Genomexcell

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Center of Excellence In Genomic Medicine Research

Newsletter

Center of EXCELLENCE In Genomic Medicine Research

Special Features

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Highly Cited Researchers Visit CEGMR

Highly Cited Researcher's Visit to CEGMR, KAU

As part of the strategic goal to encourage and enhance its multidisciplinary research programmes and initiate strong collaborations with other leading institutions around the globe, the Highly-cited Researcher Programme of the Center of Excellence in Genomic Medicine Research (CEGMR) has incorporated seven highly-cited researchers in its team. The CEGMR's Highly-cited Researcher Programme is aimed at collaborating and working in tandem with scientists and their laboratories that have made highly significant contributions to the advancement of science and technology in past decades.

CEGMR was recently honored to receive all the seven Hi Ci Professors affiliated in the Hi Ci Program (15th-17th of November 2012 (corresponding 1-3/01/1434). They were received by the CEGMR management including Dr. Mohammed H. Al Qahtani, CEGMR Executive Director and the Vice Directors, Dr. Adel Abuzenadah and Dr. Adeel Chaudhary. The three-day visit was commenced by group meetings involving the CEGMR researchers' affiliates with each one of the Hi Ci Professors followed by cutting edge seminars given by all the Hi Ci Professors on the second day of their visit. They also met Prof. Adnan Zahed, Vice President for Graduate Studies and Scientific Research, to discuss aspects of mutual cooperation in scientific research in line with KAU's policy of seeking to provide a high-quality education and taking advantage of the capacity and expertise of other local and foreign universities and scientists.

The seven highly cited (HiCi) researchers include;

Prof Jr. Albert Joseph Fornace, Director of Waters Center of Innovation for Metabolomics at GTU; previously Director of the John B. Little Center for the Radiation Sciences and Environmental Health at Harvard University, Washington DC, USA. He works in the fields of DNA damage and Repair, Stress response (oncogenic and genotoxic), functional genomics and metabolomics in stress response. As professor of biochemistry and molecular & cellular biology and oncology, Prof Forance is committed to researching how environmental stresses can cause normal cells to become cancerous—and is developing ways to stop this from occurring or to exploit differences between normal and malignant cells therapeutically. His innovative work in the area of cellular responses to radiation and other environmental toxins has earned him the Molecular Cancer Research Chair at the Lombardi Comprehensive Cancer Center.

Prof Norbert Perrimon, Professor of Genetics at the Department of Genetics, Harvard Medical School Howard Hughes Medical Institute Boston, USA. Prof Perrimon has thirty years of experience in the fields of developmental genetics, signal transduction and genomics. He has trained more than 80 students and postdoctoral fellows with most of them currently holding academic positions.

Prof. Thomas C. Südhof, of the Department of Molecular & Cellular Physiology, Department of Neurology and of Psychiatry & Behavioural Sciences, Stanford University, School of Medicine, Stanford, USA. He is currently the Avram Goldstein Professor in the School of Medicine as well as a Professor of Molecular & Cellular Physiology, Psychiatry, and Neurology. During his career, he defined himself as a clear leader in synaptic research, and his work remains on the frontiers of discovery to this day.

Prof. Jerry W. Shay, Director, Cancer Biology Department of Cell Biology University of Texas Southwestern Medical Center, Dallas, USA. He is a Professor of Cell Biology and Neuroscience at the University of Texas Southwestern Medical Center in Dallas. Dr. Shay has served as a panel member of the NIH Mammalian Genetics Study Section, the National Executive Council of the Tissue Culture Association, and he was recently elected to a four year term on the Board of Directors of the International Society of Differentiation.

Prof. Paolo Sassone-Corsi, Director, Center for Epigenetics and Metabolism, Distinguished Professor and Chair, Department of Pharmacology, University of California, Irvine School of Medicine, California, USA. He is responsible for key findings on the genetic activity behind a number of human molecular functions, has joined University of California, USA as distinguished Professor and Chair of Pharmacology. Earlier this year, his research group published its discovery that one of these regulator proteins -- named CLOCK -- modulates changes in histones, which influence the rhythm in which the DNA issues genetic messages. These changes are tightly linked to cellular energy levels and are important because about 15 per cent of all human gene activities oscillate in a circadian manner.

Prof. Foo Y Liew, Gardiner Professor and Senior Research Fellow at the Institute of Infection, Immunity and Inflammation, College of Medicine, Veterinary and Life Sciences, University of Glasgow, UK. He is unique in the West of Scotland's life science community. He is one of the highly cited researchers in Immunology as well as highly regarded and experienced figure in the area of inflammation research, particularly in relation to allergy, asthma and rheumatoid arthritis.

Prof. Hirokawa, Nobutaka, Distinguished Project Professor at the Department of Cell Biology and Anatomy, and the Department of Molecular Structure and Dynamics, University of Tokyo, Japan. He is also the President of International Human Frontier Science Program (HFSP). Highly respected in international scientific circle, he has been invited as editorial board member of international top journal such as Cell, Science, Neuron, Dev Cell, JCB, and EMBO J.

The Hi Ci Professors' visit was concluded after the Scientific Advisors Board (SAB) meeting on the 17th of November, 2012. All the Hi Ci Professors displayed pleasure with the available facilities at the CEGMR and commended its activities. At the end of their visit, they thanked CEGMR for the hospitality shown to them, and wished everybody success in his endeavors.

CEGMR Research Presentations in International Conferences

A. The Third European Congress of Immunology, Glasgow, UK, 2012

Prof.Foo Yew Liew FRS, who is currently affiliated with the Hi Ci program at CEGMR, was the President of the recently held European Congress of Immunology in Glasgow from 5th to 8th of September, 2012. Prof.Liew presented the ECI President's address as well as a lecture and Nobel Laureate Prof. Martin Evans, has presented the keynote speech in the first day of the ECI 2012.

ECI 2012, with the theme "A healthier future through research, education and innovation" had four parallel tracks with equal emphasis on basic and clinical immunology. There were 24 symposia and 72 workshops covering all aspects of immunology: Innate immunity, Adaptive immunity, Diseases of the immune system, and Immune interventions.

The speakers were leading basic and clinical immunologists from all over the world. Several European Societies dealing with immunology, mostly in applied sciences have shown enormous enthusiasm in organizing additional symposia.

The congress, with more than 5000 participants, the largest gathering of the European immunologists, provided a unique environment to network with colleagues from all over Europe and beyond. The Scottish Exhibition and Convention Centre, a modern and well planned facility, provided a relaxed and interactive venue for exciting scientific exchange and to view the latest developments offered by related industry.

Over 500 speakers from 30 countries around the World have presented their research at ECI2012. Dr.Peter Natesan Pushparaj has presented two oral and two poster presentations at the 3rd European Congress of Immunology (ECI 2012) held in Glasgow, UK, 5-8 September 2012. The congress, under the auspices of EFIS (the European Federation of Immunology Societies), was organized by the British Society for Immunology (BSI). This congress, which is held every three years,

follows the two hugely successful congresses held in Paris (2006) and Berlin (2009).

B. CEGMR Student Ms. Aisha Elaimi's Presentation at Preimplantation Genetic Diagnosis International Society (PGDIS) held in Austria

CEGMR is pleased to announce another contribution by a distinguished Saudi postgraduate student in her final year of PhD at the renowned University College of London, UK. Ms. Aisha Elaimi's work titled: 'The Use of FISH and TUNEL' to study the effect of GM-CSF on aneuploidy and DNA fragmentation in murine blastocyst cultured in-vivo and in-vitro was accepted for poster presentation at 11th, Preimplantation Genetic Diagnosis International Society (PGDIS) held during 21– 23 May, 2012 in Bregenz, Austria. Keeping in view the high standards of research ethics embedded in this student, Ms. Aisha was kind enough to show her CEGMR and KAU affiliations in this poster as continuation of her valuable contributions.

C. CEGMR Presentations at the 1st BD Middle East FACS User Group Meeting in Dubai

The Becton-Dickinson Biosciences, USA conducted its first FACS User Group Meeting from 11th to 12th of December 2012 at the Jumeirah Creekside Hotel in Dubai. Many eminent researchers and research students from all over the Middle East have presented seminars in the area of Stem Cell Biology and Clinical Research as well as in Basic, Translational and Clinical Immunology. Over 100 participants attended this meeting including the CEGMR Directors Dr. Mamdooh Gari and Dr. Adeel Chaudhry as well as the Principal Investigators, Dr. Farid Ahmed and Dr. Peter Natesan Pushparaj.

Dr. Farid presented his research work in the area of cancer stem cells on December 11 and Dr. Peter Pushparaj presented his work in the area of autoimmune diseases and the applications of flow cytometry on December 12.

Scientific Highlights of the First International Conference (continued)

International Scientists' Presentations in the Conference

1st International Genomic Medicine Conference in Saudi Arabia
CEGMR KING ABDULAZIZ UNIVERSITY - JEDDAH

Professor Vladimir B. Bajic

Affiliation: Computational Bioscience Research Center (CBRC), King Abdullah University of Science and Technology (KAUST), Kingdom of Saudi Arabia

Title: Knowledge Extraction and Integration of Information From Biomedical Repositories

Current deluge of information in the life science field opens new avenues to search for and discover useful information. Currently there are over 3000 public databases and numerous data from thousands of experiments. Thus, the search for useful information on any targeted topic is becoming increasingly complex despite new systems for information retrieval being made available.

Even more complex and exciting is the search for information not contained in any single resource but spread across multiple repositories. We present a technology, KES, for mining information from biomedical resources

including literature.

KES allows for multifaceted insights into biomedical information, such as: extraction of the cause-consequence relationships, generation of ranked hypotheses, automated extraction of topical information based on user definable sentence templates, and variety of information on association of biomedical entities identified within the searched resources. KES utilizes a number of public databases from which it gathers relevant information.

It contains precompiled human and mouse promoter annotation and transcription regulatory networks that include miRNA regulatory sub-networks. KES allows for connecting biomedical concepts with inferred pathway information, diseases, transcription regulation, and SNP data. Moreover, it is possible to explore potential effects of various compounds on cellular processes.

The system is capable of automatically generating biomedical databases on desired topic with the built-in query system ready for web publishing. Through specific examples we illustrate how the use of this sophisticated technology can lead to new discoveries.

Scientific Highlights of the Second National Genomics Day (continued)

Genomic Day Presentations

NPM1 Mutations in AML Cases from Saudi Arabia

Wafa Mohammed Abu-Nuqira ,Farid Ahmed ,Abdul Ali Peerzada ,Mamdooh Abdullah Gari ,Adel Abuzenadah ,Fatin M Al-Sayes ,Ghazi Damanhouri

Acute myeloid leukemia (AML) is a clonal malignant hematopoietic disease. It usually progresses rapidly and has an extremely variable clinical outcome. Although cytogenetic analysis provides useful diagnostic and prognostic tool in the management of AML, in about 40% to 50% of adults and 25% of children with AML, no microscopically detectable chromosomal abnormality can be found. These cytogenetically normal AML (CN-AML), are the most poorly understood subgroup.

A number of somatically acquired mutations have been observed in CN-AML patients, including internal tandem duplications (ITD) or tyrosine kinase domain (TKD) mutations of the FLT3 gene, partial tandem duplication (PTD) of the MLL gene, and mutations in the NPM1, CEBPA, NRAS and WT1 genes. NPM1 mutations alone are observed in almost 50% of adult CN-AML. Currently there is no study from the Kingdom of Saudi Arabia on the NPM1 mutations and their prognostic utility in the local population. This work is aimed at detecting mutations in NPM1 gene in CN-AML patients.

Genomic DNA extracted from well characterized AML patients is subjected to PCR amplification and sequencing of the exon-12 of NPM1 gene. Preliminary work done in a few patients resulted in the identification of a novel 4-bp insertion in the hot-spot region of the NPM1 in an AML patient. We are currently recruiting a large number of AML patients in the study with the aim of getting a clear understanding of the disease in Saudi patients.

Research Papers in Peer Reviewed Journals

Dr.Peter Natesan Pushparaj, European Congress of Immunology, 5-8 September 2012, Glasgow, Scotland. Oral and Poster Presentations and Publications in the IMMUNOLOGY Journal (Impact Factor: 3.32)

List of Conference Paper Publications in the IMMUNOLOGY Journal (Impact Factor 3.32):

1. **Pushparaj PN**, Aarthi JJ, Biswas S, Jayapal M, Narasimhan K, Al-Qahtani MH. Decrypting Genetic Signatures Associated with Experimental Colitis in Mice. **Immunology**, 137 (Suppl. 1), W12.003.
2. **Pushparaj PN**, Kumar SD, Narasimhan K, Jayapal M, Al-Qahtani MH. Deciphering the Role of Resveratrol in DSS-induced Colitis in Mice. **Immunology,** 137 (Suppl. 1), W70.004.
3. **Pushparaj PN**, Jayapal M, Narasimhan K, Kumar SD, Al-Qahtani MH. Dissecting the Disease Associated Functions of S1PR/SPHK Axis in DSS-induced Colitis in Mice. **Immunology,** 137 (Suppl. 1), P1084.
4. Elmesmari A, **Pushparaj PN**, Reilly J, Kerr S, Kurowska-Stolarska M, McInnes IB. The Role of SPHK and S1PRs in Rheumatoid Arthritis. **Immunology**, 137 (Suppl. 1), P0218.

CEGMR New Staff Presentations

CEGMR new staff gave their presentations in the Center's Meeting Room on December 18, 2012.

Dr. Gauthaman Kalamegam has presented his talk on stem cells in biology and regenerative medicine. Furthermore, he has explained about Wharton-Jelly mesenchymal stem cells in basic, translational and clinical research and its future therapeutic potential. He wanted to establish normal and abnormal human Wharton's Jelly stem cells (hWJSCs) for research and clinical applications, co-Banking of hWJSCs with cord blood for management of blood related disorders and the formulation of Wharton's Jelly Cream for management of non-healing diabetic wounds and burns. He further mentioned about his recent publications as well as future applications to Strategic as well as Large KACST grant calls from CEGMR.

Dr. Muhammad Abu-Elmagd presented his talk on Molecular Embryology. He explained further that his research interest lies in the field of gene expression and regulation during embryonic development aiming to understand the mechanism that underlies the early events of embryogenesis. He emphasized the importance on the clues about cancer that can be driven from understanding embryonic development due to changes in the gene expression levels and the activation of gene silencing machinery. He also explained that he would explore more about this link to try to add impacts in cancer therapy by discovering key molecules that play vital role in embryonic development as well as in cancer. His specific areas of interest include the investigation of Neurogenesis (Nervous System formation), Myogenesis (Skeletal Muscle formation), Cardiogenesis (Heart formation) and Chondrogenesis (Cartilage formation) using Mouse embryos, Chick embryos and Xenopus laevis embryos as model systems. He further explained about the current and future collaborations in UK and KSA as well as future research grant applications to KACST in the area of Molecular Embryology.

Dr. Mourad Assidi has given a seminar on the Obstetrics, Gynecology and Molecular Reproductive Biology. He further explained his research plans and project submissions in Preimplantation in Genetic Diagnosis, Reproduction and Development Biology, Ovarian, Breast and other Cancers, Fertility preservation, Infertility treatment, Functional genomic Analysis and Stem Cell Multi-Potency in Basic, Translational and Clinical Research.

Dr. Syed Kashif Zaidi presented his talk on anti-oxidants and ageing using in vitro and in vivo model systems. He further explained about his future research strategies at CEGMR that include the study of the role of Peroxiredoxins, Glutaredoxins and Thioredoxins (Antioxidants) in regulating the oxidative stress mediated damage in aging animals, evaluation of NDGA, a Lipoxygenase inhibitor, on the antioxidant properties and function as a potent anti-hyperlipidemic agent in rodent models and the assessment of the efficacy of NDGA in oxidative stress induced damage and its anti-diabetic and anti-cardiovascular defects in terms of signaling pathways.

Meet the CEGMR Staff

Dr. Shakeel Ahmad Ansari interviewed by Dr. Syed Ehsan Ahmad

Dr. Syed: You have done your PhD from Aligarh Muslim University (India). What really attracted you to the Kingdom of Saudi Arabia & CEGMR?

Dr. Shakeel: The Kingdom of Saudi Arabia has been serving as a platform for international researchers dedicated to advancing science and technology. CEGMR was established to address scientific challenges of regional and global significance through interdisciplinary research, and it promotes a culture of cross-discipline collaboration and encourages faculty to break the barriers between fields. Furthermore, Jeddah is a very friendly and multicultural city which is located near Makkah and Madinah. I strongly believe that CEGMR will make high-impact scientific discoveries through bold and ambitious initiatives and provide outstanding education to graduate and post-graduate students.

Dr. Syed: How did you find CEGMR and what are your expectations?

Dr. Shakeel: CEGMR has recruited faculty members from the best universities and research institutions in the world. One of the main attractions for faculty is the excellent support structure from technology, facilities and economic development expertise. This blend of custom-built infrastructure enables new ideas to move faster and further than in a traditional setting. I have joined CEGMR as an independent investigator in "Nanoparticle based drug delivery system". I am sure that it will provide me an array of opportunities to have my own research group as well as to establish a research program in nanotechnology for biomedical applications.

Dr. Syed: What is the main focus of your research group?

Dr. Shakeel: My Research group will deal with the synthesis, characterization and surface functionalization of nanocomposites for biosensor/biomedical applications.

Dr. Syed: What made you choose your research and what really motivates you to move forward?

Dr. Shakeel: I want to dedicate my potential and endeavor in scientific field for the welfare of human beings with the help of dedication, intellectual maturity and strength of purpose.

Dr. Syed: What are your achievements or contributions, so far, in Academia and Research?

Dr. Shakeel: It was a blessing on me to get a Faculty position at CEGMR after I completed my PhD in 2011. During my research, I published several papers in international journals with high impact factor. Moreover, I have three years of experience in teaching practical and theory classes to post-graduate students in the Department of Biochemistry, Aligarh Muslim University. Besides, I have qualified Graduate Aptitude Test in Engineering (GATE) – a highly reputed national level test in India.

Dr. Syed: What are your future plans at CEGMR?

Dr. Shakeel: I am blessed with a rare opportunity to be part of a new and ambitious Department with a clear vision. We are fortunate enough to have many highly cited researchers in basic, translational and clinical research in HiCi research program at CEGMR to work with them in order to facilitate the flow of research-based technological innovation from idea and discovery to market.

Dr. Syed: What is your advice or suggestion for students and junior researchers?

Dr. Shakeel: Hard work and patience will provide exceptional resources for continued success to young researchers towards their goals. They should always strive to demonstrate intelligence, drive and creativity, coupled with an exceptional initial output from their mentors to draw early signs of recognition within their academic community.

Joining of New Staff

Dr. Murad Assidi

Dr. Mourad Assidi has recently joined the *Center of Excellence in Genomic Medical Research (CEGMR)* as an Assistant Professor, *King Abdulaziz University*. He was born in Thala, Tunisia. In Tunisia, Dr. Assidi had very brilliant academic accomplishments. Remarkably, he was the National laureate and award winner of the Engineers Schools Entrance Exam (1999), Ministry of Higher Education, Tunisia. After that, he was the Laureate of the biological engineers who graduated in 2002, INAT, *Carthage University, Tunisia*. These distinctions allowed him to be awarded the sole Excellence Scholarship of Tunisia for graduate studies in Advanced Biotechnologies in Canada in 2003.

Dr. Assidi got his Master degree (M.Sc.) in Reproductive Biology & in Vitro fertilization (IVF) from Laval University, Quebec, Canada (2005). He did research for his Ph.D. degree on Genomics and Reproductive Molecular Biology (2010). In addition to his valuable publication record on high-impact international journals and lectures presented in several renowned national and international meetings, Dr. Assidi's major achievement is

his discovery of a gene-based treatment of infertility using biomarkers (patent number 2011/057 411). The most important finding in this invention is the identification of key biomarkers that can quantitatively and accurately predict the oocyte quality, which is still, unfortunately, predicted mainly using subjective morphological parameters. This discovery is an important breakthrough and a window of hope for couples suffering from infertility.

Soon after, Dr. Assidi joined McGill University Health Center as a postdoctoral researcher and a teaching fellow in Obstetrics & Gynecology department, Medicine Faculty, McGill University, Montreal, Canada. Thanks to his research experience and collaborations with many worldwide well-known laboratories, Dr. Assidi has acquired crucial skills associated to research methodology, IVF, early embryo development, Ob/Gyn, infertility treatment, Oncology; using cutting-edge molecular biology tools and the OMICS approaches (Genomics, Transcriptomics, Proteomics, Oncogenomics, Metabolomics, Reactomics, and Nutrigenomics)

It should be noted that Dr. Assidi's research projects were funded by the Canadian Institute of Health Research (CIHR) and the Natural Sciences and Engineering Research Council of Canada (NSERC). (Thanks to research grants provided by the two most famous Canadian funding agencies) In addition to his valuable research experience at both McGill & Laval Universities, Dr. Assidi has received key teaching and laboratory training of several courses and supervised the research projects of undergraduate students.

Besides his publication records, Dr. Assidi has valuable oral and poster presentations in many Canadian and international meetings, in addition to achieving very successful international collaborations with well-known researchers from Canada, United States of America, Germany, Netherlands and other countries.

News Updates

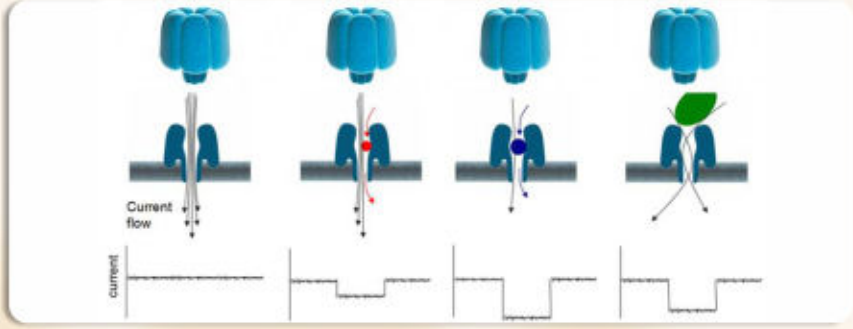
NANOPORE SENSING IN BIOLOGY AND GENOMIC MEDICINE

A nanopore is, essentially, a nano-scale hole. This hole may be biological (formed by a pore-forming protein in a membrane such as a lipid bilayer), solid-state (formed in synthetic materials such as silicon nitride or graphene), or hybrid (formed by a pore-forming protein set in synthetic material). The concept of using a nanopore as a biosensor was first proposed in the mid-1990s when nanopores were used in research in academic institutions such as Oxford, Harvard and UCSC. Recently, eminent scientists from UK and other parts of the World have established a company called as Oxford Nanopore in UK along with Oxford, Harvard and UCSC as collaborators. In an industrial setting, Oxford Nanopore was founded in 2005 to translate nanopore science into an electronics-based technology. The end-to-end system includes sample preparation, molecular analysis and informatics, and is designed to provide an array of useful applications to the end users in Biology and Medicine. A nanopore may be used to identify a target analyte as follows: Transformational products for research, human health and safety, and the environment

Oxford Nanopore's sensing system offers a new generation of direct, electronic analysis of single molecule. Capable of exquisite specificity and sensitivity, digital readouts, miniaturisation or industrial-scale installations, real-time analysis and low cost systems, this is a technology designed to truly disrupt science and medicine.

Personalised Medicine

Personalised Healthcare is the use of information about an individual to provide the best possible healthcare



(Adopted and Modified from Oxford Nanopore Technologies, UK)

for that person. This information may include physical measurements such as blood pressure or weight, biochemical measurements such as blood glucose, presence of a specific protein within blood/body tissue, or the interrogation of that person's genetic code. These measurements are often referred to as Biomarkers, a term that is commonly used when referring to levels of proteins that indicate disease status. These individual measurements may be used in a number of ways, including:

Diagnosis and treatment

Genetic analysis may enable the diagnosis of a disease or a disease sub-type. This information can facilitate rapid and appropriate treatment for individuals based on their own genomes, or in the case of cancer, the genome of their cancer cells.

In oncology (cancer care), it is now more common to classify a disease by its molecular characteristics than morphology. Overall treatment strategies can be planned more effectively. For example, for tumours that have been identified as fast-growing, clinicians may choose an aggressive treatment first, or, for a less aggressive disease, the clinician may choose to start therapy with a less aggressive treatment with a more favorable side effect profile. The choice of drug can also be tailored to the molecular characteristics of the disease. These techniques are currently best developed in non-solid cancers. For example, Acute Myeloid Leukemia, where mutations in the genes FTL3 and KIT provide useful information to the clinician, or Acute Lymphoblastic Leukemia, where understanding of specific genes relating to receptors guides treatment strategy. The cost of sequencing an exome or full genome is decreasing, towards a similar cost to an MRI scan. As a consequence, the use of large scale genetic analyses to inform treatment protocols is increasingly being used in oncology and other areas. In this way a large number of genetic indicators in that patient's cancer can be examined. The number of available clinical tests that measure protein biomarkers is also increasing; these tests may monitor progression of a disease, whether a drug is being well tolerated by a patient, or a number of other clinical scenarios.

Pharmacogenomics

In some cases it may be possible to use a patient's genotype to predict their response to a drug treatment. In this way a patient's treatment can be tailored for the best possible efficacy and lowest risk of side effects. For example, warfarin is an effective anti-clotting medicine, but difficult to manage as patients respond differently to different dosages. Studies to determine the genetic roots of the warfarin response have resulted in new genetic tests that allow physicians to tailor the dose to the patient, minimizing dangerous side effects and maximizing the therapeutic efficacy of the drug. Many available treatments are already stratified so that they are only prescribed to patients most likely to respond to that treatment. These are often used with a 'companion diagnostic', a protein or DNA based test to determine if that patient is suitable for the treatment. For example, the breast cancer drug trastuzumab (Herceptin), has the best efficacy in the 20% of patients whose tumour cells show high expression levels of the

HER2 gene. Another cancer drug, Imatinib (Glivec) is used for chronic myelogenous leukaemia. However, it is most effective in patients who have the 'Philadelphia chromosome', a genetic abnormality created when part of chromosome 9 wrongly attaches to chromosome 22 during cell division. Although an emerging field, personalised medicine is still in its infancy. Cheaper and faster genome sequencing technology will facilitate the development of personalised medicines in two ways. Firstly, a more complete understanding of the genotype-phenotype relationship will allow researchers to understand the biochemical pathways of disease in more depth. This, and the stratification of patients into subtypes, is expected to support the development of new drugs and early markers for disease detection. Cheaper and faster sequencing will also allow companies developing new drug treatments to understand more fully the relationship between genotype and drug response, increasing therapeutic efficacy and decreasing the risk of adverse events.

Prevention

Identification of genetic risk factors for disease may have clinical utility in early screening or prevention programmes. Despite major advances in recent years, research into the relationship between genomic variation and disease risk is still in its infancy. Of the diseases with some genetic influence, few are 'Mendelian' - inherited and attributed to a variation at a specific locus or point in the genome. In most cases, genetic contribution to a disease is complex and these elements are likely to interact with additional environmental factors. In the case of common diseases such as cardiovascular disease or diabetes, genetic factors are frequently outweighed by lifestyle factors. However there are examples where genetic characteristics can provide important guidance on a risk of disease. For example, the COGS study is evaluating the use of genetic information incorporated into risk screening for breast cancer, prostate cancer and ovarian cancer. Currently, most work of genetic testing services is focused on families where there appears to be risk of inherited disease and the relevant genetic markers can predict the disease with a high level of confidence. This is done in the context of a genetic counselling programme with informed consent of the family. In these circumstances the restult of the test will be actionable in some way, whether using prophylactic treatment or a screening programme or other life decisions important to that patient. For example, families with a history of breast or ovarian cancer may be tested. Women with certain mutations in the BRCA1 gene have a higher risk of developing breast, ovarian, and possibly colon cancers. Or, testing for the APC gene may be indicated where families have a history of bowel cancers.

(Adopted and modified from the Oxford Nanotechnologies, UK)

Readers should feel free to write their views, opinions and feelings directly to the Editor by email: cegmr.info@kau.edu.sa