Healthcare Initiatives for National Economic Strategy

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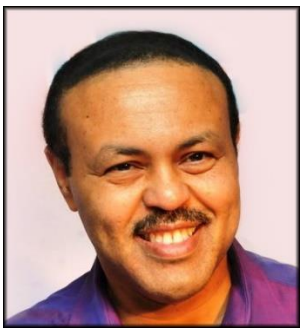

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Summary
Sudan is moving towards a transitional phase where priorities must be directed towards reconstruction, rehabilitation of the country's infrastructure and development of emergency and alternative health plans, education and basic life needs. In the area of health, there is an urgent need for public health including primary, secondary and tertiary health, care free of charge, to the Sudanese people. However, in the long run, this will be a significant burden on the government and budget unless there is real investment in the health system to bring high revenues to the state and significantly reduce the cost of health services as well as accuracy and safe management.

In this context, we must think deeply into the future with a distant vision to prepare for modern efficient medical care. While fulfilling the essential needs of the health system, we must think in parallel about how to achieve cost-effective medical services and build an infrastructure in preparation. This can be achieved through health care initiatives of the national economic strategy. In this regard, I have prepared two detailed initiatives to achieve sustainable health care that contribute to the construction of the Sudanese economy and achieve optimal and effective health care without burden on the citizen:

1. Establish a health biotechnology complex built to improve health services, education and research and make scientific and productive activities economically sustainable while contributing to the country's economy using advanced science, especially health biotechnology and molecular medicine. This project provides the latest health system in research and redevelopment (R&D) for the discovery and production of health materials for health service providers and to the market, education, and management of modern non-communicable and infectious molecular diseases (diagnosis, treatment and prevention) in close collaboration with academia, industry and health authorities.
2. Pave the way for precision medicine in Sudan through the establishment of the Sudanese Genome Project. The creation of the human genome and advances in proteomics have led to rapid advances in health care where it is informed by each person's unique clinical, genetic, genetic and environmental information. These scientists and doctors have helped develop targeted diagnoses and therapeutic approaches for more personalized management by identifying individual susceptibility to disease and responding to treatment. Thus, precision medicine can help:
    a) Elimination of redundant treatments
    b) B) Reducing the side effects of medicines
    c) Prevention and prediction of the disease
    d) Early intervention and reduction of health care costs

In this context, precision medicine has become an important medical practice in the developed world benefiting in all directions towards wellness and enabling approaches designed for prevention and care of the individual or community. In the most efficient way and at the lowest cost.

The Sudanese Genome Project is a pioneering initiative that requires high-level government support and in partnership with international pharmaceutical companies developed through academic innovation and partnership between national and international universities. The project provides a unique opportunity to create a model business platform for innovation and personalized medicine on a population scale - in a global context. By integrating and creating genomic and clinical biological data. The Sudan Genome Project will: 1) develop a globally competitive academic research program; 2) strengthen government programs to improve human health and provide accurate health services; 3) establish a reference facility to serve the clinical genome and service to Sudan; and 4) drive innovation in the discovery of medicines that will have 5) build new and cost-effective screening tools and clinical decision-making at the population level; 6) build capacity and create jobs.

ملخص
يمضي السودان نحو مرحلة انتقالية حيث يجب توجيه الأولويات نحو إعادة الإعمار وإصلاح البنية التحتية للبلد ووضع خطط طارئة وخطط بديلة في مجال الصحة والتعليم واحتياجات الحياة الأساسية. ي مجال الصحة ، هناك حاجة عاجلة للصحة العامة بما في ذلك الرعاية الصحية الأولية والثانوية والثالثة مجاناً للشعب السوداني. ولكن ، على المدى الطويل ، سيشكل هذا عبئًا كبيرًا على الحكومة والميزانية ما لم يكن هناك استثمار حقيقي في النظام الصحي لجلب إيرادات عالية للدولة وتقليل تكلفة الخدمات الصحية بشكل كبير بالإضافة إلى توخي الدقة والإدارة الآمنة.

في هذا السياق ، يجب أن نفكر بعمق في المستقبل مع رؤية بعيدة للتحضير لرعاية طبية تتسم بالكفاءة الحديثة. أثناء القيام بالاحتياجات الضرورية في النظام الصحي ، يجب علينا التفكير بشكل متوازٍ في كيفية تحقيق خدمات طبية مناسبة من حيث التكلفة وبناء بنية أساسية لها في الإعداد. يمكن تحقيق ذلك من خلال مبادرات الرعاية الصحية للاستراتيجية الاقتصادية الوطنية . وفي هذا الشأن أعددت مبادرتين مفصلتين لتحقيق رعاية صحية مستدامة تسهم في بناء الإقتصاد السوداني وتحقق الرعاية الصحية المثلى والناجعة دون عبء على المواطن:

1. إنشاء مجمع للتكنولوجيا الحيوية الصحية مبني بشكل كبير لتحسين الخدمات الصحية والتعليم والبحوث وجعل الأنشطة العلمية والإنتاجية مستدامة اقتصاديًا مع المساهمة في اقتصاد البلد وذلك باستخدام العلوم المتقدمة ، وخاصة التكنولوجيا الحيوية الصحية والطب الجزيئي ، حيث يوفر هذا المشروع أحدث نظام صحي في مجال البحوث وإعادة التطوير (R&D) لاكتشاف وإنتاج المواد الصحية لمقدمي الخدمات الصحية وإلى السوق ، والتعليم ، وإدارة الأمراض الجزيئية الحديثة غير المنقولة والمعدية (التشخيص والعلاج والوقاية) بالتعاون الوثيق مع الهيئات الأكاديمية والصناعة والسلطات الصحية.
2. تمهيد الطريق للطب الدقيق في السودان من خلال إنشاء مشروع الجينوم السوداني. فالمعروف أن إنشاء الجينوم البشري والتقدم في علم البروتينات فد أدى إلى تقدم سريع في مجال الرعاية الصحية حيث يتم احاطته بالمعلومات السريرية والوراثية والجينية والبيئية الفريدة لكل شخص. وقد ساعد هؤلاء العلماء والأطباء على تطوير تشخيصات وأساليب علاجية مستهدفة لتحقيق إدارة أكثر تخصيصًا من خلال تحديد القابلية الفردية للأمراض والاستجابة للعلاج. وبالتالي ، يمكن أن يساعد الطب الدقيق في:
   أ- القضاء على العلاجات زائدة عن الحاجة
   ب- خفض الآثار الجانبية للأدوية
   ج- الوقاية والتنبؤ بالمرض
   د- التدخل المبكر وخفض تكاليف الرعاية الصحية

في هذا السياق ، أصبح الطب الدقيق ممارسة طبية مهمة في العالم المتقدم يستفاد منه في كل الاتجاهات نحو العافية وتمكين النهج المصممة للوقاية والرعاية للفرد أو المجتمع. بأجع الطرق وبأقل تكلفة.

أما مشروع الجينوم السوداني فهو مبادرة رائدة تحتاج إلى دعم عالي المستوى من الحكومة وبالشراكة مع شركات الأدوية العالمية وضعت من خلال الابتكار الأكاديمي والشراكة بين الجامعات الوطنية والدولية. يوفر المشروع فرصة فريدة لإنشاء منصة تجارية نموذجية للابتكار والطب الشخصي على نطاق السكان - في سياق عالمي. من خلال دمج وإنشاء بيانات بيولوجية جينومية وسريرية . سيقوم مشروع السودان للجينوم بما يلي: 1) تطوير برنامج بحث أكاديمي تنافسي عالميًا ؛ 2) تعزيز البرامج الحكومية لتحسين صحة الإنسان وتوفير الخدمات الصحية الدقيقة ؛ 3) إنشاء مرفق مرجعي لخدمة الجينوم السريري وخدمة للسودان ؛ 4) دفع الابتكار في اكتشاف الأدوية التي سيكون لها أهمية عالمية ؛ و 5) بناء أدوات فحص جديدة وفعالة من حيث التكلفة وصنع القرار السريري على مستوى السكان ؛ 6) بناء القدرات وتوفير فرص عمل.

Introduction:

Sudan is proceeding to a transitional period were priorities are directed towards reconstruction and reforms of the infrastructure of the country and establishment of emergency and alternative plans in the field of Health, Education and Basic life needs. While in the field of health, urgencies are on Public Health including primary, secondary and tertiary health care, free of charge to the Sudanese people. But, on the long run, this will put a lot of burden on the government and the budget unless a real investment in the health system to bring high revenues to the state and to significantly reduce the cost of the health services and furthermore, provide accurate and safe management.

In this context, we must think deeply in future with a far vision to prepare for a modern efficient medical care. While doing the necessary needs in the health system we should consider in parallel how to achieve an appropriate cost-effective medical services and build infrastructure for it in the set up. This can be achieved by:

1. **Establishing a highly built-up Health Biotechnology Park to improve health services, education and research and to make the scientific and productive activities economically sustainable and contribute to the economy of the country.**

2. **Paving the way for Precision Medicine in Sudan by establishing the Sudan Genome Project**

I. Health Biotechnology Park

Health Biotechnology deals mainly with Molecular Medicine, which is a highly specialized part of Medicine. It explains the fundamental principles vital to an understanding of molecules that are keys to the normal functioning of the body and those related to the fundamental mechanisms of diseases. It also deals with the manipulation of these molecules to improve diagnosis, treatment, and prevention of disease. Human genome, gene regulation and expression in health and disease, genetic engineering and stem cells technology are important parts of Molecular Medicine. Principles of Molecular Medicine are then applied to the diagnosis and treatment of human disease.

Vision and Mission:

Using the advancement of science, in-particular Health Biotechnology and Molecular Medicine, this project provides a compressive state-of-the-art health organization system in Research and Redevelopment (R&D) for discoveries and production of health materials to health providers and to the market, Education, and Modern Molecular Disease Management (diagnosis, treatment and prevention) in close collaboration with Academia, Industry and Health Authorities.

Objectives:

To achieve this vision, the project adopts the following objectives:

1. *Promotion of Healthtech Start-ups and Innovation (R&D):* An area where discoveries in health care are made and developed for the benefit of all humankind. Also, producing Cost-effective Results by focusing on a particular specialized area for developing countries as for example, recombinant vaccines are relatively easy to reproduce and can be much more cost-effective way to deal with infectious diseases than drugs. Recombinant proteins to be used in investigations, therapy and evasion are generated within the park and no need to import such expensive materials.
2. *Front line Education:* By working closely with Universities programs providing B.Sc., M.Sc. and PhD in Health Biotechnology and Molecular Medicine can be developed to prepare candidates for a career in R&D, Education and Health Promotion and Management.
3. *Establishment of Molecular diagnosis, treatment, and prevention of disease:* This is achieved by the use of modern Health Biotechnology and Molecular Medicine tools to provide:

    a) diagnostic services for gene regulated (e.g. diabetes, hypertension, cardiovascular, neurological diseases, etc.) and heritable disorders including chromosome, DNA, and biochemical analyses,
    b) clinical genetics services and specialty clinics for evaluation, diagnosis, counseling, and education for the adult and pediatric population,
    c) screening in consultative and/or treatment of disorders included in newborn screening programs
    d) prenatal genetics services in pregnancy including pre-conception counseling and pre-implantation genetic testing (PGT), risk assessment, teratogen information, and prenatal diagnosis,
    e) identify and describe the patterns of birth defects and to assess the impact of congenital malformations upon infants, families, and to collaborate with others to determine factors involved in their etiologies in order to develop insights into primary prevention.

4. *Development of Stem Cells Banking and Therapy:* This innovative goal will provide the capability for performing all types of stem cell transplantation, cellular and gene therapy
5. Promotion of laboratory participation in quality assessment schemes
6. Set up a reference library of genetic disorders to serve clinical practice, education and research

Strategic evaluation:

The geographical and the cultural site occupied by the Sudan on its various applications is the most important success factor that can be relied upon to establish a specialized Health Biotechnology Park. In addition to the presence of a strong infrastructure and a variety of medical specialties and Sudanese scientific expertise around the world contribute significantly to maximize the chances of success and excellence in the field of Health Biotechnology and Molecular Medicine.

However, with these strengths, there is a series of challenges that lie ahead in achieving the goals in this project, the nature and modern of Health Biotechnology and Molecular Medicine techniques, curricula, programs and research, which requires a great deal of flexibility and innovation and follow-up and provide the resources and capabilities to keep abreast of the latest developments and the achievement of excellence required. In addition, the availability of financial resources is the greatest challenge of such pilot projects, particularly in such times.

Therefore, the project should rely on the support of the country higher authority in the first years with continuing work to diversify income sources in order to achieve greater financial independence.

In view of the modern field of Health Biotechnology and Molecular Medicine with the lack of specialized personnel within the country and the high cost of recruitment of outstanding scientific personnel, there is a strong need to build the cadre of technical and scientific staff for continuous development in this area. All these challenges open minds to explore opportunities that could be seized to achieve the vision and objectives of the project. Examples of these opportunities:

1. Develop strategic partnerships with the centers of excellence abroad where our people have been working or have strong contacts.
2. There are great opportunities for cooperation with institutions in the field of sports anti-doping in Africa and Middle East.
3. The need of many of the hospitals working in the area of specialized centers for the purposes of scientific analysis, molecular and genetic diagnosis.
4. There are some individuals from businessmen and others who wish to support research projects and the quality of health and molecular medicine.
5. The absence of similar projects in the developing countries, making the project a comfortable competitive position to provide biotechnology materials, diagnostic and therapeutic services.

Strategic directions:

This plan shows four strategic directions, namely:

1. The promotion of integration between the various disciplines of Health Biotechnology and Molecular Medicine in the areas of R&D, Education and community service.
2. Focus on capacity-building in Africa and Middle East in the field of scientific research, Education and diagnostic and treatment services in the fields of molecular genetics and hereditary diseases.
3. Promote scientific research and applied external consultations and cooperation with regional and global centers, and link status similar centers in the region and the world.
4. Seeking for financial independence by diversifying sources of traditional and non-traditional income.

Strategies:

To achieve the objectives of the project, the following strategies were developed:

1. The preparation of the infrastructure of the Park and the processing according to the latest techniques and international standards.
2. Setup of Healthtech regulations.
3. Setup of Healthtech Accelerators.
4. Setup of Digital Health Academy.
5. Strengthen the partnership with the Governmental departments and private sector, such as industries, hospitals and educational institutions and the exploitation of knowledge in the strategic deployment of the concept of Health Biotechnology and Molecular Medicine.
6. Prepare a marketing campaign to define the status and activities of the park.
7. Develop a strategic and working plan by a Houses of global experience to strengthen the own resources of the park.
8. Strengthening the competitive environment and integration with local centers and regional and global levels.
9. Development of programs and platforms for Health Biotechnology and Molecular Medicine on a continuous basis.
10. Preparation and presentation of professional development programs in the field of Health Biotechnology and Molecular Medicine.
11. Promotion of scientific publishing and doctoral theses in the field of Health Biotechnology and Molecular Medicine.
12. Building a database of all workers in the field of Health Biotechnology and Molecular Medicine in the country.

Values

The strategic values believe of the following:

This proposal working to serve the country, the continent and the Middle East in the field of Health Biotechnology and Molecular Medicine in an atmosphere of cooperation and integration. Also, to care about quality, professional and ethical controls and proficiency in all aspects of research, education and services. We also believe that every area of science and especially Health Biotechnology and Molecular Medicine challenges, therefore there have to be creativity and innovation and responsibility to provide excellent service and leadership in the field of Biotechnology and Molecular Medicine.

Scope of activities and funding sources

Action Plan of the centre is divided into three stages as follows:

1. Foundation stage (3 years):
   At this stage the focus is on the establishment of the site/building and the provision of equipment and key personnel. The financing of the park will be adopted during this phase mainly on the government and founders.

2. The first operational phase (3 years):
   This stage is to execute activities in research education and services and it focus mainly on the local level with the public sector of the Ministry of Health and sports organizations and the private sector. It is anticipated at this stage to increase reliance on self-financing of the Center through the services provided by the Center to increase to 50%.

3. The second operational phase (4 years):
   This stage is the final of the plan, the focus is on expanding services to include the African Continent and the Middle East, the financing of the park depends on self-financing for services to be provided by local and regional levels.

Project Costs, Budget, Budgeting, Process, Partnerships, Clients, Revenue Streams, Stakeholder Analysis Identification, Performance Dashboard, SWOT Analysis and Proposed Operational Budget should be included in the final strategic and work plan to be performed by a house of experience.

Disciplines and Functional Units

(A) DIAGNOSTICS SERVICES

1. MOLECULAR DIAGNOSTICS UNIT

Background:

Genetic influences on disease cover a wide range of disorders extending from single-gene diseases such as sickle cell anemia to more complicated diseases such as Huntington's disease, breast cancer and asthma. In some cases, genetic tests are available today that detect variants in genes that contribute to disease risk, such as the test for defects in a gene called BRCA1 that is associated with breast cancer. In the future, many more genetic tests will become available to check for gene variants that increase risk to develop other diseases. Due to consanguinity (intermarriage custom), and the possible influence of environmental and life style factors, many genetic and inherited disorders are highly prevalent.

Services:

Increased health personnel and public awareness of the relationship between genetics and personal health has increased the demand for molecular genetic testing. This unit offers clients diagnostic services for vast arrays of diseases and conditions. Testing is generally divided into hematology, cancer, infectious, HLA/identity, inherited diseases and pharmacogenetics testing. Our laboratory satisfies the following diagnostic demands:

- Diagnosis and classification of neoplastic diseases to aid in prognosis, therapeutic monitoring and detection of minimal residual disease of cancer patients.
- Diagnosis of coagulation defects, genetic analysis of hematologic malignancies and evaluation of stem-cell transplant patients.
- Risk assessment for familial cancer syndromes.
- Aids in the diagnosis of acquired infectious diseases.
- Diagnosis of genetic disorders.
- Prenatal and pre-implantation diagnosis.

To achieve these goals, the Molecular Diagnostics Laboratory uses progressive molecular biology technologies and procedures including the use of PCR and specialized PCR techniques, DNA hybridization, RFLP, blotting techniques, sequencing, microarrays as well as other innovative techniques. A board certified laboratory director and trained staff should continually strive to develop new test methods to improve assays by increasing detection rates and decreasing turn around time which will result in improved patient care. Below are testing goals in the first operational phase:

Hematology
FVL
MTHFR
PT
Rh genotyping
Beta globin SCE
Alpha-Thalassemia

Hematopathology
BCR-ABL (Quant 9,22)
PML-RAR (Quant 15,17)
TEL-AML (12,21)
BCL1 (11,14)
BCL2 (14,18)
JAK2 (Quant)
T-Cell Clonality
B-Cell Clonality

Cancer Genetics
BRCA1/BRCA2
MSH2, MLH1, MSH6
MSI (LOH)

Medical Genetics
Connexin 26
Connexin 30
Fragile X
CF
FGFR3
Huntington disease
PWS/AS methylation
Y chromosome markers (AZF)
Focused tiling array

ID
DNA viruses (HSV1, HSV2, EBV, CMV, Parvovirus, etc)

HIV Qual/Quant
HCV Qual/Quant
GC/CT/HPV
GBS
MRSA

Pharmacogenomics
CYP 2D6
CYP 2C19
Warfarin
UGT1A1 (Irinotecan toxicity)

HLA Genotyping:
- HLA class I (A, B, C)
- HLA class II (DP, DQ, DR)

Gene Polymorphisms:
- Factor V-Leiden (G1691A)
- Factor II
- Factor VIII
- Factor VII
- Prothrombin G20210A
- Methylenetetrahydrofolate Reductase (MTHFR): C677T and A1298C.
- Apolipoprotein E (Apo E) genotyping
- Von Willebrand Factor (VWF)
- Alpha 1 antitrypsin

Analysis of Minimal Residual Disease (MRD)

Mitochondrial gene disease

2. CYTOGENETICS UNIT

Background:

Cytogenetics involves the study of human chromosomes in health and disease. Chromosomes studies are an important laboratory diagnostic procedure in prenatal diagnosis, in certain patients with mental retardation and multiple birth defects, in patients with abnormal sexual development, and in some cases of infertility or multiple miscarriages. Cytogenetics analysis is also useful in the study and treatment of patients

with malignancies and hematologic disorders. New techniques allow for increased resolution of chromosome banding patterns, permitting differentiation of a greater number of abnormalities.

It is evident now that abnormalities are responsible for a great deal of embryo wastage, which is reflected, partially, in increased miscarriage in older women. Higher incidence of chromosomal anomalies has been found in cases suffering from severe male infertility. Such abnormalities can be passed onto their off springs if patients are not screened by cytogenetics and molecular genetics means.

Services:

- Karyotyping by cell culture system: Post-natal and Pre-natal
- Fluorescent *In-Situ* Hybridization (FISH): Pre implantation, Pre-natal, Post Natal
- Detection of structural and numerical chromosomal abnormalities

- Single Cell PCR for: preimplantation genetic analysis (SCPCR)

1. Embryo selection with regard to HLA typing
2. Diagnosis of single gene defects in indicated families
3. Triplet repeat disorders.
4. Embryo Sexing.
5. qfPCR for aneoploidy (pre-/post-natal)
6. Y microdeletion (azospermia/severe oligospermia)
7. Fragile X

3. METABOLIC AND BIOCHEMICAL UNIT

Background:

An inborn error of metabolism (IEM) involves an interruption of normal biochemical processes. Sir Archbald Garrod first coined this term in 1908, when he observed that certain diseases are due to heritable blocks in normal metabolic flow. Since that time, many specific disorders have been found to be due to inborn error of metabolism. Most of these are inherited as single gene disorders. Others are inherited as mitochondrial gene disorders. The world wide incidence is 1/5000 live birth.

One of the major problems facing the physician in attempting to diagnose an IEM is the lack of a specific phenotypic presentation because of clinical variability. Patients will usually be referred for evaluation with one of the following presentation, infants admitted to rule out sepsis. Infants or children present with failure to thrive, developmental delay, mental retardation, seizures and sudden infant death syndrome (SIDS). School age children with loss of developmental mile stones may be tested for broad spectrum of neurodegenerative disorders. Additionally children with unusual stature may be evaluated for metabolic causes of inherited bone disease.

Services:

1. Neonatal Screening Section: this is intended to detect disorders in newborns for which interventions shortly after birth have obvious benefits.

   - Phenylketonuria
   - Maple syrup urine disease
   - Tyrosinemia type I
   - MCAD (medium-chain acyl-CoA dehydrogenase) deficiency
   - Glutaric aciduria type I
   - HMG-CoA lyase (3-hydroxy-3-methylglutaric acid-CoA lyase) deficiency
   - Long-chain hydroxyacyl-CoA dehydrogenase deficiency
   - Very-long-chain acyl- CoA dehydrogenase deficiency
   - 3-methylcrotonyl-CoA carboxylase deficiency
   - Isovaleric academia

   2. Specialized Clinical Biochemistry Section: This is intended to develop and set up the new tests that are not available in Sudan and in the region where samples are sent abroad for analysis.
   - Porphyrins in urine
   - Bile acids in serum
   - Monitoring oxidative stress: malondialdehyde in plasma/serum a lipid peroxidation marker, vitamin C in plasma/serum, glutathione in whole blood, coenzyme Q10 in serum/plasma or whole blood
   - Vitamin profiling: Vitamin B1 in whole blood, vitamin B2 (Riboflavin) in whole blood, vitamin B6 in plasma/serum and whole blood, vitamin A and vitamin E in serum and plasma
   - Risk factor for arteriosclerosis: homocysteine in plasma
   - Osteoporosis diagnosis: Urinary crosslinks, 25-OH-vitamin D3 in serum/plasma
   - Biogenic Amines: catecholamines in urine/plasma; urinary vanillylmandelic acid (VMA) and homovanillic acid (HVA), serotonin in urine/ serum, plasma, and whole blood
   - Hemoglobin testing: hemoglobin variants, ß-thalassemia
   - Therapeutic drug monitoring: Itraconazole and hydroxyl- Itraconazole in derum/plasma, amiodarone and desethylamiodarone in serum/plasma, benzodiazepines and tricyclic antidepressants, antiepileptic drugs in serum/plasma, levetiracetam in serum/plasma, mycophenolic acid in plasma/serum, Olanzapine and quetiapine in serum/plasma
   - Occupational Medicine: Hippuric acid, p-methylhippuric acid, and mandelic acid
   - Chronic Alcohol Abuse
   - Drug Abuse and anabolic steroids abuse for sport duping control authorities analyzed by HPLC and GC-MS

- Mutations analysis using Denaturing High-performance Liquid Chromatography
- Denaturing high-performance liquid chromatography (DHPLC), the new technology, based on temperature-modulated liquid chromatography and a high-resolution matrix is now implemented in a way suitable for clinical application.
- The examples of mutation analysis using DHPLC include:
- Phenylalanine hydroxylase gene in patients with phenylketonuria.
- Dystrophin gene in Duchenne Muscular Dystrophy
- BRCA1/2 gene and their loss of heterozygosity (LOH) in patient s with a strong family history of breast cancer
- p53 and ST7 tumor suppressor genes in gastric carcinoma
- Mutation analysis of the entire mitochondrial genome
- CFTR gene in cystic fibrosis patients
- Identification of mutations affecting male fertility and preimplantation genetic diagnosis

(B) CLINICAL GENETICS SERVICES

1. POPULATION GENETICS UNIT

Background:

This unit deals with community and clinical genetic issues. The main objective is to establish a program for the control of Hereditary and gene regulated Disorders in the country that include educational and service providing. The program needs to match the unique demographic, religious and cultural features of the population of the region. It will be conducted in collaboration with the National Programs introduced by the Ministries of Health in the region and will provide services covering disease prevention, health promotion and case management activities.

Services:

1. Establishing community genetic health education program
2. Preparing health education material to develop, produce and disseminate information materials on relevant genetic disorders, appropriate for the needs of health professionals, families with affected members, genetic defect carriers and general public.
3. Establishing Genetic registers for the rare genetic diseases detected in the population in the country coordinated with the unit of Birth Defect (see below)
4. Training of health care personnel in specific areas:
   Such as training in Clinical Genetics, Molecular Biology, Cytogenetics, Registration of Congenital Abnormalities, and Management of Common Genetic Disorders and Prenatal pathology and fetal medicine
5. Diploma and/or Master Course in Community Genetic for Physicians
6. Genetic Counseling Course for Nurses, Health educators etc.

2. BIRTH DEFECTS UNIT

Background:

Birth Defects are abnormal conditions that are present at birth. They can result in physical and/or mental disability and can be fatal. The number and type vary, depending on family history and the parents' age, race, ethnicity, diet, medical care, and exposure to harmful substances. Some common birth defects are spina bifida, cleft lip, and Down syndrome.
Birth defects are a serious problem, internationally; three out of every 100 pregnancies are affected by a medically concerning birth defect. For many birth defects, we do not yet know the cause.

For birth defects with known causes, most are the result of a chromosomal or genetic abnormality. Less than five percent of all birth defects are caused by known environmental agents

This program will involve efforts to achieve the following goals:

1. To identify, report, investigate, and monitor various annual trends in birth defects, high-risk populations, and high-risk locations.
2. Perform nationwide epidemiological surveillance for all reportable birth defects and evaluate possible association of birth defect with exposure to areas containing environmental hazards.
3. To reduce the incidence of birth defects in Sudan from preventable causes.
4. Monitor trends to learn more about the occurrence of birth defects in Sudan.
5. Provide an accurate, unduplicated, count of children with birth defects to other programs and agencies for program planning
6. Provide statistics to public health programs and health care professionals for use in planning and evaluation
7. Identify potential areas of unmet need
8. Help prevent secondary disabilities by making recommendations concerning special services needed in local communities
9. Promote education activities for the prevention of birth defects

Services:

As to Birth Defect Program, an immediate collaboration with the Ministry of Health to develop the SUDAN BIRTH DEFECT REGISTRY (SBDR), which is a surveillance program that provides reliable, valid, and timely information on the number of infants and young children with birth defects in Sudan.
This knowledge is essential to the efficient operation and evaluation of effective prevention programs, development of sound health policy, and appropriate allocation of resources. In addition, it provides the foundation for epidemiological studies into the causes of birth defects. The Registry is an early warning system for discovering excessive occurrences of birth defects and is the foundation for the epidemiological research needed to evaluate the clusters. Reducing the human and economic costs of birth defects represents an important public health opportunity to improve the overall quality of life for all of families.

Thus, the service will be conducted accordingly as follows:
When a child is identified through the Birth Defects Registry, staffs work to ensure that the child and the family are provided with best medical care, and are referred to appropriate services that will assist the family with medical and other needs.

SBDP also maintains information about a variety of support groups, nonprofit health agencies, and government programs for information and assistance.

Other services provided include professional and public education. Expert staff members should be available to address community and professional groups about birth defects.

2. PRENATAL DIAGNOSTIC UNIT

Background:

The majority of patients seen at department of Obstetrics and Gynecology are high risk cases. These are either to fetal causes such as intrauterine growth restriction or fetal abnormalities such as chromosomal, congenital structural abnormality or metabolic disorder. There are also abnormal pregnancies due to medical conditions complicating pregnancy like haemoglobinopathies which are common in certain areas of the country.

Services:

- Prenatal diagnosis using prenatal invasive procedures (chorionic villous sampling, amniocentesis /+/ - drainage, drainage of fetal abdominal or thoracic fluid collection, cordocentesis and intrauterine fetal transfusion
- Performing advanced ultrasound for high risk patients associated with fetal congenital abnormalities

(C) MEDICAL BIOTECHNOLOGY SERVICES

Background:

Recent rapid advances in genetic engineering, protein engineering, cell culture and molecular biology have generated a virtually unlimited potential for altering the capabilities of living systems, greatly expanding the scope of biotechnology, generating new applications for biological products and allowing an unprecedented ability to control life processes.

1. MOLECULAR AND CELLULAR IMMUNOLOGY UNIT

Molecular and Cellular Immunology provides the latest information and advancements in our understanding of the immunological response of the host, both *in vitro* and *in vivo*, by featuring basic research and clinical applications.

2. PHARMACOGENETICS UNIT

Pharmacogenetics is generally regarded as the study or clinical testing of genetic variation that gives rise to differing response to drugs, while pharmacogenomics is

the broader application of genomic technologies to new drug discovery and further characterization of older drugs. Cellular &

2. STEM CELLS BANKING AND THERAPY UNIT
This innovative unit will provide the capability for performing all types of stem cell transplantation, cellular and gene therapy

Services:
1. Development of new drugs and medical treatments by the use of Cellular and Molecular Immunology knowledge to obtain and produce new biopharmaceutical products utilized in the treatment of several disorders as cancer and chronic diseases and introduce them to the Public Healthcare System.
2. Umbilical Cord Blood Stem Cell Transplant for the treatment of several diseases as for example: Leukemia, Sickle Cell anemia, Thalassemia, Aplastic anemia, Thrombocytopenia, Lymphomas, Multiple Myeloma, Metabolic Disorders
3. Provide Education and Research in:
   - Molecular and Cellular Immunology
   - Pharmacogenetics
   - Stem cells technology including drug development and toxicity tests studies on development and gene control.

(C) RESEARCH AND EDUCATION SEVICES

Background:
This unit coordinates and arranges facilities for research conducted by all units and will establish basic science and clinical research in relation to Health Biotechnology and Molecular medicine. The research projects address mechanisms in biological systems, computational biology, structural biology and approaches to diagnostics, prophylaxis and therapy. The projects deal with molecular mechanisms behind regulation, activation and dysfunction in gene regulated disorders. The goals are to understand these mechanisms in order to achieve better management of devastating diseases. The projects will focus in signaling pathways, which include stimulants, receptors, second messenger systems, transcription factors, gene expression, protein synthesis and production of effector molecules.

The Unit will coordinate the education of Molecular Medicine and the PhD program

Services:
Contract Research:
This is intended to provide special analytical services to facilitate research in the academic institutions, biological, biochemical and pharmaceutical industries.

Full Method Validation:
We offer a complete method validation service examining all aspects of the methodology from accuracy and precision through to long term analyte stability. Provide a full validation protocol designed for bio-analytical methods.

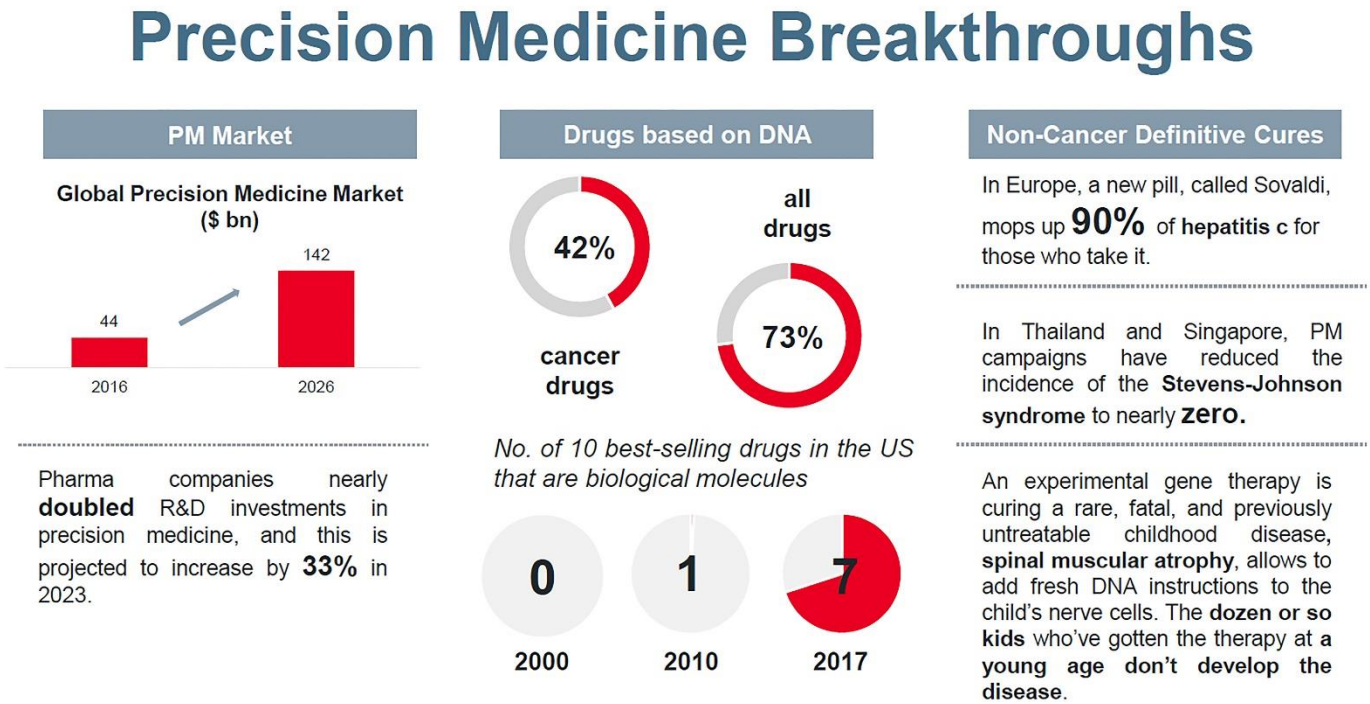
II. Paving the way for Precision Medicine in Sudan

Precision Medicine

Establishment of the human genome and advances on proteomics has led to a rapidly advancing field of healthcare that is informed by each person's unique clinical, genetic, genomic makeup, and environmental information. These helped scientists and doctors to develop targeted diagnostics and therapeutic approaches to achieve more personalized management by identifying individual susceptibility to disease and response to treatment. Thus, Precision Medicine can help in:

1. Elimination of redundant treatments
2. Decreasing side effects of drugs
3. Prevention and prediction of disease
4. Earlier intervention and reduction of health care costs

In this context, Precision Medicine has become an important medical practice in the advanced world that capitalizes on the trends toward wellness and consumerism to enable tailored approaches to prevention and care for an individual or a community.



The Sudan Genome Project

Sudan Genome Project is a leading initiative needs a high level support from the Government. Developed through academic innovation and partnership between national and international universities. The project provides a unique opportunity to create an exemplar commercial platform for innovation and personalized medicine at population scale—in a global context. By integrating and creating genomic and clinical biodata on up to 100,000 individuals from Sudan, Sudan Genome Project will: 1) develop a globally competitive academic research programme; 2) foster government programs to improve human health and provide precision health services; 3) create a reference clinical genomics facility and service for Sudan; 4) drive innovation in drug discovery that will have global relevance; and 5) build novel and cost-effective screening and clinical decision making tools at population level.

Background

The last decade has seen widespread implementation of electronic health records (EHRs) globally. Large-scale real-world data provide unique opportunities to better understand predictive factors of clinical outcomes, and stratify individuals by risk of disease to allow for better personalized care, and potentially cost savings to health services and providers. According to the National Academy of Medicine (formerly the Institute of Medicine), the U.S. health care system spends almost a third of its resources — $750 billion annually — on unnecessary services and inefficient care. New predictive analytics and personalized medicine tools promise to improve cost efficiencies and improve care by forecasting the likelihood of an event and allowing providers to tailor treatments and services accordingly. These tools can be applied across the continuum of care, from disease surveillance to chronic disease prevention to identifying patients who are at risk of deterioration. Personalized medicine solutions in this field can have an enormous impact in areas where medical expertise is sparse or non-existent. As on-line data and predictive systems become pervasive, they allow for faster and more precise decision-aide tools for healthcare providers.

The demographic history of the human population with extensive migration, separation, and historical bottlenecks followed by expansions has resulted in distinct patterns of human genetic variation across the world. This has resulted in high levels of population or continent-specific genetic variation, providing unique opportunities to understand the genetic determinants of phenotypic traits across populations. Genetically diverse population resources provide unique opportunities to identify loci functionally important in regulation of different biological mechanisms and traits, thereby providing candidate drug targets.

The Sudanese population is diverse, and includes multiple ethnic groups from across the middle east. Furthermore, the high levels of consanguinity have resulted in a high burden of recessive genetic disorders in the population. Consanguinity can lead to autozygous stretches within the genome that are likely

to harbor loss of function (LoF) mutations leading to complete inactivation or dysfunction of genes. These regions provide a unique opportunity to examine gene function by directly examining clinical syndromes among individuals with gene knock outs in these regions of the genome. Therefore, examination of genomic variation in Africa has direct impact on global drug discovery and development.

Examine genome sequences in consanguineous populations can also greatly facilitate examination of mendelian disorders. Very rare recessive mutations are present in every population but since they rarely achieve homozygosity in outbreeding populations (consistent with Hardy-Weinberg equilibrium), they are unlikely to be associated with clinical phenotypes. However, consanguinity dramatically increase the probability of being homozygous at any genetic locus in the offspring. Studying these populations considerably increase the probability of observing homozygous gene knockouts within the population. This applies to the study of mendelian disorders, where recessive causes of highly penetrant syndromes are likely to be enriched within consanguineous pedigrees. However, examining monogenic causes of complex disease (e.g. familial cholesterolaemia) can provide important insights into biological mechanisms underlying complex disease.

However, despite the importance of consanguinity to genetic research, these populations remain understudied and underrepresented in large-scale sequencing efforts globally.

Vision
To use genomic analysis to clinically implement Precision Medicine and tailor novel and improve patient care

Mission
The mission of the project is to identify genetic basis of disease in the Sudanese population and to accelerate discovery in genomics to enhance translational research into effective and safe individualized health care.

Objectives
The main aim is to use next generation technology and bioinformatics to whole genome sequence of Sudanese population.

Specific objectives:

1. Create a reference clinical genomics facility and service for Sudan.
2. Promote the clinical practice of genomic medicine, as a paradigm for health care in Sudan
3. Drive innovation in drug discovery that will have global relevance.
4. Build novel and cost-effective screening and clinical decision making tools at population level.
5. Contribute to training and capacity building through:

a) Continuing Medical Education (CMEs) on Precision Medicine, Genomics and their tools for physicians, genetic counselors, medical geneticists and other health staff and furthermore, qualify them to be able to discuss Precision Medicine with patients;
b) Medical Curriculum Reform to ensure that medical students get additional up-to-date exposure and training in topics related to genetics and genomics;

6. Integrate education, research- development and healthcare in an advanced manner for the benefit of the community;
7. Strengthen partnership with local, regional and international organizations and institutions.

Methods

As the Genomic/Precision Medicine is rapidly advancing, it is very important to ensure that medical professionals have the knowledge and skills needed to keep up with the technical intricacies of genetic and genomic information including genetic testing specifics, screening options, Precision Medicine and risk assessment.

The above objectives are accomplished through the following activities.

A. Well-established and recognized educational platforms.
B. Training courses on Bioinformatics and learn how to do genome analysis.
C. Workshops.