This Week in Genomics
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Genomic medicine: collaborating for diverse data
https://www.pmlive.com/pharma_intelligence/Genomic_medicine_collaborating_for_diverse_data_1496021

Genomic medicine has the potential to revolutionise healthcare by changing how we understand, diagnose and treat diseases. **However, to fully realise its benefits,** ***extensive and high-quality data is needed*****. Currently,** ***most genomic research data doesn't include the Middle East and North Africa (MENA) region or the MENA population*****, leaving under-represented populations at a disadvantage.** This lack of diversity hinders the application of genomic research findings for a large portion of the global population. ***Additionally, certain regions, like Arab countries, face a disproportionate burden of genetic diseases, leading to high healthcare costs and significant societal impact.***

The world's largest genome programme

The ***United Arab Emirates was quick to recognise this issue of diversity*** in genomic data and, as a result, ***launched the Emirati Genome Programme***, now the largest programme of its kind in the world. The ***programme invites UAE citizens to voluntarily take part in the study*** by ***providing a blood sample***, which is then ***sequenced and analysed*** with the help of artificial intelligence. Over ***430,000 genome samples have been sequenced*** so far.

The ***potential value*** of this data set is ***vast***. By analysing genetic information – ***characterising novel genetic phenotypes*** and ***identifying the genes responsible*** – researchers may be able to ***identify new pathways involved in complex diseases***, suggest ***new therapeutic targets***, evaluate ***adverse drug effects*** and identify ***populations for which a drug would be most effective.***

If ***programmes*** such as this one can help ***address the diversity of data*** being used for ***genomic research***, then international collaboration is the ***next key step on the road map to better access to genomic medicine***. The ***Emirati Genome Programme*** provides the ***perfect platform for such collaboration***, with such a wide data set ready to be taken advantage of.

Collaborating on diverse data

With the pressing need for global partnership in mind, the ***Department of Health – Abu Dhabi (DoH***) recently announced the ***signing of a Declaration of Collaboration with Mass General Brigham's International Center for Genetic Disease (iCGD).***

The agreement will ***allow researchers to combine our cutting-edge genomic capabilities with iCGD's world-class clinicians and scientists in multidisciplinary research***. The resulting collaboration will focus on analysing ***whole genome sequencing data from the Emirati Genome Programme*** and on ***translational studies***, with the ***aim of advancing our knowledge of genomic medicine and global genetic diseases***. This ***data*** will then be ***leveraged by iCGD*** to research the ***causes, consequences, prevention and treatment of disease*** and ultimately ***develop novel therapies in*** four specific disease areas. Each of these focus disease areas is included in the World Health Organization's (WHO) list of the top ten causes of death:

- ***Metabolic disease*****:** while common metabolic diseases have traditionally been treated as ***individual entities, their shared molecular basis means multiple diseases could be treated simultaneously and***

effectively. A better understanding of the biology of these diseases could also potentially help us reverse disease progression and prevent further complications

- ***Oncology*:** as one of the areas of medicine most impacted by genomics, oncology has benefited greatly from rapid advances in genomic technologies. Genomics research has ***vastly improved our understanding of various cancer***s, allowing us to ***better diagnose, manage and treat the disease***. In fact, it is the ***disease area that has made the most progress in developing personalised medicine***

- ***Cardiology*:** although there are conventional small-molecule treatments for common cardiovascular problems, there are ***various acquired and inherited cardiovascular diseases*** that have unmet ***clinical needs***. Gene therapy is seen as a ***potential treatment option*** for ***severe cardiac and peripheral ischaemia***, ***heart failure, vein graft failure and some forms of dyslipidemia***. Recent developments in gene sequencing also mean that cardiovascular patients may soon have access to personalised medicine

- ***Neurology*:** the complex nature of neurological diseases means there are ***major challenges in diagnosis, management and treatment***. However, there have been recent developments in the use of gene therapy for spinal muscular atrophy (SMA) and trials are underway for mucopolysaccharidosis, Batten's disease and other genetic conditions.

From bench to bedside

Genome data plays a crucial role in developing novel therapies in several ways:

- **Identification of drug targets:** ***genetic mutations*** and variations are ***often associated with diseases***. By studying the genome, scientists can ***identify these genetic markers*** and ***develop drugs specifically designed to target them***.

- **Gene therapy:** genome data can guide the ***development of gene therapies***, where ***faulty genes causing disease are replaced or edited to function properly***.

- **Understanding disease pathways:** by ***comparing genomes from healthy and diseased individuals***, researchers can gain insights into disease pathways and mechanisms. This can lead to novel therapeutic strategies.

- **Biomarker discovery:** genome data can help in identifying biomarkers, which are ***often used in early diagnosis or prognosis of disease,*** leading to ***earlier and more effective interventions***.

There are also applications further down the pipeline. ***With the use of genome data and collaboration with different healthcare providers, we can identify genes that have risk factors that may lead to certain diseases if not managed beforehand.*** Not to mention that the genome project can be used to ***diagnose patients in a very comprehensive and specific manner*** – different gene panels can be analysed in a very short time, therefore speeding up the turnaround time for diagnosis.

In terms of ***management strategies***, different ***people can respond differently to the same medication due to their unique genetic makeup***. With genome sequencing, ***doctors can predict how a patient might respond to a certain drug*** and adjust that patient's treatment plan accordingly.

These advances in genomic medicine have the potential to revolutionise healthcare by making it more effective, personalised and preventive.

Accessing genomic medicine for global health

Overall, there is still much work to be done to address the diversity gap in the data that is used to inform genomic research. However, DoH's partnership with iCGD is just one example of how established resources and knowledge could help to expand the diversity of data used in genomics. The more partnerships like this that take place, the more representation will be seen in research.
By working together, the international community will be poised to take full advantage of wider data sets and ultimately develop better therapies that could help save more lives globally.

To learn more about Abu Dhabi DoH's partnership with MGB iCGD, please visit **https://www.doh.gov.ae/en/**