Saudi Human Genome Project
Executive Summary

B - Executive Summary

This project will identify normal and disease causing genetic variants in the Saudi people. This historic project is the largest and most comprehensive gene discovery effort ever undertaken anywhere in the world, and provides a model for other nations to follow in applying genomics to make a major impact on national healthcare.

Goals, Scope and Impact

The project high level goals are to:
- Create the foundation for personalized medicine in the Kingdom
- Build infrastructure and capacity in the rapidly advancing field of genomics

The major project scope is:
- Three year duration
- ~SAR300 Million budget
- Create a national network of 7 genome sequencing centers
- Read ~20,000 Saudi genomes representing normal and disease conditions
- Analyze data to find the Saudi-specific gene variants
- Create a Knowledgebase of gene variants, both normal and disease-causing

The project major impacts will be:
- Enabling genetic tests for disease risk in the Saudi population, which can be used to lower healthcare costs and improve quality of life, either by screening to prevent disease-impacted births, (pre-natal, newborn, pre-marital), or to screen the population and guide prevention or therapy (e.g. for Diabetes).
- Providing a rapidly scalable genomics capacity, in the form of a modular genome sequencing lab that can be rapidly deployed into universities and healthcare centers not involved in the initial project
- Establish world leadership for Saudi Arabia and KACST in genomics for genetic disease research and personalized medicine

Project Major Value Proposition: Immediate Healthcare Savings

- The current national healthcare expenditure is ~SAR100 Billion per year
- A substantial fraction of this is due to 8% of births impacted by severe inherited disease, and 30% of the population impacted by common genetic-related diseases such as Diabetes and cardiovascular disorders
- Genetic screening could reduce these burdens and repay the cost of the effort in just a few years, once the underlying causal genes and variants are known
- For example, treatment for a newborn with a severe metabolic disorder can cost SAR500,000 per year, and approximately 400 such children are born each year in the Kingdom, which alone results in SAR200 Million per year in healthcare expenditure due to new cases alone adding in
other severe genetic diseases, the healthcare cost of impacted newborns easily exceeds SAR1 Billion per year. In the few cases where pre-natal or pre-marital screening is currently available, action is usually taken to avoid having an impacted child. Thus annual savings of SAR500 Million could be realized as soon as the genetic factors are discovered and deployed into a proper screening program, such as the existing pre-natal or pre-marital screening efforts.

- Similar large, immediate saving could result from screening and preventative actions to delay the onset of common diseases such as Diabetes

Project Major Deliverables:

1. **Establish Network of 7 Sequencing Laboratories:**
   - 1 training laboratory for genome sequencing capacity building
   - 1 central genome sequencing production laboratory at KACST
   - 5 satellite genome sequencing laboratories at sites across the Kingdom

2. **Catalogue Saudi-Specific Mutation in KNOWN Disease Genes**
   - Sequence 3000 normal and disease patient samples on panels of known disease genes for all major sever inherited genetic disease categories (metabolism, vision, hearing, skeletal formation, muscle weakness, cognitive, neurological, immune system, breathing, blood) and record the mutations found in these known genes

3. **Catalogue Normal Genetic Variation in the Saudi Population**
   - Sequence 1000 normal subjects from the general population

4. **Catalogue the Mutations for Recessive and Common Genetic Disorders of UNKNOWN CAUSE in the Kingdom**
   - Sequence 19,000 subjects representing recessive and common, and carry out a major gene discovery effort to identify these new disease causing variants

5. **Create a National Knowledgebase of Genetics Findings**