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Medical Genetics: Ever Changing with Innovations

Whatever knowledge we have of our biological system, we are still incomplete in our understanding of their make-up and functioning. Newer technologies are unravelling the details out of tangled puzzle. Even if we add those pieces of information, the picture is far from clear especially for very common conditions like Diabetes, asthma, obesity and hypertension. It has become so obvious now that Medical Genetics is still evolving and changing. New innovations and exploratory nature of mankind is contributing new information almost daily.

If we just look back, the pace of innovations about 2-3 decades was very slow. If someone just explore the happenings in last decade, he will be surprised to find huge accumulation of information in molecular and genetic research. Innovations may include unflinching, new platforms that look primed to rev up discovery in basic biology, drug development, and clinical labs. But may also include products that speak to the important, but often underappreciated, new ideas that drives life science innovations. Often the innovations are becoming more clinically relevant.

The year 2016 saw few exceptional innovations which are going to change the way we are going to apply biotechnology skills in human sufferings. Few of these are as follows:

1. ExVive Human Kidney Tissue from Organovo is a replica of the kidney proximal tube created using 3-D bioprinting. With the tight regulation on experimental trials on humans and limited value of animal results, it is difficult to test any new drug for its toxicity. This Human kidney tissue will provide aanswers to our most to the query.
2. The Sequel System latest offering in single molecule, real-time (SMRT) sequencing by Pacific Biosciences. This is small, affordable and generates the same long reads and single-molecule resolution accomplished by the company's older SMRT sequencer, called the PacBio RS II.
3. Gene editing is s buzzword these days. CRISPR-Cas9 is a genome editing tool that is creating a thrill in the science world. It is faster, cheaper and more accurate than previous techniques of editing DNA and has a wide range of potential applications. If one can create a change in a gene, either in a cell line or a whole organism, it is possible to then study the effect of that change to understand what the function of that gene is. A popular company's new LentiArray CRISPR Libraries, introduced last year, make applying the tool in screening assays even more accessible to researchers. It will pave way for studying effects of single gene and their product.
4. Another tool from the same company is a recombinant Streptococcus pyogenes Cas9 protein purified from E. coli, the GeneArt Platinum Cas9 which contains a nuclear localization signal that aids in delivery to the nuclei of target cells.
5. ZipChip is a microfluidic device that radically speeds up mass spectrometry, requires minimum sample volumes, and broadens the range of materials that a mass spectrometer can handle. The device uses capillary electrophoresis to separate sample components in just two to three minutes when liquid chromatography columns would require up to an hour. The device provides better separation for samples, such as proteins, antibodies, and antibody-drug con-jugates than otherwise possible with any other method.

Along with these innovations and new technology, scientists are promoting the applications of these ideas in Clinical medicine. Mtochondrial disorders (MDs), for example, are a clinically heterogeneous group of disorders caused by a dysfunction of the mitochondrial respiratory chain. They can be related to mutation of genes encoded using either nuclear

DNA or mitochondrial DNA. The advent of next generation sequencing and whole exome sequencing in studying the molecular bases of MDs will bring about a revolution in the field of mitochondrial medicine, also opening the possibility of better defining pathogenic mechanisms and developing novel therapeutic approaches for these devastating disorders.

In this issue of the Journal, there are articles from basic science to clinical applications. Post-menopausal women are at increased risk of fractures due to osteoporosis and it is enhanced in the setting of Diabetes Mellitus. In the their study, Zenith *et al* demonstrated that pioglitazone has more potent effect on the adultskeleton in the context of low estrogens level and ismore susceptible to increased fracture risk. The study is an exploration of cause and effect using cell isolation, culture, assays in-vitro and their correlations with diabetic status and estrogen levels.

In an another study on Herpes simplex infection, Sharma *et al* studied HSV 1 and 2 in CSF samples from patient suspected with Herpes meningo-encephalitis.

Turner syndrome is the most common cause of primary amenorrhea in adolescent girls. Author have described a rare entity with pure 46,XX Gonadal Dysgenesis, a rare, autosomal recessive disorder of germline differentiation and migration.XX-GD when combined with neuro-sensory deafness is known as Perrault Syndrome.

In another review, Singh *et al* have briefly reviewed the current understanding of obesity in the era of genomic medicine. Obesity is a multifactorial disorder, definite genes have been implicated in some. But despite our research on obesity, the factors responsible genes to be the real culprit is still not clear. Epigenetic phenomenon have been implicated but not proved yet. Obesity provides a rich fertile ground to study root causes for multifactorial disorders which will have far more implication for prevention and for designing personalized therapies.

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