MEDGENOME

GeKNOWme
Internal Quarterly Newsletter
Vol 31 | Oct 2024

Pharmacogenomics

A clinical tool for personalized medicine

Featured article

Precision medicine: transforming healthcare in India amidst significant challenges

Book review

Women in science: 50 fearless pioneers who changed the world

WORDS FROM THE **FRONTLINE**

Dr. Arvind Murali Venkatesan, PhD
Associate Director- Strategic Initiatives & Partnerships

From Lab Rats to Corporate Cats: My Journey to MedGenome

My mantra of life...

**"Anyone who stops learning is old, whether at twenty or eighty.
Anyone who keeps learning stays young."**

This Henry Ford quote was a favorite of my dad, who often shared it with me from a young age. Over time, it became etched in my mind and has since evolved into the mantra of my life.

My journey to MedGenome...

Over the past 35 years, my journey has felt like that of a passenger on a hop-on, hop-off bus-spanning 9 schools, 3 companies, 13 cities, and 2 countries. From attending government schools in rural Maharashtra to earning a PhD in Life Sciences from the University of Massachusetts and an MBA from the Indian School of Business, I've had the privilege of experiencing diverse cultures and gaining multifaceted learnings.

My professional journey, though relatively short, has been equally eventful. I began in the R&D function of the pharma sector at Syngene, transitioned to the strategy function in a management consulting firm at L.E.K., and now find myself in a commercial role at MedGenome, a leading diagnostics company.

It has been close to three years at MedGenome, and the learning has been unparalleled. MedGenome has been a remarkable platform for growth and development.

- Working with the leadership on the strategic roadmap for the organization gave me a 30,000-foot view of the market and its opportunities.
- Assessing the in-licensing of new technologies to bring to the eastern hemisphere and commercializing MedGenome's intellectual property to create value in the western hemisphere allowed me to stay connected with science and technology.
- Managing revenues across domestic and international markets taught me the nuances of sales and sales processes.

Few roles offer such rich cross-functional exposure, and I am forever indebted to Ram and Mahesh for trusting me with this opportunity.

My few cents on MedGenome and the path forward...

Every organization stands on three pillars: **People, Technology, and Processes.** While all three are critical, each plays a dominant role in different phases of growth. Here's how I see it at MedGenome:

MedGenome's 0 to 1 journey

This phase was driven by an exceptional set of founding members, which includes not just the founders but everyone who ensured the company's survival and pivoted it in the right direction.

MedGenome's 1 to 10 journey

This phase was powered by the development of proprietary technology, such as our LIMS and analytics software, which became the backbone of the organization.

MedGenome's 10 to 100 journey

As we embark on this next phase of growth, scaling and sustaining success require robust, foolproof processes. It is now imperative that we consciously work toward transforming from a people-dependent unit to a process-oriented organization.

One piece of wisdom my dad often shared comes to mind: **"Act as if you're in your manager's shoes, but think as if you're in your CEO's shoes."** I encourage each reader to reflect on this advice. Applying this will not only benefit MedGenome, but it will also accelerate the professional growth of the individual.

Here's to building a future that's innovative, inclusive, and impactful.

Contents

MedGenome Connect

REPRODUCTIVE GENOMICS

This quarter, we launched several initiatives to boost awareness and engagement. We created explainer videos on KaryoSeq, Karyotyping Reflex KaryoSeq, and KaryoSeq HD, featuring Dr. Priya Kadam. Additionally, we featured KOL testimonials on NIPT and other reproductive genetic tests, emphasizing how MG's services are transforming clinical practice. We had done a KaryoSeq HD video and made carousel posts on advanced NIPT, Karyotyping Reflex KaryoSeq, CNVsure, and more. KOLs also launch new tests like Karyotyping Reflex KaryoSeq and KaryoSeq HD at major events. Our efforts were complemented by informative blog posts on topics like NIPT for twins and the KaryoSeq vs. Karyotyping comparison, along with online polls on LPWGS and KaryoSeq HD. Digital mailers were also sent to clinicians, highlighting offerings. These initiatives aim to strengthen awareness and reinforce MG's leadership in reproductive genetics.

Dr. Priya Kadam, Karyotyping Reflex KaryoSeq - Explainer Video

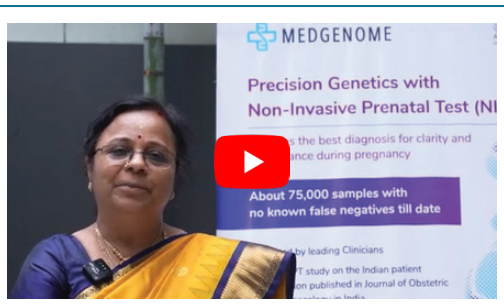
Dr. Priya Kadam, Director of Reproductive Genomics at MedGenome Labs talked about the clinical benefits of KaryoTyping Reflex KaryoSeq test, an NGS-based whole genome sequencing test combining the benefits of KaryoSeq with reflex analysis for a comprehensive understanding of chromosomal abnormalities.

#MedGenome #Genetics #WholeGenomeSequencing

Dr. Priya Kadam, Director of Reproductive Genomics at MedGenome Labs reveals the clinical utility and benefits of KaryoSeq (Low Pass Whole Genome Sequencing). This genetic test helps identify aneuploidies and copy number variations, and can be used as an alternative to chromosomal microarray. Watch the video to learn more.

#MedGenome #Genetics #KaryoSeq #LowPassWhole GenomeSequencing

Dr. Priya Kadam, KaryoSeq - Explainer Video



Dr. Chitra Ganesh, HOD - Fetal Medicine and Perinatology at Kauvery Hospital, Bangalore - Video

Dr. Chitra Ganesh, HOD - Fetal Medicine and Perinatology at Kauvery Hospital, Bangalore, highlighted MedGenome Labs' unwavering support and dedication to improving patient health outcomes.

#MedGenome #Genetics #10YearsofPioneeringGenomics

Dr. Anu Joseph, Senior Consultant - Obstetrics & Gynaecology & Fetal Medicine at Kauvery Hospital, Bangalore, shared her insights and experiences collaborating with MedGenome.

#MedGenome #Genetics

Dr. Anu Joseph, Senior Consultant - Obstetrics & Gynaecology & Fetal Medicine at Kauvery Hospital, Bangalore - Video

Dr. Ashok Khurana, Mentor Emeritus of the Society of Fetal Medicine, Chairman and Consultant in Reproductive Ultrasound at the Ultrasound Lab, New Delhi - Video

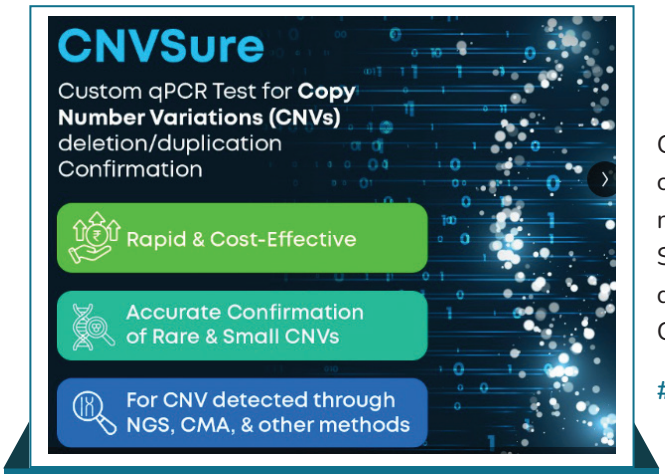
We've launched Karyotyping Reflex Karyoseq, a genetic test that combines the benefits of KaryoSeq with reflex analysis for a deeper and more accurate understanding of chromosomal abnormalities. Watch the unveiling ceremony by **Dr. Ashok Khurana,** Mentor Emeritus of the Society of Fetal Medicine, Chairman and Consultant in Reproductive Ultrasound at The Ultrasound Lab, New Delhi.

#MedGenome #Genetics #ChromosomalAbnormalities

Dr. Meenu Vaish, President of UKSOG, Gynecologist and Obstetrician at Vaish Nursing Home, Dehradun - Video

Karyotyping Reflex KaryoSeq, our latest genetic test merges the benefits of KaryoSeq with reflex analysis for a comprehensive understanding of chromosomal abnormalities. Watch the unveiling ceremony by **Dr. Meenu Vaish,** President of UKSOG, Gynecologist and Obstetrician at Vaish Nursing Home, Dehradun.

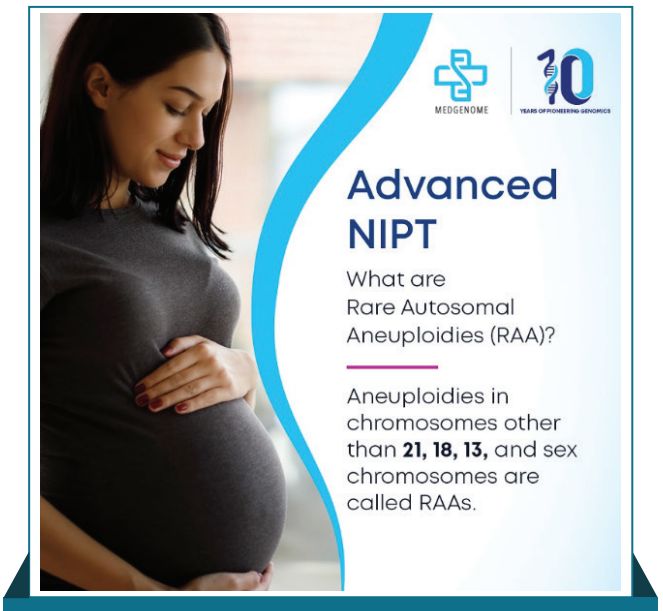
#MedGenome #Genetics #ChromosomalAbnormalities



Social Media Poster

CNVSure: Reliable solution for CNV confirmation accurately confirms rare and small CNVs with our custom qPCR test. Faster, more flexible and cost-effective than traditional methods, CNV-Sure ensures precise results for even single exon deletions or duplications. Ideal for confirmation of CNVs detected by NGS, CMA, and prenatal testing.

#Genetics #Diagnostics #CNVs #Genomics



Social Media Poster

Advanced NIPT offers crucial insights into Rare Autosomal Aneuploidies.

Detecting RAAs early is essential, as they can affect pregnancy outcomes, ranging from normal birth in 50% of cases to outcomes such as fetal growth restriction, congenital anomalies, or fetal demise. This advanced test allows for early, non-invasive detection, leading to timely interventions like genetic counseling, ultrasound evaluation, and confirmatory testing.

With validated data enhancing our test's accuracy, MedGenome supports expectant parents with compassionate, and comprehensive prenatal screening.

#MedGenome #Genetics #NIPT #RareAutosomalAneuploidies

Social Media Poster

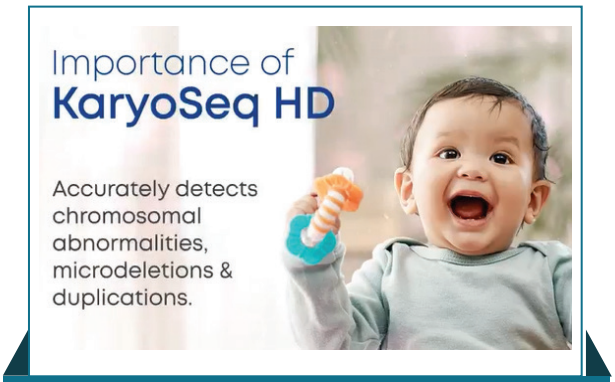
When the culture for Karyotyping fails to grow, Karyotyping Reflex KaryoSeq is employed as a more precise alternative. This low-pass Whole Genome Sequencing test combines the benefits of KaryoSeq with Reflex Analysis, to deliver comprehensive chromosomal insights while eliminating the need for resampling.

#MedGenome #Genetics #KaryotypingReflexKaryoSeq

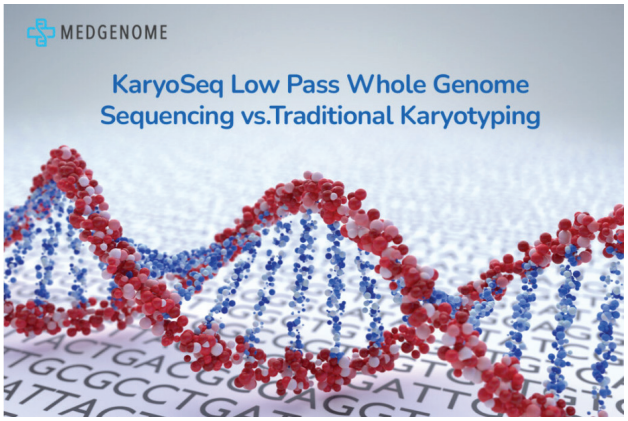
Elevate Your Genetic Insights with KaryoSeq HD!

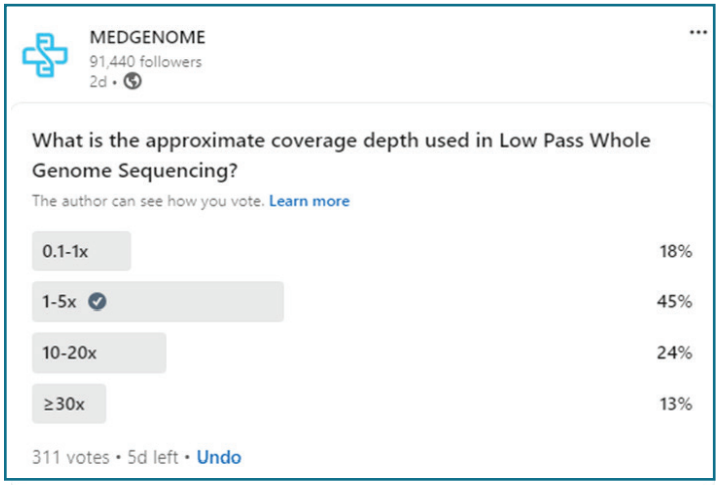
Experience high-definition low-pass whole genome sequencing that detects aneuploidies and copy number variations ≥ 50 kilobases. The test offers enhanced diagnostic yield and comprehensive genome-wide insights for both prenatal and postnatal testing.

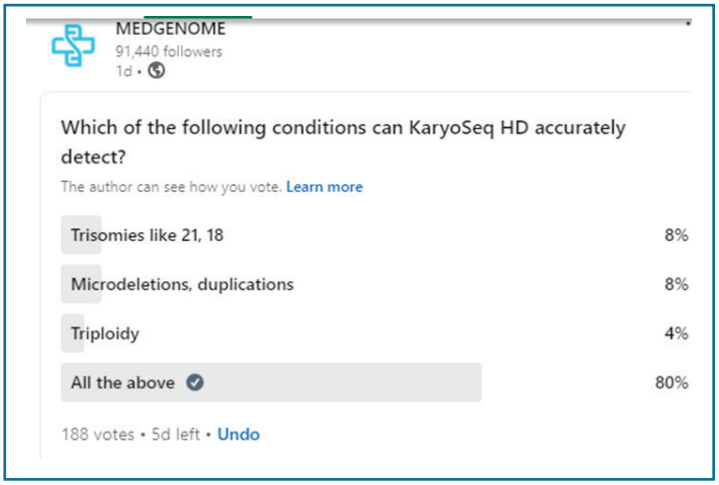
#MedGenome #Genetics #KaryoSeqHD



Social Media Poster







RARE INHERITED DISEASE GENOMICS

Apart from observing and commemorating several important health days, we dedicated significant efforts to raising awareness about various inherited diseases. These initiatives were designed to educate both the general public and healthcare professionals on the importance of early detection and prevention, particularly in relation to genetic disorders. Through multiple channels, we shared valuable information to help people better understand the risks, symptoms, and available treatment options for these conditions.

A key focus of our outreach efforts was newborn screening tests, for which we emphasized the numerous benefits through a series of videos, blog posts, and other awareness materials. These resources were created to highlight the critical role of early screening in identifying potential health issues in newborns before they become more severe. By informing parents and healthcare providers about the importance of screening, we aimed to ensure that babies receive the necessary care and attention right from the start, improving their overall health outcomes.

Additionally, we took an important step in improving healthcare access for underserved populations by launching a subsidized Whole Exome Sequencing (WES) test in collaboration with Twist Bioscience. This initiative was specifically designed to support underprivileged patients by making cutting-edge genomic testing more accessible.

World Sight Day - Video

This #WorldSightDay, discover how MedGenome is transforming eye health through advanced genetic testing. With over 675 genes covered, the Comprehensive Ophthalmic Genetic Disorder Panel provides precise diagnoses for inherited eye diseases, empowering tailored treatment options.

Let's pave the way for a future where every life is enriched.

#MedGenome #Genetics

At MedGenome, we know every parent's first promise is to protect their child. Our BabySecure Newborn Screening Test detects potential health concerns early, allowing for timely treatment and peace of mind for you and your little one. Detect early, protect early - because every baby deserves the best start in life.

#BabySecure #BabySecure #NewbornScreeningTest

BabySecure - Video

BabySecure - Social Media Poster

Introducing MedGenome's BabySecure Lysosomal Storage Disorders (LSD) Panel! This test detects a diverse group of Lysosomal Storage Disorders (LSDs) through enzyme activity assays using Liquid Chromatography–Mass Spectrometry (LC-MS). By identifying genetic mutations early, we help ensure your baby receives the right support and care from the very beginning. Give your little one the best start with BabySecure!

#MedGenome #Genetics #BabySecure #NewbornScreeningTest

BabySecure, an advanced newborn screening test helps in screening over 100 inherited metabolic disorders. The test helps in screening disorders like:

1. Fatty Acid Oxidation Disorders
2. Amino Acidopathies
3. Organic Acid Disorders
4. Severe Combined Immunodeficiency (SCID)
5. X-linked Adrenoleukodystrophy (X-ALD)
6. Congenital Hypothyroidism (CH)
7. Congenital Adrenal Hyperplasia (CAH)

Newborn Screening Test - Social Media Poster

Hypercholesterolemia - Social Media Poster

Hypercholesterolemia is high amounts of cholesterol in the blood, and it often runs in families due to genetics. To explore genetic testing and expert counselling to understand your risks, test details and results.

#MedGenome #Genetics #Hypercholesterolemia

Empowering lives through genomics.

MedGenome and Twist Bioscience have joined hands to bring affordable Whole Exome Sequencing Test to underprivileged patients in India through the HOPE for Rare Diseases Program. This initiative aims to ease the diagnostic journey of rare disease patients, offering them access to advanced genetic testing and the chance for better care and treatment options.

#MedGenome #TwistBioscience #HOPEForRareDiseases #WholeExomeSequencing #Genomics #RareDisease

MedGenome & Twist Bioscience - Social Media Poster

CANCER GENOMICS

In the past quarter, a wide array of activities was carried out, each focusing on various types of cancers. These initiatives are designed to engage both clinicians and patients, with the goal of enhancing awareness, knowledge, and collaboration. As part of these efforts, we developed a detailed case study that examines the impact of our Hereditary Cancer Panel, Lung Cancer Panel, and Liquid Biopsy tests. The case study highlights how these advanced diagnostic tools have significantly influenced treatment decisions, leading to more tailored and effective care for patients across different cancer types.

In addition to the case study, we organized several key events to foster deeper engagement within the medical community. We invited prominent clinicians to share their insights and experiences with these diagnostic tests, providing a platform for knowledge exchange. We also hosted a series of informative webinars, allowing clinicians, patients, and other stakeholders to learn about the latest advancements in cancer diagnostics and treatment.

Moreover, in partnership with GenX Diagnostics, we organized a Continuing Medical Education (CME) event in Bhubaneswar. This event was designed to further educate healthcare professionals on the clinical applications of our cancer panels and the role of genetics in oncology.

Dr. Shona Nag, Director of Oncology at Sahyadri Super Speciality Hospital - Video

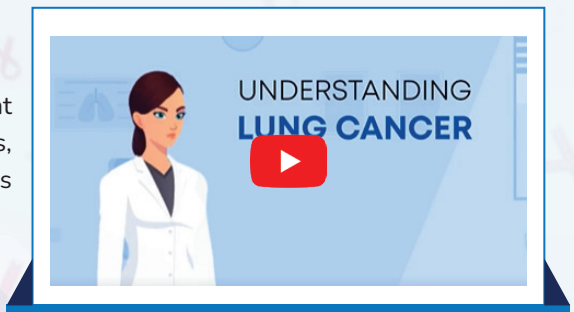
Dr. Shona Nag, Director of Oncology at Sahyadri Super Speciality Hospital, Pune, sheds light on the significance of genetic testing in breast cancer this Breast Cancer Awareness Month.

While all cancers are genetic, only a small percentage are hereditary. About 6-7% of breast cancers are inherited, often linked to specific gene mutations. Genetic testing can be vital for patients under 50, those with breast cancer, or those with a strong family history.

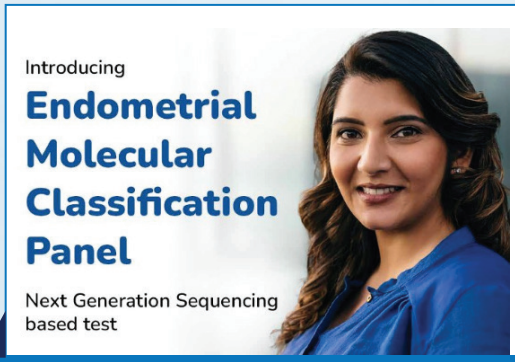
#BreastCancerAwareness #GeneticTesting #EarlyDetection

Lung cancer remains one of the most common cancers, often diagnosed at advanced stages. Watch our video to learn about lung cancer, its causes, symptoms, Metastases and how MedGenome NGS-based genetic tests provides precise diagnosis for more effective management.

#LungCancer #GeneticScreeningTests #Genomics



Understanding Lung Cancer - Video



Dr. B.R. Shrivastav, Endometrial Molecular Cancer Panel - Social Media Poster

MedGenome introduced the Endometrial Molecular Cancer Panel at MP GYNEC-ONCON 2024, with **Dr. B.R. Shrivastav,** Director of the Cancer Hospital & Research Centre, Gwalior, unveiling the test. This next-generation sequencing (NGS)-based test provides genomic profiling for the molecular classification of endometrial cancer. It detects genomic alterations in various endometrial cancer-related genes, including mutations in POLE, *TP53*, *MMR* genes, tumor-agnostic biomarkers, and genes linked to germline predisposition and prognosis.

#MedGenome #Genetics #EndometrialMolecularCancerPanel

CANCER GENOMICS

MedGenome's Comprehensive Hereditary Cancer Gene Panel helps identify genetic mutations in Hereditary Breast & Ovarian Cancer Syndrome and Cancers such as Prostate, Endocrine, Retinoblastoma and more.

It is an NGS-based CAP-accredited test that covers not only hotspots but the complete coding regions, covers variants in the non-coding regions reported in clinical databases, and precisely detects SNVs, CNVs and InDels.

#MedGenome #HereditaryCancerTesting #EarlyDetection #GeneticTesting #CancerPrevention

Endometrial Molecular Cancer Panel - Social Media Poster

Dr. Palanki Satya Dattatreya, Precision Oncology - Social Media Poster

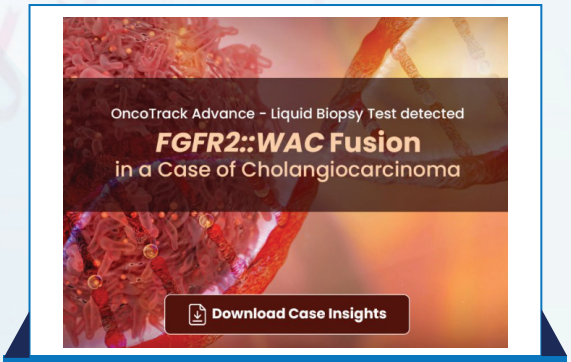
An insightful session on "Precision Oncology" with **Dr. Palanki Satya Dattatreya,** as we explore the foundation and advancements in cancer treatment. Learn about how precision medicine is shaping the future of oncology.

#precisiononcology #cancertreatmentinindia #medicaloncology #OncologyInnovation #HealthcareEvent #GenXDiagnostics #MedGenome #cancerresearch #BhubaneswarEvents #SwostiPremium #FutureofHealthcare

Here is a remarkable example demonstrating the OncoTrack Advance Liquid Biopsy Test's effectiveness. It identifies 117 actionable pan-cancer genes across SNVs, InDels, and CNVs, plus 15 genes for fusions, as recommended by guidelines.

In this case, the FGFR2::WAC fusion at 73X depth was found in a 34-year-old male with intrahepatic cholangiocarcinoma (ICC) using the OncoTrack Advance test. After chemotherapy progression, **Dr. Senthil Rajappa,** HOD, Medical Oncology, Basavatarakam Indo American Cancer Hospital & Research Institute, Hyderabad, referred the patient for genomic testing to explore precision medicine targets.

#genetictesting #medgenome #liquidbiopsy



Dr. Senthil Rajappa, Liquid Biopsy - Social Media Poster

Lung Cancer - Social Media Poster

Here is a remarkable case highlighting the utility of the MedGenome Lung Tumor Panel by NGS, which detects SNVs, InDels, fusions, and CNVs in actionable genes. A 46-year-old male with NSCLC was detected with a PRKAR1A::ALK fusion and showed a complete response to the targeted treatment selected based on the ALK fusion partner. This case underscores the importance of identifying ALK fusion partners, which may be targeted with different ALK inhibitors.

Referred by **Dr. B. S. Ankit** (MBBS, MD, DM Medical Oncology), Consultant Medical Oncologist, HCG Cancer Centre, Jaipur.

#genetictesting

INFECTIOUS DISEASE GENETICS

The past quarter has been incredibly encouraging for the infectious disease segment. We focused on engaging clinicians effectively through targeted initiatives, including test-specific mailers and brochures for panels like the SPIT SEQ and Sepsis AMR Panel etc. Additionally, we amplified our outreach with social media posts, CMEs, and the successful launch of new tests such as the Comprehensive Transplant Panel and the Febrile Neutropenia Panel.

A key highlight was the successful execution of the World Sepsis Day campaign in September, which included impactful mailer dissemination. Moreover, we strengthened our digital presence with a series of testimonial videos featuring esteemed clinicians like Dr. Supraja K, Dr. V. Anil Kumar, Dr. Rajib De and Dr. Gopinathan shared across all social media platforms. Complementing this, we published insightful blogs and a detailed case study on SPIT SEQ, reinforcing our commitment to delivering value-driven solutions in infectious disease segment.

Dr. Supraja K Testimonial - Video post on Metagenomics

Highlighting the alarming rise in respiratory fungal infections post-COVID-19, **Dr. Supraja K,** Director of Pulmonology at the Medway Institute of Pulmonology, shares her insights on utilising NGS-based metagenomics for detecting and analyzing these infections.

#MedGenome #Genetics

Dr. Rajib De, New Test - Febrile Neutropenia Panel Launch - Video

Prof. **Dr. Rajib De,** Professor of Haematology at NRS Medical College & Hospital, Kolkata, and also Clinical Lead at the BMT Programme, Narayana Superspeciality Hospital, unveils MedGenome's Febrile Neutropenia Panel. This all-inclusive panel targets gram-positive and gram-negative bacteria, AMR genes, crucial fungi, and viruses, setting a new benchmark in managing hematological malignancies and febrile neutropenia, particularly in Bone Marrow Transplant (BMT) facilities.

#MedGenome #Genetics

Dr. Gopinathan M, New Test - Comprehensive Transplant Panel Launch - Video

Dr. Gopinathan M, Consultant Hemato-Oncologist & BMT Specialist at MGM Cancer Institute, Chennai, unveils MedGenome's Comprehensive Transplant Panel. This all-inclusive panel targets bacteria, AMR genes, crucial fungi, and viruses, setting a new benchmark in managing hematological malignancies.

#MedGenome #Genetics

Dr. Vikas Oswal, World Sepsis Day - Video

Discover how SPIT SEQ by MedGenome, an NGS-based test for drug-resistant TB, played a crucial role in treating a 33-year-old woman. Watch the case study by **Dr. Vikas Oswal,** M.B.B.S, D.T.C.D, Chest Medicine, Consulting Chest Physician & Pulmonologist, to see how this innovative test guided the effective treatment for TB.

World Sepsis Day - Video

Sepsis remains a critical global challenge, with 48 million cases annually; 11 million of which are in India. Early detection is key to saving lives. The Sepsis-AMR Panel empowers clinicians with rapid, accurate diagnosis from just 1ml of Whole blood, identifying 30 critical targets - Bacteria, Viruses, AMR Genes and Fungi.

With CE-IVD certification and results within a day, it's a game-changer in **#SepsisCare.**

#MedGenome #Genetics #WorldSepsisDay

Dr. V. Anil Kumar, Professor & HOD of Department of Microbiology at Amrita Institute of Medical Sciences, Kochi, highlights the significance of NGS-based Whole Genome Sequencing test by MedGenome for Non-Tuberculous Mycobacteria (NTM) at PULMOCON 2024. This test, with a turnaround time of 14 days, offers comprehensive insights on the species, subspecies, and drug resistance of NTM.

#WholeGenomeSequencing #NGS #MedGenome #Genetics

Dr. V Anil Kumar Testimonial - Video

SPIT SEQ Blog - Social Media Poster

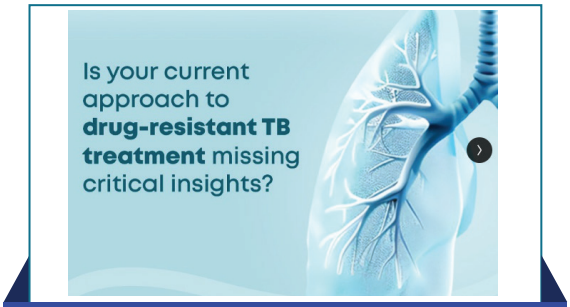
Introducing SPIT SEQ – an innovative tool on the frontline of combating drug-resistant TB! SPIT SEQ is an NGS-based test for drug resistance testing in TB patients, facilitating the development of effective treatment plans. Discover how MedGenome's SPIT-SEQ is transforming and enhancing patients' lives.

#tuberculosis #TBpatients #SPITSEQ #drugresistancetesting #MedGenome

Could current approach to Drug-Resistant TB testing be overlooking key insights?

SPIT SEQ, our NGS-based test, offers fast and accurate resistance detection directly from clinical samples. With extensive drug resistance profiling and peer-validated precision, it empowers clinicians to make informed decisions for better patient outcomes.

#MedGenome #SPITSEQ #GeneticTesting



SPIT SEQ Carousel - Social Media Poster

CME on Recent update on Multiplex PCR for infectious diseases at Victoria Hospital, Bangalore - Event Snapshot

GENOMIC WELLNESS

The past quarter has been highly encouraging for the Genessense segment, marked by continued growth and evolution of the diverse tests within our portfolio, particularly through digital platforms and our website. We strengthened our outreach with engaging social media content, including videos, carousels, and case studies, highlighting tests such as Kardiogen, Curegen, Neurosense, and more. A standout achievement was the successful World Heart Day campaign in September, dedicated to promoting Kardiogen. This included a range of impactful activities, such as a video post, a testimonial video, online polls, and an engaging blog. Additionally, we spotlighted Oncosense during Breast Cancer Awareness Month and Diabetogen on World Diabetes Day with compelling video and carousel posts across digital platforms. The Kardiogen mailer was also shared as part of our focused efforts to drive awareness and engagement.

World Heart Day - Kardiogen Video

Your heart's story begins long before the first beat. This World Heart Day, hashtag#StartFromTheSource; uncover the Genetic Risks shaping the future of your heart. Take control of your heart health with Kardiogen by Genessense.

#Genessense #Kardiogen #WorldHeartDay #StartFromTheSource

On this World Heart Day, **Dr. Ramesh Menon,** Associate Director, Genomic Medicine and Bioinformatics, MedGenome Labs, shares his insights, he urges all to take control of their heart health.

Kardiogen test by Genessense screens 1.3 million genetic markers and calculates a personalized risk score, giving you insights tailored to your DNA.

It's your heart. Your story. Your future. Take control of it.

#KardioGen #WorldHeartDay #MedGenome #HeartHealth #GeneticTesting #StartFromtheSource

Dr. Ramesh Menon, World Heart Day - Video

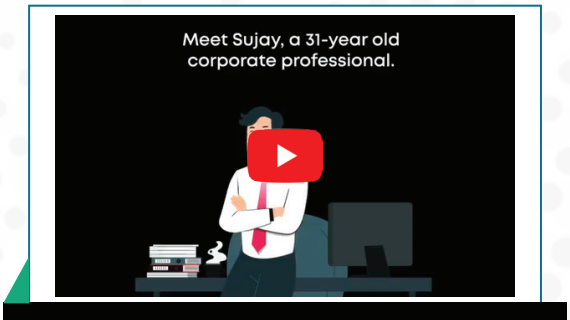
Genessense Portfolio - Video

At Genessense, we empower you to take control of your health with the power of genomic insights. Our preventive genetic screening tests help you predict and manage chronic and lifestyle diseases before they manifest. Embrace genomic wellness today. Your genes hold the key to a healthier future.

When heart health is a family concern, early genetic screening can make all the difference. Sujay's case shows how the KARDIOGEN test empowered his family to understand their genetic risk for Coronary Artery Disease (CAD) and take proactive steps towards a healthier future.

Know your risk, take control, and protect your heart health today.

#Genessense #Kardiogen



Kardiogen Case Study - Video

World Breast cancer Awareness Month - Oncosense Video

A legacy of strength deserves the power of knowledge.

This Breast Cancer Awareness Month, embrace your legacy of resilience. With Oncosense screening test, uncover hidden hereditary risks and take charge of your health and future with confidence.

#Genessense #BreastCancerAwareness #Oncosense

Knowing your genetic profile can be a powerful ally in safeguarding brain health. The *APOE* gene, crucial for cognitive function, can also elevate the risk of Alzheimer's, especially for those with two *APOE4* copies—a significant finding from recent research. Early genetic insights can be pivotal, offering a clearer path to proactive health choices.

Genessense's NEUROSENSE genetic screening empowers you with these essential insights, helping you plan for a healthier future.

Take control of your health with NEUROSENSE.

#Genessense #Genetics #Alzheimers #Neurosense #BrainHealth #APOE4

Neurosense - Video

World Diabetes Day - Diabetogen Video

India holds the title of the Diabetes Capital, with millions at silent risk. With advanced genetic screening, we can predict and prevent diabetes. Family history shouldn't determine your future.

Know your hashtag#SugarLines and take control early.

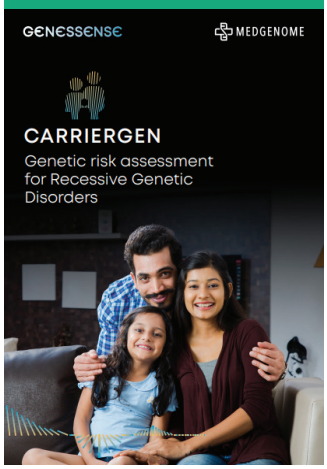
#Genetics #WorldDiabetesDay #Genessense

Family history of diabetes? Discover your risk with an early genetic risk screening test. This World Diabetes Day, take control of your health. Choose the screening test and help shape a healthier future for yourself and your loved ones.

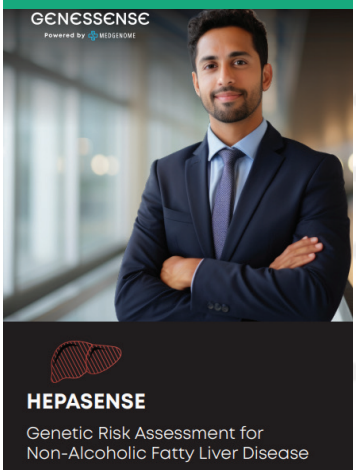
#SugarLines #Genessense #GeneticTesting

World Diabetes Day - Static Social Media Poster

Carriergen Brochure



Hepasense Brochure



What's new

Research Publications

KBG Syndrome in 16 Indian Individuals

Journal : *American Journal of Medical Genetics Part A* Read more

Movement Disorders in Megalencephalic Leukoencephalopathy with Subcortical Cysts - A Case Series

Journal : *Parkinsonism & Related Disorders* Read more

Spectrum of Alport Syndrome in an Indian Cohort

Journal : *Pediatric Nephrology* Read more

rAbDesFlow: A Novel Workflow for Computational Recombinant Antibody Design for Healthcare Engineering

Journal : *Antibody Therapeutics* Read more

Navigating the Clinical Landscape: Update on the Diagnostic and Prognostic Biomarkers in Multiple Myeloma

Journal : *Molecular Biology Reports* Read more

Identification of Nine Novel HLA Alleles by Next-Generation Sequencing in Individuals from India

Journal : *HLA* Read more

GNE Myopathy: Genotype - Phenotype Correlation and Disease Progression in an Indian Cohort

Journal : *Journal of Neuromuscular Diseases* Read more

Mosaic Embryo Transfer After Preimplantation Genetic Testing for Structural Rearrangement: A Case Study

Journal : *Journal of Biochemical and Clinical Genetics* Read more

Extending HLA Allele Sequences Using Next-Generation Sequencing Technology

Journal : *HLA* Read more

Tests Launched

- CNVSure - CNV confirmation by qPCR
- BabySecure Lysosomal Storage Disorders (LSD) Panel
- Blended Genome Exome
- AML Focus panel by NGS (SNVs, InDels & Fusions)
- Low Pass WGS
- FISH for t(1;19), TCF3::PBX1 (q23;p13), ALL
- Comprehensive Transplant Panel

Proud Moment

We are thrilled to announce that MedGenome has been honored with the **2024 ASK Private Wealth HURUN INDIA Future Unicorn Award** for **Innovation in Genomics** at the prestigious **India Future Unicorn Awards** held on November 20, 2024, in Bengaluru.

Our **CFO, Surajit Chakrabartty,** had the privilege of accepting this award on behalf of the team at MedGenome. This recognition fuels our determination to continue pushing boundaries in the field of genomics and work tirelessly to fulfil our vision of making innovative genomic solutions affordable and accessible across the world.

From Our US Office

In September and October, the U.S. office celebrated several milestones, including the appointments of **Dr. Felix Olale, MD, PhD, as President and CEO,** and **Jennifer Rose as Chief Commercial Officer,** bringing transformative leadership to our team. The U.S. office came together for a vibrant Diwali celebration, highlighting the diversity and unity within our team. The festivities were a reminder of the global culture that powers MedGenome and its mission. October also saw the sales team visit to our Foster City HQ for in-depth training sessions. The team capped off their visit with a trip to the California coast, blending learning and team-building to strengthen collaboration and alignment on future goals. The U.S. team participated in the American Society of Human Genetics (ASHG) conference, showcasing MedGenome's cutting-edge multiomics capabilities to a global audience. The event provided valuable opportunities to connect with researchers and biopharma leaders who share our vision for advancing precision medicine.

Dr. Felix Olale, MD, PhD
President and CEO

Jennifer Rose
Chief Commercial Officer

New Leadership Appointments: Their expertise and vision are poised to lead us into a transformative phase of innovation and expansion.

Diwali Celebrations: The team came together to celebrate Diwali, embracing the spirit of unity and cultural diversity that defines MedGenome.

Sales Team Training: The sales team gathered at our Foster City HQ for comprehensive training sessions, concluding with a team-building trip to the stunning California coast.

From Our US Office

Showcasing at ASHG: At the American Society of Human Genetics (ASHG) conference, we highlighted MedGenome's cutting-edge multiomics capabilities, engaging with researchers and biopharma leaders to drive precision medicine forward.

We continue to share insights and updates through our blog. Check out the latest articles here: https://research.medgenome.com/blog/

Recent Blog Highlights:

Single-cell sequencing: applications, methods, and insights

This article explores the latest advancements in single-cell sequencing, highlighting its applications in gene expression, disease research, and personalized medicine.

Comprehensive cancer profiling with Illumina's TSO 500 assay

This post discusses how Illumina's TruSight Oncology 500 (TSO-500) assay enables comprehensive genomic profiling of tumor samples, aiding in cancer diagnosis and therapeutic strategies.

Immune repertoire diversity: key to understanding disease processes and creating novel therapies

The article delves into the importance of immune repertoire diversity in understanding disease mechanisms and how it can be leveraged to create innovative therapeutic solutions.

Lung cancer: molecular insights and emerging therapeutic approaches

This blog provides an overview of the molecular insights into lung cancer and highlights emerging therapies aimed at improving treatment outcomes for patients.

Exploring the new Human Cell Atlas release and its impact on genomic research

The article examines the latest release of the Human Cell Atlas, its contributions to genomic research, and its potential to revolutionize our understanding of human biology.

Explore our latest blog articles at **MedGenome Research Blog.**

We value your insights and invite you to share your viewpoints and articles of interest at mgus-blog@medgenome.com.

Pharmacogenomics:

A clinical tool for personalized medicine

Dr. Venu Seenappa, PhD
Senior Scientist, MedGenome

Pharmacogenomics (PGx), also known as pharmacogenetics, is the study of how an individual's genetic makeup influences response to different drugs or medications. Individuals with the same disease will respond differently to the same drug prescription. We also often hear people say that a particular drug does not work too well for them or that the drug has no effect.

Similarly, it is common to hear of adverse effects of drugs on some individuals (Figure 1). This is now explained by science - individuals process different drugs differently.

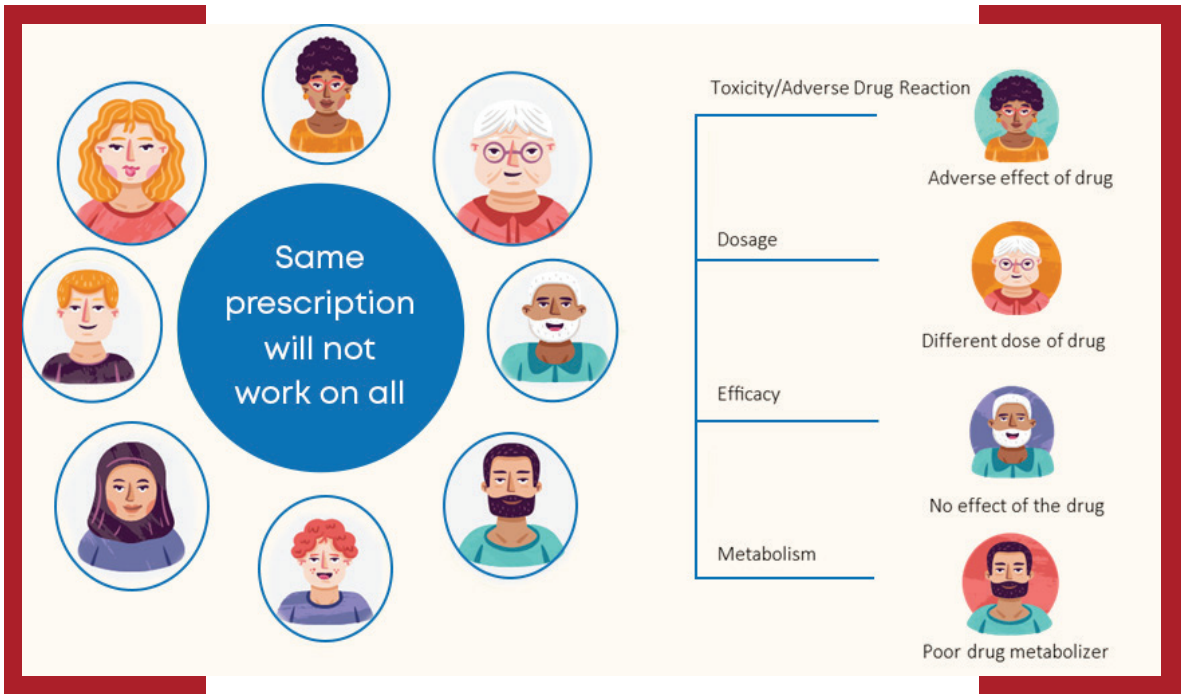


Figure 1: Schematic representation of individuals with varied drug responses to the same drug prescription.

In addition to age, gender, and weight, genetic factors play an important role in drug response. Pharmacogenomics studies have revealed the relationship between genetic variation and drug responses. The genetic variants such as SNPs, Indels, and CNVs in the genes encoding for enzymes lead to changes in the function or abundance of proteins, leading to the gain of function or loss of function phenotypes implicating varied responses to drugs. Based on the functional status of the allele and its impact on enzyme activity, individuals will be categorized as rapid, normal, intermediate, or poor metabolizers for a specific gene/drug pair (1).

Drug Metabolism

The metabolism of drugs is a process of altering the molecules chemically after they enter the body. Most drugs have lipophilic centers and are converted to hydrophilic centers during biotransformation, which increases their water solubility to allow elimination in urine or bile (2). The cytochrome P450 (CYP) families of enzymes are major players in the metabolism of many drugs; it is estimated that 1/3rd of all drugs are metabolized by CYP3A and 1/4th of all drugs are metabolized by CYP2D6 enzymes (3). Although more than 50 CYP enzymes are reported, 6 enzymes (CYP1A2, CYP2C9, CYP2C19, CYP2D6, CYP3A4, and CYP3A5) metabolize 90 percent of drugs, where CYP3A4 and CYP2D6 are most significant enzymes (4).

The History of Pharmacogenetics

The term Pharmacogenetics (PGx) was coined by Friedrich Vogel in 1959 (5). The history of PGx dates back more than 2000 years when Pythagoras described a trait now known as favism and advised against the consumption of fava beans. Later, scientists discovered deficiency of G6PD leads to the development of red blood cell hemolysis or acute hemolytic anemia in some individuals upon eating fava beans, indicating the important observation made by Pythagoras on non-tolerating of fava beans in certain individuals is prominent in a subset of the population (5, 6).

Furthermore, PGx gained importance about 60 years ago with the discovery that the metabolism of the muscle relaxant succinylcholine and a deficient N-acetylation of isoniazid (antituberculosis drug) has a genetic component (7,8). Looking at the succinylcholine history, doctors sometimes encountered patients in whom the paralyzing effects of succinylcholine lasted considerably longer than normal, putting the patients in prolonged apnea. Later Werner Kalow showed that the prolonged apnea was caused by the presence of a variant in BCHE (9).

Drug Metabolizers and Transporters

Multiple drugs and genes under many therapeutic areas are reported in the literature on their role in drug response. Drug metabolizers like CYP2C9 for non-steroidal anti-inflammatory drugs (NSAIDs), CYP2D6 for Opiods, CYP2B6 for Sertraline, CYP2C19 for Tricyclic antidepressants (TCAs), and many more drugs are reported in individuals with multiple metabolizer phenotype based on the allelic variation in these genes (10). For example, CYP2C9 *3/*3 is a poor metabolizer of NSAIDs, CYP2D6 *4/*4 is a poor metabolizer for Opiods, CYP2C19 *2/*17 is an intermediate metabolizer for TCAs (Figure 2a).

Similarly, drug transporters such as SLCO1B1 for Statins, which facilitates the hepatic uptake of all statins, and ABCG2 for Rosuvastatin, which modulates the absorption and disposition of rosuvastatin, are well cited in the guidelines. For example, SLCO1B1 *5/*5 shows poor function phenotype for statins, where it carries two no-function alleles (Figure 2b) and carries one ABCG2 c.421 C/A variant shows decreased function phenotype for Rosuvastatin (11).

The therapeutic area covered under Pharmacogenetics is expanding as more scientific evidence gets accumulated. Therapy areas such as cardiovascular, oncology, psychiatry, neurology, transplantation, and many more benefit patients both in cost savings and by improving the quality of health (12, 13).

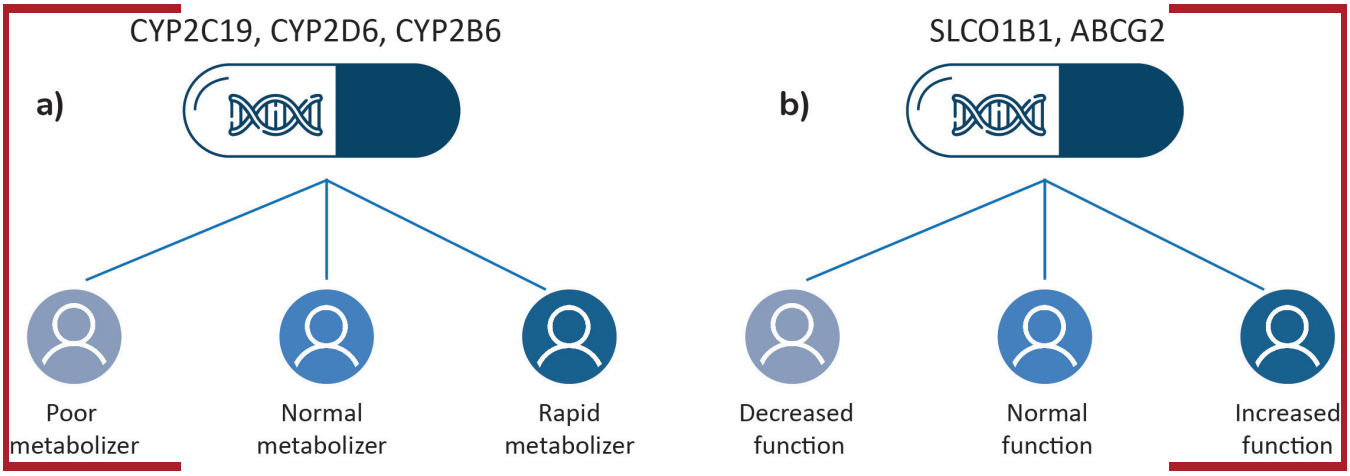


Figure 2: Phenotype for drug-metabolizing enzymes and transporters.

Guidelines for Drug Dosing

Although the PGx testing showed a promising outcome in preventing adverse drug reactions, multiple drug selection, multiple hospital visits, and cost savings, not much of this knowledge has been translated into clinical practice (6). The reasons for being lack of guidelines for dosing recommendation, lack of awareness in real-world clinical settings, and limited evidence of clinical validity have delayed PGx progress to clinical settings. However, upon accumulation of guidelines, scientific statements from consortiums like the Clinical Pharmacogenetics Implementation Consortium (CPIC), The Pharmacogenomics Knowledge Base (PharmGKB), PharmVar, AHA, and approvals in the FDA for testing specific gene and drug combinations lead to the development of more clinical PGx tests. Further, the growing body of evidence is helping to integrate PGx testing into daily clinical practice (10, 14, 15).

CPIC is an international consortium established in 2009 with the objective of clinical implementation of pharmacogenetic tests by creating, curating, and posting freely available, peer-reviewed, evidence-based, updatable, and detailed gene-drug clinical practice guidelines for the benefit of patient care. CPIC is a close partner of PharmGKB, an interactive tool and knowledge database to investigate how genes affect drug response (16). CPIC has published guidelines for nearly 100 drugs and covers up to 20 genes with dosing recommendations for different phenotypes. CPIC provides recommendations for the level A and B drug/gene combinations, where these pairs have sufficient research evidence for at least one prescribing action to be recommended. Levels C and D are not considered to have adequate evidence or actionability to have prescribing recommendations (10). Similarly, levels 1 and 2 are considered as high evidence drug/gene pairs in PharmGKB.

Advantages of Pharmacogenetics Test

The PGx test has multiple advantages on specific gene/drug pairs, which indicates,

- Whether the medication will be effective for an individual
- Whether an individual needs different doses than standard
- Whether an individual needs alternative therapy
- Whether an individual is at risk for serious side effects or adverse drug reactions due to altered metabolism of the medication

Methodology

The PGx test can be performed using multiple technology platforms to identify the variants in the gene of interest. Platforms such as Sanger sequencing, Real-time PCR, Genotyping arrays, Next generation sequencing (NGS) are utilized in the research and clinical setting for variant detection (Figure 3). Each platform has its own advantages, hence the objective for the testing must be clear before opting for the test.

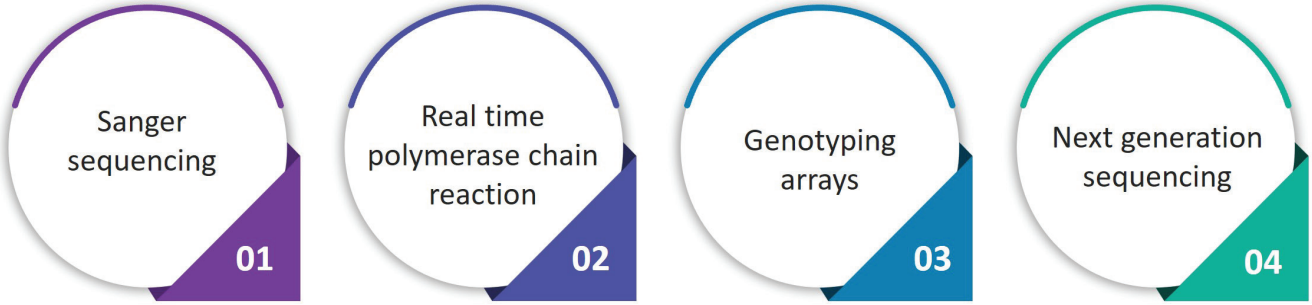


Figure 3: Different methodology for PGx testing.

For example, if variants of interest are restricted to single or 2 variants in a gene, it's always better to go with Sanger sequencing, which saves both time and cost. On the other hand, the most robust and advanced technology, like NGS, can opt for the multi-gene analysis, which is rapid, saves time, and provides comprehensive analysis for multiple drug/-gene pairs in a shorter time, which will always be beneficial for the patient from the clinical point of view. Many genes like CYP2C19, CYP2C9, CYP2D6, DPYD, etc. are reported with multiple certain function variants and haplotypes (2 or more SNPs in a single allele) in the genes, indicating the whole gene analysis for accurate diplotype (represent two-star alleles) calling and to categorize precise phenotype.

Direct-to-consumer PGx Test

Though PGx tests are more suitable and appropriate for clinical settings, few companies offering direct-to-consumer (DTC) health packages include PGx as one of the screening components to provide a complete wellness package. Consumers willing to take DTC tests as a proactive step are encouraged, however, they must be cautious and ensure they consult the clinician and follow the directions of the physician before taking or changing any medication based on such DTC reports.

PGx Testing at MedGenome

Being a global leader in the genomic industry, many PGx tests are offered by MedGenome. Single gene to multiple gene/drug pair tests are performed using various technology platforms. For example, Clopidogrel-CYP2C19, 5- Fluorouracil-DPYD, Tacrolimus-CYP3A5, and multi gene/drug pair tests like Curegen advanced, which reports up to 100 drugs and checks 20 genes are offered. Multiple therapy areas are covered in MedGenome PGx tests to provide a comprehensive analysis of various drugs and genes (Figure 4).

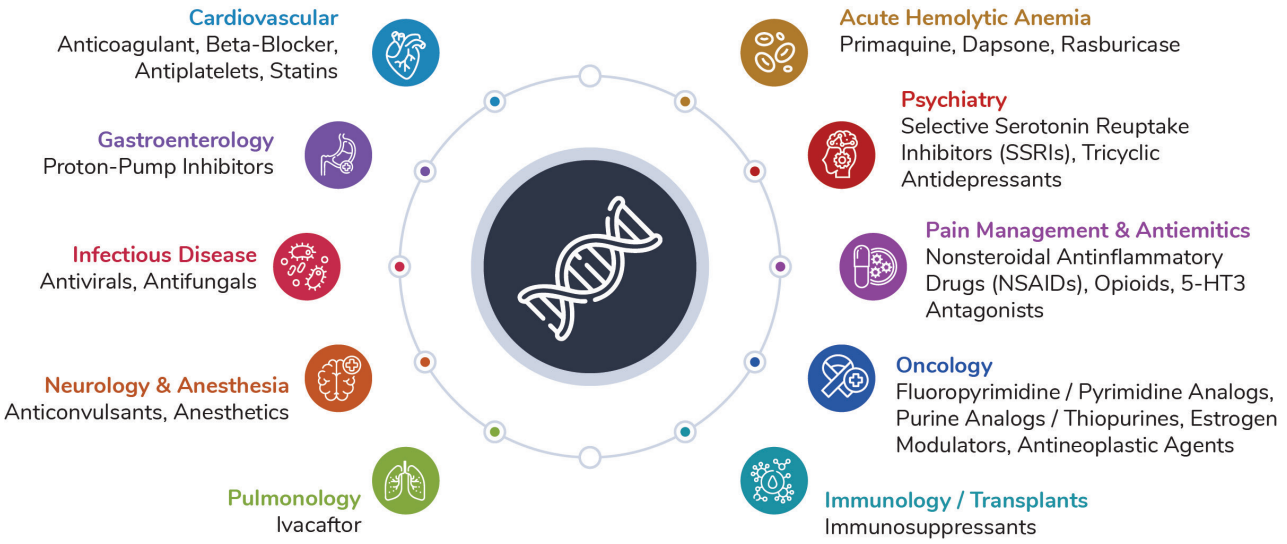


Figure 4: Therapy areas covered under pharmacogenetics test at MedGenome.

Conclusion

Pharmacogenomics is a combination of pharmacology and genetics, which is used effectively to profile an individual's DNA to prescribe suitable drugs with appropriate dosages. Pharmacogenetic testing informs us there is a better way to prescribe medicine than a hit-and-miss strategy. PGx is a clinical tool for personalized medicine, which must be utilized in primary to tertiary clinical settings by clinicians to i) combat adverse drug reactions, ii) avoid multiple visits to hospitals by patients for non-responsive therapy, iii) to reduce the financial burden, and iv) to improve the quality of patient health. Enhancing the knowledge of clinicians by continuous education on the advantages of PGx testing at clinics and awareness programs for the public may boost the utility of testing and, in parallel, it helps holistically on true well-be-

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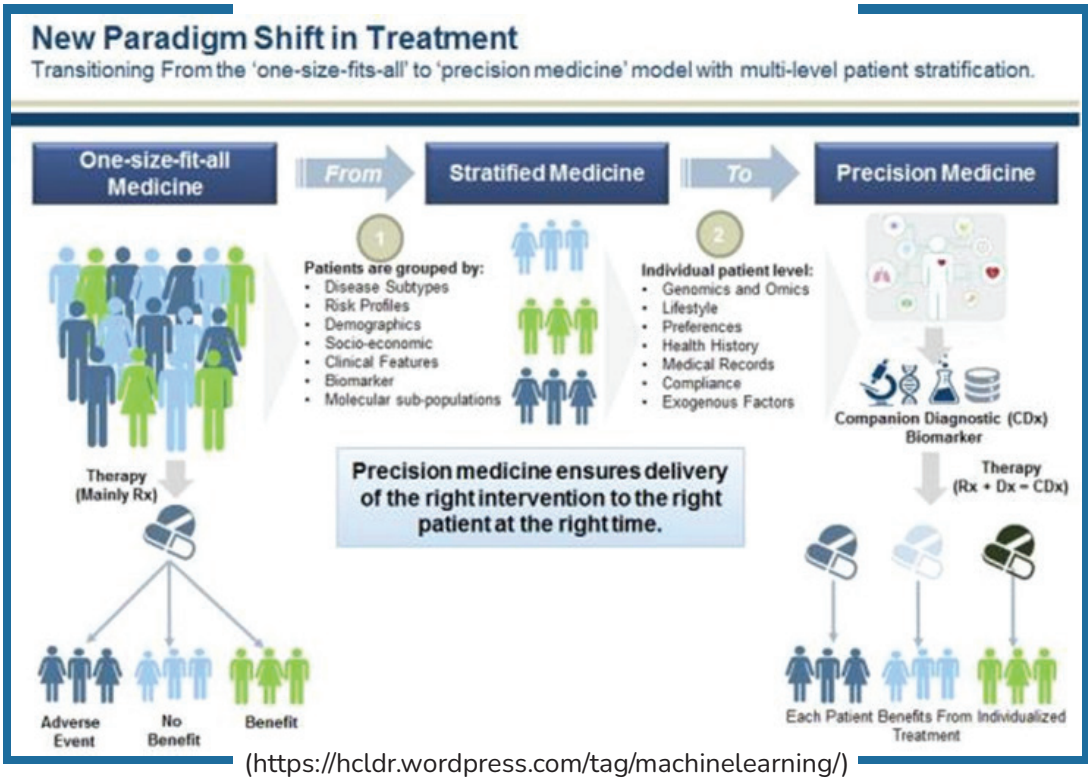
Precision medicine: transforming healthcare in India amidst significant challenges

Dr. Suruchi Aggarwal, PhD
Head- Scientific Affairs & Technical Support

Precision medicine, often referred to as personalized medicine, represents a transformative approach to healthcare that tailors medical treatment to the individual characteristics of each patient. By leveraging genomic, environmental, and lifestyle information, this field aims to move beyond the one-size-fits-all approach of traditional medicine. While precision medicine has made significant strides globally, its adoption in India holds immense potential to address the country's diverse healthcare challenges which may hinder its widespread implementation. This article explores the multifaceted challenges of adopting precision medicine in India, including economic barriers, infrastructural issues, regulatory hurdles, and ethical considerations.

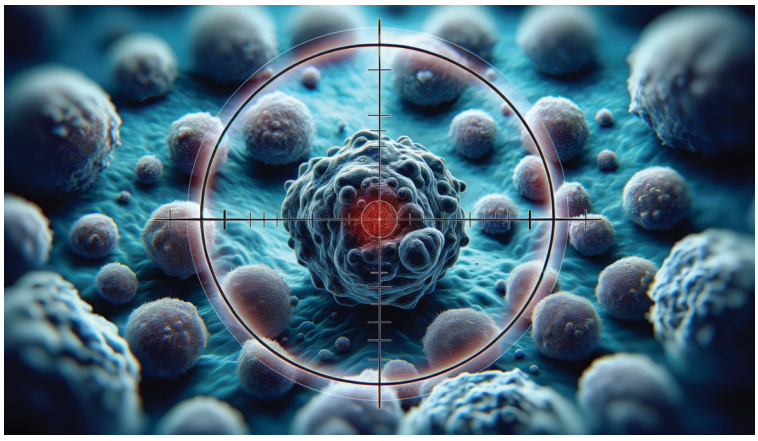


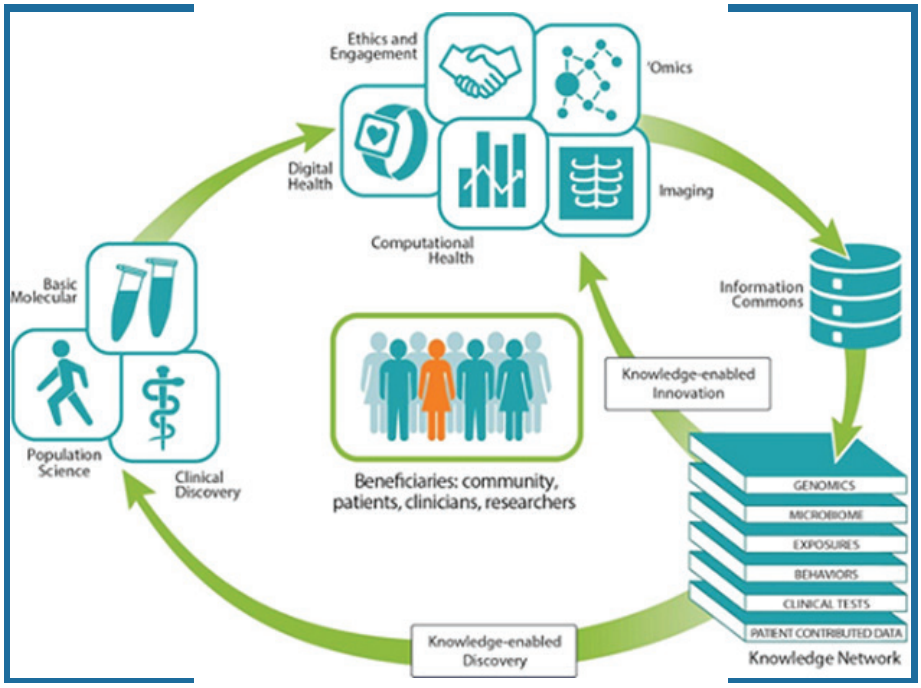
(https://hcldr.wordpress.com/tag/machinelearning/)

The Promise of Precision Medicine

Precision medicine is revolutionizing healthcare by emphasizing predictive, preventive, personalized, and participatory approaches to patient care. It enhances disease prevention through advanced genetic testing and risk profiling, enabling individuals to adopt early interventions and lifestyle changes to mitigate risks of conditions like cancer, diabetes, and cardiovascular disorders. Targeted therapies, such as biomarker-driven treatments and immunotherapy, provide tailored options for diseases that traditionally lacked effective solutions, particularly in oncology.

Pharmacogenomics optimizes drug efficacy by personalizing dosages and reducing adverse reactions, accounting for the unique genetic variations seen in Indian populations. While initial costs can be high, precision medicine offers long-term savings by minimizing trial-and-error treatments and prioritizing preventive care. Furthermore, it empowers patients by delivering personalized health insights, fostering engagement, and enabling informed decision-making, marking a transformative shift in modern healthcare.





The Indian Context

Genetic Diversity in India

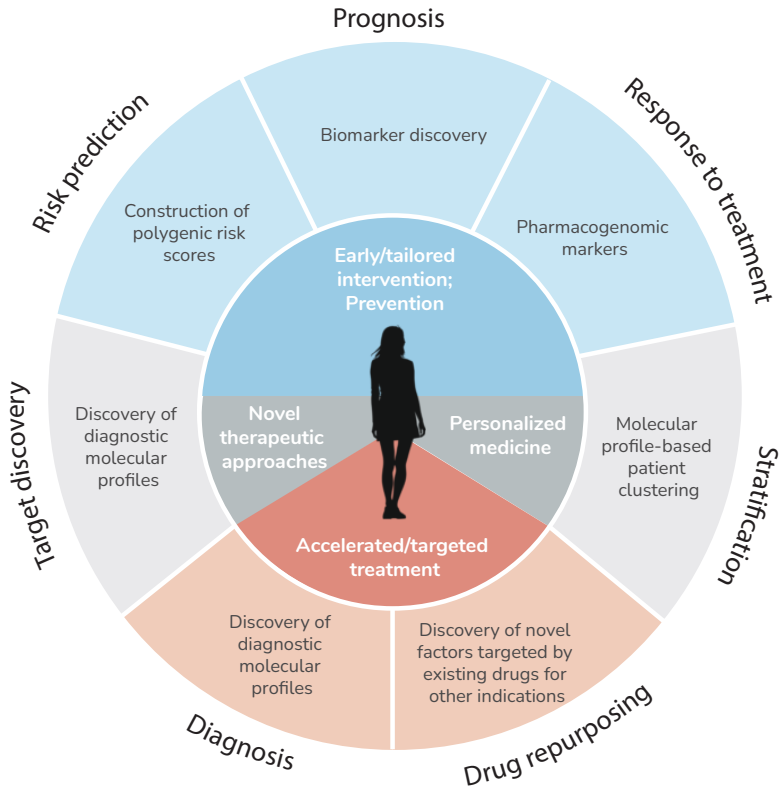
India's healthcare landscape is uniquely positioned to benefit from precision medicine. The country is home to over 1.4 billion people with rich genetic diversity spanning ethnic, linguistic, and geographical lines. This diversity offers a treasure trove of data for understanding disease patterns and developing region-specific treatments.

India has seen remarkable advancements in genomic research and healthcare infrastructure over the past decade. Initiatives like the GenomeIndia Project, launched in 2020, aim to map the genetic makeup of diverse Indian populations, providing a robust foundation for disease research and therapeutic development. Alongside this, public and private healthcare organizations are increasingly investing in next-generation sequencing (NGS) technologies, making genomic testing more accessible to clinicians and patients alike.

Leading the way in this genomic revolution is MedGenome Labs, based in Bangalore, a pioneer in genomics-driven diagnostics and research. MedGenome has played a crucial role in integrating cutting-edge genetic testing into routine clinical care. The company offers a comprehensive portfolio of diagnostic solutions, including cancer genomics, reproductive health testing, and rare disease diagnostics. The company's collaborations with global pharmaceutical giants and research institutions further position India as a hub for precision medicine. There are other genomic diagnostic companies like Strand Biosciences, which work in a similar way.

Additionally, the clinical expertise of India's healthcare providers complements advancements in genomics. Physicians across major hospitals and specialty centers are increasingly incorporating genomic insights into patient management. For example, in oncology, tumor molecular profiling has become a cornerstone of personalized cancer care, enabling oncologists to select targeted therapies with greater precision. Similarly, clinical genetics specialists are leveraging insights from genomic testing to diagnose rare genetic disorders, guiding families with actionable health information. This synergy between innovative diagnostic Labs and the growing adoption of genomic tools by clinicians is creating a robust ecosystem for precision medicine in India.

Precision Medicine – Opportunities



Despite its promise, the implementation of precision medicine in India faces significant challenges.

01 Inadequate Infrastructure

India's healthcare infrastructure is unevenly distributed, with rural areas often lacking basic facilities. Precision medicine relies on advanced diagnostic tools, sequencing technologies, and specialized laboratories, which are scarce in remote regions.

02 Cost and Affordability

One of the most significant challenges in adopting precision medicine in India is the high cost associated with genomic testing and personalized therapies. Advanced diagnostic tests can be prohibitively expensive for many patients, especially those from lower socioeconomic backgrounds. For instance, the total cost of treatment plans can range from ₹12 lakh to ₹24 lakh per patient annually, which is unaffordable for a large segment of the population. Furthermore, limited insurance coverage exacerbates this issue, leaving many patients without financial support for necessary treatments. While costs have decreased over the years, they are still out of reach for many in a country where healthcare expenditure is largely out-of-pocket.

03 Limited Awareness and Education

A significant knowledge gap exists among healthcare providers and patients regarding precision medicine. The successful implementation of precision medicine requires a workforce skilled in genomics and bioinformatics. Unfortunately, there is a shortage of trained professionals capable of interpreting complex genetic data and integrating it into clinical practice. This skills gap poses a barrier to effectively utilizing precision medicine technologies in patient care. Educational institutions must enhance their curricula to include training in genomics and

04 Lack of Trained Professionals

The successful implementation of precision medicine requires a workforce skilled in genomics and bioinformatics. Unfortunately, there is a shortage of trained professionals capable of interpreting complex genetic data and integrating it into clinical practice. This skills gap poses a barrier to effectively utilizing precision medicine technologies in patient care. Educational institutions must enhance their curricula to include training in genomics and personalized healthcare.

05 Limited Access

Access to advanced diagnostic facilities is uneven across India. While metropolitan areas may have access to cutting-edge genomic testing and precision therapies, tier-2 and tier-3 cities often lack such facilities. This disparity limits the reach of precision medicine and creates inequities in healthcare access. The government has initiated programs like the National Genomic Grid to improve data sharing and access to genomic information; however, significant gaps remain in infrastructure development across different regions.

06 Lack of Standardized Guidelines

India lacks standardized protocols for genetic testing, data interpretation, and patient management. The absence of regulatory frameworks raises concerns about the quality, reliability, and ethical use of genetic information. Current regulations struggle to keep pace with rapid advancements in genomics and biotechnology. The complexity of designing clinical trials for personalized therapies poses additional challenges; smaller trial designs often face scrutiny regarding their statistical validity. There is a pressing need for clearer guidelines from regulatory authorities to facilitate the approval process for new precision medicine therapies.

07 Data Privacy and Ethical Concerns

Genomic data is highly sensitive, and its misuse can lead to discrimination and stigmatization. The collection and storage of vast amounts of genetic data raise significant privacy concerns. Patients may be apprehensive about sharing their genetic information due to fears of misuse or discrimination based on their genetic predispositions. Establishing robust data protection regulations is crucial to gaining public trust and encouraging participation in genomic studies.

08 Genetic Diversity and Research Gaps

While India's genetic diversity is a strength, it also poses challenges. Many existing genomic databases are skewed toward Western populations, leading to underrepresentation of Indian genetic variations. Addressing this requires large-scale, inclusive research initiatives.

09 Integration of data systems into Healthcare

Effective implementation of precision medicine relies on integrating various data sources, including electronic health records (EHRs), genomic databases, and clinical data repositories. However, logistical barriers exist that prevent seamless data sharing among different healthcare institutions. Overcoming these barriers requires investment in health information technology infrastructure and collaboration among stakeholders.

Despite these challenges, there are promising developments on the horizon for precision medicine in India. A multi-faceted approach involving collaboration among various stakeholders—including government bodies, healthcare providers, pharmaceutical companies, and academic institutions—is essential. By working together, these entities can create an ecosystem that supports research, education, and equitable access to precision medicine solutions.

Investments in healthcare infrastructure, especially in rural areas, can bridge the urban-rural gap by establishing genetic testing centers and training laboratories in Tier 2 and Tier 3 cities. Reducing costs through public-private partnerships and government-backed insurance schemes can make personalized treatments more accessible to economically weaker sections. Education and training programs must incorporate genomics into medical curricula and provide continuous learning opportunities for healthcare professionals, supported by public awareness campaigns to educate individuals about genetic testing.

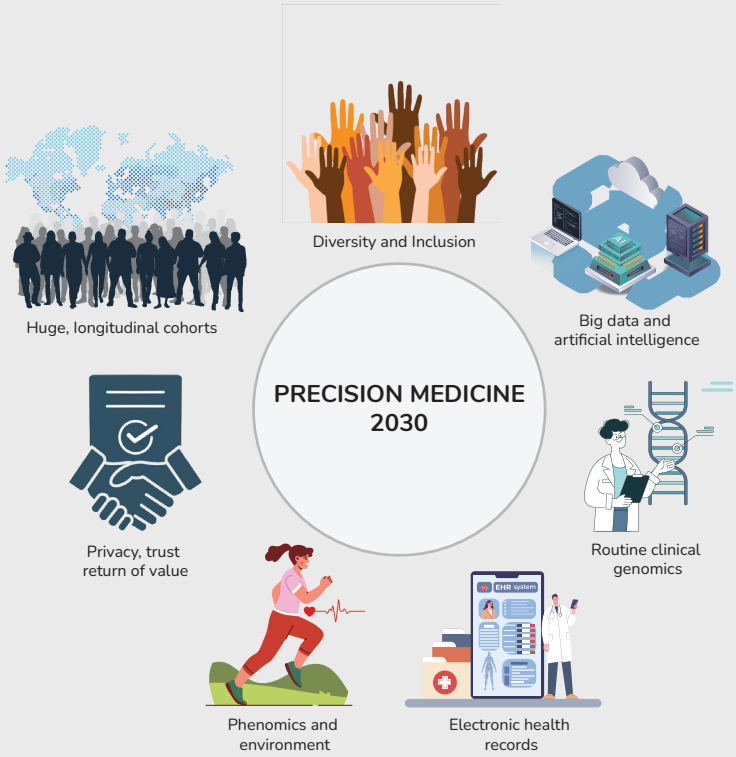
Raising public awareness about the benefits of precision medicine can also play a crucial role in its adoption. Educating patients about available options and encouraging participation in clinical trials can help build a more informed patient population that advocates for their healthcare needs.

Robust guidelines and regulatory frameworks are needed to standardize genetic testing, address ethical concerns, protect data privacy, and ensure equitable access.

Additionally, leveraging advanced technologies like artificial intelligence and telemedicine can enhance the scalability of precision medicine, enabling remote genetic counseling and expanding access to underserved populations.

Most importantly, maintaining comprehensive health records is a cornerstone of precision medicine, as it provides the foundation for tailoring medical care to individual needs. Accurate and up-to-date health records, including genetic profiles, medical history, and lifestyle information, enable healthcare providers to deliver personalized treatments and predict disease risks with greater accuracy. These records facilitate seamless care coordination across multiple specialists, reduce redundancies in testing, and enhance the efficacy of targeted therapies. Moreover, aggregated and anonymized health data can drive research in genomics and pharmacogenomics, accelerating the development of region-specific solutions and fostering innovation in precision medicine.

The journey toward adopting precision medicine in India is fraught with challenges ranging from economic barriers to ethical considerations. However, with concerted efforts from all stakeholders involved—government agencies, healthcare providers, researchers, and patients—these hurdles can be addressed effectively. As India continues to invest in genomic research and infrastructure development, there lies an opportunity to harness the full potential of precision medicine for improving health outcomes across diverse populations. By fostering an inclusive approach that prioritizes equity and accessibility, India can pave the way for a future where personalized healthcare becomes a reality for all its citizens.



As aptly said by Sir Charles Darwin – 'It is not the strongest of the species that survives nor the most intelligent, it is the one most adaptable to change.

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Case study

Atypical fusion transcript detected by Next Generation Sequencing in Acute Promyelocytic Leukemia

Ritika Chauhan, PhD
Lead Genome Analyst

A 33-years old male patient who was a diagnosed and treated case of APML in 2019. In 2024, the patient showed symptoms and was suspected to have relapsed. His blood profile reflected Hemoglobin (Hb) = 9.3 gm%, TLC = 2110/c-mm, Platelets = 80,000/cumm, corrected reticulocyte count = 1.1%. The peripheral blood smear showed Anisopoikilocytosis. The bone marrow examination revealed hypercellular marrow with Blast- 0.1%, Promyelocytes- 22%, Myelocytes- 22%, Metamyelocytes- 10%, Neutrophils- 12%, Lymphocytes- 0.8%, Monocytes- 0.4%, Eo-Baso 10%, Plasma cells- 0.1%, and Erythroid precursors- 10%. There was a marked reduction in Erythropoiesis and Megakaryocytes, and an increase in Promyelocytes.

The patient was cytogenetically tested for *PML::RARa* translocation t(15;17) which is the characteristic of Acute Promyelocytic Leukemia (APML). However, the Fish result turned out to be negative for *PML::RARa* translocation.

Peripheral Blood sample was sent to MedGenome Laboratory for NGS Targeted Sequencing (NGS) to identify pathogenic Single Nucleotide Variations (SNVs), small INDELs, Copy Number Variations (CNVs), and gene fusions.

Nucleic acid extracted from blood was used to perform targeted gene capture by custom capture kit, and the QC passed library was sequenced on a validated Illumina sequencing platform >250X depth for SNVs, Indels and CNVs, 500X for gene fusions. The in-house developed pipeline was used to analyze and report genomic alterations (SNVs, Indels, CNVs and fusions) as per AMP/CAP/ASCO guidelines.

The molecular analysis by Next generation sequencing (NGS) revealed the presence of atypical transcript with a translocation of t(11;17) *ZBTB16::RARa* which tends to represent similar morphological features as classic Acute Promyelocytic Leukemia (APL). The *ZBTB16::RARa* gene fusion was detected at 53X read depth, and a total of 69 unique reads were detected for this junction sequence. The fusion breakpoint was at chr11:114156434 of *ZBTB16* gene at exon 3 of 5' gene, and the fusion breakpoint was detected at chr17:40348316 of *RARa* gene at exon 3 of 3' gene. This fusion was confirmed by Arriba tool (Figure 1) as well as blatting of read sequences between *ZBTB16* and *RARa* in Integrative Genomic Viewer (IGV). No clinically significant SNVs, Indels, and CNVs were identified in the patient sample.

APML is characterized by the presence of atypical promyelocytes in bone marrow, and favorable overall survival with retinoid-based therapy. However, a very small percentage of APML cases lack classic t(15:17), and show variant translocations involving *RARA* such as t(11;17) (q23;q21) which forms *ZBTB16::RARA* gene fusion. The studies suggest that *ZBTB16::RARA* gene fusion is most prevalent, accounting for approximately 1% of all APML cases. This genetic APML fusion variant is associated with resistance to ATRA (all-trans retinoic acid), and poor prognosis. The studies also suggest that a better Complete remission (CR) has been observed in the patients who receive ATRA plus intensive chemotherapy regime.

Key Takeaways

- The clinical presentation of the patient harboring atypical *RARA* transcript is usually similar to a patient with *PML::RARA APL*, but the *ZBTB16::RARa* is characterized by a distinctive cytomorphology and expression of CD56.
- In the absence of *PML::RARA* gene fusion in suspicion of APML cases, genetic variants which are susceptible to ATRA treatment should also be considered.
- Early recognition of APML cases with t(11;17) is very critical due to ATRA resistance and poor overall survival. Therefore, a combination of morphology analysis, clinical features, cytogenetic and molecular testing is recommended to achieve rapid identification.
- Due to lack of studies based on MRD monitoring for *ZBTB16::RARa* APL genetic variant, the clinical management and disease monitoring by molecular techniques such as RT-PCR and NGS test is very beneficial due to their sensitivity.

Test Results

Result - POSITIVE CLINICALLY RELEVANT VARIANT/S DETECTED in *ZBTB16/RARA* (fusion)			
Gene/AMP Classification ^	**Clinical relevance**	**Therapeutic relevance $**	**Interpretation**
FUSION Variants			
***ZBTB16/RARA* (FUSION)** Total Read depth - 53x			
Tier IB (Variant of strong clinical significance & well documented literature)	Prognostic	NA	Poor prognosis in ATRA resistant APML cases

Gene Fusion Description

ACTIONABLE BIOMARKER DETAILS

***ZBTB16/RARA* (FUSION)**		
Gene Fusion: *ZBTB16:RARA*	**5'Gene** *ZBTB16*	**3'Gene:** *RARA*
Total Read Depth: 53x	**Ensemble Gene ID:** ENST00000335953.9	**Ensemble Gene ID:** ENST00000254066.10
	Exon/Intron: E:3	**Exon/Intron:** E:3
	5'Chromosome Breakpoint (hg38): chr11:114156434:+	**3'Chromosome Breakpoint (hg38):** chr17:40348316:+

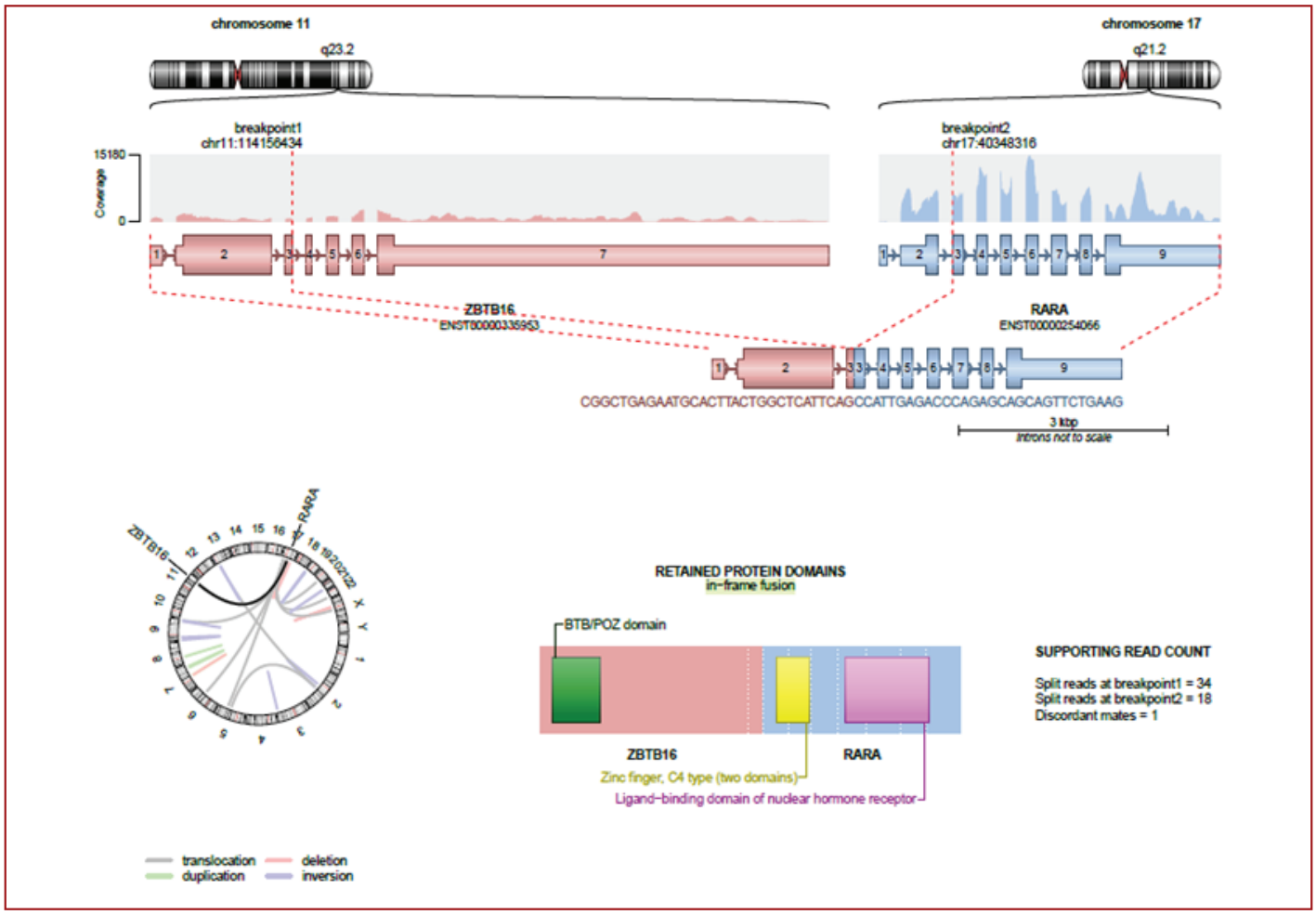

Figure 1. Graphical representation of an in-frame gene fusion occurring at exon 3 of *ZBTB16* of Chr 11 and exon 3 of *RARa* of Chr 17.

Figure 2. Pictorial representation of IGV which confirms the blatting of read sequences from *ZBTB16* gene at Chr11 to *RARa* at Chr 17 and vice- versa.

Complied by: Dr. Ritika Chauhan, Lead Genome Analyst, NGS Hemato-oncology CRT, MedGenome Labs., Ltd.

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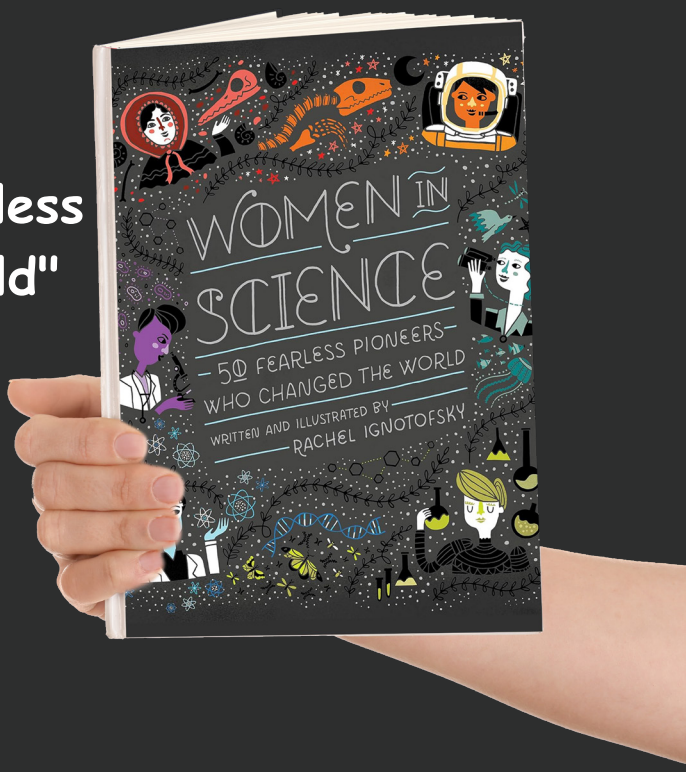
Book Review

"Women in Science: 50 Fearless Pioneers Who Changed the World"



The Unsung Women Heroes of Science

By

Avinash Pradhan, Ph.D
Associate Scientist

Cytogeneticist

This book honors the lives and achievements of women scientists across history, from ancient times to the present. It spans a wide array of scientific fields, such as physics, chemistry, biology, astronomy, mathematics, and computer science. Each profile offers a concise biography of the featured scientist, emphasizing her background, scientific contributions, and influence on the world. Vibrant illustrations accompany the biographies, bringing to life the essence of each scientist's work and character. I would like to highlight the achievements of one of the foremost cytogeneticists, Barbara McClintock.

Barbara McClintock was an American cytogeneticist renowned for her ground breaking work in genetics, particularly her discovery of "jumping genes" or transposons. She was born in 1902 in Connecticut and grew-up in New York City. She loved boxing, riding bikes, and playing baseball. She didn't fit in with the girls, and the boys didn't want to play with her. Against her mother's wishes, but with her father's support, she got a PhD in botany from Cornell University. At Cornell she started her revolutionary work with corn and chromosomes. In 1936, she started working in genetics at the University of Missouri. She was spunky, direct, and much more intelligent than many of her male peers and this made them nervous. The dean threatened to fire her if she ever got married or if her male research partner left the university. Barbara realized they would never give a woman a full-time faculty position, so she quit in order to find her dream job. She got down to business at a research facility in Cold Spring Harbor, New York. Barbara knew that corn was a perfect tool to explore genetics-she was fascinated by corn kernels of different colors growing on the same plant. She planted a field of corn and spent hours gazing at corn cells under a microscope.

She discovered that different colored kernels have the same genes, but they are rearranged in a different order. This meant that a gene could "jump" to a different part of a chromosome and turn on and off. The discovery of jumping genes, or "transposons," explained why there is so much variation in the world and how animals, people, and plants can evolve to react to their environment. Excited by her discovery, Barbara gave a lecture in 1951 at the Cold Spring Harbor symposium, but no one believed her. She didn't mind, because, as she said, "When you know you're right, you don't care." Almost 20 years later, the scientific community caught up with Barbara, and she finally received the recognition due to her. She was awarded a Nobel Prize in 1983, over 30 years after her initial discovery. The span of Barbara's work includes some of the greatest discoveries made in genetics. Her legacy as a pioneering scientist has continued to inspire generations of researchers, and she remains a figure of great importance in the history of genetics.

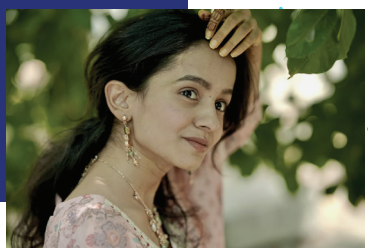
"When You have that joy, you do the right experiments. You let the material tell you, where to go and it tells you at every step what the next has to be."

- Barbara McClintock

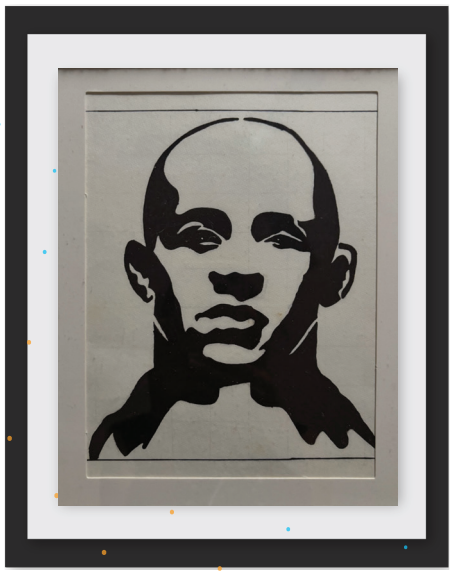
From our Colleagues

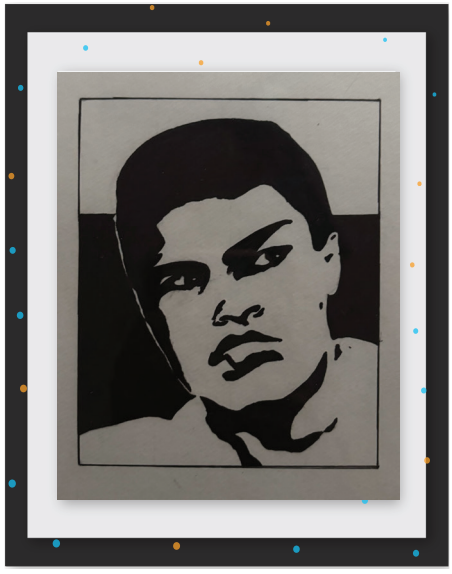
Art meets Science

The most beautiful thing we can experience is the mysterious. It is the source of all true art and science.
— Albert Einstein



By:
Ms. Riya Kulkarni
GA Trainee - Lab Operations





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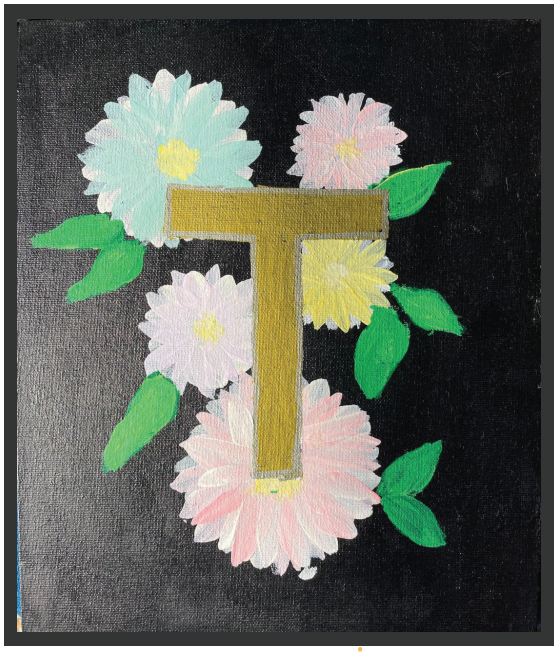
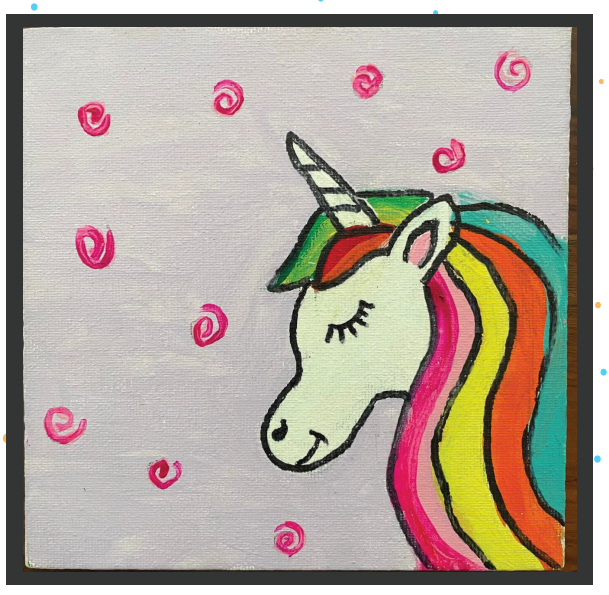
Our employee's little Picasso:)

By:
Trisha, 6 years
DNA of Shalmali Sardesai,
Senior Manager - PR and IR, Corporate





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By:
Arunkumar Ravindran
Research Associate - Lab Operations

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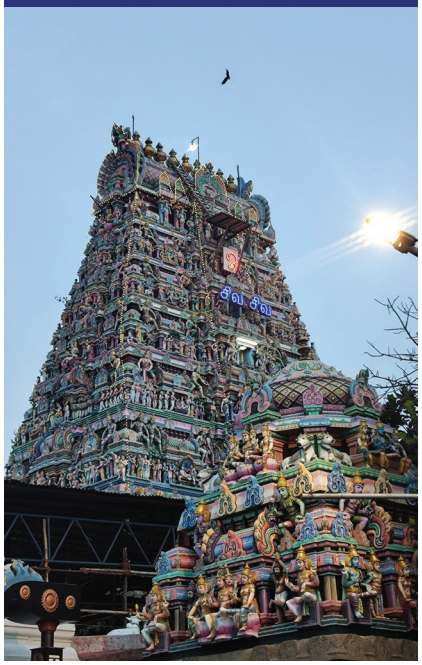
Kerala

Kapaleeshwarar temple
Tamilnadu



Tiruvannamalai temple
Tamilnadu

Shree Sakhi Gopal temple
Odisha

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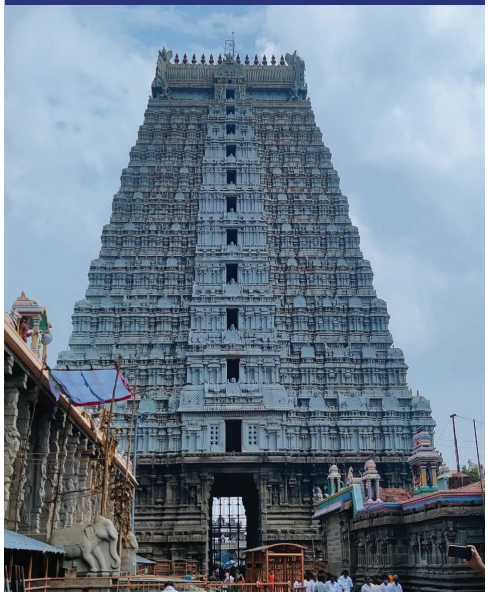
Tamilnadu

Tiruvannamalai temple Main Gopuram
Tamilnadu



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Somireddi Pavan Kumar

Sonu Jain

Sreelakshmy R

Sreelaskhmi Sivan

Srinivasa B T

Sriramula Riya

Subrat Kumar

Suresh R M

Sushmita A

Sweta Nayak

Tamanna Golani

Thotakura Pragna Lakhmi

Vaishnavi D

Vikash Kumar Singh

Vinay Kumar S V

Walter Lawrence

Yash Ninad Tiwarekar

Yashaswini N

Yasheswinee R

Photo Feature

Celebrations of Onam, Diwali & Kannada Rajyotsava

The past few months have been a delightful celebration of festivity, color, and togetherness, as our engagement team and volunteers joined forces to plan and execute vibrant events like Onam, Kannada Rajyotsava, and Diwali. Adorned in their finest attire, surrounded by scrumptious snacks and stunning decorations, everyone basked in an atmosphere brimming with fun and positivity.

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