014

objectives

- Supporting human genome research for implementation of personalized medicine ($10 Million)

Direction in 2014

① Fundamentals for clinical genomics
   - Development of personalized medicine system
   - Innovative research for integrative genomics
   - International collaboration
   - Building research center for clinical genomics data analysis

② ELSI (Ethical, Legal and Social Implications)
   - Research & Development support for social understanding & consensus about controversial issues of genomics research

③ Production and utilization of Korean research resources for genomics
   - Production of cohort-based Korean genome data & information

④ Legislating genome research Act and technical roadmap for precision medicine are under planning
Contents

1. Post-genome Multi-ministry Genome Project

2. Plan for Genomic Medicine

3. Projects for Genomic Medicine in 2014

4. Infrastructure for Genomic Medicine: Center for Genome Science in KNIH
   - Current projects of CGS
   - Perspectives and issues
Infrastructure for Genomic Medicine in Korea NIH

Center for Genome Science

Division of Epidemiology and Health Index
Korean Genome and Epidemiology Study (KoGES)

Division of Structural and Functional Genomics
Korean Genome Analysis project (KoGAP)
Korean epigenome reference

Division of Biomedical Informatics
Korean Reference Genome(KRG) Bioinformatics

Division of Biobank for Health Science
Korea Biobank Project (KBP)
As a national institute...

Genomic data  Epidemiological data  Biobank

Center for Genome Science, KNIH

PROVIDING VALUABLE RESOURCE FOR BIOMEDICAL RESEARCH AND PERSONALIZED MEDICINE
Mission

- To provide infrastructure for the genome and epidemiological studies
  - Producing genomic and epidemiological data
  - Collecting and sharing epidemiological data and biospecimen
  - Establishing guideline and regulation

- To identify genetic and environmental factors, gene-environmental interaction influencing disease outcome
  - GWAS, cohort study
  - Factors associated with the Korean-specific traits
  - Factors associated with intermediate and final disease outcomes
Current projects of CGS

Korean Genome and Epidemiology Study (KoGES)

- Large-scale population-based prospective cohort study
  Targeted Disease: T2DM, hypertension, obesity, metabolic syndrome and osteoporosis
- collecting epidemiological data and biospecimen
  - Past medical history, life style (smoking, drinking, physical activity, diet, etc)
  - longitudinal repeated follow-up data
  - blood, urine etc.
- Providing resources
  - Questionnaire
  - Survey protocols (FFQ)
  - Data quality control guidelines

Korea Biobank Project (KBP)

Korean genome analysis project (KoGAP)
Korean Genome and Epidemiology Study (KoGES)

Community-based cohorts
- Ansan, Ansung cohorts
- Rural area cohorts

Health examinee cohorts
- Regular health check up-based cohorts

Gene-environment interaction cohorts
- Twin & Family Cohort
- Korean Emigrant Cohort
- Asian Immigrant Cohort
- Asian Collaborative Cohort

~ 2013: total 240,000 participants
Studies using whole genome analysis of KoGES samples

Nature Genetics (2009) 41:527-534
Science (2009) 326:1541-1545
Nature Genetics (2011) 43:531-538
Nature Genetics (2011) 43:539-538
Nature Genetics (2011) 44:67-72
Nature Genetics (2012) 44:302-306
Nature Genetics (2012) 44:307-311
Nature Genetics (2012) 41:527-534
Nature Genetics (2012) 44:904-909
Nature Genetics (2013) 45:25-33
Nature Genetics (2013) 45:621-631
Current projects of CGS

- **Korean Genome and Epidemiology Study (KoGES)**

- **Korea Biobank Project**
  - A network of National biobank of Korea and 17 regional biobanks
  - Providing biospecimen for biomedical research
    - DNA, serum, plasma
    - *Other omics data in future*
  - Providing
    - Sample QA/QC guideline
    - Education for standardization and harmonization of biobanks
    - A consensus for ELSI (Ethics, legal, and social Implication)

- **Korean genome analysis project (KoGAP)**
Biobanks of Korea Biobank Project

- **NBK**: National Biobank of Korea
- 17 regional Biobanks

- **National Biobank of Korea (NBK)**: National Control tower
  - Development of QC Tech.
  - Management of sample
  - Distribution

**Locations**:
- Seoul NUH
- Ajou University Medical Center
- Seoul Asan Medical Center
- Korea University Guro NUH
- Soonchunhyang Bucheon Medical Center
- Kangwon NUH
- Chungbuk NUH
- Chungnam NUH
- Kyungpook NUH
- Keimyung University Dongsan Medical Center
- Pusan NUH
- Gyeongsang NUH
- Inje University Busan-Paik Hospital
- Jeju NUH
- Chonnam NUH Hwasun Hospital
- Chonbuk NUH
- Wonkwang University Hospital

* NUH: National University Hospital*
KBP Biospecimen Collection: 618,958 participants

- New collected
- Accumulated

Regional Biobanks

NBK
Distribution of human biospecimens

As of Dec. 2013

NBK

(258,208 vials)

201 Research projects

DNA
Serum
Plasma
Other Biospecimens

17 Regional Biobanks

(146,510 vials)

712 Research Project

Tissue
Serum
Buffy Coat
Plasma
Other Biospecimens

Total: 345,432 vials (913 Research Projects)
Current projects of CGS

- Korean Genome and Epidemiology Study (KoGES)
  - Large-scale population-based prospective cohort study
  - 8 cohorts

- Korea Biobank Project
  - National biobank and 17 regional biobanks

- Korean genome analysis project (KoGAP)
  - Finding genetic factors associated with the traits (anthropometric, chronic disease outcomes)
  - Korean Reference Genome, epigenome reference
Research Objectives

Discovery of risk factors
- To identify risk factors influencing a wide range of quantitative traits and lifestyle-related diseases of major biomedical relevance including Type 2 Diabetes, dyslipidemia, hypertension, and obesity

Construction of reference information infrastructure
- Korean Reference Genome
  - whole genome sequencing on > 400 samples

- Korean Reference Epigenome
  - to produce 50 epigenome maps until 2017 with a budget of 10 million dollars. Produced data will be shared with IHEC
Perspectives and Issues

- Needs
- Funding
- Legal & ethical issues
- High quality data and samples
- Data sharing
- Promoting R&D
- Providing Genome Research Resources
- Public genomics
- Gene-environment Interaction
- Prediction model
- Improve Genomic Medicine
Thank you