بسم الله الرحمن الرحيم

وبعد حمد الله والثناء عليه والصلوة على رسوله محمد وآله فان
هذه الكتب التي صنعناها في الطب التي الاول منها هو في الاحكام
الكلية من الطب والثاني منها هو هذا الكتاب المجموع في الادوية
المفردة وقسمنا هذا الكتاب مقالتين الاول منهما في
القوانين الطبيعية التي يجب ان يعرف من امر الطب في قوى الادوية
الجزويه اما الاول فقسمناها الى ستة فصول ا في تعرف الامزجة
الادويه المفردة ب في تعريف امزجة الادوية المفرده بالتجربة
ج في تعريف امزجة الادوية المفردة بالقياس د في تعريف
افعال قوي الادوية المفردة ه في احكام عرض الادوية
في حارج ف في التقاط الادوية وادخارها واما الثانية
فان جعلت الادوية المفردة فيها الواحا ا الاسماء الادوية
المفردة وتعريف ماهياتها ب لاختيار الجيد منها ج لذكر
كيفياتها وطبايعها د لخواص افعالها واحوالها الكليه
مثل التحليل ومثل الانضاج والتقريه والتحدير وما اشبه ذلك
من الافعال التي ذكرناها في المقالة الاول وخواص احواد كانت
لها وجعلت لكل واحد منها كتابه نصبغ
حتى يسهل التقاطه ه في افعالها
التي يتعلق بالزنه وعلمت على كل شي

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# KUWAIT GENOME PROJECT



Fahd Al-Mulla B.Sc., M.B.Ch.B., Ph.D. PCTM, FRCP

Professor of Molecular Pathology at Kuwait University

Head of Functional Genomics at Dasman Diabetes center

Director of Genomic Medicine Center "GENATAK" "جيناتك"

# Kuwait




Area:
Total: 17,820 sq km
Population:
Total: 3,442,945
Kuwaiti nationals: 1,102,485
Non-nationals : 2,340,460
(Public Authority of Civil Information, 2009 )

6-Governorates

One public many private Universities
Centralized specialized Free public Health Service:
-Kuwait Cancer Control Center
-Kuwait Medical Genetics Center/maternity
-Private free Dasman Diabetes Center

# Research and External funding

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http://kfas.org/index.html


- Kuwait Foundation for the Advancement of Sciences. Directed Dr. Adnan Shihab Eldin

  -1% of profit from local businesses
  -Research, programs and educational-based funding
- Kuwait institute for Scientific research another source of research Funding
- Research funding dropped $30M-300k 2017

# Research funding not a priority

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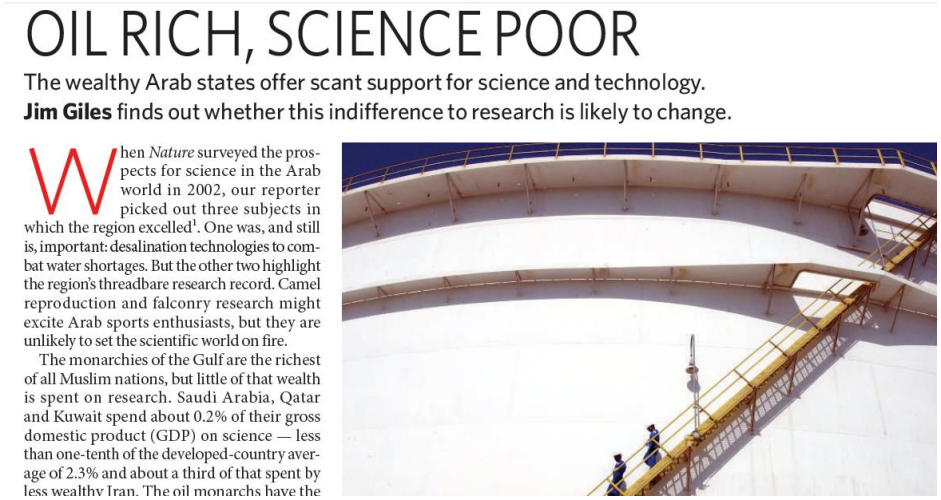
- Nature 2006 issue on Islam and Science
- Most Gulf sates spend 0.1-1% of GDP towards research and development
- Better trends now with Qatar 5% and Saudi Arabia matching this and more
- Kuwait-Bahrain-UAE-Oman remain on low spending side



OIL RICH, SCIENCE POOR

The wealthy Arab states offer scant support for science and technology.
**Jim Giles** finds out whether this indifference to research is likely to change.

When *Nature* surveyed the prospects for science in the Arab world in 2002, our reporter picked out three subjects in which the region excelled[1]. One was, and still is, important: desalination technologies to combat water shortages. But the other two highlight the region's threadbare research record. Camel reproduction and falconry research might excite Arab sports enthusiasts, but they are unlikely to set the scientific world on fire.

The monarchies of the Gulf are the richest of all Muslim nations, but little of that wealth is spent on research. Saudi Arabia, Qatar and Kuwait spend about 0.2% of their gross domestic product (GDP) on science — less than one-tenth of the developed-country average of 2.3% and about a third of that spent by less wealthy Iran. The oil monarchs have the

# Research barriers

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TheScientist
MAGAZINE OF THE LIFE SCIENCES

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March 2011 » Critic At Large

## Another Revolution Needed?

Counting the many plagues that threaten research in the Middle East and North Africa region

By Fahd Al-Mulla | March 1, 2011

Comment | Like | Link this | Stumble | Tweet this

NASA

Scientists around the world face obstacles during their research—a rejected manuscript, a failed funding application, an illegible electrophoresis gel. But these annoyances are simply par for the course when doing science. In the Middle East, however, scientists are up against much steeper challenges—a lesson I learned all too well when I finished my PhD and postdoc at the University of Glasgow in the United Kingdom and returned to my native Kuwait.

In Glasgow, research was facilitated and encouraged. We had several well-trained technicians, up-to-

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5. Immune Role in Brain Disorder?

Current Issue
TheScientist
Medicating CHILDREN

- Very difficult research environment
- Delayed tenders/ethical approvals etc
- Please read our cry for help in theScientist

http://www.the-scientist.com/?articles.view/articleNo/29545/title/Another-Revolution-Needed-/

# Rare allele avenue!

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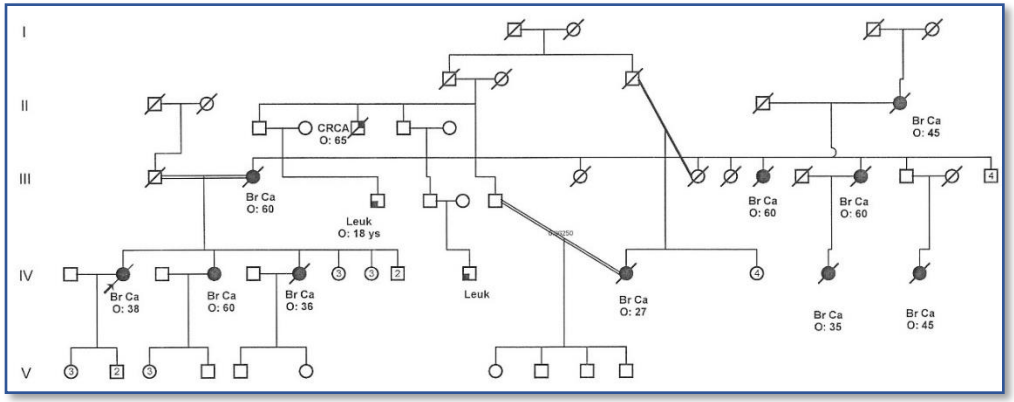



This family cost Government +3 million KD
Genetic screening/ prediction costs only few thousands

- Enormous time depth of human habitation (second only to sub-Saharan Africa)

- 50-70% Consanguineous marriages
- Source of major genetic discoveries
  - Autozygosity mapping
  - Linkage analysis
- Clusters of familial disorders and cancers
  - 50% of 50-years aged adults T2DM
  - 30% of the population are obese

- Arabs are not represented in the HapMap, 1000 Genome project

# Genome Arabia

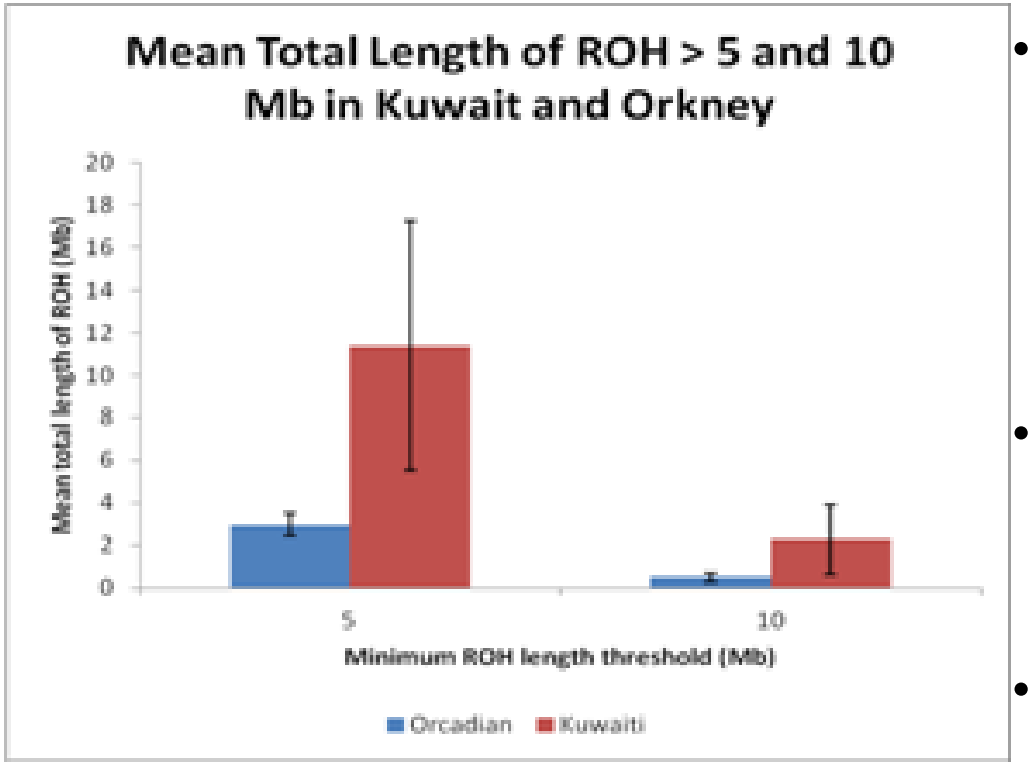


**Original Article**

*Journal of Human Genetics* **59**, 615-622 (November 2014) | doi:10.1038/jhg

The influence of admixture and consanguinity on population genetic diversity in Middle East

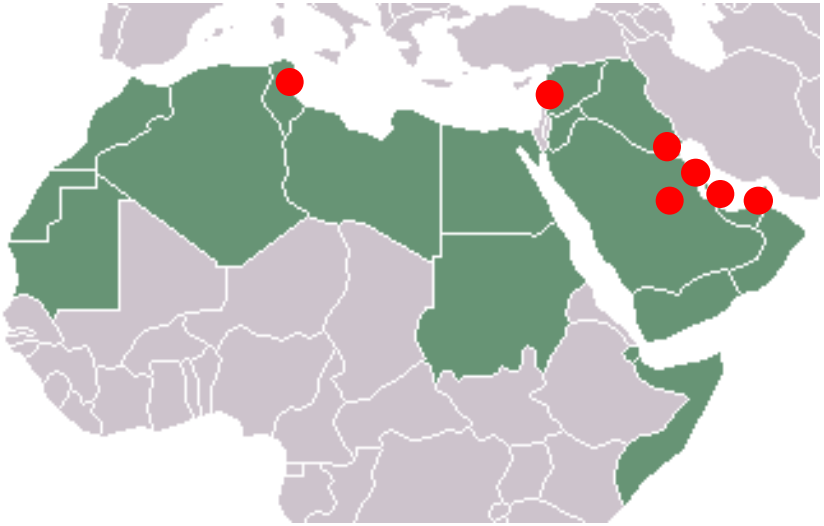
Xiong Yang, Suzanne Al-Bustan, Qidi Feng, Wei Guo, Zhiming Ma, Makia Marafie, Sindhu Jacob, Fahd Al-Mulla and Shuhua Xu




- **Considerable higher proportion of ROH over 5 Mb was observed in the Kuwaiti samples when compared to the Orkney population.**
- **Over three times as many Kuwaitis had at least one ROH > 10 Mb in length than Orcadians.**
- **Indicating recent inbreeding loops in their pedigrees.**

# Genome Arabia



- In 2012:

Set up Genome Arabia working group to whole genome sequence 1000 normal Arabs.

Grant funded by QNRF

2013-stopped??

# Qatar Genome

www.genatak.com

Gulf | Qatar

## Qatar Genome launched

Project is road map for future treatment of personalized medicine

By Habib Toumi, Bureau Chief
Published: 12:08 December 10, 2013

GULF NEWS

10

Manama: Shaikha Moza Bint Nasser, the Chairperson of the non-profit Qatar Foundation for Education, Science and Community Development has announced the launch of the 'Qatar Genome'.

"In Qatar, when we strived to build our all-inclusive culture of health, we transformed our health centers into research and academic centers, which incorporate hands on experience," Shaikha Moza said.

"As a result of the integration of scientific research and the clinical realities, I am pleased to announce the project 'Qatar Genome', a project that consists of a road map for future treatment of personalized medicine," she said as she opened the World Innovation Summit for Health (WISH) in the Qatari capital Doha on Tuesday.

## Mapping the Qatari genome to prevent inherited diseases

January, 2014

Dr. Ron Crystal, chairman of genetic medicine at WCMC-NY, is leading a group of research projects investigating the Qatari genome

A Weill Cornell Medical College study that analyzed the DNA of Qatar's native population has discovered genetic variations that could help doctors target interventions to reduce the prevalence of a variety of debilitating hereditary disorders.

Researchers at WCMC-Q and Weill Cornell Medical College New York (WCMC-NY), working with colleagues from Cornell University in Ithaca and Hamad Medical Corporation, identified 37 genetic variants in 33 genes known to play causal roles in a total of 36 diseases, including such devastating conditions as cystic fibrosis, sickle cell anemia and muscular dystrophy. The study points the way to more

# Saudi Genome

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9 December 2013 Last updated at 01:33 GMT

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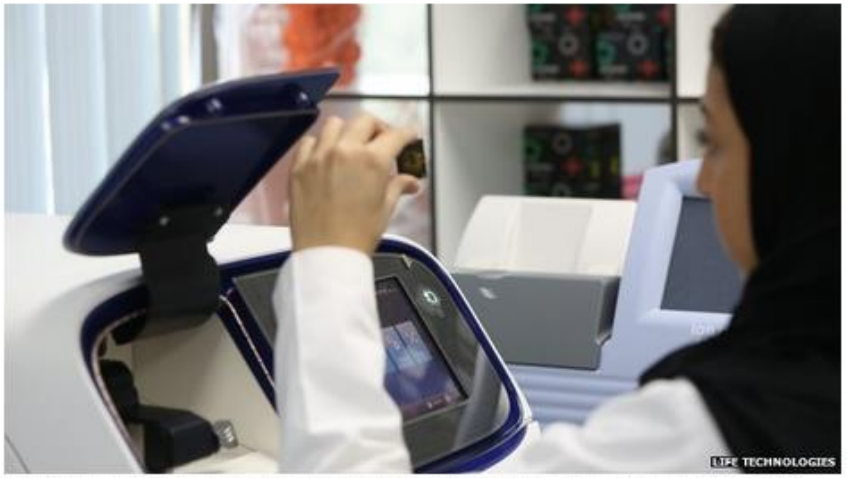
## Hundred thousand genomes to be mapped in Saudi Arabia

By Helen Briggs
BBC News




Saudi Arabia is launching a national research project to study the genetic basis of disease in its population

**Up to 100,000 people in Saudi Arabia are to have their genetic codes mapped in a new human genome project.**

Related Stories

European Journal of Human Genetics (2017) 25, 176–182
Official journal of The European Society of Human Genetics
www.nature.com/ejhg

EJHG Open

ARTICLE

# Clinical exome sequencing: results from 2819 samples reflecting 1000 families

Daniel Trujillano*[,1,10], Aida M Bertoli-Avella[1,10], Krishna Kumar Kandaswamy[1,10], Maximilian ER Weiss[1], Julia Köster[1], Anett Marais[1], Omid Paknia[1], Rolf Schröder[1], Jose Maria Garcia-Aznar[1], Martin Werber[1], Oliver Brandau[1], Maria Calvo del Castillo[1], Caterina Baldi[1], Karen Wessel[1], Shivendra Kishore[1], Nahid Nahavandi[1], Wafaa Eyaid[2,3], Muhammad Talal Al Rifai[3,4], Ahmed Al-Rumayyan[3,4], Waleed Al-Twaijri[3,4], Ali Alothaim[3,5], Amal Alhashem[6], Nouriya Al-Sannaa[7], Mohammed Al-Balwi[3,4], Majid Alfadhel[2,3], Arndt Rolfs[1,8] and Rami Abou Jamra*[,1,9]

**We report our results of 1000 diagnostic WES cases based on 2819 sequenced samples from 54 countries with a wide phenotypic spectrum. Clinical information given by the requesting physicians was translated to HPO terms. WES processes were performed according to standardized settings. We identified the underlying pathogenic or likely pathogenic variants in 307 families (30.7%). In further 253 families (25.3%) a variant of unknown significance, possibly explaining the clinical symptoms of the index patient was identified. WES enabled timely diagnosing of genetic diseases, validation of causality of specific genetic disorders of *PTPN23*, *KCTD3*, *SCN3A*, *PPOX*, *FRMPD4,* and *SCN1B*, and setting dual diagnoses by detecting two causative variants in distinct genes in the same patient. We observed a better diagnostic yield in consanguineous families, in severe and in syndromic phenotypes. Our results suggest that WES has a better yield in patients that present with several symptoms, rather than an isolated abnormality. We also validate the clinical benefit of WES as an effective diagnostic tool, particularly in nonspecific or heterogeneous phenotypes. We recommend WES as a first-line diagnostic in all cases without a clear differential diagnosis, to facilitate personal medical care.**

*European Journal of Human Genetics* (2017) **25,** 176–182; doi:10.1038/ejhg.2016.146; published online 16 November 2016

# Top-to-bottom model



- Kept knocking
- No major reaction from policymakers
- Unlike Qatar GP. We have no champion.
- Few doctors/specialists understood our GM initiative
- Doctors/specialists send-out

# GM and the private sector

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- Bottom-to-top model
- Direct to customer model
- Deliver state-of the art services
- Limited bureaucracy

# Kuwait Public and Private Genome Projects

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Private sector

Counseling
Phenotyping
1000 genomes 30x
400 rare diseases

KU
Breast cancer

200 Families

Genomes

KU
Multiple Sclerosis

176 samples/families

Dasman Diabetes Institute
T1DM/T2DM
MODY

25,000 samples Biobank
Plan for 10,000 genomes
Functional studies

# Bioinformatics/EHR made simple

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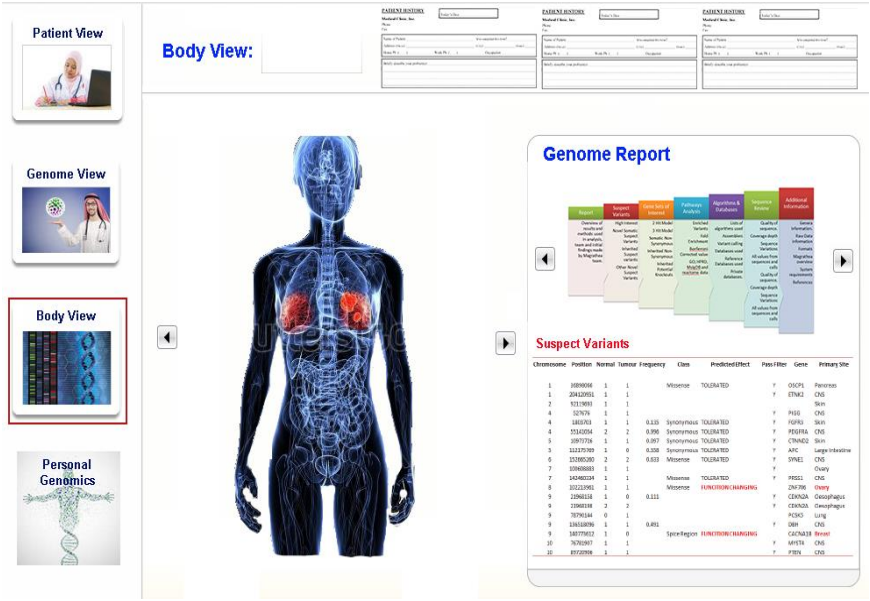


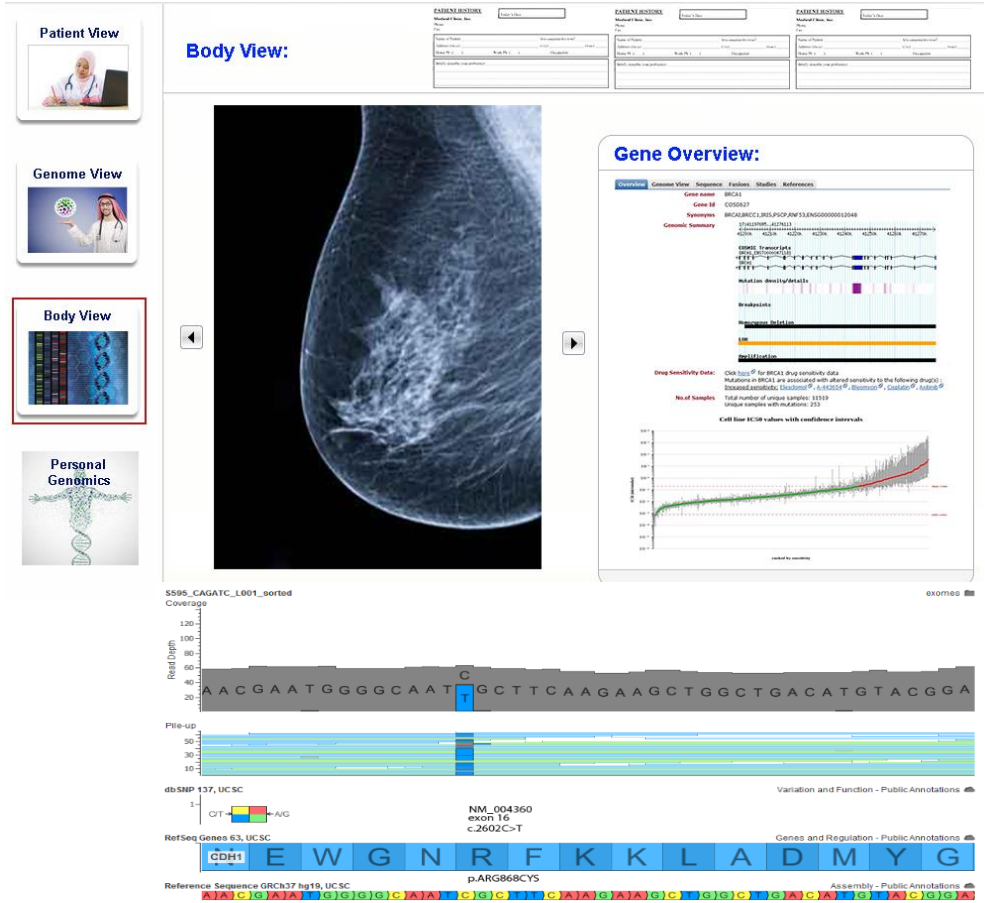


Association with Dr. Darrol Baker
Golden helix

# Data generation and databases

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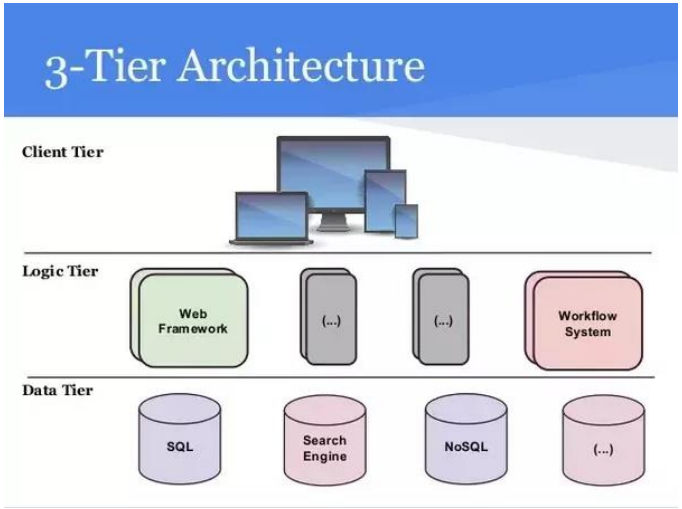


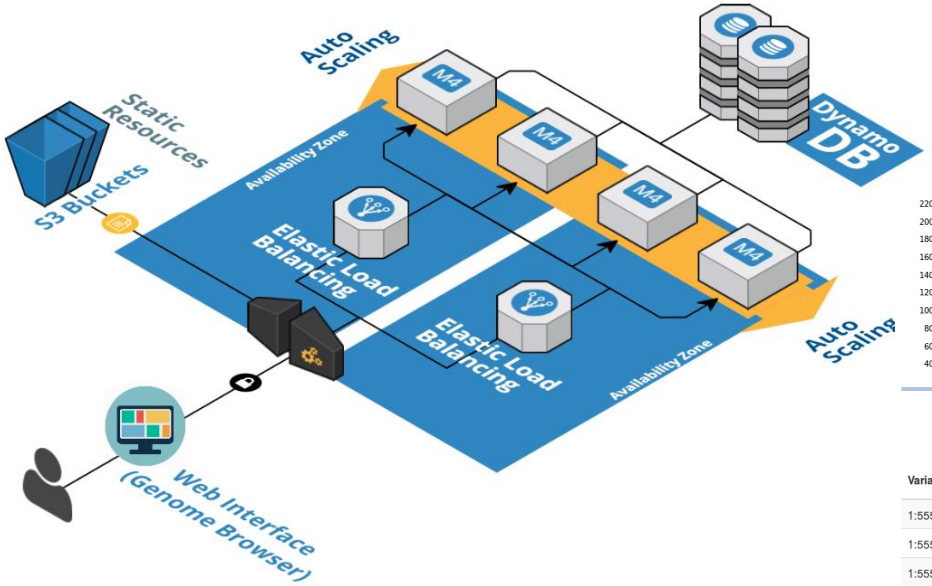


- Building online Kuwait genomics portal


| Variant | Chrom | Position | Consequence | Filter | Annotation | Flags | Allele Count | Allele Number | Number Of Homozygotes | Allele Frequency |
|---|---|---|---|---|---|---|---|---|---|---|
| 1:55512190 C / T | 1 | 55512190 | c.400-6C>T | PASS | splice region | test | 1 | 121374 | 0 | 0.082 |
| 1:55512192 G / T | 1 | 55512192 | c.400-6C>T | FAIL | splice acceptor | | 1 | 121374 | 0 | 0.082 |
| 1:55509511 G / T | 1 | 55509511 | c.400-6C>T | FAIL | splice acceptor | | 3 | 121374 | 119198 | 0.838 |
| 1:55505537 C / T | 1 | 55505537 | p.Ser9Ser | PASS | synonymous | | 5 | 23606 | 166 | 0.838 |

# Kuwait Genome Project

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| Inherited disease | Inheritance mode | SNP ID | Mutation | KGP_MAF | ExAC MAF |
|---|---|---|---|---|---|
| **21-hydroxylase deficiency** | Autosomal recessive | rs6471 | p. V282L | 0.0056 | 0.01 |
| **Adenosinedeaminase 2 allozyme** | Autosomal recessive | rs73598374 | p.D8N | 0.087 | 0.13 |
| **Alport syndrome** | X-linked recessive | rs104886192 | p.M898V | 0.0034 | 0.0003 |
| **Arrhythmogenic right ventricular cardiomyopathy type 8** | Autosomal dominant | rs121912998 | p.V30M | 0.0056 | 0.0031 |
| **Metachromatic leukodystrophy** | Autosomal recessive | rs2071421 | p.N352S | 0.24 | 0.21 |
| **Metachromatic leukodystrophy** | Autosomal recessive | rs74315479 | p.E384K | 0.0028 | 0.000025 |
| **Asphyxiating thoracic dystrophy 2** | Autosomal recessive | rs138004478 | p.G104R | 0.0028 | 0.00009 |
| **Autoimmune lymphoproliferative syndrome type 2** | Autosomal dominant | rs80358239 | p.I406L | 0.0085 | 0.0049 |
| **Congenital ichthyosis 1** | Autosomal recessive | rs41295338 | p.S42Y | 0.0056 | 0.004 |
| **Bartter syndrome antenatal type 2** | Autosomal recessive | rs59172778 | p.M338T | 0.0056 | 0.008 |
| **Becker muscular dystrophy** | X-linked recessive | rs1800279 | p.H2798R | 0.051 | 0.026 |
| **Bethlem myopathy** | Autosomal dominant/recessive | rs117725825 | p.P932L | 0.0028 | 0.0029 |
| **Biotinidase deficiency** | Autosomal | rs35976361 | p.I296V | 0.011 | 0.004 |

# Kuwait Genome project: Breast Cancer

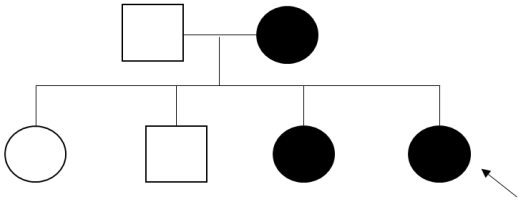
www.genatak.com

## Next-generation sequencing in familial breast cancer patients from Lebanon

Nadine Jalkh[1], Eliane Chouery[1], Zahraa Haidar[1], Christina Khater[2], David Atallah[3], Hamad Ali[4,5], Makia J. Marafie[6], Mohamed R. Al-Mulla[7], Fahd Al-Mulla[5,8*] and Andre Megarbane[9*]

| Gene | Variation | BIC database Clinically Importance/ Clinical Classification | COSMIC | Leiden Open Variation Database (LOVD) | BRCA Exchange |
|---|---|---|---|---|---|
| *BRCA1* | c.G131T p.C44F FOUNDER | unknown/ pending | Not found | Affects function | Not found |
| | c.A536G p.Y179C | unknown/ pending | Not found | Does not affect function | Benign |
| | c.C4327T p.R1443* | yes/ class 5 | Neutral | Affects function | Not found |
| | c.A1067G p.Q356R | unknown/ pending | Pathogenic | Does not affect function | Benign |
| | c.5090_5093del GTTA p.L1697fs | Not found | Not found | Not found | Not found |
| *BRCA2* | c.C65T p.A22V | unknown/pending | Not found | Effect unknown | Not found |
| | c.G223C p.A75P | unknown/ pending | Not found | Does not affect function | Benign |
| | c.658_659delGT p.V220I* | yes/ class 5 | Not found | Affects function | Not found |
| | c.C4061T p.T1354M | unknown/ pending | Neutral | Does not affect function | Benign |
| | c.G4258T p.D1420Y | no/ pending | Neutral | Does not affect function | Benign |
| | c.C5744T p.T1915M | no/ class 1 | Neutral | Does not affect function | Not found |
| | c.G8775C p.Q2925H | unknown/ pending | Not found | Effect unknown | Not found |
| | c.A1114C p.N372H | no/ class 1 | Neutral | Not found | Benign |
| | c.C1151T p.S384F | no/ pending | Not found | Not found | Benign |

Family 13



13/B36/Age 32-years

***BRCA1*: p.C44F** ***BRCA1*: p.C44F**

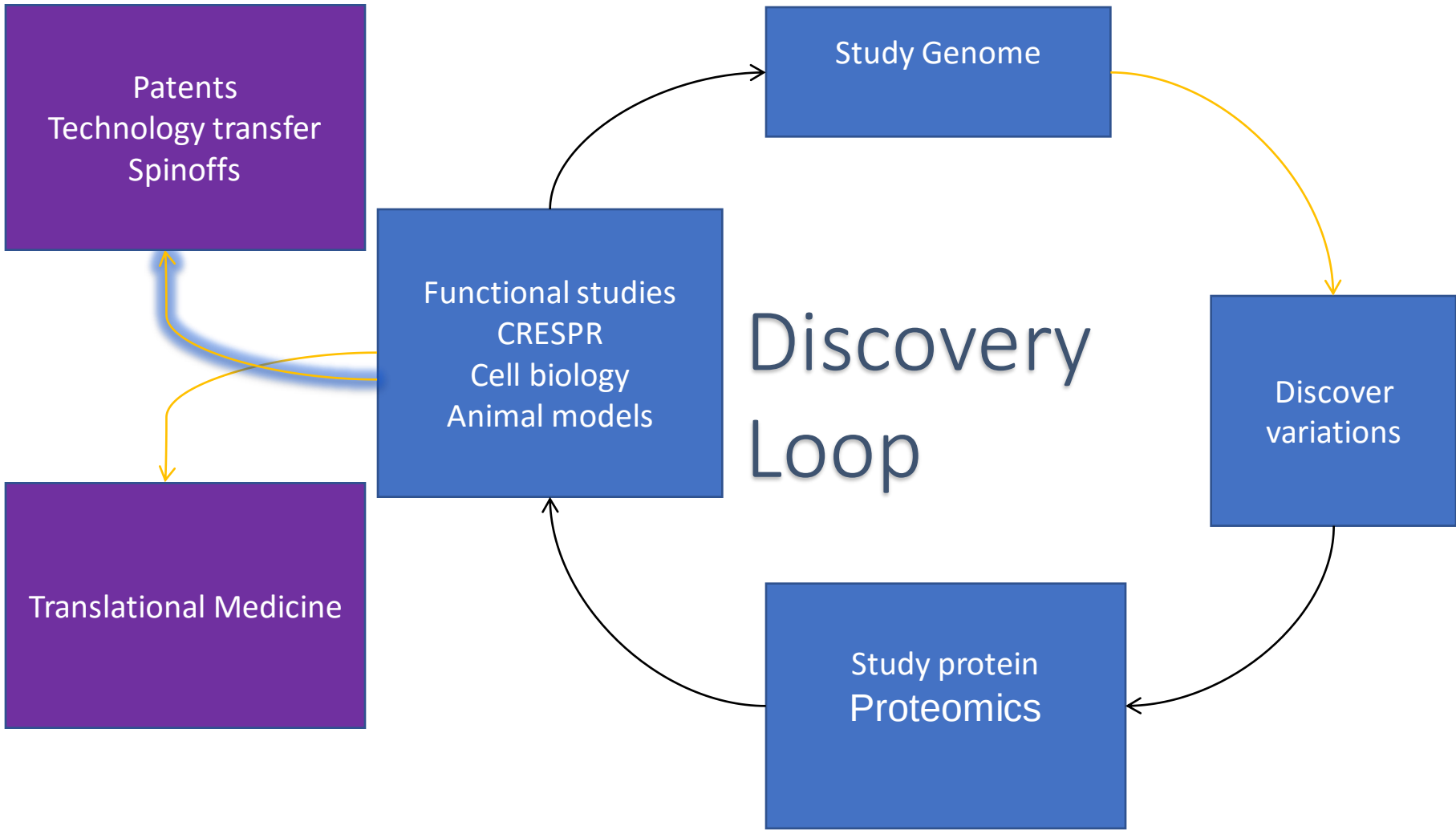
*PALB2*: p.G998E

***SLX4*: p.G141W**

***STK11*: p.S404F**

# Discovery Loop





# Lessons Learned



Implementation of GM in the Kuwait public sector is challenging

Collaborations should be based on sincere partnerships

Significant opportunities for GM and discovery in the area