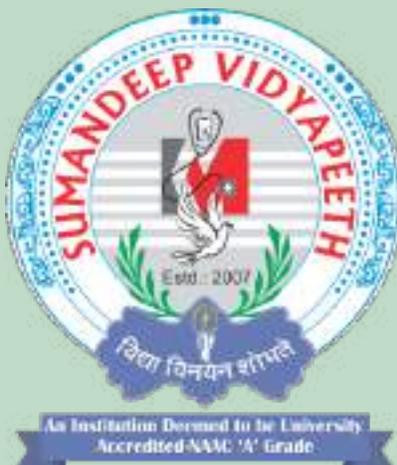


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Research Newsletter

7th ISSUE

Feb-Apr 2018

RESEARCH CELL,

2ND FLOOR, DEPARTMENT of PHARMACY ,

SUMANDEEP VIDYAPEETH AN INSTITUTION DEEMED TO BE UNIVERSITY

It is our pleasure to release the 7th issue of Research Newsletter. The theme of the present issue is **“Biomedical Research & Molecular Intervention”**

Biomedical Research has opened new vista and possibilities for the scientists and clinicians towards treatment of complex health issues. Such research breakthrough cannot be imagine without cellular-molecular intervention and a team efforts of clinicians, para-clinicians and biologists.

This news letter attempted to summarize this theme to explore this vital tool for research up-

Research Cell aims to nurture research ecosystem in all constituent institutes through various research updates and discussion with faculty & researchers of Sumandeep Vidyapeeth. Research Cell believes that students, faculty and clinicians should come forward with hypothesis based research project.

Research Cell feels that this issue of Newsletter will update the faculty and researchers with regard to this theme. Suggestions are always welcome to make this communication more meaningful.

- Dr. A. K. Seth



- **Medical research and molecular intervention**
- **Medical Biotechnology**
- Recent bio-molecular intervention by medical professionals
 - Biomedical research in epilepsy
 - Biomedical research in ophthalmology
 - Biomedical research in diabetes
 - Biomedical research in orthopedics
- **From the view point of our faculty**
- **Upcoming conference on molecular biology and medicine**
- **Scientific work/communication by our faculty**
- **High impact research of our faculty**
- **Recently organized scientific seminar**
- **Buzz around the world**

MEDICAL RESEARCH AND MOLECULAR INTERVENTION

The impact of molecular biology on medical sciences is without precedent.

There is a strong need to bridge the gaps in translating novel concepts in molecular biology into robust applications for use in the clinic.

Research is either discovery of new facts, enunciation of new principles, or fresh interpretation of the known facts or principles. Research is a step in relentless search for truth - it is an organized and systematic way of finding better answers to questions. The basic function of research is to answer why and how of a phenomenon, but searching answers to what, when, how much, etc., is also part of research endeavours. All these questions have relevance to any discipline but medicine seems to have special appetite for such enquiries.

The goal of medical research is to improve health, and the purpose is to learn how systems in human body work, why we get sick, and how to get back to health and stay fit. It is a systematic process to better determine etiology, patho-physiology, diagnosis, therapy and

prognosis. Research is the very foundation of improved medical care. It can also provide evidence for policies and decisions on health development.

MEDICAL BIOTECHNOLOGY

Medical biotechnology is an application of biotechnology that touches the lives of individuals every day. Both wellness and illness have ties to biotechnology. Advances in biology over the last 20 years have generated new insights into the causes of disease. This new level of understanding has, in turn, created opportunities for the development of new therapies, drugs, diagnostic tools and research/clinical instrumentation. Medical biotechnology is one of the fastest growing opportunities for employment in the medical research field.

Scientists are looking at the genetic causes of diseases, genetic links among family members, and individualized cures. As the Human Genome Project continues to map the locations of genes on human chromosomes, more solutions to the cause, prevention and cure of diseases will be discovered. Students will enjoy many aspects of medical biotechnology as they study genetic diseases and relate them to the medical experiences of family and friends.

http://www.lonestar.edu/departments/biotech/medical_biotechnology_chapter_wlinks.pdf

Recent Bio-molecular Intervention by Medical Professionals

BIOMEDICAL RESEARCH IN EPILEPSY

Method

Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation

Joshua Traynelis,^{1,7} Michael Silk,^{1,7} Quanli Wang,² Samuel F. Berkovic,³ Liping Liu,⁴ David B. Ascher,⁵ David J. Balding,⁶ and Slavé Petrovski^{1,8}

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Gene panel and exome sequencing have revealed a high rate of molecular diagnosis among diseases where the genetic architecture has proven suitable for sequencing approaches, with a large number of distinct and highly penetrant causal variants identified among a growing list of disease genes. The challenge is, given the DNA sequence of a new patient, to distinguish disease-causing from benign variants. Large samples of human standing variation data highlight regional variation in the tolerance to missense variation within the protein-coding sequence of genes. This information is not well captured by existing bioinformatic tools, but is effective in improving variant interpretation. To address this limitation in existing tools, we introduce the missense tolerance ratio (MTR), which summarizes available human standing variation data within genes to encapsulate population-level genetic variation. We find that patient-ascertained pathogenic variants preferentially cluster in low MTR regions ($P < 0.005$) of well-formed genes. By evaluating 20 publicly available predictive tools across genes linked to epilepsy, we also highlight the importance of understanding the empirical null distribution of existing prediction tools, as this varies across genes. Subsequently integrating the MTR with the empirically selected bioinformatic tools in a gene-specific approach demonstrates a clear improvement in the ability to predict pathogenic missense variants from background missense variation in disease genes. Among an independent test sample of case and control missense variants, case variants (0.83 median score) consistently achieve higher pathogenicity prediction probabilities than control variants (0.02 median score; Mann-Whitney U test, $P < 1 \times 10^{-16}$). We focus on the application to epilepsy genes; however, the framework is applicable to disease genes beyond epilepsy.

Dr. Joshua Traynelis, Department of Medicine, University of Melbourne and his colleagues have optimized genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Evaluating the clinical relevance of a novel missense variant found in an established disease gene is recognized as one of the central challenges facing modern medical genomics. Although probabilistic bioinformatics tools are unlikely to resolve this problem completely, they can optimize the triaging of candidate variants by identifying the empirical bioinformatics signatures of pathogenicity—properties found to be significantly enriched among variants that have been described to be clinically relevant.

(<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5630035/>)

BIOMEDICAL RESEARCH IN OPHTHALMOLOGY

Omics in Ophthalmology: Advances in Genomics and Precision Medicine for Leber Congenital Amaurosis and Age-Related Macular Degeneration

Anneke I. den Hollander

Departments of Ophthalmology and Human Genetics, Radboud University Medical Center, Nijmegen, The Netherlands

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Submitted: September 10, 2015

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Citation: den Hollander AI. Omics in ophthalmology: advances in genomics and precision medicine for Leber congenital amaurosis and age-related macular degeneration. *Invest Ophthalmol Vis Sci*. 2016;57(1):378-1387.

The genomic revolution has had a huge impact on our understanding of the genetic defects and disease mechanisms underlying ophthalmic diseases. Two examples are discussed here. The first is Leber congenital amaurosis (LCA), a severe inherited retinal dystrophy leading to severe vision loss in children, and the second is age-related macular degeneration (AMD), the most common cause of vision loss in the elderly. Twenty years ago, the genetic causes of these diseases were unknown. Currently, more than 20 LCA genes have been identified, and genetic testing can now successfully identify the genetic defects in at least 75% of all LCA cases. Gene-specific treatments have entered the clinical trial phase for three LCA genes, and for seven LCA genes gene-specific therapies have been tested in model systems. Age-related macular degeneration is a multifactorial disease caused by a combination of genetic and environmental factors. Currently, more than 40 loci have been identified for AMD, accounting for 15%-65% of the total genetic contribution to AMD. Despite the progress that has been made so far, genetic testing is not yet recommended for AMD, but this may change if we move to clinical trials or treatments that are dependent on an individual's genotype. The identification of serum or plasma biomarkers using other "omics" technologies may further improve predictive tests and our understanding of the disease mechanisms of AMD. Ultimately, it is anticipated that predictive tests will help to stratify patients for the most suitable therapy, which will enable the development of precision medicine, tailored to individual needs.

Dr. Anneke I. den Hollander, M.D. mentioned that genomic revolution had a huge impact on our understanding of the genetic defects and disease mechanisms underlying ophthalmic diseases. During the past 20 years, we have witnessed huge technological advancements in the genetics field. Dr. Hollander has worked on two diseases during his career, are Leber congenital amaurosis (LCA), a rare inherited disorder, and age related macular degeneration (AMD), a common, multifactorial disease. Twenty years ago, the genetic causes of LCA and AMD were unknown, but with the genomic revolution major progress in understanding the genetic causes, offering genetic testing and developing new treatments through Omics technology.

(<http://iovs.arvojournals.org/article.aspx?articleid=2506481>)

Genomics and the Eye

Val C. Sheffield, M.D., Ph.D., and Edwin M. Stone, M.D., Ph.D.

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This article (10.1056/NEJMra1012354) was updated on May 19, 2011, at NEJM.org.

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THE EYE HAS HAD A PIVOTAL ROLE IN THE EVOLUTION OF HUMAN GENOMICS. At least 90% of the genes in the human genome are expressed in one or more of the eye's many tissues and cell types at some point during a person's life. Consistent with this impressive genomic footprint is the observation that about a third of entries in the Online Mendelian Inheritance in Man database for which a clinical synopsis is provided include a term that refers to the structure or function of the eye.¹ Moreover, the phenotypic effects of even small genetic variations are made readily apparent by the many layers of amplification in the human visual system. For example, a single-nucleotide change in PAX6 can cause an anatomic abnormality of the macula less than a millimeter in diameter that results in noticeably reduced visual acuity and nystagmus.²

The heritable inability to correctly perceive the color green, known as Daltonism (after the English chemist John Dalton, who himself was affected), was the first human trait mapped to the X chromosome.³ (See Fig. 1 for a timeline of historic discoveries.) The Coppock cataract was the first human trait mapped to an autosome,⁴ and Leber's hereditary optic neuropathy was the first human disease shown to be caused by a mutation in mitochondrial DNA.⁵ More recently, age-related macular degeneration (AMD) and glaucoma^{6,7} — two common causes of human blindness — have been shown to be largely genetic, as has Fuchs' endothelial dystrophy,⁸ the most common cause of corneal transplantation in developed countries. Here, we review discoveries in mendelian and complex ophthalmic disorders and their implications for genetic testing and therapeutic intervention.

Dr. Val and Dr. Edwin reviewed that the eye has had a pivotal role in the evolution of human genomics. The eye has figured prominently in the development of genetic and genomic approaches to human disease. Vision is critically important to most activities of daily living, and cures for blindness will remain an important goal for medicine for many years to come. Physicians and scientists will be aided in the pursuit of this goal by the optical and anatomic accessibility of the organ, as well as by the large amount of visual cortex devoted to the interpretation of the neural information originating in the retina. This latter attribute will be a tremendous advantage for clinician scientists seeking to translate all the recent progress in gene-transfer and stem-cell biology into effective therapies for their patients with genetic eye diseases.

(<https://www.nejm.org/doi/full/10.1056/NEJMra1012354>)

BIOMEDICAL RESEARCH IN DIABETES

The Application of Genomics in Diabetes: Barriers to Discovery and Implementation

James S. Floyd¹ and Bruce M. Psaty^{1,2,3}

Diabetes Care 2016;39:1858-1865 | DOI: 10.2337/dci16-0728

The emerging availability of genomic and electronic health data in large populations is a powerful tool for research that has drawn interest in bringing precision medicine to diabetes. In this article, we discuss the potential application of genomics to the prediction, prevention, and treatment of diabetes, and we use examples from other areas of medicine to illustrate some of the challenges involved in conducting genomics research in human populations and implementing findings in practice. At this time, a major barrier to the application of genomics in diabetes care is the lack of actionable genomic findings. Whether genomic information should be used in clinical practice requires a framework for evaluating the validity and clinical utility of this approach, an improved integration of genomic data into electronic health records, and the clinical decision support and educational resources for clinicians to use these data. Efforts to identify optimal approaches in all of these domains are in progress and may help to bring diabetes into the era of genomic medicine.

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See accompanying articles, pp. 1854, 1870, 1874, 1879, 1889, 1896, 1902, 1909, and 1915.

James and Bruce from Dept. of Epidemiology and Medicine, Seattle, USA, discuss about the potential application of genomics to the prediction, prevention and treatment of diabetes to illustrate the challenges involved in conducting genomics research. The increasing availability of genomic data in large populations linked with electronic health data may become a powerful resource for genomic discovery, and examples from other areas of medicine offer lessons about the limitations of these data that can help guide the direction of future research. Whether genomic information should be used in clinical practice requires a framework for evaluating the validity and clinical utility of this approach, an improved integration of genomic data into electronic health records, and the clinical decision support and educational resources for clinicians to use these data.

(<http://care.diabetesjournals.org/content/39/11/1858>)

GENOMIC MEDICINE

W. Gregory Feero, M.D., Ph.D., and Alan E. Guttmacher, M.D., Editors

Genomics, Type 2 Diabetes, and Obesity

Mark I. McCarthy, M.D.

TYPE 2 DIABETES, THOUGH POORLY UNDERSTOOD, IS KNOWN TO BE A DISEASE characterized by an inadequate beta-cell response to the progressive insulin resistance that typically accompanies advancing age, inactivity, and weight gain.¹ The disease accounts for substantial morbidity and mortality from adverse effects on cardiovascular risk and disease-specific complications such as blindness and renal failure.² The increasing global prevalence of type 2 diabetes is tied to rising rates of obesity³ — in part a consequence of social trends toward higher energy intake and reduced energy expenditure. However, the mechanisms that underlie individual differences in the predisposition to obesity remain obscure.

Failure to understand the pathophysiology of diseases such as type 2 diabetes and obesity frustrates efforts to develop improved therapeutic and preventive strategies. The identification of DNA variants influencing disease predisposition will, it is hoped, deliver clues to the processes involved in disease pathogenesis. This would not only spur translational innovation but also provide opportunities for personalized medicine through stratification according to an individual person's risk and more precise classification of the disease subtype. In this article, I consider the extent to which these objectives have been realized.

From the Oxford Centre for Diabetes, Endocrinology and Metabolism; the Oxford National Institute of Health Research Biomedical Research Centre; and the Wellcome Trust Centre for Human Genetics, University of Oxford — all in Oxford, United Kingdom. Address reprint requests to Dr. McCarthy at the Oxford Centre for Diabetes, Endocrinology, and Metabolism, University of Oxford, Oxford OX3 7LJ, United Kingdom.

N Engl J Med 2010; 363:2339-50.

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Dr. W. Feero and Dr. Alan Guttmacher, M.D., Oxford National Institute of Health Sciences mentioned that to give the substantial time to translate basic biomedical discoveries into clinical tools in the genetic basis of common diseases is probably an underestimate. An improved understanding of fundamental disease mechanisms is already emerging; this will underpin future therapeutic advances. But the expansion of personalized medicine beyond monogenic forms of disease awaits a more complete description of predisposition. The boundaries of personalized medicine will be much clearer in a few years, after large-scale genome wide resequencing efforts (now under way) provide a systematic, comprehensive description of the relations between genome sequence variation and major clinical phenotypes.

(<https://www.nejm.org/doi/full/10.1056/NEJMra0906948>)

BIOMEDICAL RESEARCH IN ORTHOPEDICS

TEAM APPROACH: THE MANAGEMENT OF INFECTION AFTER TOTAL KNEE REPLACEMENT

Chun Hoi Yan, MBBS, FRCS

Carla Renata Arciola, MD, PhD

Alex Soriano, MD, PhD

L. Scott Levin, MD

Thomas W. Bauer, MD, PhD

Javad Parvizi, MD, MS, FRCS

Investigation performed at the Department of Orthopaedic & Traumatology, The University of Hong Kong, Hong Kong SAR, People's Republic of China, and the Department of Orthopaedic Surgery, Rothman Institute at Thomas Jefferson University, Philadelphia, Pennsylvania

Abstract

> Diagnosis and management of infection after total knee arthroplasty are challenging. They require a multidisciplinary team approach, much like the management of musculoskeletal tumors.

> Patients presenting with suspected infection after total knee arthroplasty require diagnostic confirmation, medical optimization, comprehensive surgical care that may include measures to cover the soft tissues, administration of long-term antibiotics, and extended rehabilitation to improve outcome.

> Surgeons should work closely with infectious disease specialists or microbiologists at every step to minimize the perioperative risks of reinfection, should decide on the most appropriate surgical modality and antibiotic regime, and should monitor the response to therapy.

> The current evidence on the best surgical management of infection after total knee arthroplasty (debridement and retention of prostheses compared with 1-stage exchange or 2-stage exchange arthroplasty) is lacking. Randomized, prospective studies that are under way may provide this much-needed information.

Dr. Chun Yan, M.D., and his colleagues from different countries found that the battle against infection is as old as the human civilization. Periprosthetic joint infection, with all of its disastrous consequences, continues to pose a challenge to the orthopaedic community. The management of patients with periprosthetic joint infection after total knee arthroplasty requires the expertise of professionals from different disciplines. The orthopaedic surgeon is responsible for orchestrating the involvement of various specialists and experts who can help to address the challenge of periprosthetic joint infection. Perhaps the multidisciplinary team approach, when properly established, calls for centers specialized in fighting against musculoskeletal infection.

(https://journals.lww.com/jbjsreviews/Abstract/2018/04000/Team_Approach_The_Management_of_Infection_After.1.aspx)

Basic research in orthopedic surgery: Current trends and future directions

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Musculoskeletal problems continue to represent a growing source of death and disability world-wide, particularly with the growing burden of disease associated with an aging population and increase in the rates of road traffic accidents. To address the societal and economic burdens presented by musculoskeletal disorders, research in the normal biology of musculoskeletal tissues, the diseases and injuries associated with these tissues, and the underlying mechanisms of musculoskeletal tissue regeneration continue to gain importance. These investigations often require multidisciplinary approaches ranging from basic cellular and molecular biology, bioengineering, biomechanics, and clinical research. It is clear that collaboration between disciplines and centers with expertise in biology, mechanics, and clinical research is essential to continue to advance the field. The purpose of this review is to address issues that may be of interest to the development of new basic science research programs and initiatives, including a brief review of current and developing areas of orthopaedic research, and the resources required for the successful creation of new biology and mechanical research laboratories.

Chuanrong LU and his colleagues at San Francisco General Hospital, San Francisco, USA found that the field of orthopedic research will continue to grow in order to address the increasing global burden of musculoskeletal injury and disease. New basic scientific discoveries in biological and mechanical research will continue to advance rapidly, and present opportunities to bring these new discoveries to the clinic. The complex nature of the musculoskeletal system requires multi-disciplinary collaborations between investigators that possess a wide diversity of expertise. Although the development of research laboratories and opportunities require extensive planning and resource development, ultimately basic discoveries have the potential to develop into translational projects that can impact patient care.

(<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2762563/>)

FROM THE VIEW POINT OF OUR FACULTY

"In order to improve the quality of research (high impact) and to get in-depth resolution of medical problems, a clinical and para-clinical faculty/researchers need to expand their research horizon in collaboration with Cellular-Molecular biologists".

Dr. R. Balaraman, Professor, Department of Pharmacy explained this very clearly. He said that Medical Science is big enigma where we are yet to understand etiology and pathophysiology of many dreadful diseases like cancer, AIDS, auto-immune diseases. Therefore, it is imperative for the medical fraternity to work with the biologist to find factors responsible for several diseases. This is called as Translational Research. Whatever the clinician get to know about the clinical

findings, it should be fed to the laboratory scientists. In this process, a clinician (Medical & paramedical) should consult with molecular biologists to discuss the possibility of the etiology of the disease. Molecular biologist in turn should work on the molecular level of the disease process. The finding of the biologist should be updated to the clinician who should try to develop their own strategies to evolve a method to treat the disease.

In this, a process is called bed side to bench and bench to bed side.

UPCOMING CONFERENCE ON MOLECULAR BIOLOGY AND MEDICINE



ME
CONFERENCES

International Conference on Molecular Biology and Medicine

August 27-28, 2018

Registration & Timings
August 27, 2018 (6:00AM to 6:00PM)
August 28, 2018 (9:00AM to 8:00PM)

Group Photo Time
10:00AM - 10:10AM, August 27, 2018

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Speakers PPT
You may submit your presentation to any of our onsite organizers on the day of your talk. If your presentation is not compatible with our laptops, then you may use your own laptop.

Venue
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SCIENTIFIC WORK/COMMUNICATION BY OUR FACULTY

[Dr. Anshula Deshpande](#), Professor, Department of Paedodontics and Preventive Dentistry, KMSDCH was invited as a speaker for the conference entitled "2nd Case-report in Pediatric Dentistry" of "Oral Injuries in Children" held on 20-21st April, 2018 at Selangor, Malaysia.



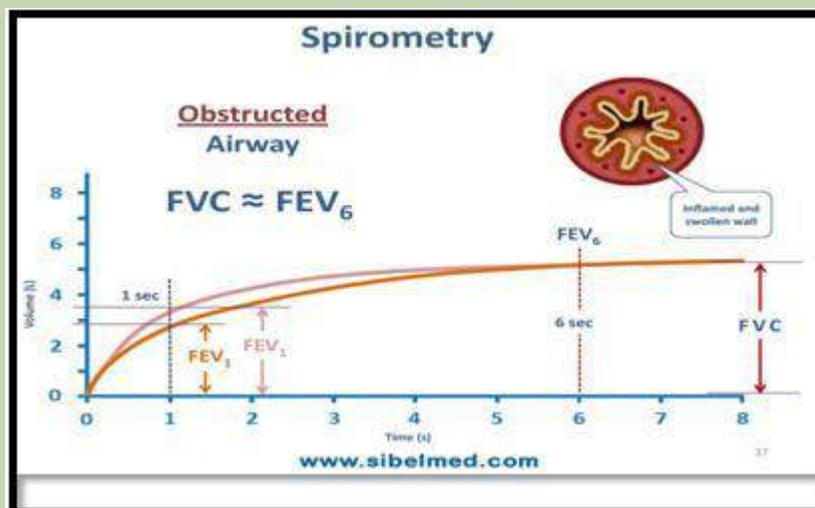
Recent research work by SVDU Faculty- Dr Geetanjali Purohit.

Time to update the Gold standard of forced spirometry (FEV1% or FEV6%)

Medicine is an updating science and Asian countries need to set their own standards and reference values. The American Thoracic Society (ATS)/European Respiratory Society (ERS) standards for the diagnosis and management of patients with chronic obstructive airway disease (COPD) recommend a fixed proportion of forced expiratory volume in 1 s and forced vital capacity (FEV1/FVC) of 0.7 as the cutoff and considered as gold standards. Poor subjective efforts, frustration, time taken and complications as syncope associated with forced spirometry inspires researchers to find out the surrogate of FVC and FEV1%. Recently, I studied respiratory parameters during pregnancy. This study is first extensive study in India on lower socioeconomic class.

Dr. Geetanjali Purohit, Assistant Professor, Department of Physiology, SBKS MI & RC performed a study on total 400 participants (100 in each trimester and 100 nonpregnant control) attending antenatal clinic of Obstetrics and Gynecology Department, Dhiraj General Hospital, SVDU. Data analysis found that results of FVC and FEV6 (forced expiratory volume in 6th second) remain within physiological limit. The ratio of FEV1/FVC (FEV1%) and FEV1/FEV6 (FEV6%) were similar and comparable. FEV6 requires short exhalation time, thus effectively used in place of FVC in evaluation of lung function test. Psychological exertion made pregnant female more conscious for the longer exhalation. Compared with measurements of FVC, using FEV6 reduces the test time, frustration and can reduce the complication as syncope during the test. The FEV1/FEV6 may be applied as a proxy for FEV1/FVC. Further extensive studies to generate Indian reference values are required to replace the Gold standard of forced spirometry. The reference values of

FEV₆ and FEV_{6%} indeed provided researchers with opportunity to use shorter FVC maneuvers during spirometry.



HIGH IMPACT RESEARCH PUBLISHED BY OUR FACULTY

1. Dr. Nirmal Shah, Associate Professor, Department of Pharmacy has published the research article entitled "Oral bioavailability enhancement of raloxifene by developing microemulsion using D- optimal mixture design :optimization and in vivo pharmacokinetic study" in the high impact journal of "Drug Development and Industrial Pharmacy" having [2.29 impact factor \(Clarivate Analytics\)](#).

2. Dr. Sunil Doshi, Associate Professor, Department of Forensic Medicine, SBKS MI&RC has published the review article entitled "Paraphilic infantilism, diaperism and pedophilia: a review" in the journal of "Journal of Forensic and Legal Medicine" having [1.135 impact factor \(Clarivate Analytics\)](#).

RECENTLY ORGANIZED SCIENTIFIC SEMINAR

Internal Quality Assurance Cell (IQAC) has organized a Student and Faculty Development Programme on title "Complexities in Designing Simplistic Research Entities in Bio-medical Science", held on 10th May, 2018 at auditorium, SBKS MI&RC at 10 am to 12.30 pm.

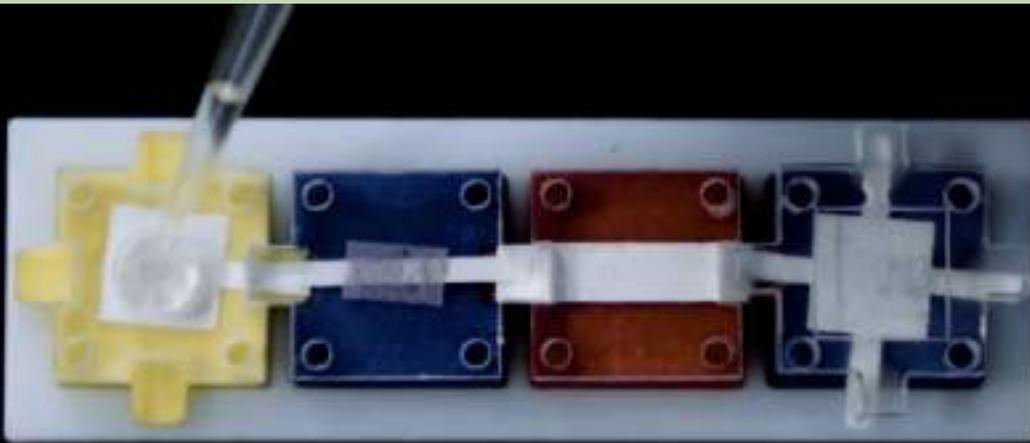


BUZZ AROUND THE WORLD

The first medicine designed to prevent **migraines** was approved by the **US Food and Drug Administration** on 17th May, 2018. The drug, **Aimovig**, made by **Amgen** and **Novartis**, is a monthly injection with a device similar to an insulin pen. Aimovig blocks a protein fragment,

CGRP, that instigates and perpetuates migraines.

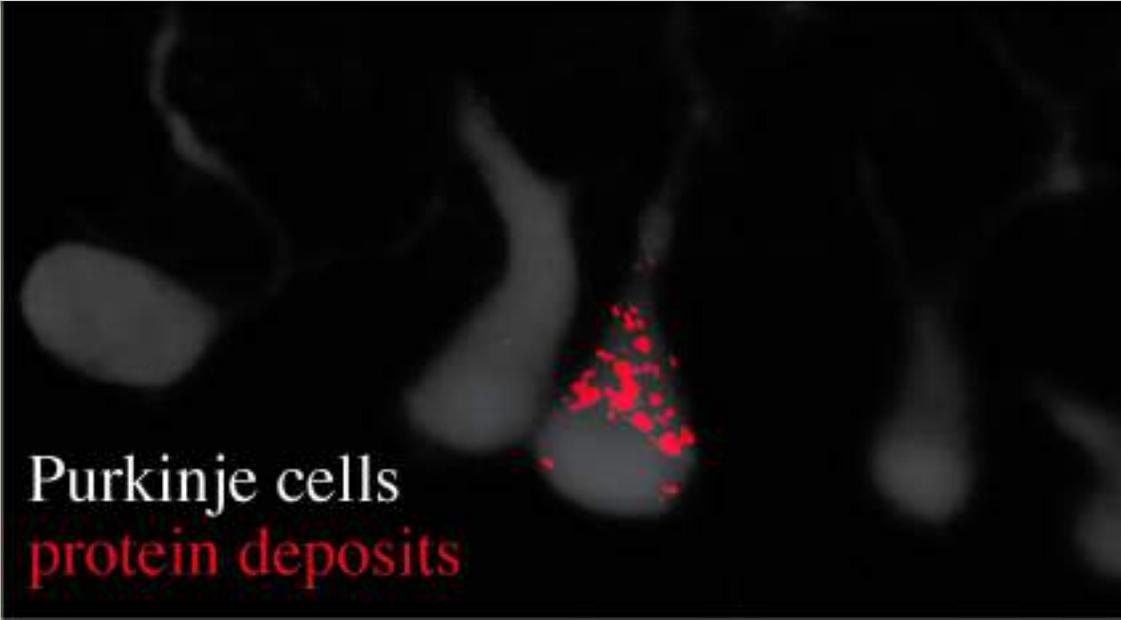
(<https://timesofindia.indiatimes.com/home/science/now-a-drug-to-prevent-migraine/articleshow/64230894.cms>)



Modular blocks could enable labs around the world to cheaply and easily build their own diagnostics

Researchers at MIT's Little Devices Lab have developed a set of modular blocks that can be put together in different ways to produce diagnostic devices. These "plug-and-play" devices, which require little expertise to assemble, can test blood glucose levels in diabetic patients or detect viral infection, among other functions.

(<https://news.mit.edu/2018/plug-and-play-diagnostic-devices-0516>)



Purkinje cells
protein deposits

Researchers Identify Gene That Helps Prevent Brain Disease

Ackerman, Paul Schimmel (Scripps Research Institute) My-Nuong Vo (Scripps Research Institute) and Markus Terrey (UC San Diego) identified that Ankrd16 rescued specific neurons called Purkinje cells that die when proofreading fails. Without normal levels of Ankrd16, these nerve cells, located in the cerebellum, incorrectly activate the amino acid serine, which is then improperly incorporated into proteins and causes protein aggregation.

The levels of Ankrd16 are normally low in Purkinje cells, making these neurons vulnerable to proofreading defects. Elevating the level of Ankrd16 protects these cells from dying, while removing Ankrd16 from other neurons in mice with a proofreading deficiency caused widespread buildup of abnormal proteins and ultimately neuronal death.

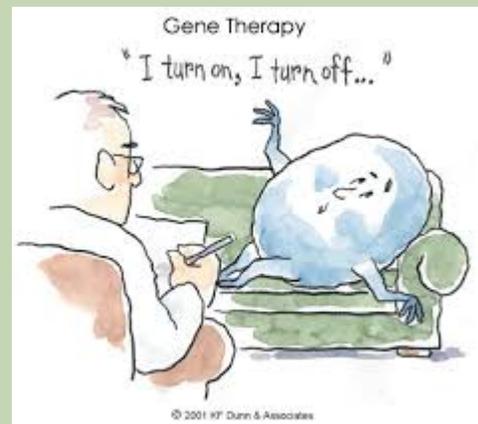
<https://www.nature.com/articles/s41586-018-0137-8>



Researchers from Drexel University [reversed symptoms of Alzheimer's disease in fruit flies by restoring the balance between two epigenetic enzymes](#) that regulate gene expression.

Priyakshmi Panikker, a PhD student, and Felice Elphand, Ph. D, an associate professor, both in Drexel's College of Arts and Sciences, performed tests in flies and found that if they added extra Tip60 HAT in the brain of flies that displayed symptoms close to Alzheimer's disease, the balance between the enzymes could be successfully restored. When that balance came back, behaviors the team had taught the flies were able to be learned again and remembered. Their findings strongly support the concept of exploring the efficacy of specific Tip60 HAT activators, as well as identifying and manipulating additionally misregulated Tip60 target genes," Elefant said.

<http://www.jneurosci.org/content/early/2018/04/13/JNEUROSCI.2840-17.2018>



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Dr. Maneesh Jaiswal, Chief Research Officer, SVDU