Personalized Medicine: How It May Soon Save Your Life

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Modern medicine has come a long way from where it was 100 years ago. With newer knowledge down to the genetic level, accurate diagnostic tools, and decent research and therapy, we now understand more about illnesses and how they come about; we are even treating some of the previously evasive diseases like Hepatitis C, some cancers and just recently, genetic diseases. Yet modern medicine is not perfect. One of the reasons is that when it comes to treatments, a lot of the drugs prescribed for a certain illness are ineffective for up to 50%-75% of cases!

Drugs are manufactured based on their effectiveness on the "average person". Drugs get approved after many clinical trials that benefit a large number of people with similar symptoms or diagnosis. If the drug is safe, and the majority of this population shows improvement, the drug is eventually approved for mass-market use. Outliers- people who showed little or no benefit- always exist in such trials; this may be why in the general population, there are always those who don't get better with a standard therapy.

Recent discoveries in human genetics have shown that each of us, our immunity, illnesses we get, and even how we respond to threats, is unique. It is therefore implied that 1 drug prescribed en masse for a disease may be outdated. Instead, a newly emerging approach- called "precision medicine" or "personalized medicine"- may be the answer. This approach uses patient data in detail, including the genetic makeup of the patient, to tailor the best treatment for that individual.

Sounds futuristic? Maybe. Yet, it was the main topic of last year's M.E.M.A. conference at AUB-MC, and the subject keeps making headlines in scientific magazines these days. If you are wondering about the price tag for such an approach, here is some data:

- The human DNA contains around 3.4 billion units: basically, a hundred books, each with 1000 pages.
- First sequenced in the US (government funded) in 2000, cost was 3 billion US$. It took 15 years to complete. The data was made public immediately.
- Current cost of a human genome sequence is ~ 1400 US$ with "high throughput sequencers". Time needed is around 20 days! This is a major improvement in cost and turnaround time.
- Not everything about our genome is known today. The function of more than 50% of our genome is still unknown. We can sequence the chromosomes, and read the letters inside it, but we don't yet understand what it means fully. It's like a person who knows English, and thus, can read Spanish, but won't understand what a Spanish sentence means fully. A work in progress.

Once the genetic make-up of a patient is known and understood, a personalized and targeted treatment is given. In cancers today, some centers are already using genome sequencing of their patients to decide on the best treatment available. Currently in the West, personalized medicinal approaches are preventive in nature. The idea is to know the genetic make-up of a patient, add to it family history and present data, and try to evaluate what illnesses this patient might encounter in the future… and prevent it. This approach is different from the one physicians study for in med-school: identify a disease already present ("chief complaint") to diagnose and treat it. This is why most insurance companies are still shy of covering gene sequencing for a personalized approach. Although many health-care workers are vowing to change this- favoring personalized medicine- it might still be 10-15 years for us to see it as a standard trend. Still, today, there are more than 350 products in clinical trial utilizing genomic data for preventive medical science.

In conclusion, genome based research will eventually lead to the development of targeted diagnostic tests to better understand the health needs of patients based on their individual genetic make-ups. Genomic data can also help tailor highly effective preventive approaches and treatments against diseases in the near future. With faster and cheaper technologies, and more understanding of the human genome, this approach can be achieved in our lifetime.

The "High Hopes" Center in Dubai

Majid and Lynn Jafar's life changed when their daughter was born with a rare genetic disorder. Today, beautiful Alia is 3 years old and struggles with almost daily epilepsy and other difficulties, unable to walk or talk. Motivated to do all they can to help her, her parents travelled the world in search of the best therapies. Inspired by all they learned and the daily challenges of Alia and other children with complex special needs, the Jafars have now established "High Hopes" Dubai Pediatric Therapy Center. It is the first center of its kind in Dubai and the region to cater for the needs of these children with an interdisciplinary, integrated approach in a loving and caring environment. With a handpicked international team of physiotherapists as well as occupational and communication therapists from across the four continents and the latest educational equipment, the center is run on a non-profit basis with the social objective of helping these children truly maximize their potential while having fun.

We admire and celebrate the courage and perseverance of all these children and their families each day and we all aspire for Dubai and the UAE to provide the love and supportive environment to help them lead good lives. Their determination is an inspiration to us all.

P.S: CDKL5(1)

CDKL5 is a gene that provides instructions for making a protein called cyclin-dependent kinase-like 5 also known as serine/threonine kinase 9 (STK9) that is essential for normal brain development. It is an independent clinical entity caused by mutations in an X-linked gene While CDKL5 is primarily associated with girls, it has been seen in boys as well(2). This disorder includes developmental problems, loss of language skills, and repeated hand wringing or hand washing movements, recurrent seizures beginning in infancy.

There are currently no approved drugs to treat CDKL5 Deficiency, save for Anti-Epileptic Drugs (AEDs) to treat the epileptic seizures. These have limited efficacy, pointing to a strong need to develop new treatment strategies for patients(3) A clinical trial of Ataluren has been announced(4). A CDKL5 protein replacement therapy is in development (5.)

(1) Wikipedia
(2) Wong VC, Kwong AK (April 2015). "CDKL5 variant in a boy with infantile epileptic encephalopathy: case report". Brain & Development. 37 (4): 446–8. doi:10.1016/j.braindev.2014.07.003. PMID 25085838.
(3) Müller A, Helbig I, Jansen C, Bast T, Guerrini R, Jähn J, et al. (January 2016). "Retrospective evaluation of low long-term efficacy of antiepileptic drugs and ketogenic diet in 39 patients with CDKL5-related epilepsy". European Journal of Paediatric Neurology. 20 (1): 147–51. doi:10.1016/j.ejpn.2015.09.001. PMID 26387070.
(4) Clinical trial number NCT02758626 for "Ataluren for Nonsense Mutation in CDKL5 and Dravet Syndrome" at ClinicalTrials.gov
(5) "Preclinical Program for Cyclin-Dependent Kinase-Like 5 (CDKL5) Deficiency". Amicus Therapeutics Press Release. 6 July 2016.

The "High Hopes" Center in Dubai

Lynn Bargouth Ja'far with HH Princess Haya and Sheikha Boudour Al Qassemi