MEDGENOME

Vol 16 | Jan 2021

GeKNOWme

Internal Quarterly Newsletter

SPECIAL FEATURE

Heart to Heart with Dr I C Verma

Pioneer in Genetics

Scientific Articles

Inherited Retinal Degenerative Diseases

Rare form of Cardiomyopathy in multiple families from India

Featured Article

Nutrition, Health & Weight Management

From the CHAIRMAN'S DESK

Hello Team MedGenome,

Wish you all a Happy New Year!

As we march into 2021, it would normally be a time for some annual resolutions. But for us at MedGenome, even in the midst of the ongoing pandemic, our path has hardly changed – our vision has only been further validated, our strategy has succeeded and our execution has improved. My intention is not to indulge in chest thumping, but I feel we should acknowledge the reality of MedGenome's leadership in genomics in India and our role in putting India in the genome data map of the world. Our success is the result of the combined effort of all the employees who work every day towards providing diagnostics services for patients, bioinformatics analysis for researchers and keeping MedGenome's technological capabilities at the cutting edge. At the same time, we fully realize that we have a long way to go – both for MedGenome as a company and for India as a country in leveraging genomics for solving the health problems of our people.

When I started SciGenom in 2009, Sekar (Dr. Somasekar Seshagiri), then at Genentech, was my scientific consultant and guide. We also convinced a couple of scientists to move from the U.S to India (Kochi) to run the wet lab, and Ravi (Dr. Ravi Gupta) to move from the Wistar Institute in Philadelphia to lead the Bioinformatics effort. While SciGenom was getting operational, Sekar, Ravi and a team of willing scientists focused on completing the analysis and publication of the first female Whole Genome from India. Sekar also encouraged me to start an effort in parallel to promote Scientific Research and Education in India and thus the non-profit unit SciGenom Research Foundation (SGRF) was born in 2010. The first NGBT conference by SGRF was a small affair with around 150 participants at RGCB, Trivandrum in Dec 2011, compared to the gala events that most of you would have participated in recent years.

Meanwhile SciGenom had started Sanger Sequencing services and we moved into NGS by buying the first MiSeq from Illumina (their first installation in India) – and guess who sold it to me – none other than Ram (Dr. Ramprasad) who was working for Illumina then. Ram and Sakthi (Dr. Sakthivel) soon joined SciGenom and we started incubating MedGenome with some single gene tests for EGRF, KRAS, BRCA etc. Dr.Prasanna joined to drive marketing and we created an initial sales team to focus on cancer which I thought would be a growth area for genetic testing. Paul George came over from Calsoft (my earlier company) to head our IT and Software team.

My close friend Peema (Mahesh Pratapneni) – we had worked closely together for 10 years building Calsoft (1994-2004), had continued to be in touch with me. He was intrigued with what I was trying with SciGenom and started collaborating closely. We spun off MedGenome as a separate company in 2013 and set up a new lab in the Narayana Netralaya building at Bangalore, which all of you are familiar with. Peema had started his VC company Emerge Ventures in Singapore and they brought in our first round of funding. It may surprise many of you that Ray (Rayman Mathoda) was one of our first investors through Emerge. Ray then continued to closely follow MedGenome and guide us till she formally joined MedGenome, U.S in Jan 2020.

Last but not least, our growth would not have been possible without the right financial backing. Over the last seven years, marquee investors like Sequoia Capital, Sofina, HDFC and Leapfrog, and advisors like Neeraj Bhargava and Kris Gopalakrishnan have provided invaluable strategic advice and unwavering support which kept us on the right track. Over the years, every member of the MedGenome family – employees, consultants, advisors, and investors – has helped the company overcome hurdles and lead with our strengths.

Well, why this long story now? Firstly, many of you may not know the story of MedGenome's beginning and the key people behind it; Secondly, not only are all our key people been part of the team from early stages but some even more actively devoting their time and energy into MedGenome such as Ray in the US side. Finally, and most importantly, you can visualize the strong bonds that tie all of us. This has been of great help especially during the Covid-19 pandemic when most of us could not travel or meet in person. I feel very privileged and humbled by this world class team of friends who are now working hard to fulfill our vision. I am excited at the opportunities in front of us and confident that MedGenome will continue to play its part in India's future growth trajectory. Over the next few months, Ram and Ray will articulate our aggressive growth plans post Covid, in India and U.S respectively.

I thank you all for joining us in this journey.

Keep safe and healthy.

Yours truly,
Sam Santhosh,
Founder Chairman and Global CEO

Contents

Most Talked About

The News

MEDGENOME NEWS | October to December 2020 | MEDGENOME NEWS

ACTIA • CLARIA • PRIMA • MICRA • Business • Research • Awards • Genetic Counselling • Health Care

Healthworld.com

Neonatal hypoglycemia, genetic diseases are underlying cause of epilepsy in young Indian children, says study

The findings of this study highlight that neonatal hypoglycemia (low blood sugar at birth) is the most important underlying cause of infantile spasms.

ETHealthWorld • December 16, 2020, 14:15 IST

New Delhi: Neonatal hypoglycemia (low blood sugar at birth) is the most important underlying cause of infantile spasms, according to a study conducted by Medgenome Labs in partnership with doctors from Santokba Durlabhji Memorial Hospital, Jaipur, School of Medicine, University of Glasgow,

WELLBEING insider

Preventive Healthcare Using Polygenic Risk Score for Coronary Artery Disease

brainfeed

What is Genetic Counselling?

Dr. Sheetal Sharda, Senior Consultant in Clinical Genetics, MedGenome Labs Ltd.

Genetic and genomics are slowly becoming routine in clinical practices across the world and in India. Doctors are increasingly relying on genetic tests to help in the diagnosis and also the management of patients with a genetic disorder. But genetics is a complex subject, and the field is evolving quickly with new information emerging at a fast pace. Doctors must constantly update themselves with the latest information and patients also need to understand the results of the test.

Genetic counsellors act as a crucial bridge between the patient, the doctor and the genetic testing lab. Genetic counselling is a communicative process, by which the Consultand (one seeking consultation) and their families are informed about the genetic basis of their disorder, its inheritance pattern and the chances of it happening again in future generations. They are guided through available testing options based on current scientific evidence to choose a course of action in view of their family goals, ethical and religious standards.

The emergence of genetic and genomic technology has given significant importance to a rewarding career

THE HINDU

Genetic research pilot study by Delhi hospital finds 'unexpected results'

PTI

For the first time a high carrier frequency of 4.5% (9 per 200) for Cystic Fibrosis (CF) has been revealed, it claimed.

A genetic research pilot study on cystic fibrosis (CF) conducted by a leading private facility here has found "unexpected results", hospital authorities said.

CF is considered to be very rare in Indian population. This study not only shows prevalence of CF but also to a high extent, they said.

"The genetic research pilot study which was done by the Institute of Medical Genetics & Genomics at Sir Ganga Ram Hospital and has been published in BMC Medical Genetics, London has revealed unexpected results," the hospital said in a statement.

For the first time a high carrier frequency of 4.5% (9 per 200) for Cystic Fibrosis

THE TIMES OF INDIA

New tumor mutation burden test may aid clinicians

Chethan Kumar | TNN | Nov 7, 2020, 23:32 IST

TOI

BENGALURU: MedGenome Labs, a city-based genomics-driven research and diagnostics company has launched a Tumor Mutation Burden (TMB) test to help clinicians make informed decisions on immuno-therapy for cancer patients.

Immuno-therapy is a type of cancer treatment using the body's own immune system to kill the cancer cells. Therefore, a key aspect of success of the TMB test is that the body's immune system should differentiate cancer cells and normal cells.

The mechanism by which the body's immune system can differentiate between cancer cells and normal cells is based on the expression of 'neo-antigens'. Neo-antigens are altered self-proteins which result from a change in the self-protein due to a mutation. Thus, the key factor leading to expression of neo-antigens on the cell

TH All in the genes

EDUCATION

All in the genes

Dr. Sheetal Sharda

Genetic counselling, which is slowly making its mark in India, can be both a rewarding and fulfilling career.

We have entered the era of precision and personalised medicine, which has been enabled by the adoption of genetics and genomics in everyday clinical practice. Doctors are increasingly relying on genetic tests to help guide a patient's diagnosis as well as treatment strategy. But genetics is a complex subject, and the field is evolving quickly with new information emerging very quickly. Doctors must constantly update themselves with the latest information and patients too need to understand the results of the test.

In such a situation, the role of the Genetic Counsellor is crucial. Genetic counselling, which is becoming an integral part of healthcare services, is a communicative process by which the patient and his/her family are informed about the genetic basis of the disorder, its inheritance pattern, and the chances of it occurring in future generations. They are guided through available testing options based on current scientific evidence to choose a course of action in keeping with their family goals, ethics and religious standards.

For press articles, please click

https://diagnostics.medgenome.com/press/

MedGenome Connect

Claria
Reproductive Genetics

Claria events for this quarter were primarily dominated by a series of webinars with the Bangladesh Society for Medical Genetics. We did a series of 5 webinars over 2 months covering topics ranging from Basics of genetics to the role of cytogenetics in prenatal screening and NIPT with Dr. Sheetal Sharda, Dr. Meenakshi Lallar, and Dr. Priya Kadam as speakers. This webinar series was a tremendous success with many Bangladeshi doctors appreciating our content and resulting in an increased sample volume from Bangladesh. More engagements are planned for the new year. Our carrier screening paper published in BMC medical genetics highlighting the increased prevalence of certain rare genetic disorders such as Cystic Fibrosis etc. in India was picked up by leading national dailies such as The Hindu and The New India Express. This study also showed the need for an India specific database for Carrier Screening.


THE NEW INDIAN EXPRESS | BENGALURU

03 BENGALURU SATURDAY 21.11.2020

'Many carriers of genetic disorders among Indians'

Study conducted on North Indian population, were screened for pathogenic variants

EXPRESS NEWS SERVICE

A study using Next Generation Sequencing (NGS) to determine carrier frequency and to look out for any mutations in common genetic disorders in Indians, done by MedGenome Labs in Bengaluru, and Sir Ganga Ram Hospital, New Delhi, found that out of the 200 participants, 32 (16%) were found to be carriers of one or more rare genetic disorders, 12 individuals (6%) were identified to be carriers for congenital deafness, 9 individuals (4.5%) were carriers for cystic fibrosis and 2 individuals were detected to be carriers for the Pompe disease.

The study showed a higher carrier frequency for these disorders, which was contrary to the generally held view about their low prevalence in Asian Indians. "The disease causing variants observed for disorders such as deafness, cystic fibrosis, Pompe disease, Canavan disease, primary hyperoxaluria, junctional epidermolysis bullosa, galactosemia, medium chain acyl CoA deficiency etc. were different from what is seen in the Western population. Thus, this pilot study highlights the importance of having a Genetic Variant Database for the Indian population," a press release said.

The 22 month study was done on a North Indian population, where 200 individuals were screened for pathogenic variants in shortlisted 88 genes, using NGS technology. These variants were classified as per the guidelines of American College of Medical Genetics. Dr Sunita Bijarnia Mahay, author and senior consultant, Institute of Medical Genetics & Genomics, Sir Ganga Ram Hospital, said, "This study brought surprises by detecting those genetic disorders like cystic fibrosis which were not thought to be common. In future, such NGS-based screening tests will benefit not only young couples planning a baby, but also healthcare officials in charting out prevention strategies for the Indian population."

Currently, there is no carrier screening programme available in India, except for limited screening for Thalassemia and Down's Syndrome. Dr Sheetal Sharda, senior consultant, Clinical Genetics, said couples may not even be aware that they could be carrying a genetic variant. In most cases, carriers of a genetic disorder are asymptomatic and may have no family history, and unfortunately, their carrier status is often confirmed only after an affected baby is born.

Prof I C Verma, senior consultant and adviser, Institute of Medical Genetics & Genomics, Sir Ganga Ram Hospital, said those with IVF pregnancies, consanguineous couples (descendants with the same ancestor) and those with previous history of abnormal babies must get carrier testing done before their next pregnancy, to prevent the birth of babies with genetic disorders.

ALL IN A DAY'S

Health workers at a Covid clinic sort through samples collected

Emergency contra pandemic, caused

EXPRESS NEWS SERVICE

DOCTORS have observed an increase in the number of women reporting side-effects from using emergency contraception too often, instead of regular birth control.

Doctors at Fortis Hospital see about 10-15 patients every month with health issues caused by using emergency contraception pills (ECP) too often. "Emergency contraception pills have high doses (of hormones) to prevent ovulation. We are observing an increase in the number of women reporting side-effects of these pills, such as nausea, vomiting, and irregular bleeding. The pills also cause hormonal imbalances, which could that periods don't occ time," said Dr Parimala obstetrics and gynaec consultant at Fortis Hos

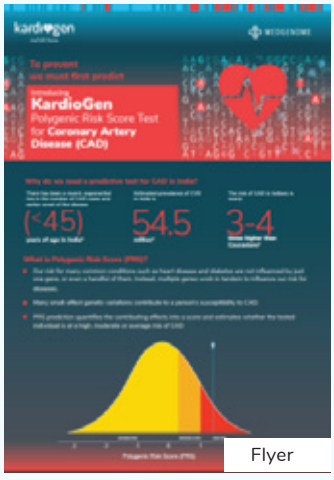
Doctors said women resort to emergency c ception often because ples are spending more time at home together due to the lockdown, and also a certain lack of recreation and entertainment.

Regular birth control includes the use of condoms, oral contraceptives (tablets taken for 21 days), and more permanent methods such as the use tra-uterine devices su Copper-T.

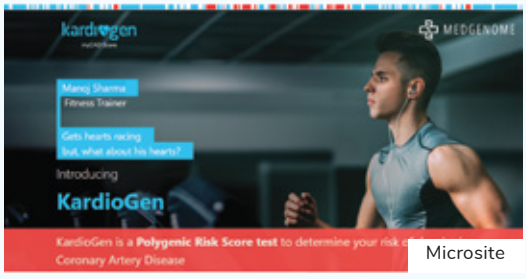
Around 20%-30% of w

ACTIA
Inherited Genetics

Actia was focused on launching the Coronary Artery Disease Polygenic Risk Score (CAD-PRS) The marketing efforts involved designing of relevant collateral, launching of microsite for lead generation and even created a focused social media campaign. A video explaining the concept was developed and shared through our social media channels and whatsapp groups. We also started actively pitching PR stories about the test to create more awareness about the test.



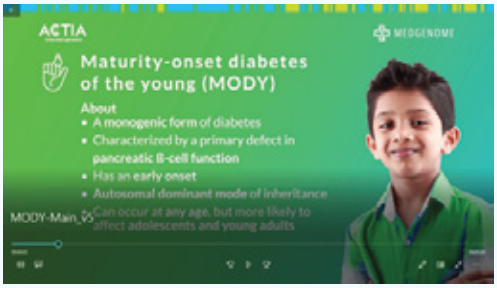
Flyer



Microsite

Video


Social Media Post



However, it was not all about KardioGen. This quarter also witnessed the World Diabetes Day falling on the 14th of November. We highlighted the need to check for Maturity Onset Diabetes of the Young (MODY) through a social media post highlighting the need for such a test in management of early onset of diabetes.

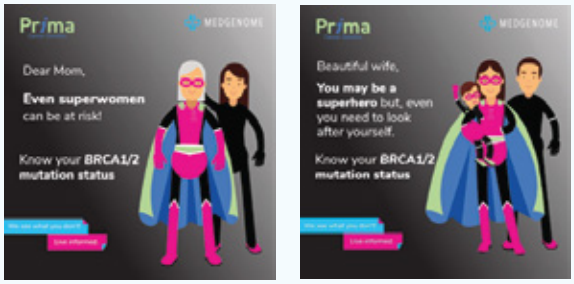
MedGenome Connect

Prima Cancer Genetics

We continued to focus on digital platforms and leveraged it for various activities such as online symposiums and digital campaigns. November being the lung cancer awareness month, we conducted a successful symposium on Lung Cancer and Genetics where eminent speakers – Dr. Boben Thomas, Dr. Tushar Patil and Dr. Mithun Shah shared their clinical experience, along with Dr. Arun Kumar who talked about MedGenome's experience with lung cancer and genetics. The symposium was well received and well attended.

Additionally, a focus on digital campaigns for public awareness was continued for October (Breast Cancer Awareness Month) and November (Lung Cancer Awareness Month).

Breast Cancer Awareness Month



Lung Cancer Awareness Month

micra Infectious Disease Genetics

As a part of our efforts in spreading awareness about our proprietary SPIT SEQ test, we conducted a webinar in December with Dr Anil Kumar (AIMS, Kerala) and Dr Lakshmi Soundararajan (MedGenome) as the speakers. The webinar was quite interactive and well-received.

What's New?

Research collaboration & Publications

01

NGS-based expanded carrier screening for genetic disorders in North Indian population reveals unexpected results - ***Published in BMC Medical Genetics***

https://bmcmedgenet.biomedcentral.com/articles/10.1186/s12881-020-01153-4

02

Phenotypic expression and clinical outcomes in a South Asian PRKAG2 cardiomyopathy cohort - ***Published in Nature***

https://www.nature.com/articles/s41598-020-77124-9

03

A pilot study reveals low blood sugar at birth and genetic diseases, are important underlying cause for a common form of epilepsy in young Indian children - ***Published in Epilepsy Behavior Reports***

https://www.sciencedirect.com/science/article/pii/S2589986420300459?via%3Dihub

AWARDS

The Top 25 **Healthcare Technology CEOs** of Asia for 2020

Healthcare Technology Report, New York announced ***Dr V L Ramprasad, CEO, MedGenome*** among the *Top 25 Healthcare Technology CEOs of Asia for 2020.*

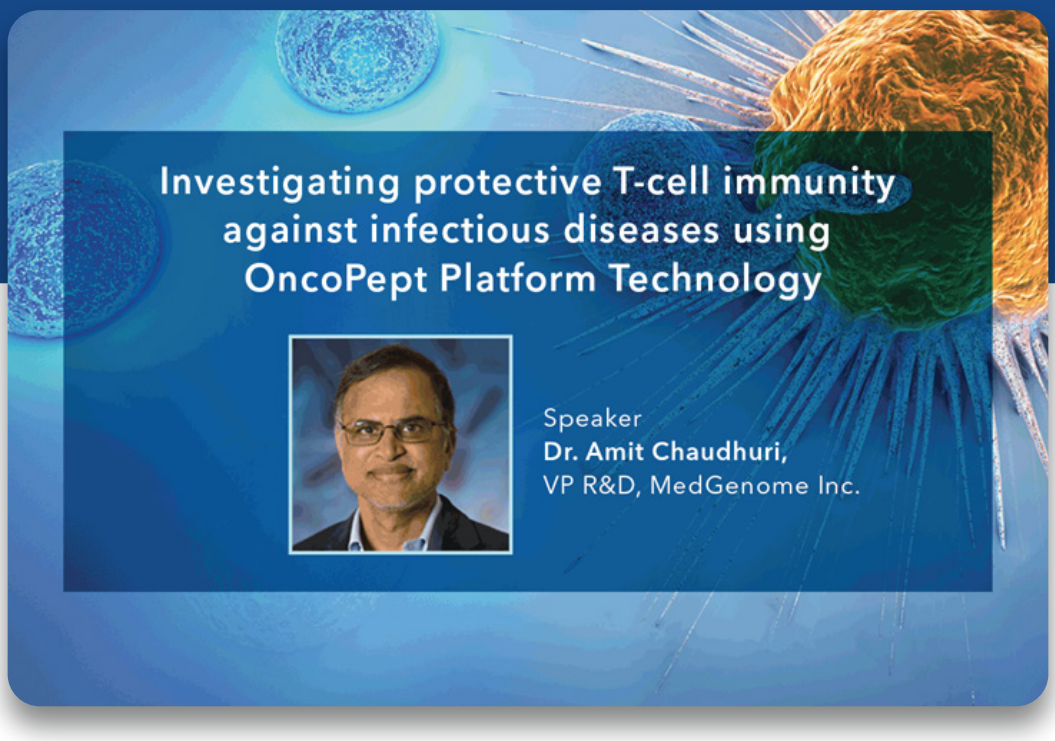
From our US Office

This quarter we saw an overall recovery in business process owing to our ability to keep all our customer base intact and the tireless efforts of our team to deliver the services in time during this tough pandemic situation.

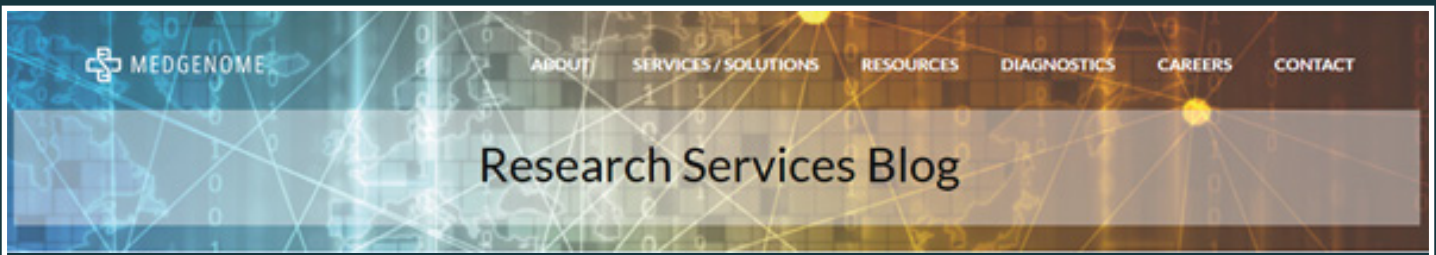
We are happy to share some of the novel insights on COVD-19 through our recent webinar which was presented by Dr. Amit and a blog article by our OncoPept Team who are actively in pursuit of finding "Approaches to Fighting COVID-19 and New Emerging Infectious Diseases". To know more, please click on the below links:

https://research.medgenome.com/videos/

https://research.medgenome.com/approaches-fighting-covid-19-new-emerging-infectious-diseases/



And, we have recently added a few more articles on our Research Blog. MedGenome colleagues are encouraged to take initiative and contribute towards the blog. You can share your viewpoints and articles with Vinay and Hiran at mgus-blog@medgenome.com



Our 2 new publications – the result of continued efforts of US and India Team are as below which appeared on "Nature Scientific Reports" and Journal of "BMC Ophthalmology" subsequently.

Links for your reference:

https://www.nature.com/articles/s41598-020-77124-9

https://bmcophthalmol.biomedcentral.com/articles/10.1186/s12886-020-01705-5

Making a Difference

kardiogen
myCAD Score

CAD-PRS detects high risk of **Coronary Artery Disease** in a 14-year-old heart attack patient

Patient information

A 14-year-old female had heart attack/MI and underwent anterior wall Myocardial Infarction on 06 October 2020. She is currently on antiplatelet, statins and heparin medication. She was suggested to undergo Coronary Artery Disease-Polygenic Risk Score Test to find out whether she was at high risk of getting Coronary Artery Disease.

Genetic History

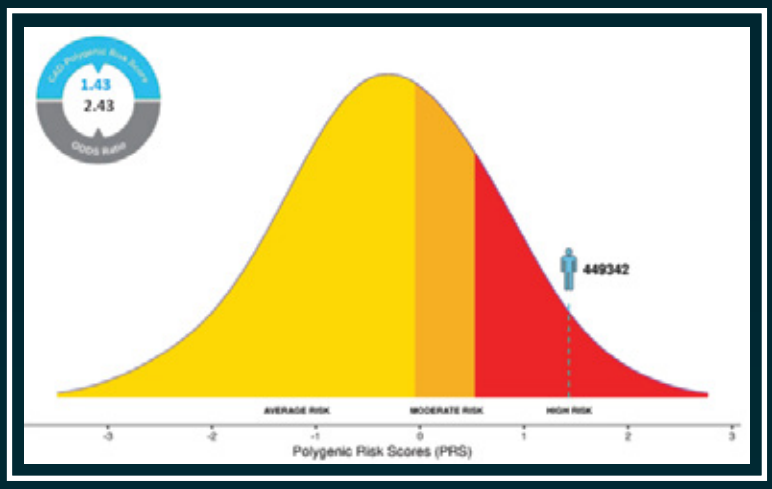
No history of heart disease, hypertension and hypercholesterolemia in the family.

Genetic testing at MedGenome

Coronary Artery Disease-Polygenic Risk Score Testing was done at MedGenome.

Result

The person has a PRS of 1.43 (blue dotted line), which indicates a population high polygenic risk, with an odds ratio of 2.43 relative to MedGenome's cohort.

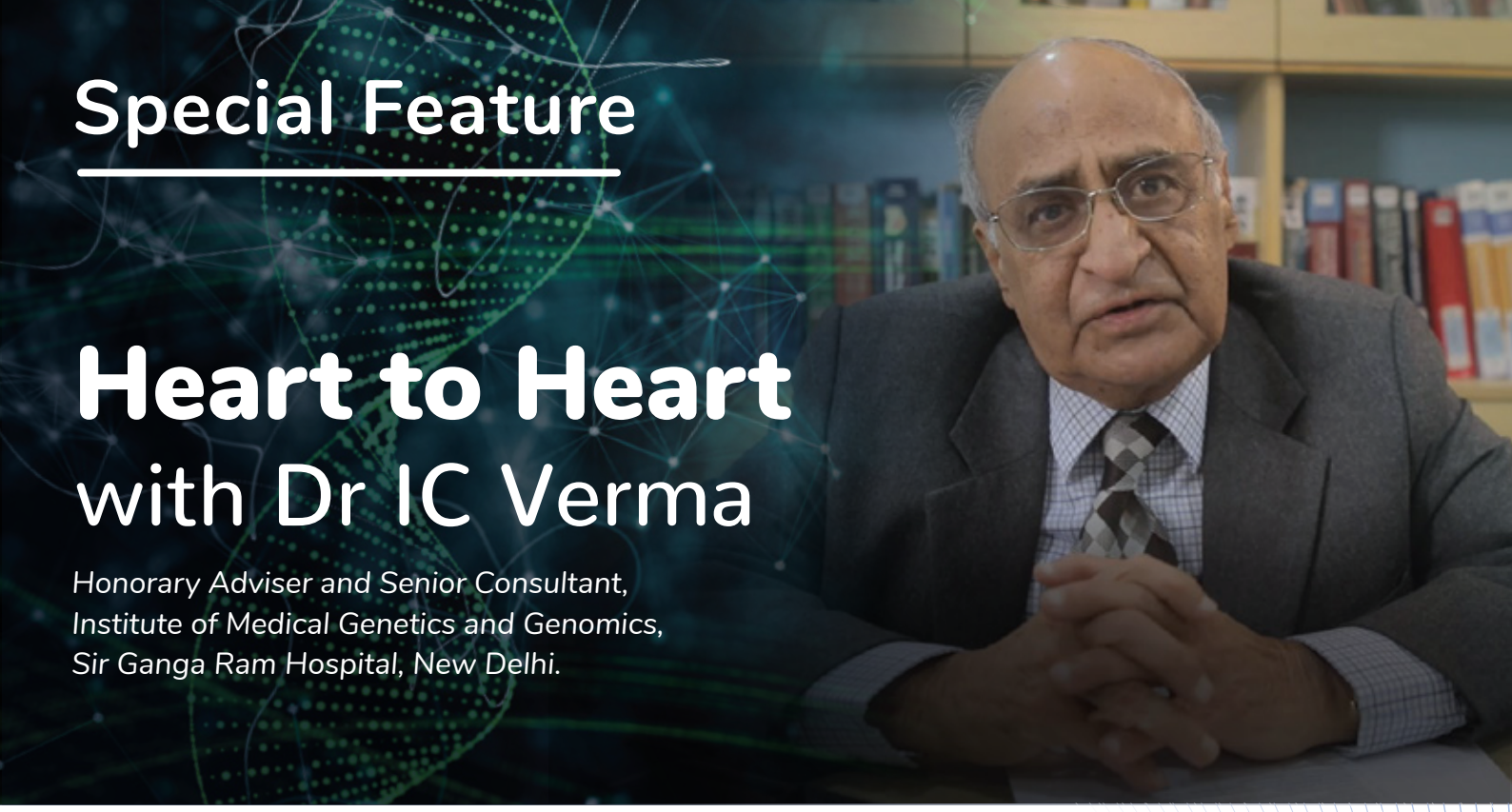


Case Discussion

The Coronary Artery Disease-Polygenic Risk Score (CAD-PRS) for the person was found to be 1.43. This score has been profiled as a high risk as observed in MedGenome's study cohort of South Asian ancestry. The person carries a high polygenic risk of getting CAD, with an odds ratio of 2.43, based on MedGenome's study cohort. Thus, the CAD-PRS test can accurately predict the risk of CAD in very young patients.

Test Details

Test Code	Test Name	Sample	TAT
MGM1473	Polygenic Risk Score for Coronary Artery Disease	3 ml blood in EDTA tubes	12 Working Days



Special Feature

Heart to Heart with Dr IC Verma

Honorary Adviser and Senior Consultant,
Institute of Medical Genetics and Genomics,
Sir Ganga Ram Hospital, New Delhi.

Dr Ishwar C Verma is Honorary Adviser in Genetics and Senior Consultant in the Institute of Medical Genetics and Genomics, and Professor of Medical Genetics at Ganga Ram Postgraduate Institute of Medical Sciences in New Delhi. He was formerly Professor in Genetics and Pediatrics at the All India Institute of Medical Sciences (AIIMS), New Delhi and established the WHO Collaborating Centre in Genetics in Developing countries at AIIMS, New Delhi. He has been an adviser in genetics to WHO Headquarters in Geneva, and SEARO for several years, and has been the Past President of India Society of Human Genetics, Society of Fetal Medicine and Indian Society of Inborn Errors of Metabolism in India.

Dr Verma has been accorded with numerous national awards / accolades such as the Indian Council of Medical Research (ICMR) award, the National Academy of Medical Sciences (NAMS) award, Dr B C Roy (Medical Council of India) National Award. **He has been mentioned in the Limca Book of Records 2003 as a Pioneer in Genetics in India.**

In this new year edition of GeKNOWMe, we are honoured to feature an interview with Dr Verma. This interview presents some interesting and useful insights from his work in genetics, his professional journey, latest research and more.

Tell us about what prompted your interests towards Genetics? How did you discover your passion for genetics?

When I qualified MBBS from Amritsar Medical College in 1961, Watson and Crick had already published their paper on structure of DNA in 1953. No one ever taught us about this during pre-medical or medical training. It is only after I started my senior residency training in hospitals in Liverpool in the UK in 1963 that I learnt about it. The monthly meetings of the Liverpool Medical Society were chaired by Professor Cyril Clarke who had great interest in genetics.

He was experimenting with butterflies to solve the riddle of Rh incompatibility and presented this data at one of the meetings. Many other topics involving genetics were also presented, such as pharmacogenomics of isoniazid and sulphonamides, and HLA in arthritis and spondylitis.

After obtaining the MRCP from the Royal College of Physicians London and returning from England, I joined the All India Institute of Medical Sciences in New Delhi in 1967. I wanted to pursue Neonatology, however Professor Ghai, who was the head of the department asked me to take up and develop the new field of genetics at AIIMS, Delhi. Given the Liverpool experience I was happy to accept this offer.

What challenges did you face during the early years as a genetic researcher since understanding genetic disorders and testing was fairly new in India?

Yes, it was a struggle in the beginning. There were a few anatomists in India who were studying chromosomes. In our laboratory we followed the book by Yunis *et al* to set up cultures of lymphocytes to study the chromosomes. At AIIMS, I had the opportunity to interact with Dr. Grewal who was working in the department of Anatomy and had a PhD in Genetics from London University. The Swiss Government had given a big grant to AIIMS to establish genetics and this included training in cytogenetics in the Genetics Laboratory of Professor Werner Schmid at the Children's Hospital in Zurich. It was good to imbibe the meticulous care with which the Swiss carried out scientific research. I still remember and follow the advice he gave me "if a technique works, do not change it, even if new techniques come along." Later, I was selected for a WHO fellowship to work in the Genetics Unit in Massachusetts General Hospital in Boston, USA. I took rotations in clinical genetics as well as in the different laboratories, as I was clear in my mind that to practice genetics in India, I would have to learn the associated laboratory techniques too. After returning to Delhi, I was awarded a fellowship of the Royal College of Physicians London, to spend three months in some of the best genetic centres in the UK- in Guys Hospital in London, The MRC Molecular Medicine Unit in Oxford, the Will ink Biochemical Laboratory in Manchester and Professor Emery's Genetic Department in Edinburgh. This gave me an opportunity to learn many new techniques in molecular and biochemical genetics from experts in these centres. The friendships I developed proved extremely useful in setting up these techniques and services in India.

> "Yes, it was a struggle in the beginning."

> "if a technique works, do not change it, even if new techniques come along."

You are a pioneer in genetics, how would you describe the change in the perceptions of the society and mindset of people in India about genetic disorders and testing?

As nutritional and infectious diseases were brought under control the mortality which genetic disorders were causing became apparent. It was realised that India has as many genetic disorders as the West, and in fact even more due to the large number of consanguineous and endogamous marriages. In recent years, the huge burden of non-communicable disorders such as coronary artery disease, diabetes mellitus, hypertension, stroke etc. became apparent, which had a large genetic component. These had to be controlled in addition to the nutritional and infectious disorders – which is the so-called dual burden of disease.

> **“It was realised that India has as many genetic disorders as the West, and in fact even more due to the large number of consanguineous and endogamous marriages.”**

What are some common myths associated with genetic testing in our country?

In the early days there were primarily three techniques – cytogenetics, biochemical genetics and enzymology and molecular genetics. Most people stuck to one of these. I had realized that to be a success in India I had to learn all the three. ***Molecular genetics was the last to come on the field and is gradually replacing other techniques.*** Looking back, the myth was that these techniques were difficult. In Oxford I saw a schoolboy performing perfect sequencing. He was initially taught by an expert and was then employed to do research sequencing. This is the pattern of learning that is followed all over the world. You learn from others and teach those who follow you. In India I noticed that if I sent a student to learn a new technique in another lab, he/she was reluctant to teach the same technique to others. That is why some of the arts that flourished in India in the past did not survive into the present e.g. the architectural expertise of building the Taj Mahal. So, the genetic techniques are easy to carry out, once you have learnt them from someone knowledgeable. Indian doctors knew little about genetics, and they thought it is a difficult subject and the techniques and tests are hard to learn. This was certainly a myth. As people did not know much about genetics, they thought all the wrong things about it. I remember during those days the US experts had forwarded a proposal for consideration to eradicate malaria in India. It depended upon creating mutant mosquitoes who would be sterile. The Ministry of Health in the Government of India shot down the project saying the US Government wanted to spy on India through this project. The atmosphere was therefore not conducive to establishment of genetics as a specialty as genetic disorders were considered very rare, and the good that genetics could do was not appreciated.

Would you like to share an interesting case study with us?

Soon it became apparent that some genetic diseases are pretty common e.g. beta thalassemia major and spinal muscular atrophy (SMA). The former has a mean carrier frequency of about 3.3 %, though there are many communities such as Aroras and Lohannas who have a much higher frequency. The rate worked out to the birth of one child with thalassemia major per 4000 births. The cost of treatment of an affected child was expensive and burdensome and most doctors realized that it is much better to screen every pregnant woman for thalassemia rather than diagnose and treat the affected child after birth. Similarly, babies born with SMA most often used to die within 6 months of birth, and we could do nothing to stop this except make a prenatal diagnosis in future pregnancies. For the last three years, or so antisense oligonucleotide therapy has been used to make more SMN protein by the SMN2 gene and this ameliorates the symptoms. In fact, if the treatment was given when the patient is asymptomatic, it makes these children not to develop the diseases at all and behave completely normally. These case examples demonstrate how the advances in genetic therapies have changed the outlook for many genetic disorders.

> “Advances in genetic therapies have changed the outlook for many genetic disorders.”

DBT has taken some initiatives by opening kendra/centres for genetic testing and tie ups for training. Do you think there is a lot more scope for the Government to be involved in this segment? What do you think are the constraints?

DBT has done a great deal to encourage and develop genetic services in India. Recently, they have introduced a program of establishing genetic centres and services in government institutes. The concept is good but what we have observed in the past is that these centers stop functioning once the DBT withdraws its financial support. A mechanism must be set up to ensure that these centers and services must continue once the DBT project is over. Two steps are essential for this – one is to appoint people who have real interest in genetics to lead in these centers, and secondly charge some money for the tests so that a fund is generated for the laboratory test to continue. Tests when offered completely free lose their value and patients then expect free tests as their right. Moreover, although the Government professes to support public-private partnership in such matters, in practice they do not follow it. The private sector must be involved in providing genetic services and tests to the public.

> “Centers stop functioning once the DBT withdraws its financial support.”

> “The private sector must be involved in providing genetic services and tests to the public.”

You have recently published a research article about carrier screening to detect genetic disorder among north Indians. How has population genomics influenced genetic diagnostics? What more lies ahead?

For this study we had used nextgen sequencing technology to sequence the whole genes rather that check for targeted mutations, as the data on mutations in various genes in India is grossly incomplete. The study sprang a number of surprises – genetic deafness turned out to be the commonest genetic condition among the cohort, secondly, cystic fibrosis which was considered to have a low frequency on India was shown to have a frequency close to the western data, and thirdly, carrier status for Pompe disease was also demonstrated to be common. The only limitation was that the study was done in only 200 subjects, and we hope **MedGenome**, our partner in this project would carry out a study on a larger number of subjects who hail from all over India. A properly designed study by MedGenome would be a great boon for India.

> **“A properly designed study by MedGenome would be a great boon for India.”**

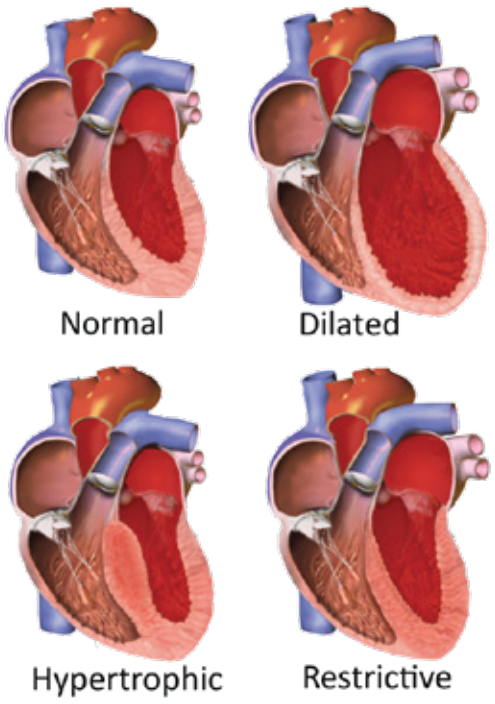
You have been an inspiration to all of us, what message would you like to give to anybody who’s interested in taking up genetics.

Genetics is currently one of the most exciting fields to work in. In terms of innovation and discovery of new genes this field is unrivalled. All those who join to undertake a journey in this field, should tie their seat belts properly as they are in for an exciting ride.

Clinical and genetic features of a **rare form of cardiomyopathy in multiple families from India**

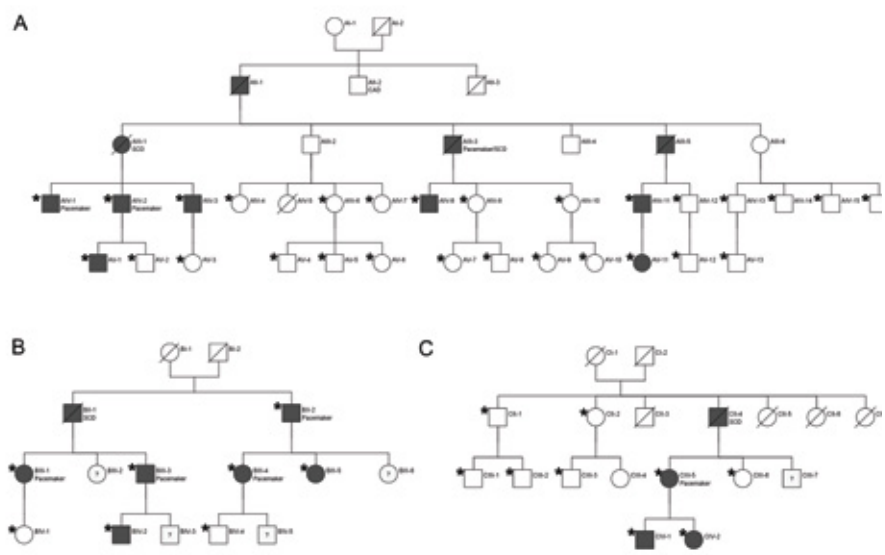
By **Sameer Phalke**, PhD
Senior Scientist, MG India

Cardiomyopathies are the group of diseases that affect the heart muscles. Hypertrophic cardiomyopathy (HCM) is the most common cardiomyopathy worldwide and a genetic origin for this heterogenous group of diseases is found in ~40-60% of HCM patients, usually with an autosomal dominant mode of inheritance. The mutations responsible for HCM often localize genes encoding sarcomeric proteins. There are several other rare genetic cardiomyopathies which are not caused by cardiac sarcomere mutations and yet they share many of the phenotypic manifestations of HCM disease. PRKAG2 cardiomyopathy is one such rare HCM phenocopy characterized by ventricular pre-excitation, progressive conduction system disease and left ventricular hypertrophy. PRKAG2 cardiomyopathy is caused by the mutation in the gene encoding PRKAG2 (Protein Kinase Adenosine monophosphate activated Gamma 2 non-catalytic subunit 2) protein. The clinical symptoms of PRKAG2 cardiomyopathies overlaps with sarcomeric HCM disease with high incidence of cardiac conduction system disease and high rates of sudden cardiac death (SCD) and often leads to misdiagnosis that hinders the appropriate patient management.



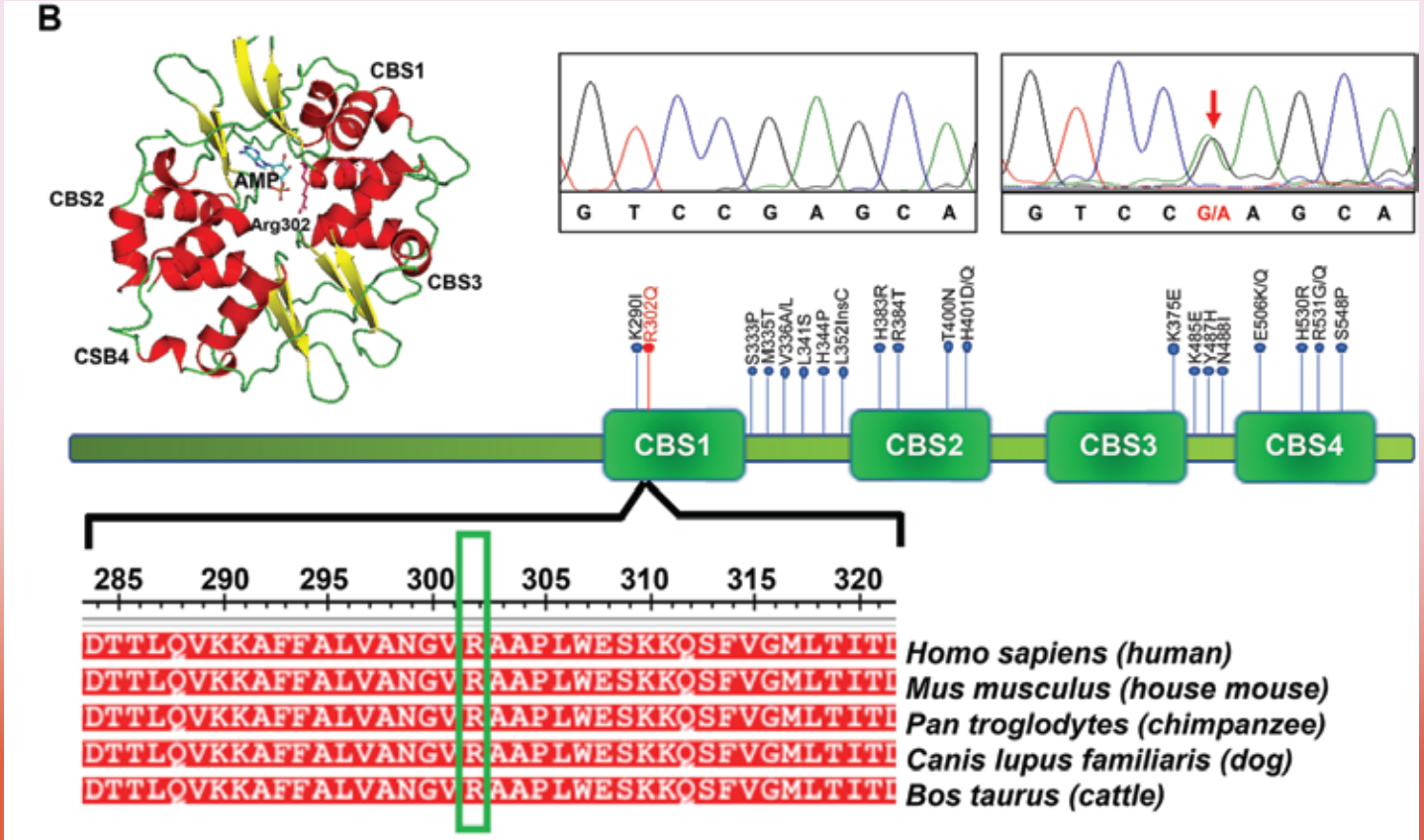
By Npatchett at English Wikipedia, CC BY-SA 3.0, https://commons.wikimedia.org/w/index.php?curid=40722856

So far, less than 300 families with PRKAG2 cardiomyopathy have been reported worldwide and none reported from South Asia. In this study, we looked at three unrelated index patients from Southern state of India, Kerala, who were presented with HCM like clinical symptoms. Further clinical evaluation of the families of these index patients revealed several other members of these families to be affected by similar clinical phenotype. Considering strong familial features, we performed the exome sequencing on the affected and unaffected members of these families to understand the genetic cause of the disease in these families. Indeed, whole exome sequencing identified a pathogenic autosomal dominant missense p.Arg302Gln mutation in PRKAG2 protein, the mutation lies in the CBS1 domain of PRKAG2 protein and impacts the AMP/ATP binding pocket. The mutation was identified as a causal variant for the disease in all 22 affected individuals in these families establishing a PRKAG2 cardiomyopathy diagnosis. There was a complete genotype-phenotype correlation in all the tested individuals. We collected the clinical, electrocardiographic, echocardiographic, and cardiac MRI data from 22 individuals with PRKAG2 variants and followed them to understand the clinical outcomes and the natural history of the disease in this South-Asian PRKAG2 cardiomyopathy cohort (68% men; mean age 39.5 ± 18.1 years) for over a period of 7-years. During this follow up period, their condition progressed with 8 out of 22 needing permanent cardiac pacing for atrio-ventricular blocks and sinus node dysfunction along and 6 out of 22 succumbing to sudden cardiac death (SCD).



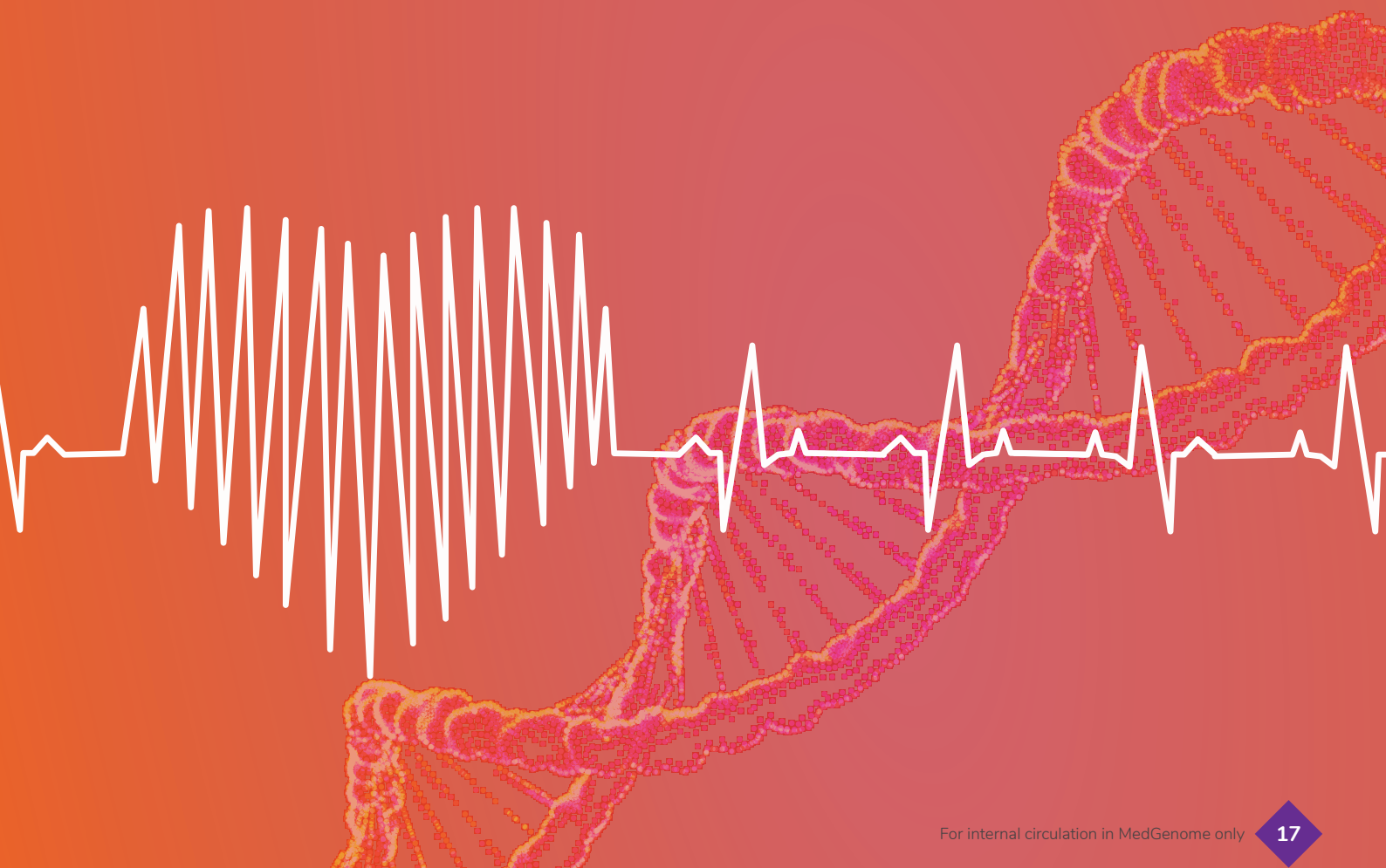
Detailed pedigree of the three families studied in this cohort. (A) Family A (FGD0128), (B) Family B (FGD0137) and (C) Family C (FGD0314)

This is the first study with comprehensive analysis of the clinical spectrum, outcomes and genetic analysis of three unrelated families affected by PRKAG2 cardiomyopathy in the South Asian population. This also is the first study with 22 affected individuals coming from a single cardiomyopathy centre. The investigation with extended clinical follow up of this PRKAG2 cohort has revealed invaluable insights into this rare disease from an entirely different demographic perspective. In this study, the judicious use of genetic testing accurately characterised the specific type of cardiomyopathy in each patient and thus led to the rapid identification of a large number of family members who inherited the same disease. This timely recognition led to the systematic risk stratification of the patients and their family members, which allowed them to receive advanced therapy which would protect them from the risk of a potential sudden cardiac death.



Identification of casual variants in the three families in the study cohort

From the studies like this it is quite clear that, at least for the inherited genetic diseases, we are now in the era of leveraging the potential of genetic testing for the prompt diagnosis of the disease as well as choosing an appropriate management strategy that will improve the quality of life and has a tremendous impact on the cost of healthcare. The unique population structure of India makes studies such as this possible. Familial disease studies in India will create a wealth of knowledge and opportunity to understand many diseases beyond HCM.



Inherited retinal degenerative diseases

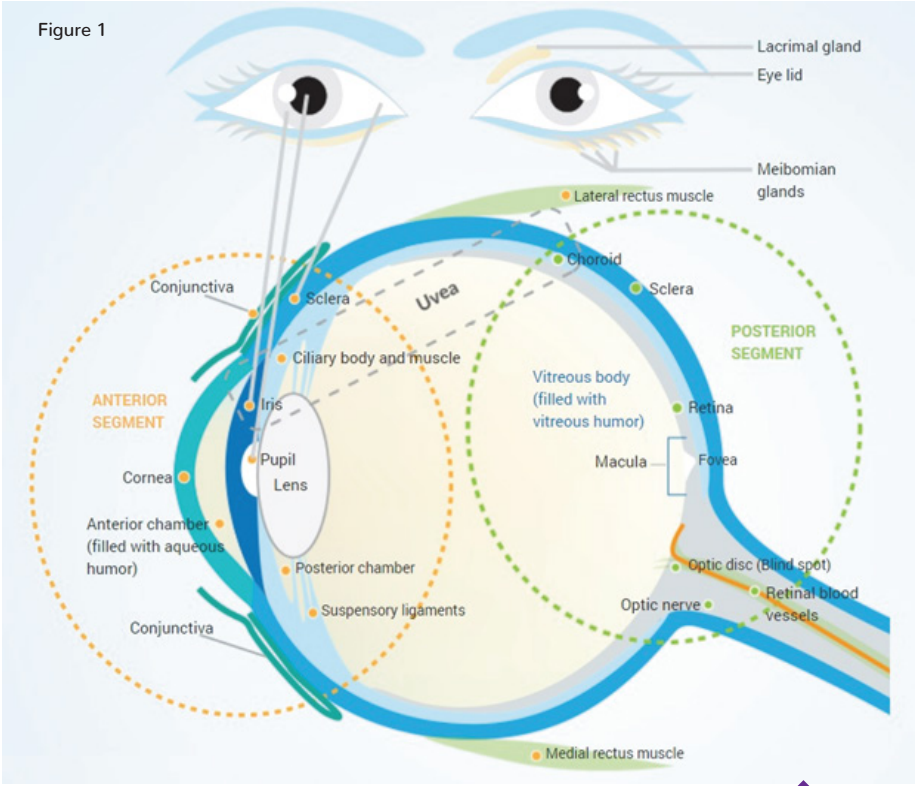
By **Soumittra N, PhD**

Disease Head - Ophthalmology, R&D Division
MG India

Human eye is a complex organ, sitting within the bony socket called the ocular orbit and attached to it through the extraocular muscles. The human eye consists of the (i) anterior segment that includes, the conjunctiva, cornea, sclera, iris, pupil, lens, and the aqueous humor, (ii) the posterior segment including the retina, retinal pigment epithelium, choroid, posterior sclera, vitreous humor, and the optic nerve (iii) ocular adnexa comprising the lacrimal and Meibomian glands, eye lashes, and the eye lids[1]. Figure 1 shows the schematic representation of the different segments and cross section of the eye.

Retina is the photosensitive layer of the human eye, captures and processes the visual information of the world that we see. Retina consists of 7 layers, the nerve fiber layer, ganglion cells, amacrine cells, horizontal and bipolar cells, muller cells, the photoreceptors; rods and cones, and the retinal pigment epithelium[1]. The light captured by the photoreceptor cells in converted to electrical signal, during the phototransduction pathway, the signals are further processed, and the image is formed in the visual cortex of the brain[2].

Figure 1

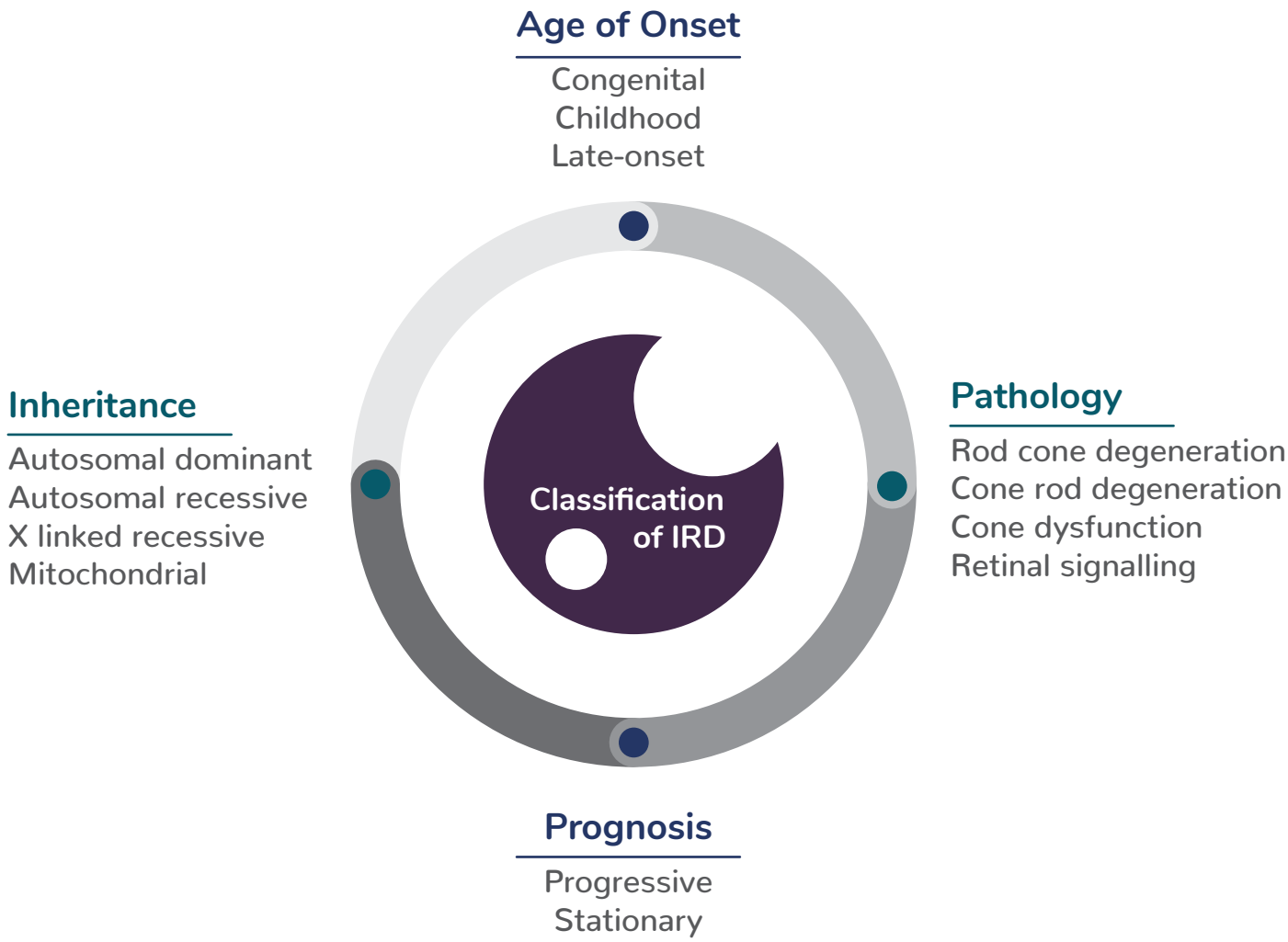


Inherited retinal degenerative (IRD) diseases are rare with a collective prevalence of 1 in 2000, caused due to the degeneration or dysfunction of the photoreceptor cells- the rod and/or the cones, and/or degeneration of retinal pigment epithelium, or retinal ganglion cells.

Classification of IRD

IRD presents with both clinical and genetic heterogeneity and can be classified in many ways. (i) The onset of the disease varies from being congenital as in Leber congenital amaurosis (LCA) to presenting in the first decade or in the 4th decade of life as in autosomal recessive (ar) or autosomal dominant retinitis pigmentosa (adRP), respectively. (ii) IRD can be stationary as in congenital stationary blindness (CSNB) or progressive as in RP. (iii) Further, it may be classified based on pathology, dysfunction of signalling from rods and cones to bipolar cells as in CSNB, or degeneration of cones as seen in Stargardts (STGD), or degeneration of rods followed by cones as in RP, cone-rod dystrophy (CRD) characterised by primary cone degeneration followed by rod. (iv) IRD show either autosomal dominant (AD), autosomal recessive (AR), or X-linked recessive (XLR) pattern of inheritance[3]. Rarely, digenic inheritance i.e. two heterozygous non-allelic mutations, on two separate genes co-inherited has also been reported in IRD[4] (Figure 2).

Figure 2 *presents the different categories of classification of IRD*



Most of the IRD show genetic heterogeneity, i.e., mutations in more than one gene can cause the same phenotype (non- syndromic RP is caused by 84 genes) as well as allelic heterogeneity i.e. mutations in same gene can cause different phenotype (eg.: mutations in *RPE65* can cause Leber congenital amaurosis (LCA) or retinitis pigmentosa (RP).

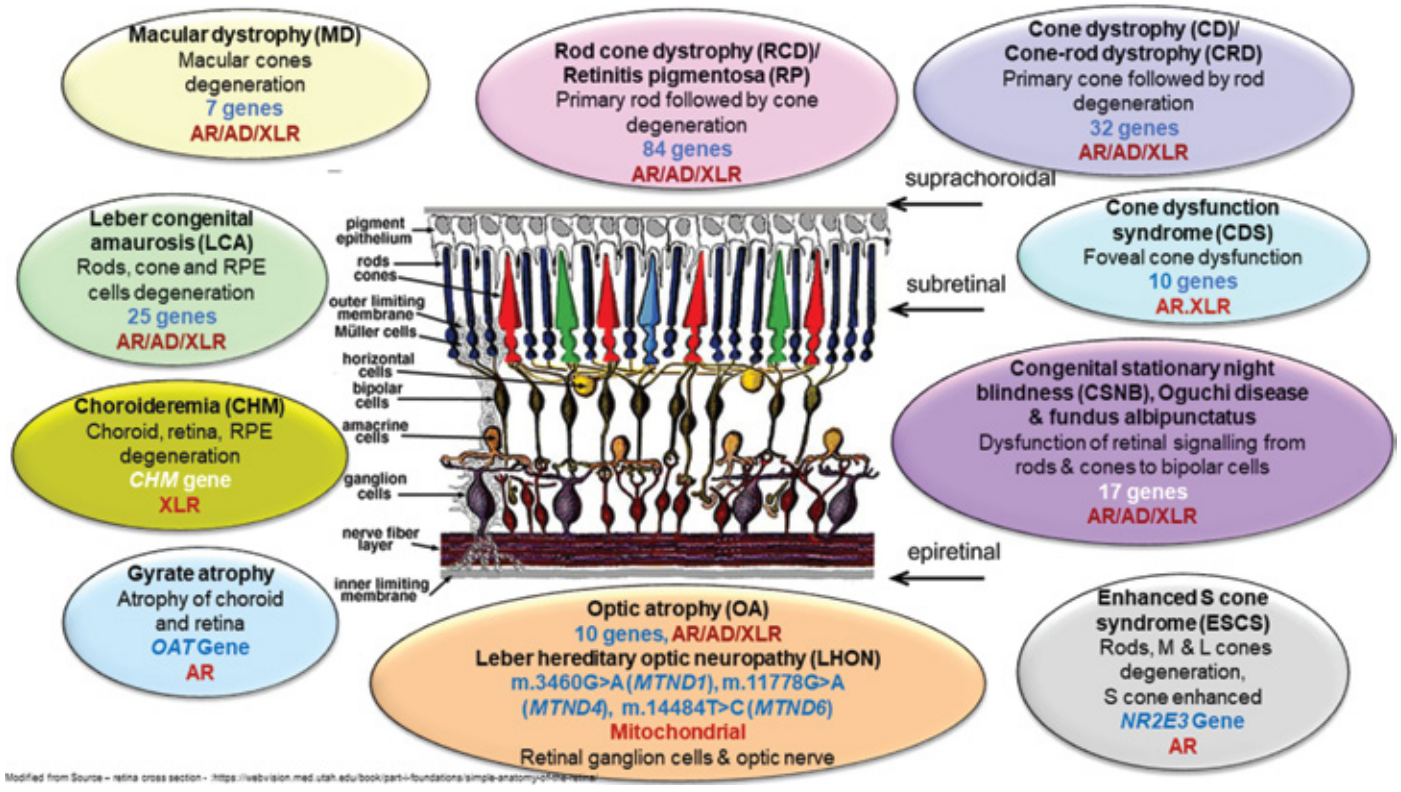
IRDs predominantly present as non-syndromic forms which constitutes 70-80% of cases, however, syndromic IRD where another organ system is affected like Usher syndrome with retinal dystrophy and hearing impairment or systemic IRD with multiple systems involved (Bardet Biedl syndrome) are seen in 20-30% of cases[3].

Types of IRD and their genetics

IRD presents with vast spectrum of phenotype variability, both inter and intra familial. The clinical presentation, fundus changes, rate of progress show considerable variability as well as overlap between different types of IRD[5]. More than 270 genes have been identified to cause IRD, genes that are involved in eye development, photoreceptor survival, phototransduction mechanisms, retinoid cycle, retinal enzymatic function, or cell structure[2].

Figure 3 shows a schematic representation of cross section of retina, the types of IRD, ocular cells/layer affected (rods/cones/ retinal ganglion cells/choroidal layer/ retinal pigment epithelium (RPE)/optic nerve), number of genes so far identified, and pattern of inheritance.

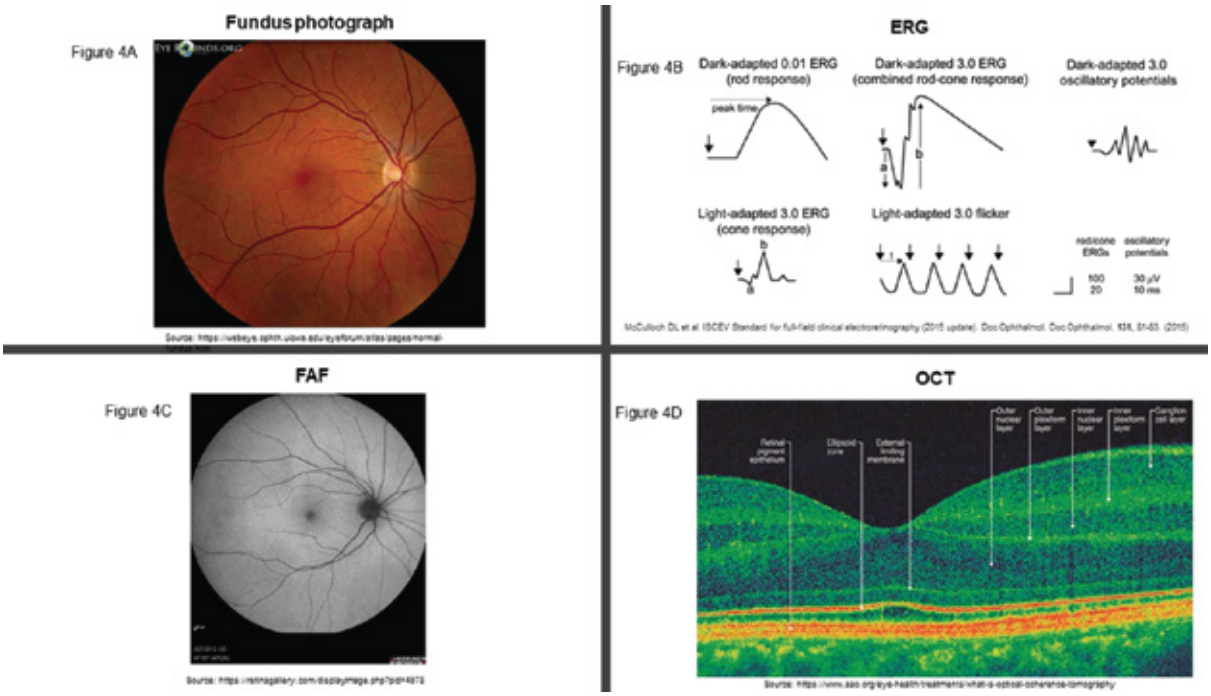
Figure 3



Clinical Diagnosis of IRD

IRD are clinically diagnosed based on slit lamp biomicroscopy, fundus photograph (Figure 4A[6]), electroretinogram (ERG)[7] (Figure 4B), fundus autofluorescence (FAF) (Figure 4C[8]), optical coherence tomography (OCT) (Figure 4D[9]). The ERG wave patterns help understand the extent of functional loss of photoreceptors, identify the types of retinal cells affected/ involved and is also diagnostic for certain types of IRD like CSNB, ESCS. All imaging tools assess the pigmentary changes, blood vessel attenuation, involvement of the various sectors of retina, integrity of the different retinal layers and these helps define the genotype-phenotype correlation. Recently, optical coherence tomography – angiography (OCTA) that captures the depth resolved image of microvasculature of choroid and retina is also being used[10].

Figure 4 (A) – Normal fundus photograph (B) Normal ERG wave patterns (C) Normal fundus autofluorescence (D) Normal OCT



Gene discovery and genetic testing

Linkage analysis, homozygosity mapping, candidate gene screening, screening the gene based on similar phenotype in existing animal models[3] have been used to identify the candidate genes in IRD. The emergence of next generation sequencing (NGS) technology has not only facilitated the identification of novel genes causing IRD[11,12] but also made the clinical genetic testing of this genetically heterogenous disease a reality as many genes can be screened simultaneously at a very reasonable cost[13]. The diagnostic yield of specific phenotype of IRD like RP or LCA or CSNB or screening various phenotype spectrum of IRD using either targeted gene panel or whole exome sequencing ranges from 40-70%[14].

Additionally, NGS based screening of IRD cohort have identified expanding phenotype spectrum for known genes, refined clinical diagnosis based on genetic tests results[4,15], thus improving our understating of disease pathology, disease progression and clinical course.

IRD – Data from India

The prevalence of each type of IRD in the Indian population is not known, however, it is expected to be higher than the world-wide numbers because of consanguinity. A prevalence of about 1 in 750 in rural central India (≥30 years)[16], 1 in 930 in urban south India and 1 in 372 in rural south India has been estimated for RP (≥40 years)[17].

Genetic studies using candidate gene screening, homozygosity mapping, targeted panels, whole exome sequencing have been published on RP, LCA, CSNB, STGD, Oguchi disease, choroideremia, retinoschisis. These studies have described the genotype-phenotype correlation as well.

Over the past four and half years (May 2015 – Nov 2019) we have screened 343 non-syndromic IRD cases. Ninety-two (27%) of these cases were reported with either a pathogenic or likely pathogenic variant(s), 94 (27%) cases as variant of uncertain significance (VUS) and the rest 157/343 (46%) as none. The mutation spectrum included 76 different genes with *CERKL* (8%), *PROM1* (8%), *ABCA4* (7%), *RHO* and *RPGR* (3% each), *EYS*, *PDE6B*, *RDH12*, *GUCT2D*, *OPA1* (2% each) and the rest at 1% frequency (unpublished data).

Though, there are considerable number of reports on genetics of different IRD types from Indian cohorts using various screening methodologies including NGS as well as clinical diagnostic cohorts (unpublished data), data from a large cohort with various phenotype spectrum of IRD is not yet available.

Treatment for IRD

IRD being a neurodegenerative disease was untreatable until 10 years ago, with only few management and rehabilitation options available. However, initiation of phase1/2 gene therapy trials using recombinant adeno associated virus (rAAV) in patients with biallelic *RPE65* mutation in 2008 to test the safety and efficacy[18,19] has now resulted in LUXTURNA, the first FDA approved prescription gene therapy drug for inherited retinal disease due to *RPE65* gene[20]. Phase I/II or II/III clinical trials of gene therapy for few other genes, *CEP290*, *MERTK*, *PDE6B*, *RPGR*, *CHM*, *MYO7A*, *ABCA4*, *CNGB3*, *CNGA3*, and exon 13 of *USH2A* are under way (https://clinicaltrials.gov/)[2].

The clinical trials have shown improvement in the vision and/or slowing of the degeneration providing some useful vision in these subjects. Research on various other therapeutic options and gene delivery methods offer hope of treatment to those suffering from IRD.

Benefits/challenges of genetic testing

The benefits of genetic testing;

(i) offers to identify the genetic cause

(ii) pattern of inheritance, thus risk prediction for sibs or future generation

(iii) prognosis based on available genotype-phenotype data

(iv) appropriate management or preventive measures of other symptoms wherever applicable/possible in case of syndromic disease

(v) possible candidature for gene therapy or other therapeutics that are being developed. All these are answered when the causative gene/variant is identified. From the patient's and their families' perspective too it is beneficial, however with an average diagnostic yield of 70%, the rest of the patients and their families where the test result is negative, the diagnostic odyssey is still unanswered[21].

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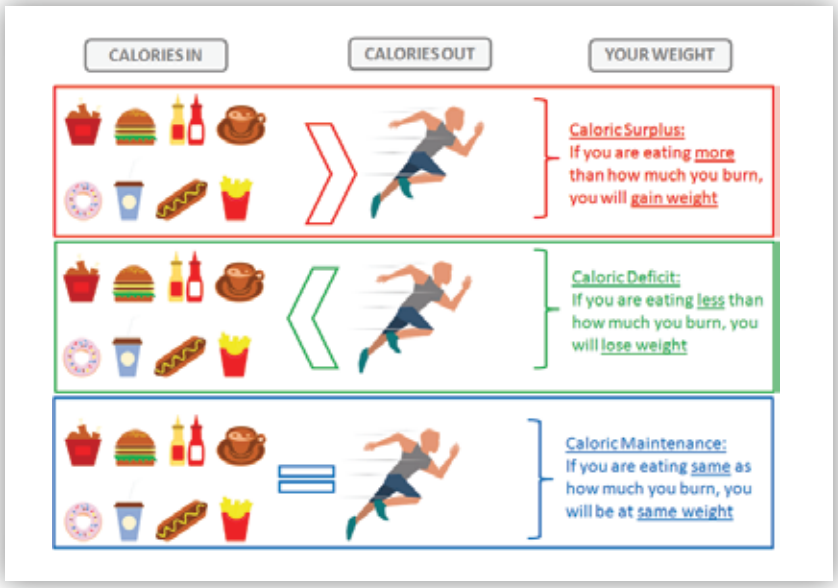
Sneak Peek into the World of Personal Genomics

Maths of **nutrition, health & weight management**

By: Vidyadhar Karmakar, PhD
Product Head, ApnaGenome

Want to lose weight? Naah, just want to maintain weight! Or you are one of the few who want to gain a few pounds? Well, it doesn't matter what your goal is, the maths of nutrition, health and weight management doesn't change!

Probably, by now you may have tried several diets and spent thousands of bucks on health check-ups. Your pocket is a bit lighter on cash, your body is still heavy or may gotten heavier! If you are struggling with your health, here I will share with you some simple maths behind nutrition, health and weight management (Figure 1). This is an open secret for anyone in fitness but less known for those who haven't read much about this topic. Hopefully, you will find this article useful. Please feel free to inbox me if you have specific questions.



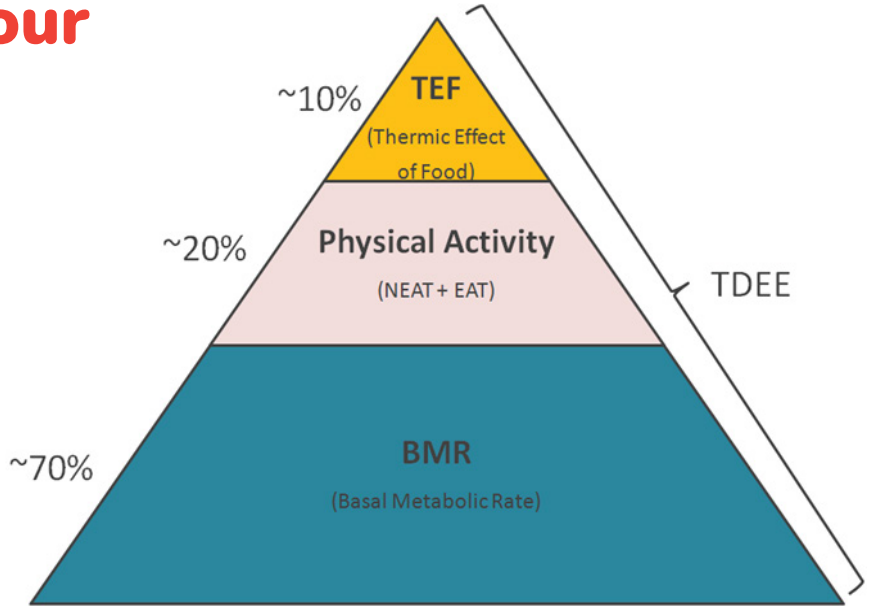
(Figure 1: Maths of Nutrition, Health and Weight Management)

So now you see how easy the math of weight management is! In financial analogy: if you earn more than how much you spend, you will be rich (i.e. money surplus); if you spend more than how much you earn, you will be poor (i.e. money deficit) and if your expenses are same as your earning, you will be financially same (i.e. maintenance income).

How do we burn our calories?

We burn our calories in 3 ways (Figure 2). The total of all calories burnt is called our Total Daily Energy Expenditure (TDEE). Based on the lifestyle, TDEE varies. Simply, TDEE increases with physical activity. Following is the math of TDEE and physical activity levels:

TDEE Very Active > TDEE Moderately Active > TDEE Lightly Active > TDEE Sedentary



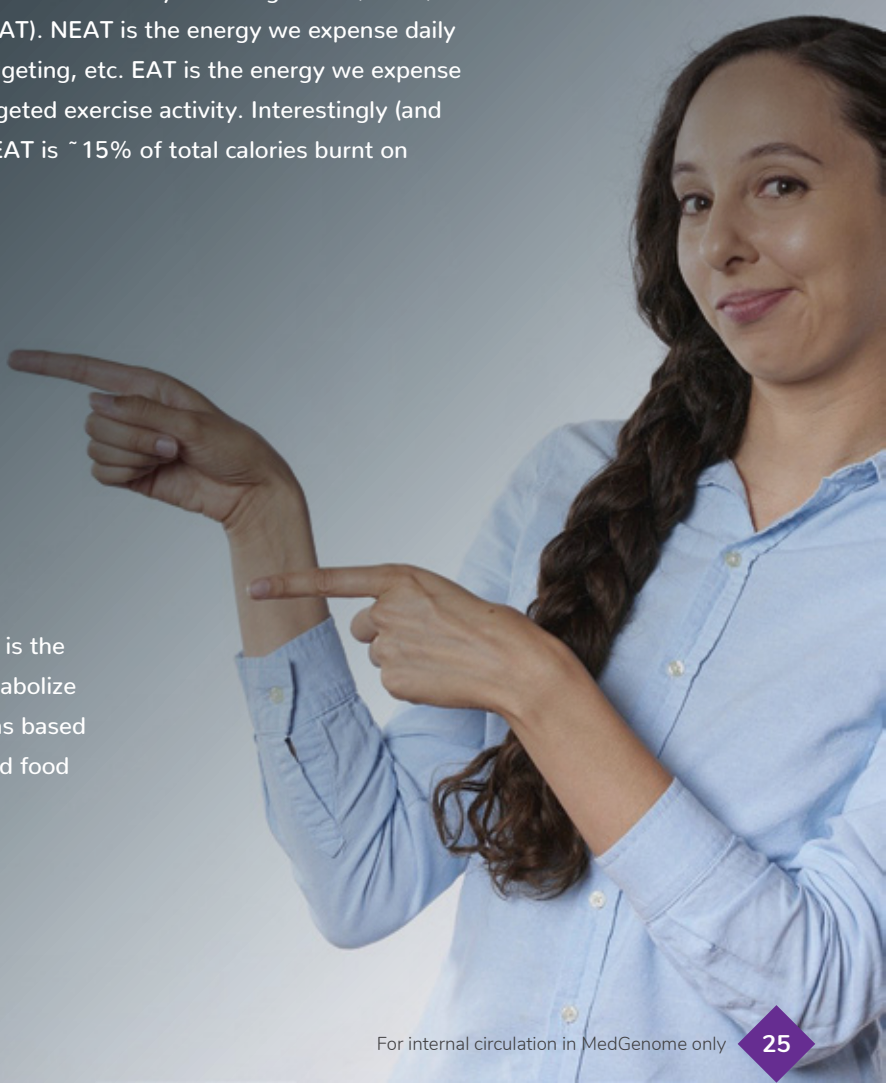
(Figure 2: Total Daily Energy Expenditure, TDEE)

The various components of TDEE are:

1. ***Physical Activity:*** This includes Non-exercise Activity Thermogenesis (NEAT) and Exercise Activity Thermogenesis (EAT). NEAT is the energy we expense daily while walking to office, taking a bus, fidgeting, etc. EAT is the energy we expense while in the gym or doing any other targeted exercise activity. Interestingly (and counter-intuitively), EAT is ~5% and NEAT is ~15% of total calories burnt on physical activity.

2. ***Basal Metabolic Rate (BMR):*** This constitutes ~70% of our energy expenditure. The various factors that affect BMR are sleep, stress, fitness, inflammation and disease.

3. ***Thermic Effect of Food (TEF):*** This is the total energy burned by the body to metabolize the food we eat. TEF can vary in humans based on fitness level, body fat percentage and food type/composition.



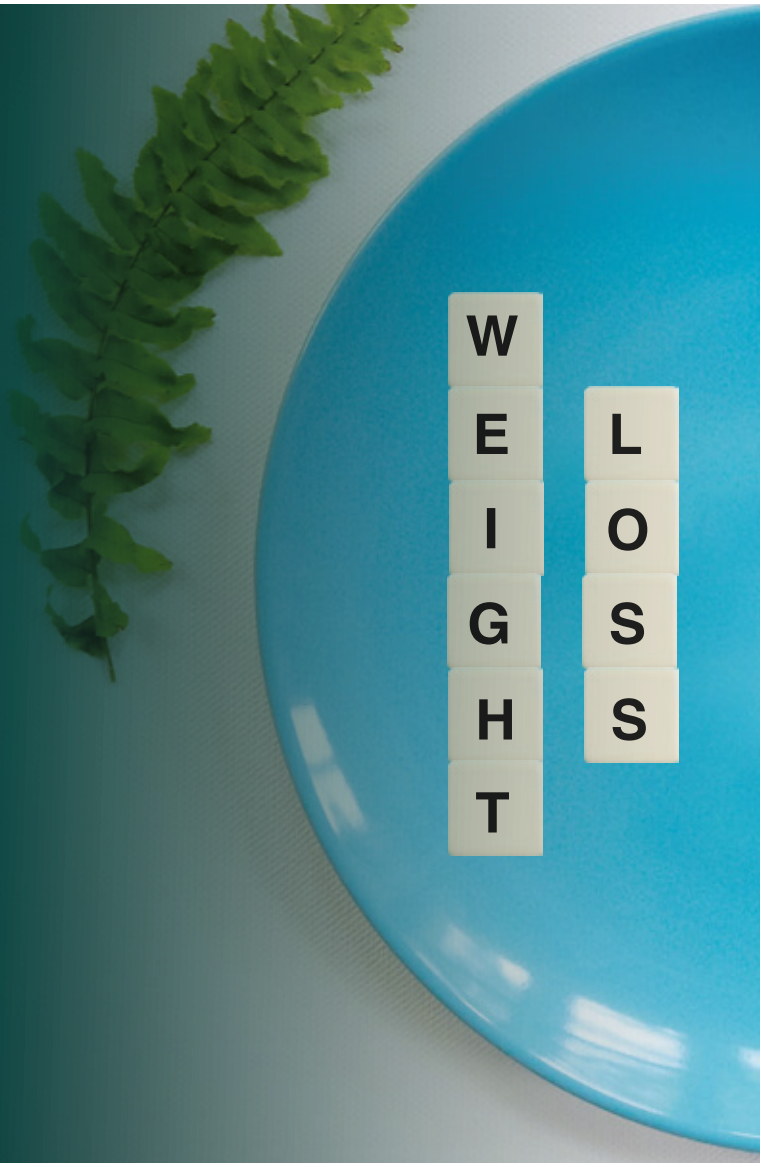
How can you lose weight?

If you have not been on a diet before, a "forward dieting approach" i.e. gradual reduction in calories along with increase in your physical activity levels should help you lose weight. However, if you have been on a diet and have reached a plateau, then different dieting strategy may be required (i.e. reverse dieting approach). Following is a practical approach to lose weight for those who have not been on any kind of diet before. This is also called "forward dieting" strategy:

"Calorie In" Strategy:

Quantified nutrition i.e. measuring weight of what you eat is ideal. But if you do not have a kitchen scale at home then you can try the following qualitative approach:

- Always use the same plate to eat for your every meal (breakfast, lunch, snack, dinner). This will ensure portion variation because of change in plate size
- Serve yourself the usual portion that you usually consume for each meal. Take a picture of the served plate
- Every week, reduce the portion size of each meal by 20% and bring down the food consumption to a level such that you are feeling satiated and not hungry the whole day. A little bit of hunger is expected when in caloric deficit.
- Stick to the 4 meals — breakfast, lunch, snack and dinner. Avoid unnecessary munching of snacks and sugary drinks between the meals
- If you follow this strategy along with the "calorie out" strategy below, you should possibly see weight loss



"Calorie Out" Strategy:

- ***Increase NEAT:*** If you have to attend any phone calls, walk while you take the calls. Take steps not the elevator. If you take bus/cab, get down a kilometer before your stop and walk rest of the distance. Likewise, you can be creative and think how to increase daily non-exercise physical activity levels.
- ***Increase EAT:*** Join gym, run, swim, zumba or do any activity that makes your heart beat faster and are out of breath to an extent that you find it a bit difficult to hold a conversation. If you want best results, then take up to resistance/weight training. And to women out there — no, you won't be muscular with weight training!

Measuring your performance:

- Monitor your weight every week. Measure your weight same time of the day. It is best to weigh in the morning after you have cleared your bowels and have not had breakfast.

- Measure your body fat percentage every month. Along with weight, it is important to deplete your body fat percentage too. This will be especially important if you are doing resistance training as you will see changes in muscle and fat mass. Hence, it could happen that one is seeing inch loss, yet the weight is constant.

Now you see, the math of nutrition, health and weight management is so simple! While a good fitness coach would always be a value for money, I hope this article helps you lose weight and not money on fad diets/meal services and expensive diet foods!

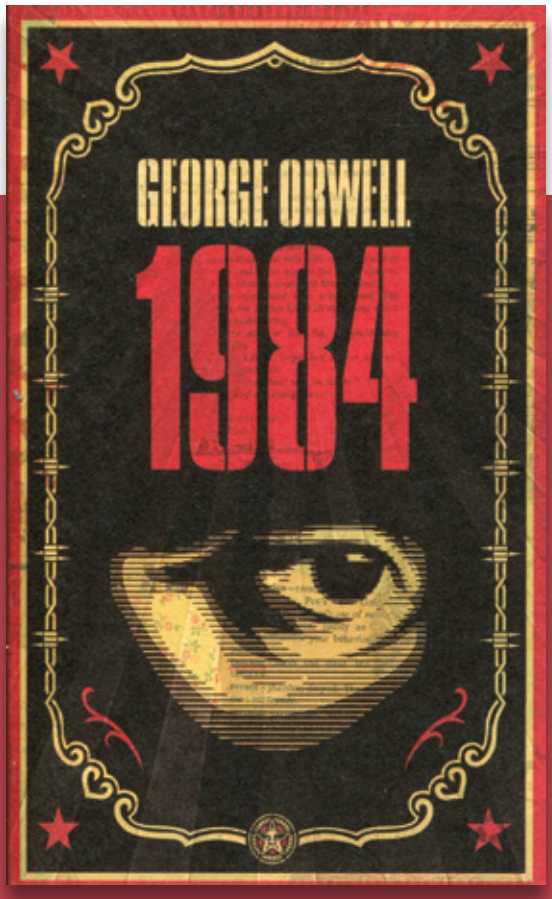
From our Colleagues

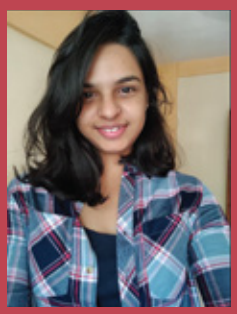
Book Review

Book

1984

by George Orwell





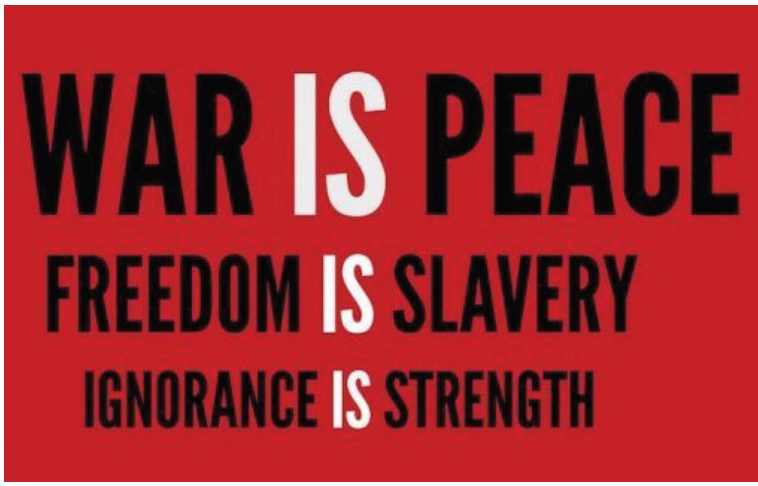
Book review by

Shweta Mahalingam,

Genetic Counsellor, Ops Dept., MG India

As Italo Calvino once said, "A classic is a book that has never finished saying what it has to say", some books can withstand the true test of time. Consuming content without any deliberation or consequences we are witnessing an era of infinite knowledge but there is little boundary to how much we are unveiling. We have not only shared our physical appearances to the world but also, what our minds are capable of – our ideas, opinions, feelings, prejudices, and thoughts. But imagine if we lived in a society which consumed each of it and basically averted your mind to form the next thought you believed was yours?

George Orwell in his last novel 1984 describes a dystopian society with the actualization of twenty-four-hours-around-the-clock surveillance by the government of Oceania - Ingsoc by every way possible: telescreens, recorders, spies, and thought polices observing every minute of life. Winston Smith, the protagonist reveals the story page by page to the readers. The society he lived in was constantly at war and there is one dominating party that controls every aspect of everybody's life. He witnesses the evolution of the domination and tries to comprehend the abolishment of freedom, human mind, and spirit as he explored in his journey the true understanding of the ruling party's agendas: war is peace, freedom is slavery and ignorance is strength.



While working as one of the lower members for the government, his job was to change the past in the print media as instructed by the Party and thereby abandon his own memory and conscience to live with the party slogan of "who controlled the past controls the future: who controls the present controls the past". He realizes his constriction every day with events such as the idea of developing a language by the party that narrows the use of words - to narrow the range of thoughts. Often tormented by the thirst to find the real truth, to find real freedom, he starts to bend the rules and then break it. He craved to find the reality and starts his THOUGHTCRIME with knowing the consequences of committing it - "thoughtcrime does not entail death: thoughtcrime IS death." echoing in his mind as he starts to pen down his thoughts.

He believed his life changed when he met Julia. Evoking from the long slumber of loneliness, they both pursue their love story, which they both knew was a ticking bomb. Love and true relationships were restricted to oppress the minds of humans to sketch a better goal in life than to rebel with a cause. One thing led to another, like a lion that tasted blood for the first time, the newfound peace and freedom explored by Winston led to his meeting with O'Brien an Inner Party Member who he believed to be his salvation. There are some itches better left unscratched, but Winston had broken that rule long ago.

O'Brien convinces Winston about Brotherhood – a way to rebel and find true freedom. As Winston explores the path, he finds THE BOOK which standardizes his thoughts and creates a better understanding of HOW the party works and yet he remains to struggle with the WHY. Each chapter explaining the slogan of the party and what these powerful institutions plan in the bigger picture. How they intend to create the epitome of totalitarianism to remain in power and promote the concept of anthropocentrism and geocentrism. How with war they intend to create a regimen that promotes stability in the current hierarchy and not let more heresies become martyrs or face of history.

As Winston finds himself closer to the truth and escapism from the false reality of limit of the human mind, he also realizes Julia is apathetic to his political views. He understands how people can be trained their entire life to believe in the smallness of their own lives and remain happy. The realization although comes with the price of him being caught by the THOUGHT POLICE and suffer the consequence that breaks him down as a human. With every bone in his bone being rearranged and weeks and months of torture only to stand in front of his mirror image that he no longer recognizes. He arguably fights his own morals and feels powerless to question the insanity of the wrong.

> **What can you do, thought Winston, against the lunatic who is more intelligent than yourself, who gives a fair hearing and then simply persists in his lunacy?**

At the end Winston finds his own heart and mind broken, only to accept **"TWO PLUS TWO IS FIVE OR THREE"** and idealizes the party slogans and beliefs and quiets his mind with the skepticism that poses as merely an illusion which he once considered the truth.

This book is almost eight decades old and yet it does not oversell the idea of human freedom. Orwell wrote this book in 1949, just after the second world war as we had already witnessed the bombing, the concentration camps, the sufferings, the loss, the rise of dictators and any book of that era will tell you, that in war, there is little left to differentiate between winning and losing. The hunger for power as it rises, only leaves the society into weaker sections which may or may not choose to revolt, thus the book considered as a piece of fiction when it stands true after decades become an oracle. But how far is that future? May be next time we see an advertisement pop in our screen based on the phone call conversation we had twenty minutes ago – we might set a timeline!

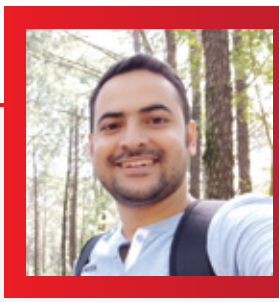
From our Colleagues

Art meets science

"Art and science have their meeting point in method." — Earl Edward George Bulwer-Lytton

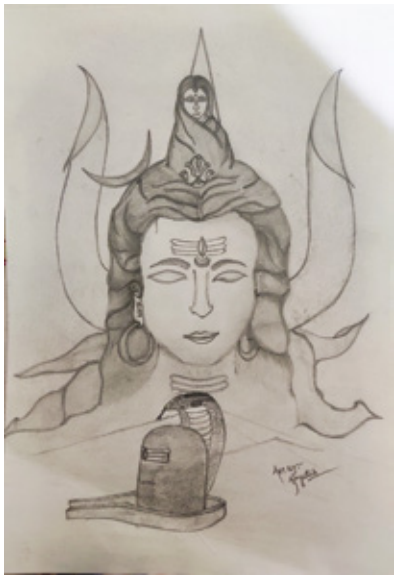
By: Sharon

Research Associate Trainee, Ops Dept
MG India



By: Ajay Katoch

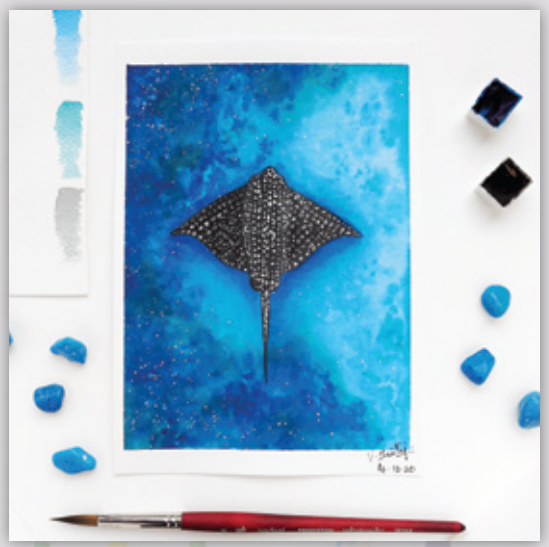
Bioinformatics Analyst,
Ops Dept, MG India



From our Colleagues

Our employee's little picasso :)

By: Shruthi V (13 Years)
DNA of Lakshmi V, Administration, MG India



Our employee's little picasso :)

By: Kirthana Warrier (11 years)
DNA of Hiranjith G H, VP & Head of Research Services, MG USA

By: Shaurya (9 Years)
DNA of Dr Sanghamitra Mishra, Senior Scientist, Ops Dept, MG India

From our Colleagues

Frozen moments - Photography

A Leap of Faith

Beetles at work

Alley of the Ants

The Lone Ranger

By: Faizal Eeman,
Bioinformatics Analyst, Ops Dept, MG India

Employee Connect

WELCOME

Our New-Joiners


Akshaya Bhat


Amit Kumar Sharma


Anjan Asaram Devkar


Ayushi Tiwari


Balasubramani


Bhuvana M


Chidananda


Dhriti C Nathan


Dinesh Kumar K


Dinesh Kumar


Divyashree V


George Thomas

Employee Connect


Himanshu Saini


Himashri Deka


Kunal Kishore Kukreja


Lokesh Singh


Menila Daniel


Monika Dass


Muthtamil Sambath


N Pramod


Parth Varangaonkar


R Padmaja


Rohit Dhaware


Roopa C


Sabariswaran R


Sakshi Sharma


Vishnu S Nair


Salaiha Rahman


Sangeeta Tripathy


Satyendra Gupta

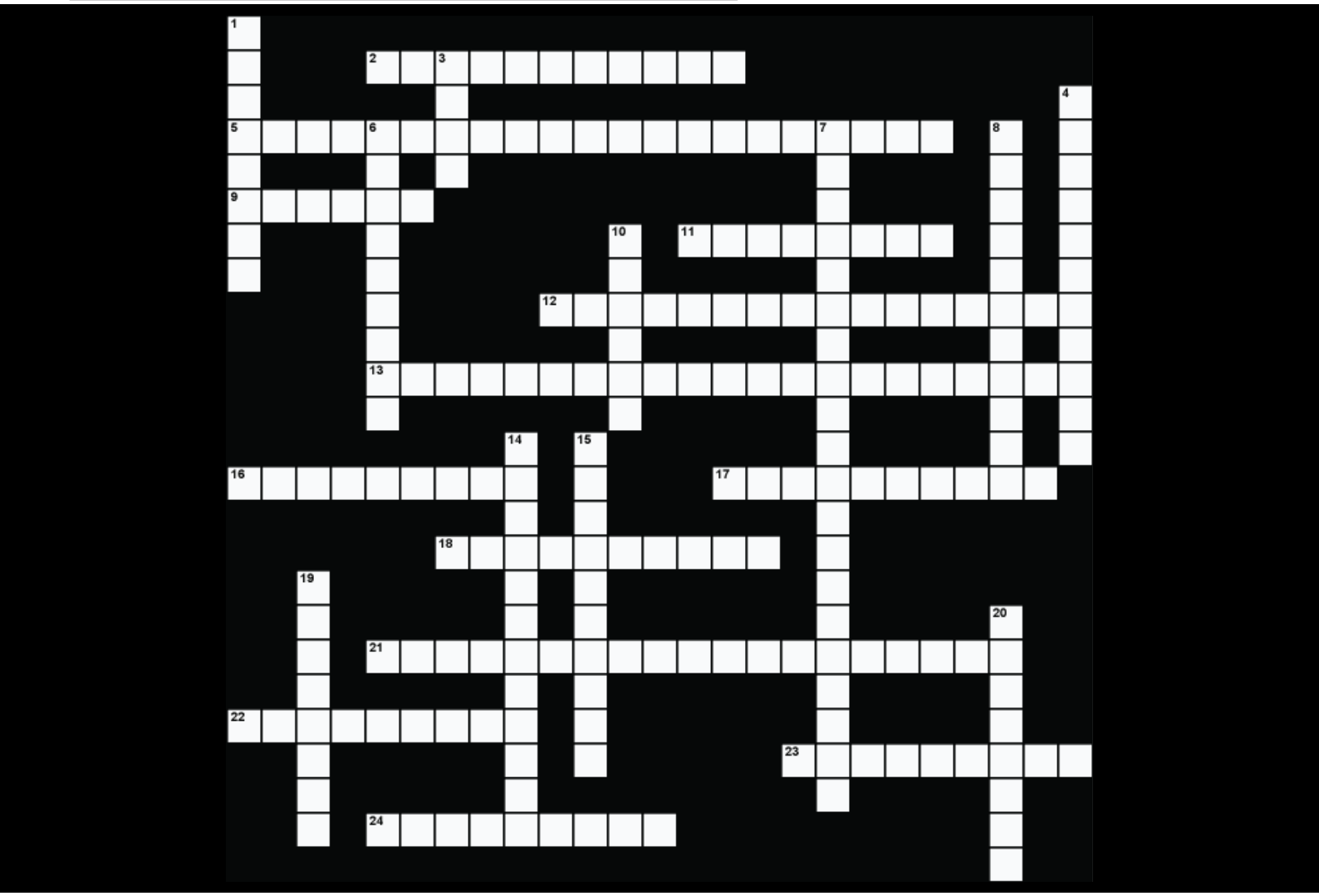

Sheikh Mohamed


Srinivas D J


Suruchi Aggarwal


Syed Tanveer A

MedGenome Connect



Across

2. Mendel's principle of genetic inheritance stating that, for any particular trait, the pair of genes of each parent separate and only one gene from each parent passes on to an offspring.
5. Mendel's principle of genetic inheritance stating that different pairs of genes are passed to offspring independently so that new combinations of genes, present in neither parent, are possible.
9. An alternate form of the same gene.
11. The genetic makeup of an individual for a trait or for all of his/her inherited traits—not the observable or detectable characteristics.
12. An inheritance pattern in which a gene will have a different effect depending on the gender of the parent from whom it is inherited.
13. Genes whose effect does not normally occur unless certain environmental factors are present.
16. The observable or detectable characteristics of an individual organism; the detectable expression of a genotype.
17. Genes that are inherited by both men and women but are normally only expressed in the phenotype of one of them.
18. The inheritance pattern in which a single allele is responsible for a variety of traits.
21. The term for a genotype in which there are two recessive alleles.
22. Genes that can alter how certain other genes are expressed in the phenotype.
23. Genes that can either initiate or block the expression of other genes.
24. The general term for an allele that is masked in the phenotype by the presence of another allele.

Down

1. The general term for an allele that masks the presence of another allele in the phenotype.
3. A unit of inheritance usually occurring at a specific location on a chromosome.
4. Twins that come from the same fertilized egg
6. A trait that is determined by the combined effect of more than one gene.
7. An inheritance pattern in which a gene has more than two alleles.
8. The inheritance pattern in which two different alleles for a trait are expressed unblended in the phenotype of heterozygous individuals.
10. He acquired his understanding of genetics mostly through pea plant breeding experiments.
14. A genotype consisting of two different alleles of a gene for a particular trait.
15. A genotype consisting of two identical alleles of a gene for a particular trait.
19. A theory that inherited traits blend from generation to generation. Most of the leading scientists in the 19th century accepted it. However, Gregor Mendel proved that it was not correct.
20. The study of gene structure and action and the patterns of inheritance of traits from parent to offspring.

Previous Winners

Fazal Mohammed

Praveena L Samson

Kindly mail your answers by 31st Jan 2021 to editor@medgenome.com. The first two people to answer the puzzles correct will be featured in the next edition of our newsletter.

Solve the Puzzle & WIN Voucher

Employee Connect

EDIFY- Healthshots

MedGenome's wellness brand for the employees.

A glimpse of banners shared on good habits and healthy practices!

Photo Feature

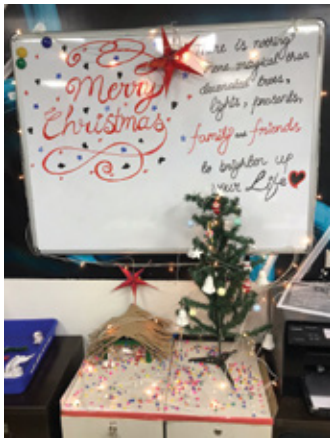
Christmas celebration

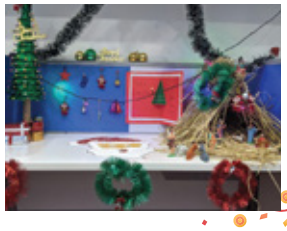
Greeting card design competition



Home office decoration

Office decoration



Tambola game



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