

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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Jan 8th, 2014

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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, KNIH**

Overview of Multi-ministry genome project

Official Name

Genome Technology to Business Translation Program

Duration

2014 ~ 2021(4+4, 2 steps, \$500Mill/8 years)

Final Goals

**Implementation of Genomic Medicine
Exploration of genomics-based biological resources**

Objectives

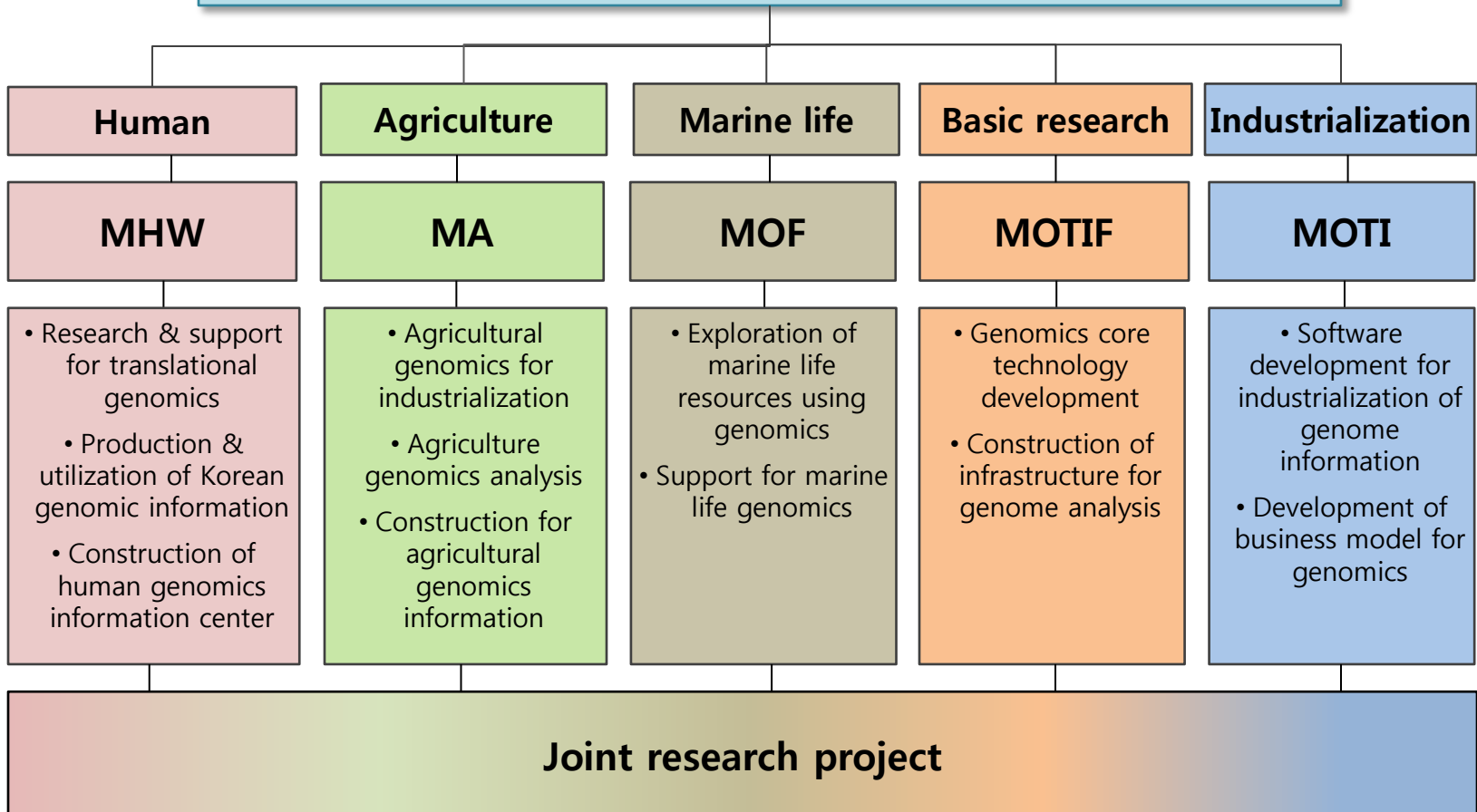
Development for novel diagnosis & treatment for personalized medicine (Human)

Exploitation of high value-added natural resources using genomics (non-Human)

Construction of infrastructure for activation of genome information industry (infra)

Establishing genomic research infrastructure for fundamental genomics technology (infra)

Multi-ministry genome project



MHW; Ministry of Health and Welfare

MA; Ministry of Agriculture, Food, and Rural Affairs

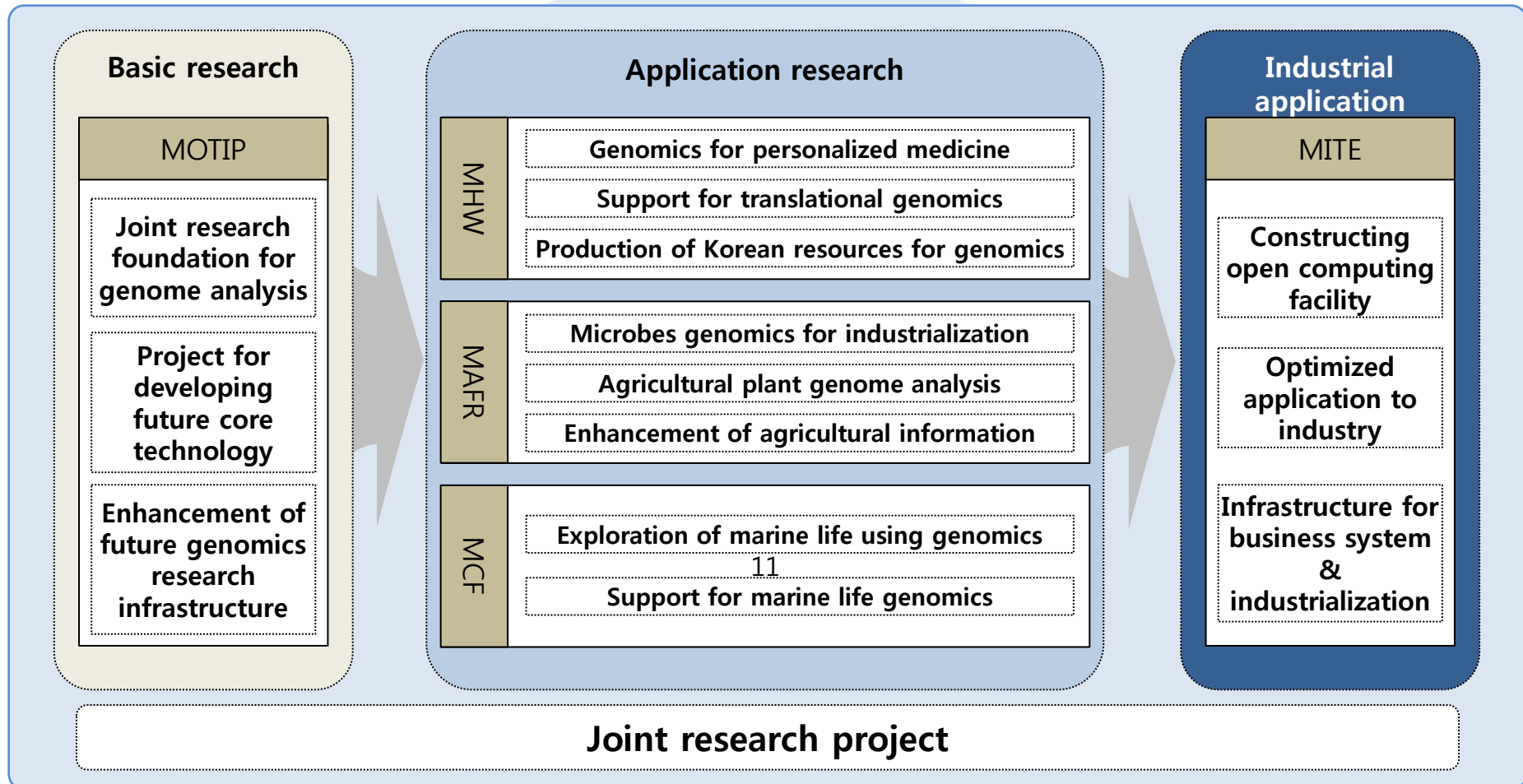
MOF; Ministry of Oceans and Fisheries

MOTIF; Ministry of Science, ICT, and Future Planning

MOTI; Ministry of Trade, Industry and Energy

Cooperative links between ministries

Genome Research to Business System



Vision & Mission for Genomic Medicine at MHW

Vision

- Promotion of public health and personalized medicine

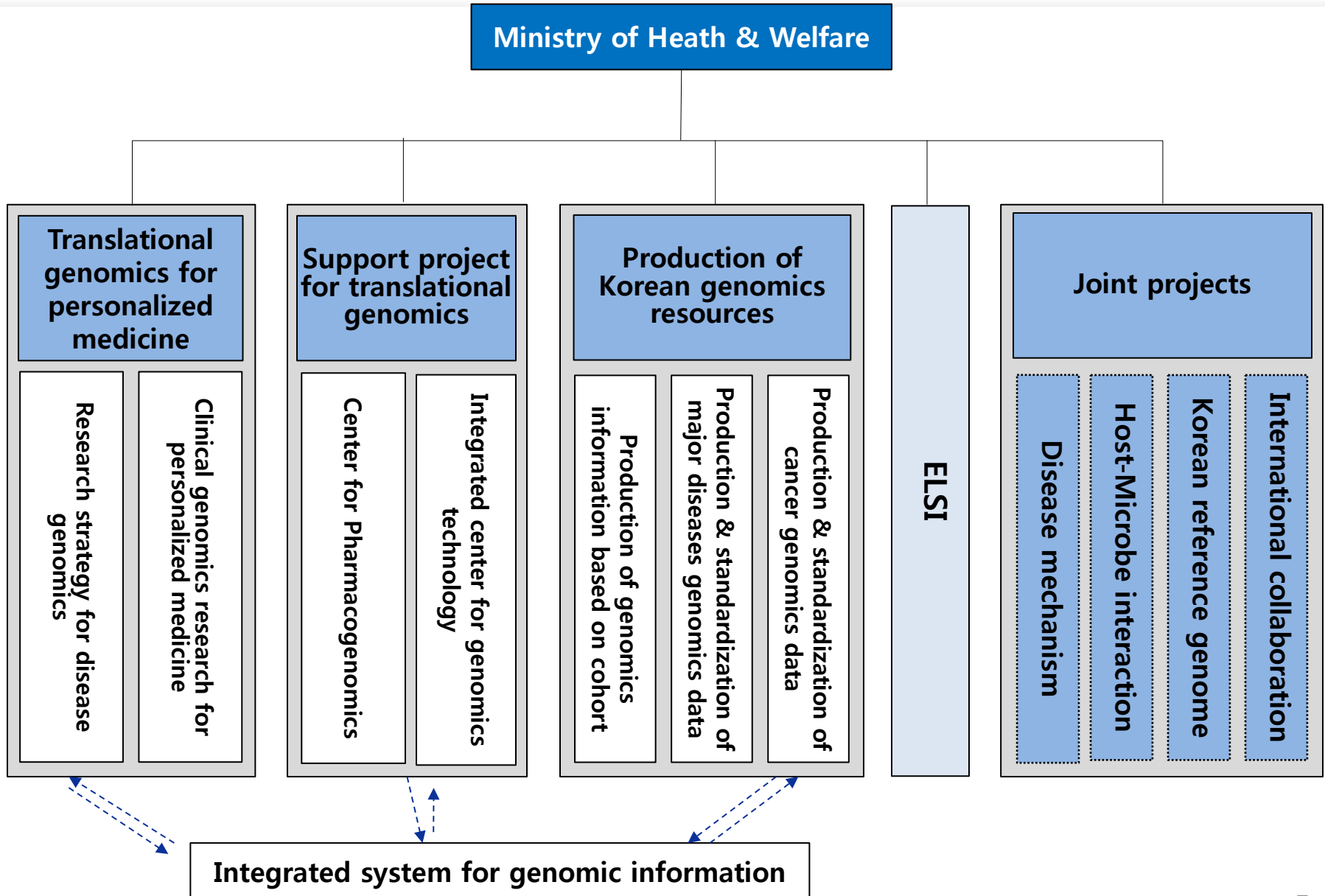
Mission

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

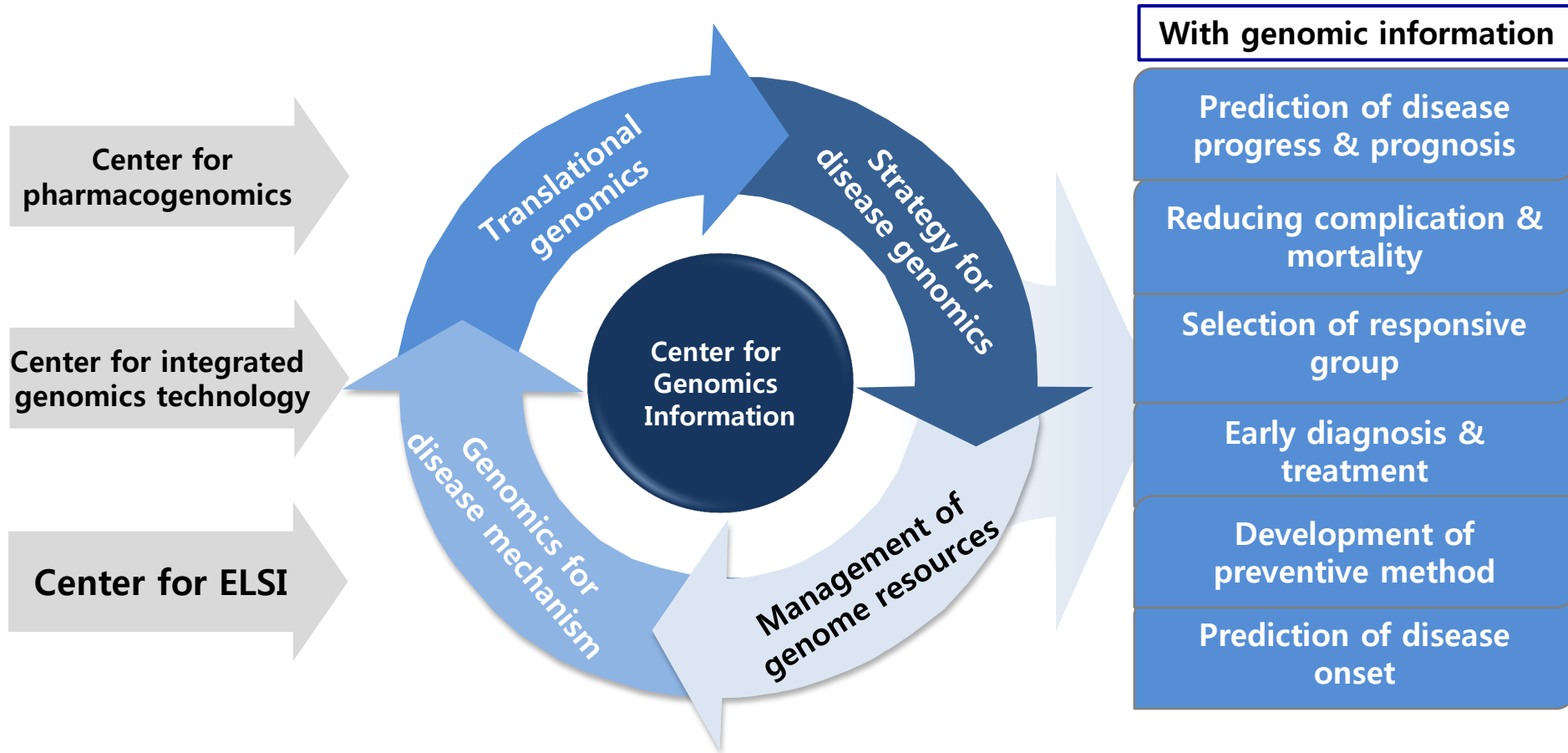
Projects

- 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



Conceptual diagram of project organization



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)**

- R & D 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**

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**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)

■ ~ 2013: total 240,000 participants

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

Studies using whole genome analysis of KoGES samples

A large-scale genome-wide association study of Asian populations uncovers genetic factors influencing eight quantitative traits

Yoon Shin Cho¹, Min Jin Go¹, Young Jin Kim¹, Jee Yeon Heo¹, Ji Hee Oh¹, Hyo-Jeong Ban¹, Dankyu Yoon², Mi Hee Lee¹, Dong-Ioon Kim¹, Miev Park¹, Seung-Hun Cha¹, Jun-Woo Kim¹, Bok-Ghee Han¹

***Nature Genetics* (2009) 41:527-534**

***Science* (2009) 326:1541-1545**

***Nature* (2010) 466:707-713**

***Nature Genetics* (2011) 43:531-538**

***Nature Genetics* (2011) 9:990-995**

***Nature* (2011) 478:103-109**

***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**

Mapping Human Genetic Diversity in Asia

The HUGO Pan-Asian SNP Consortium*†

Asia harbors substantial genetic variation. Autosomal variation shows that genetic diversity is highest in Southeast Asian populations. Haplotype diversity is highest in SEA and is a source of EA population genetic diversity.

Populations united by membership in a linguistic family, by geographic proximity, by a known history of admixture, or, especially at higher Ks, by membership in a small population isolate. The results obtained using *FastTree* (17), a maximum-likelihood...

Biological, clinical and population relevance of 95 loci for blood lipids

A list of...

Plasma triglyceride levels are elevated in European and Asian (SEA) and using the Affymetrix 50K Xba Array genotypes were single-nucleotide polymorphisms (SNPs) implicated in lipid phenotype. Individuals representing Asian HapMap populations were used to develop a genetic structure and its relationship to demographic history.

Plasma protein (HDL-C) and vascular disease of European inter-ethnic differences in these loci lipid control GWAS, remains.

Hypertension and premature...

Meta-analysis of genome-wide association studies identifies common variants associated with blood pressure variation in east Asians

Norihiro Kato^{1,2,41*}, Wan Ting Tang¹, Mitsuhiro Matsuda¹, Yoshihiro Hara¹, Jong-Young Lee⁵, Takao Sugiura⁶, Xuegong Zhang⁷, Naoharu Iwano⁸

We conducted a meta-analysis of subjects of European ancestry (which includes *TBX3*). Among loci except for those near *ALDH3B1*, ethnic specificity was observed.

LETTERS

nature genetics

Meta-analysis identifies multiple loci associated with kidney function-related traits in east Asian populations

Yukinori Okada^{1,2,41*}, Xueling Sim^{3,4,41}, Min Jin Go^{5,41}, Jer-Yuarn Wu^{6,7,41}, Dongfeng Gu^{8,41}, Fumihiko Takeuchi^{9,41}, Atsushi Takahashi¹, Shiro Maeda¹⁰, Tatsuhiko Tsunoda¹¹, Peng Chen¹², Su-Chi Lim¹²⁻¹⁴, Tien-Yin Wong¹⁵⁻¹⁷, Jianjun Liu¹⁸, Terri L Young¹⁹, Tin Aung^{15,16}, Mark Seielstad²⁰, Yik-Ying Teo^{3,12,18,21,22}, Young Jin Kim², Jong-Young Lee⁵, Bok-Ghee Han², Daehee Kang²³, Chien-Hsiun Chen^{6,7}, Fuu-Jen Tsai⁷, Li-Ching Chang⁶, S-J Cathy Fann⁶, Hao Mei²⁴, Dabeeru C Rao²⁵, James E Hixson²⁶, Shufeng Chen⁸, Tomohiro Katsuya^{27,28}, Masato Isono⁹, Toshio Ogihara^{28,29}, John C Chambers³⁰, Weihua Zhang³⁰, Jaspal S Kooner³¹, The KidneyGen Consortium³², The CKDGen Consortium³², Eva Albrecht³³, The GUGC consortium³², Kazuhiko Yamamoto², Michiaki Kubo³⁴, Yusuke Nakamura³⁵, Naoyuki Kamatani³⁶, Norihiro Kato^{9,42}, Jiang He^{24,42}, Yuan-Tsong Chen^{6,42}, Yoon Shin Cho^{5,37,42}, E-Shyong Tai^{12,38,39,42} & Toshihiro Tanaka^{40,42}

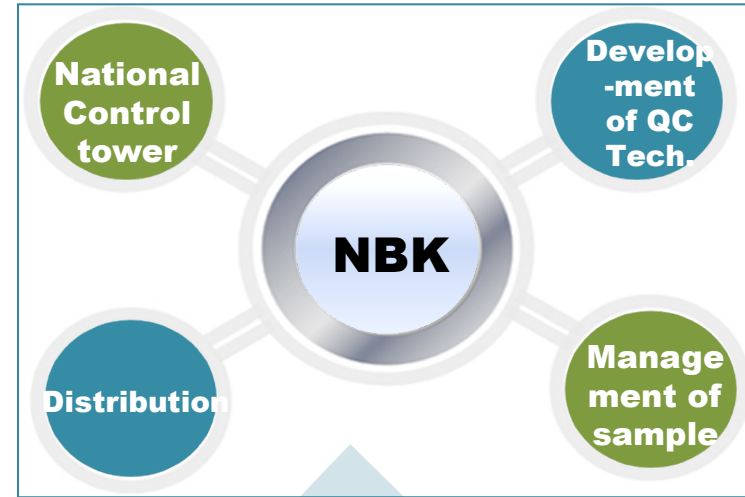
Current projects of CGS

- 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

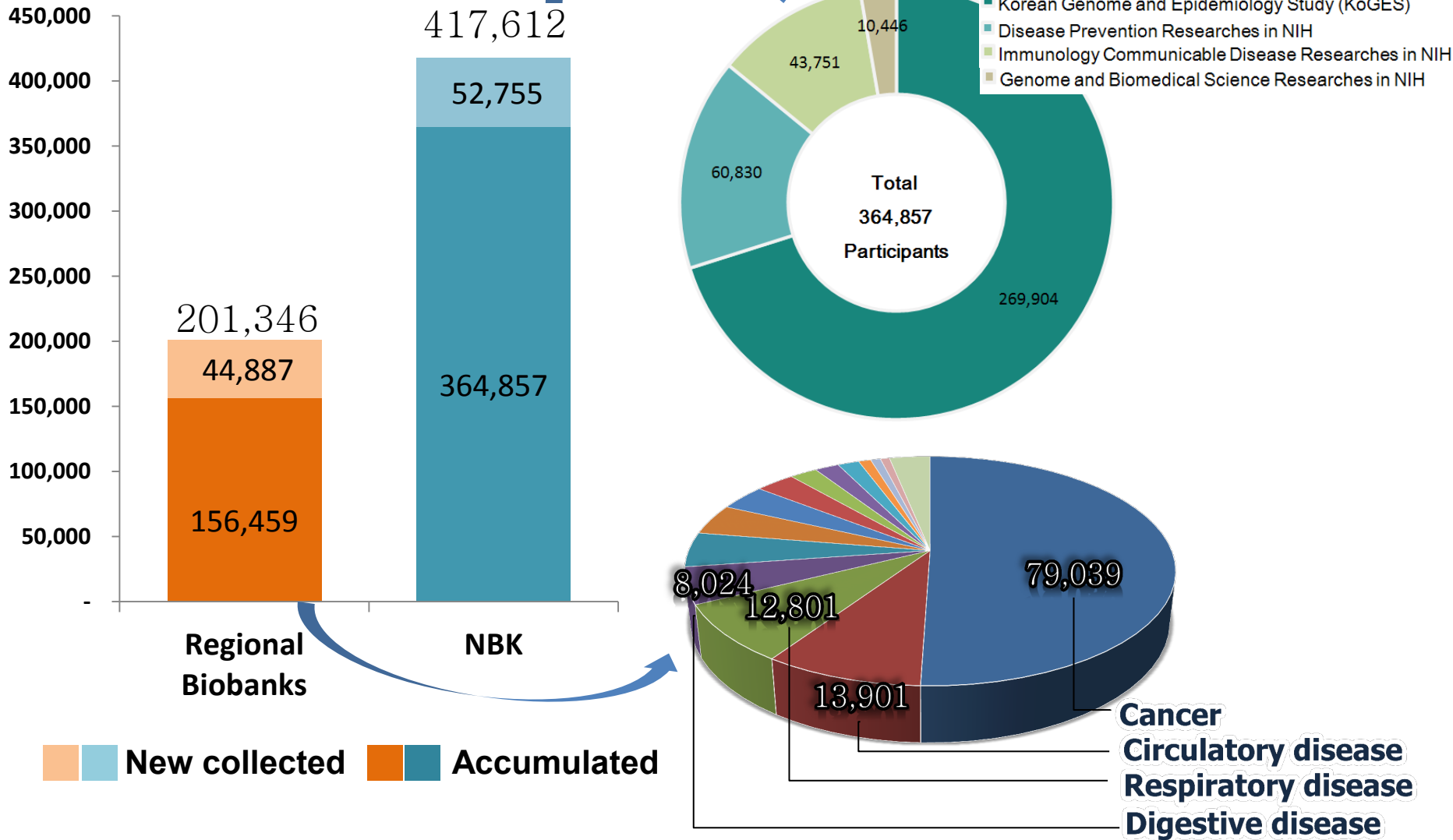
 NBK : National Biobank of Korea

 17 regional Biobank



* NUH: National University Hospital

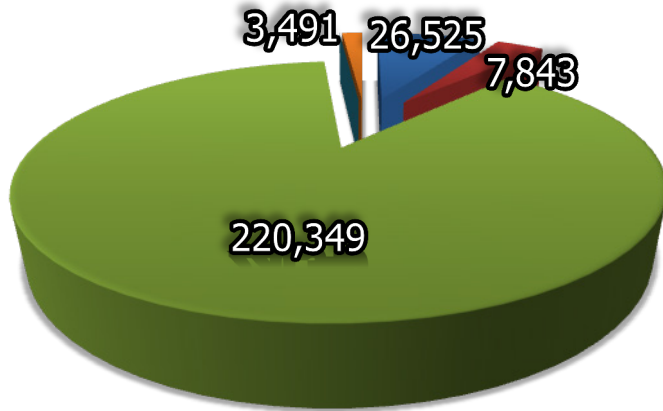
KBP Biospecimen Collection: 618,958 participants



Distribution of human biospecimens

As of Dec. 2013

NBK

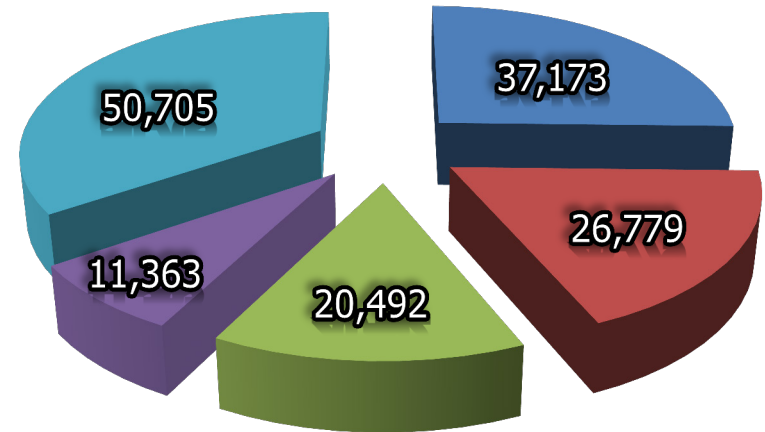


(258,208 vials)

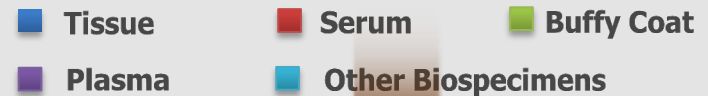


201 Research projects

17 Regional Biobanks



(146,510 vials)



712 Research Project

Total: 345,432 vials (913 Research Projects)

한국인체자원은행사업

KBN 한국인체자원은행네트워크
Korea Biobank Network

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사업목적 및 연혁**

코호트기반 인체자원

X 질병기반 인체자원

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활용성과

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- [일반공지] 생명윤리법 개정에 따른 인체유래물은행 운영방안
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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

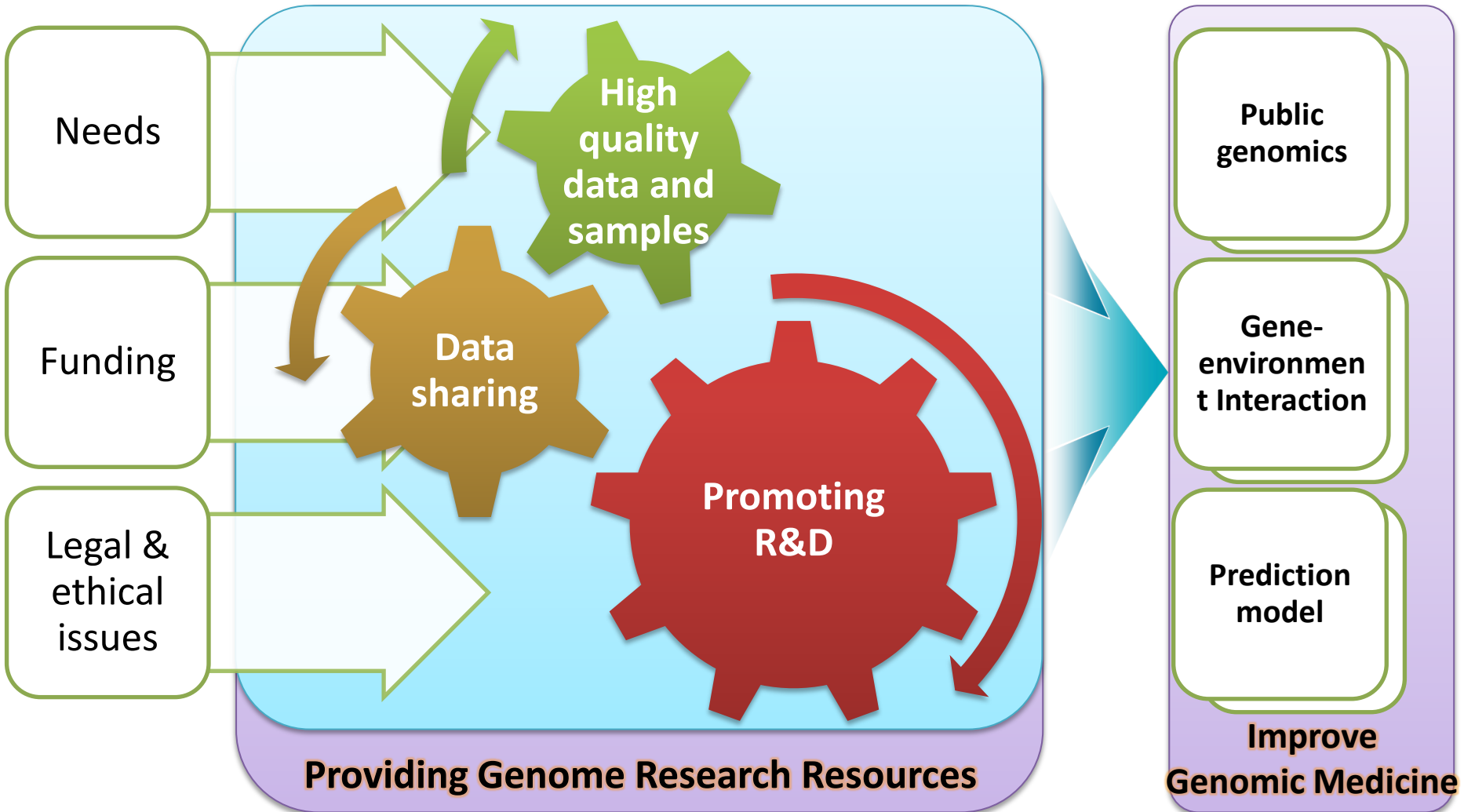
- whole genome sequencing on > 400 samples

– 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





Thank you