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Department of Biotechnology
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Subject: Priority areas of Human Genetics and Genome Analysis Task Force (HGGA)-reg.

Priority areas of Human Genetics and Genome Analysis Task Force may be categorized into three broad thematic areas underlying various unexplored issues which can provide the future roadmap of this program.

A. <u>Thematic area I- Genomics to Biology</u>
(Elucidating the structure and function of genomes)

(a) Resources development: Generation of large, publicly available, comprehensive sets of human genomic sequences and data (**scientific resources/infrastructure**) that along with other new, powerful technologies comprise a toolkit for genomics-based research (genomic maps and sequences).

1. **Infrastructure:** Creation of centres
   a. To produce large-scale sequence data on human diseases.
   b. To clinically diagnose and provide counselling on human genetic diseases.
   c. As data warehouses for open dissemination of human genomics data to a wider community of researchers and public with secured maintenance of patient privacy.
2. **Patient cohorts and Reagents:**
   a. Comprehensive collections of model organism's knockouts and knock-downs of all genes.
   b. Well-annotated patient and population cohorts for a specific clinical indication to assess the effect of gene(s) and/or variant(s) on various genetic and other disorders including healthy cohorts.
3. **Sequence Data:**
   a. Storage and generation of large and comprehensive set of genetic/genomic data on human diseases using Integrated OMICS.
   b. Generation of databases of sequence variation on human diseases.

(b) Technology development: The Human Genome Project was aided by several 'breakthrough' technological developments, including Sanger DNA sequencing and its automation, DNA-based genetic markers, large-insert cloning systems and the polymerase chain reaction. Even newer approaches, such as nanotechnology and microfluidics, are being developed, and need to be applied.

1. Human sequencing and genotyping technologies to reduce costs further and increase access to a wider range of investigators.
2. Identification and validation of functional elements in human genome.
3. In vivo, real-time monitoring of gene expression and the localization, specificity, modification and activity/kinetics of gene products.
4. Modulation of expression of all gene products using, for example, large-scale mutagenesis, small-molecule inhibitors and knock-down approaches (such as RNA-mediated inhibition)
5. Monitoring of the absolute abundance of any protein (including membrane proteins, proteins at low abundance and all modified forms).
6. Improved imaging methods that allow non-invasive molecular phenotyping.

7. Correlating genetic variation to human health and disease using haplotype information or comprehensive variation information.
8. Laboratory-based phenotyping, including the use of protein affinity reagents, proteomic approaches and analysis of gene expression.
9. Linking molecular profiles to biology, particularly pathway biology to disease.
10. Understanding evolutionary variation across species and relevance to human diseases.

B. <u>Thematic area II- Genomics to Health</u>

(Translating genome-based knowledge into health benefits)

a. Develop Network programs on the underlying categories

I. Monogenic disorders
II. Polygenic disorders
III. Chromosomal disorders
IV. Mitochondrial disorders

b. Functional genomics:

1. Identify genes and pathways and their mechanism to interact with environmental factors to cause human diseases.
2. Develop, evaluate and apply genome based diagnostic methods for the prediction of susceptibility to disease, the prediction of drug response, the early detection of illness and the accurate molecular classification of disease.
3. Develop and deploy methods that catalyse the translation of genomic information into therapeutic advances.
4. Develop strategies to identify gene variants that contribute to good health and disease resistance.

C. <u>Thematic area III- Genomics to Society</u>

(Promoting the use of genomics to maximize benefits and minimize harms)

1. Make available the products of genomics to society at large in terms of molecular diagnosis of disease types, antenatal diagnosis, new born screening, pharmaco-genomics etc.
2. Population study for disease screening with emphasis on clinical and genetic counselling.
3. Define policy options, and their potential consequences, for the use of genomic information and for the ethical boundaries around genomics research.