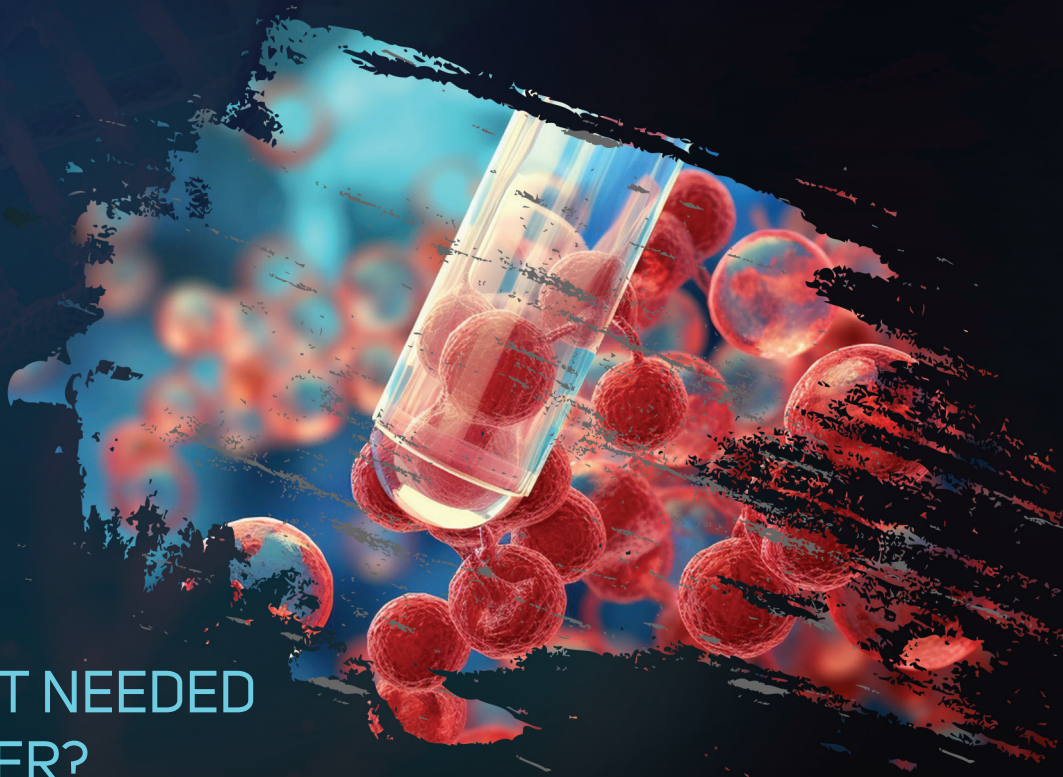
MEDGENOME

GeKNOWme

Internal Quarterly Newsletter Vol 29 | Apr 2024

RARE DISEASES – **GENETIC DIAGNOSIS** AND WAY FORWARD



NGS MRD IN LEUKEMIAS: IS IT NEEDED MORE THAN EVER?

WORDS FROM THE **FRONTLINE**

Gunisha Pasricha, Ph.D
Associate Director, Research Services and Infectious Diseases

It has been four years since I have been associated with MedGenome. What a fulfilling and profound journey it has been! I joined MedGenome in March 2020 and in the same month it was obvious that we were in the midst of the COVID-19 pandemic. The pandemic presented unprecedented challenges which required rapid adaptation and innovation. In response to the urgency of the situation, collaboration across various functions became essential and crucial to delivering results within tight deadlines, often as short as six hours. Additionally, navigating the ever-changing government regulations added another layer of complexity, requiring constant vigilance and adaptation to ensure compliance.

In the pandemic and post it, molecular techniques like RT-PCR and whole genome sequencing became household names and it set tone to expand our portfolio of test for infectious diseases. With the support of excellent MICRA lab team and of course guidance and support of Ram and Sakthi, we have almost 55 tests now. However, much needs to be done in this segment to beat the competition. Apart from adding newer tests, we are exploring use of NGS in the diagnosis of blood stream infections. Clinical metagenomics holds promise as a first-line diagnostic test. However, its clinical implementation still requires further technology refinement, addressing accreditation and regulatory requirements. Recently, there has been a very exciting development in the field of **Drug Resistance (DR)** testing for TB. WHO has formally announced that targeted NGS method could be used for DR testing. This announcement could be game changer for our SPIT SEQ test. We believe the acceptability of this test among the clinicians is going to exponentially increase in the coming months.

Furthermore, I was given an additional responsibility to manage our research services. This role has presented to me as an opportunity to have an excellent learning curve. Project management is an art in its own, although one mantra works best “CUSTOMER is SUPREME”.

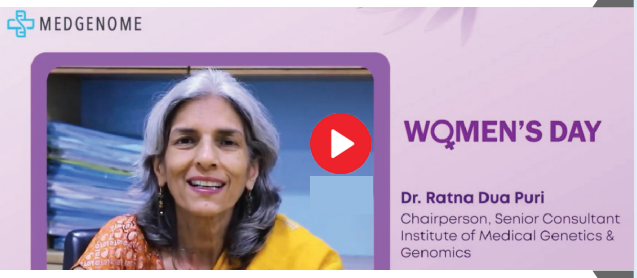
It's been a journey of personal and professional development, where each day brings new opportunities for innovation and collaboration, ultimately leading to a sense of fulfilment and accomplishment.

Contents

MedGenome Connect

REPRODUCTIVE GENOMICS

The previous quarter was centred on campaigns as we actively involved our key doctors nationwide. These engagements occurred both centrally, through corporate-driven campaigns, and locally through various initiatives. A particularly successful corporate campaign was held for Women's Day (8th March), where we collaborated with Key Opinion Leaders (KOLs) across the country's four zones. Our campaign received high praise as we engaged doctors through online polls, articles, video bytes, and a Women's Day video across all our social media channels. Additionally, we sent virtual greetings card to all female clinicians (12,000+). Other than that, KaryoSeq Carousel for social media channels were made to create awareness about the test and and its benefits for clinician. We consistently sent mailers to clinicians featuring information on reproductive genetic tests, and case studies.



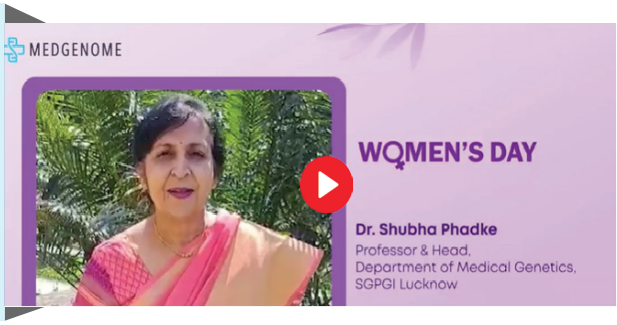
Dr. Ratna Dua Puri, Chairperson, Senior Consultant Institute of Medical Genetics & Genomics, Sir Ganga Ram Hospital, Delhi talks about the important women who motivated her to choose the medical field and later Genomics. She also delves deep into the fascinating world of genetics and highlights that there is nothing a woman can't do.

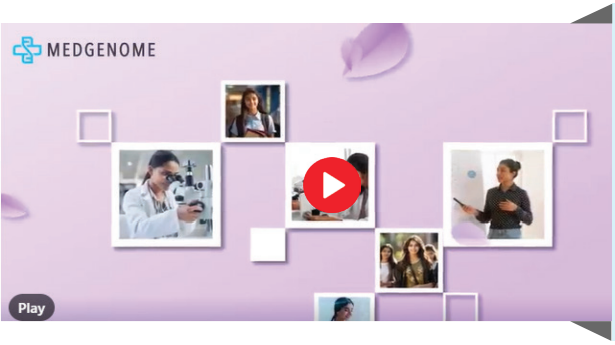
Join Dr. Ratna Dua Puri as she shares her inspiring journey and insights on International Women's Day. Watch this motivational video to learn more about the fascinating world of genetics and how women are shaping the future.

#InternationalWomensDay #WomensDay2024 #Women #Inspiration #WomenEmpowerment #MedGenome #GeneticTesting #Genes #SirGangaramHospital #DrRatnaPuri

Dr. Shubha Phadke, Professor & Head, Department of Medical Genetics, SGPGI Lucknow talks about this era of molecular genetics which is an important step for treatment and genetic counselling and encourages women to choose a career in genetics and increase women power in the field of science.

#InternationalWomensDay #WomensDay2024 #Women #Inspiration #WomenEmpowerment #MedGenome #GeneticTesting #Genes





This #InternationalWomensDay, we honor the brilliant women leading in #STEM fields. From decoding rare diseases to pioneering gene-editing tech, they're at the forefront of shaping the future of medicine. Let's champion diversity and #Inspire-Inclusion together!

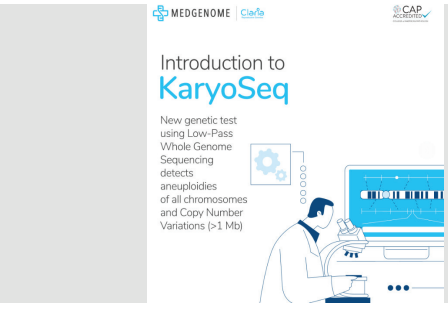
Wishing all Women a Happy Women's Day! Celebrating Women in Genetics & Genomics!

#WomensDay2024 #WomenInSTEM #ScienceForAll #MedGenome #Medicine #GeneticTesting #Genetics #Genes

Discover the crucial genetic tests every #woman should consider for her #health and #wellbeing. From Non-Invasive Prenatal Testing to BRCA mutation screening and beyond, this article dives deep into the world of genetics. Read the blog to unlock vital knowledge and empower your journey towards a healthier future!

https://lnkd.in/grAsnpSE

#MedGenome #Genetics #Healthcare #WomenEmpowerment #womanhealth #genetictesting #geneticcounseling #genetic



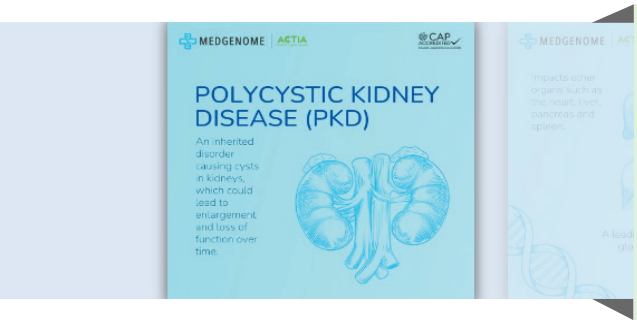
Introducing advanced genetic analysis with KaryoSeq. Our cutting-edge technology utilizes next-generation sequencing to uncover chromosomal abnormalities, aiding in precise diagnosis and personalized treatment decisions for genetic disorders, both prenatally and postnatally.

Discover how KaryoSeq empowers clinicians and patients alike with comprehensive genetic insights. From identifying aneuploidies to detecting translocations and duplications, trust KaryoSeq for accurate and reliable genetic testing.

#MedGenome #KaryoSeq #MedGenomeClaria #GeneticTesting #PrecisionMedicine #PrenatalGeneticTesting #PostnatalGeneticTesting

RARE INHERITED DISEASE GENOMICS

A successful Rare Disease and World Kidney Day campaign were the highlights of the quarter through multiple KOL videos and enlightening webinar. We launched the Whole Genome Sequencing reflex SV by OGM test at AOCN annual conference, which also had many engaging activities such as MedGenome Geneus Genetic Quiz. We also published research paper on diagnostic exome where we speak about the identification of a novel PRKG2 mutation in a proband with Skeletal Dysplasia.



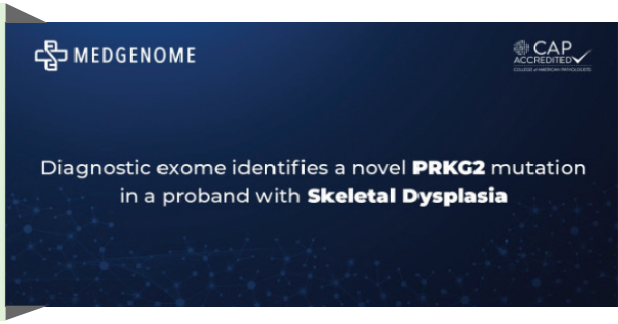
Polycystic Kidney Disease (PKD) is the most prevalent kidney disorder worldwide, impacting 12.4 million individuals, according to the PKD Foundation. Delayed diagnosis can lead to serious complications including kidney failure.

Early detection is key, and genetic testing can play a vital role in identifying PKD early.

#PolycysticKidneyDisease #PKD #InheritedKidneyDiseases #GeneticTesting #KidneyHealth #MedGenome #Healthcare #Genes #Genetics #Genomics

A remarkable collaboration between our team at MEDGENOME and clinical experts at #CMCVellore, unveils the genetic roots behind a puzzling case of skeletal abnormality and #Dwarfism. Despite initial challenges, our dedicated analysts uncovered a compelling loss of function mutation in the cGMP-dependent type II protein kinase (PRKG2) gene.

#Genetics #MedicalResearch #RareDiseases #Collaboration



Dr. Radha Ramadevi - Senior Consultant Paediatrician - Geneticist, Rainbow Children's Hospital, Hyderabad, sheds light on the challenges of identifying rare diseases in this insightful video for Rare Disease Day. She emphasizes the significant diagnostic advancements offered by new age genetic technologies compared to traditional techniques. Watch the video for deeper insights.

#RareDiseaseDay #MedGenome #RareDiseases #GeneticDisorders #GeneticTesting #GeneticDiseases

Dr. Prashant Utage, Senior Consultant in Pediatric Neurology at Utage Child Development Centre, Hyderabad, discusses the significance of karyotyping and the advancements in molecular genetics. Gain deeper insights by watching the video.

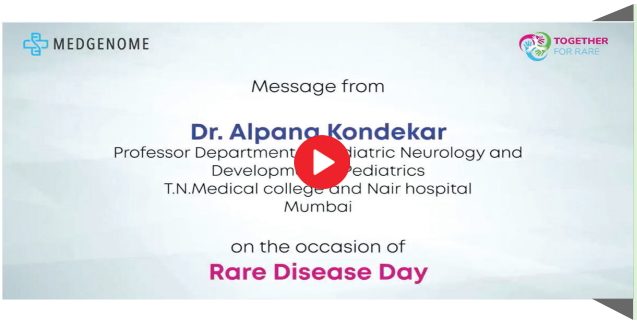
#RareDiseaseDay #MedGenome #RareDiseases #GeneticDisorders #GeneticTesting #GeneticDiseases

This #RareDiseaseDay2024, Dr. CP. Ravi Kumar, Consultant in Pediatric Neurology at Aster CMI Hospital, Bangalore, discusses the benefits of genetic testing in aiding the diagnosis of numerous rare diseases and urges parents to get genetic testing done for their children if they notice any neurological abnormalities.

#RareDiseaseDay #MedGenome #RareDiseases #GeneticDisorders #GeneticTesting #GeneticDiseases #Neurology #NeurologicalDiseases

This #RareDiseaseDay2024, Dr Sandeep Patil, Senior Consultant in Pediatric Neurology, Epilepsy Monitoring, and Surgery at Deenanath Mangeshkar Hospital & Research Center, underscores the significance of genetic testing in precisely diagnosing neurological conditions and complex epilepsies.

#RareDiseaseDay #MedGenome #RareDiseases #GeneticDisorders #GeneticTesting #GeneticDiseases #Epilepsy #Neurology #Research



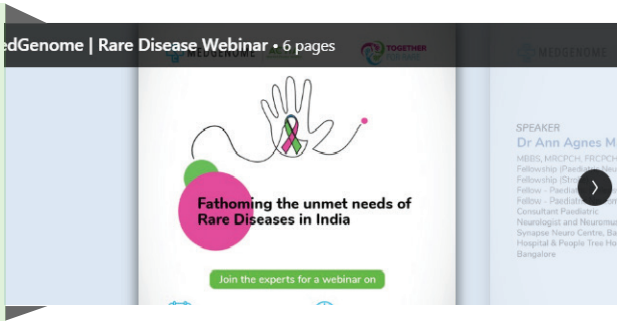
This #RareDiseaseDay2024, Dr Alpana Kondekar, Professor in the Department of Pediatric Neurology and Development Pediatrics at T.N. Medical College and Nair Hospital, Mumbai, emphasizes the significance of Carrier Screening and genetic counseling for both prenatal and postnatal cases.

#RareDiseaseDay #MedGenome #RareDiseases #GeneticDisorders #GeneticTesting #GeneticDiseases

Join us for an enlightening webinar on "Fathoming the Unmet Needs of Rare Diseases in India," which will be held on February 28, 2024 at 7:00 PM. Led by a distinguished panel of experts, this session promises to delve deep into the challenges and opportunities surrounding rare diseases in India.

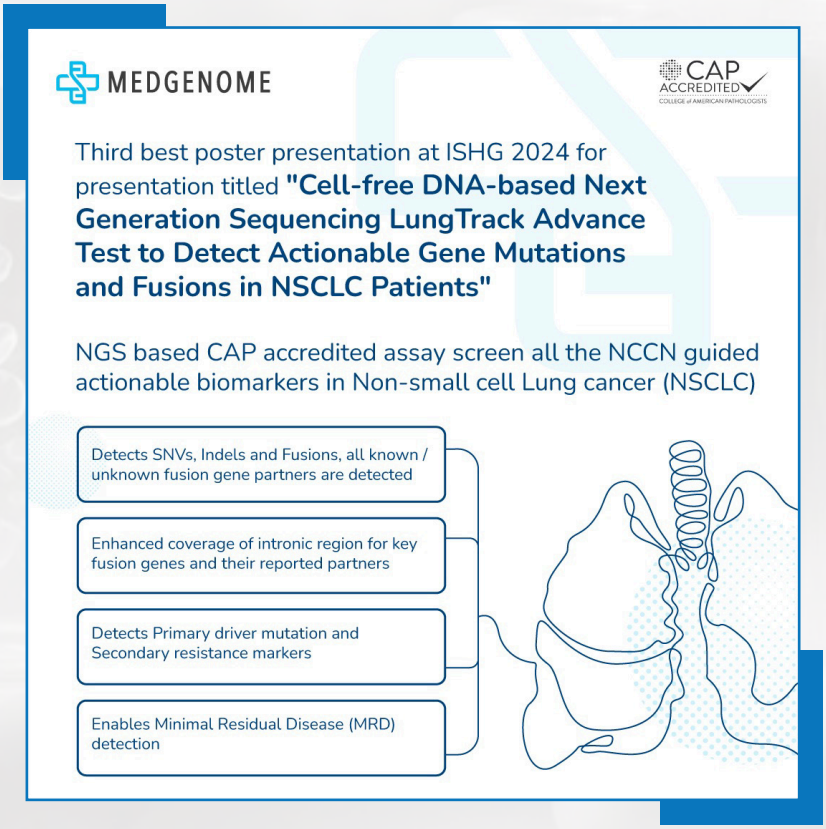
Dr Ann Agnes Mathew Sunita Bijarnia-Mahay Dr. Meenal Agarwal (MD, DM) Dr. Anup Rawool

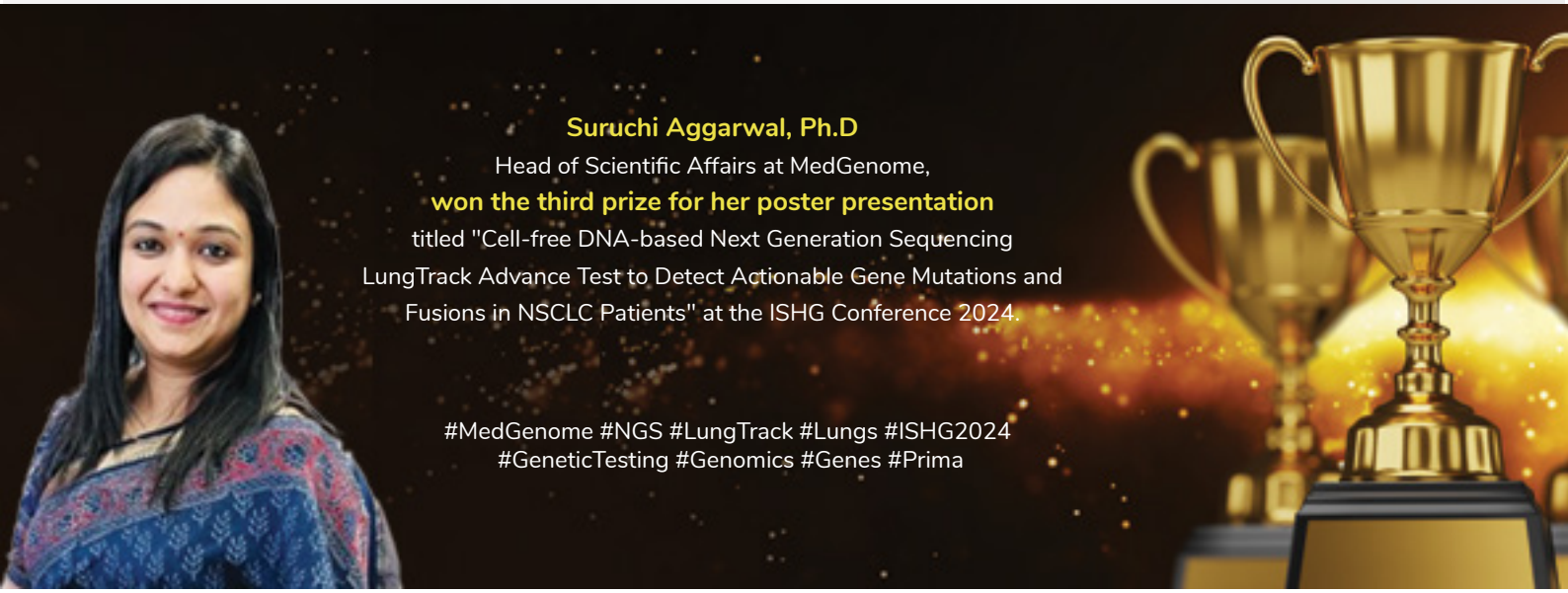
#RareDiseases #MedGenome #RareDiseases #Webinar #RareDiseaseAwareness #ExpertPanel



CANCER GENOMICS

Last quarter, the campaigns were focussed on liquid biopsy. We had a series of insightful case studies, webinar and CMEs on different topics of cancer genetics. We also presented a poster on liquid biopsy test at ISHG conference Ahmedabad which won the 3rd prize.





INFECTIOUS DISEASE GENETICS

The last quarter was very encouraging and has been highly active with a focus to engage more clinicians with CME, conference participation, and test specific mailers along with social media posts. We have also completed the World TB Day campaign in the month of March successfully. We posted various posts (Testimonials videos of Dr. Sameer Bansal, Dr. Lakshmi V Krishna, and Dr. Gunisha Pasricha and awareness video of SPIT SEQ) on all social media platforms.
The team is also working on various new webpages like Sepsis AMR Panel, CMV Drug Resistance testing for the website.

Participation at CME-Aster Hospital at Bangalore

World TB Day - Dr. Sameer Bansal

Dr. Sameer Bansal, Senior Consultant specializing in MD (Pulmonary Medicine), Apollo Hospitals, addresses the challenges posed by Drug-Resistant Tuberculosis (DR-TB) and emphasizes the shortcomings of conventional testing methods for DR-TB. He underscores the significance of rapid diagnostics in tackling this issue.

#WorldTuberculosisDay #Tuberculosis #DrugResistantTuberculosis #EndTB #AccurateDiagnosis #MedGenome #SPITSEQ

World TB Day - Dr. Lakshmi Krishna

Dr Lakshmi Krishna V, Consultant Neurologist at Manipal Hospitals, Bangalore, discusses advancements in tuberculosis detection technologies on the occasion of World TB Day. She emphasizes the efficacy of Next Generation Whole Genome Sequencing, which can identify sensitivity to over 18 drugs, facilitating prompt and precise treatment.

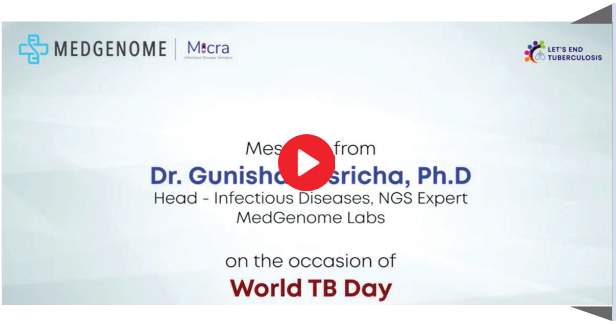
#WorldTuberculosisDay #Tuberculosis #EndTB #AccurateDiagnosis #MedGenome #SPITSEQ

World TB Day - SPIT SEQ

Today, on World TB Day, let's shed light on a pressing global issue: TB remains a significant health threat worldwide. According to WHO, India is one of the top 3 countries with 27% of MultiDrug Resistant-TB cases? It's time to take action!

SPIT SEQ test, an NGS - based Whole Genome Sequencing test can accurately diagnose MDR-TB.

#WorldTBDay #EndTB #Tuberculosis #MedGenome #NGS #TuberculosisDay #PersonalizedTreatment #Micra



World TB Day - Dr. Gunisha Pasricha

Dr. Gunisha Pasricha, Ph.D, Head of Infectious Diseases and NGS Expert at MedGenome Labs, elaborates on strategies to combat tuberculosis and underscores the significance of SPIT SEQ, a rapid test for drug-resistant TB (DR-TB) that delivers reports on sensitivity to over 18 drugs.

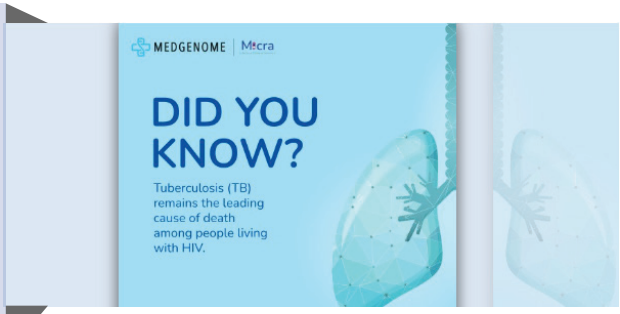
#WorldTuberculosisDay #Tuberculosis #EndTB #AccurateDiagnosis #MedGenome #SPITSEQ

Social Media Post on SPIT SEQ

Did you know? TB is the leading cause of mortality in people living with HIV, making them 18 times more susceptible. India faces a dual burden of HIV and DR-TB, posing a global health challenge.

SPIT SEQ - an NGS-based Whole Genome Sequencing Test by MedGenome - provides precise detection of Drug-Resistant Tuberculosis, especially crucial for HIV-positive patients.

#MedGenome #SPITSEQ #NGS #GlobalHealth #Micra #MDRTB #Tuberculosis #TB #HIV #DRTB



GENOMIC WELLNESS

GENESSENSE
Powered by MEDGENOME

We continued to evolve the various tests being offered under the Genessense portfolio in this quarter. We focused on forging partnerships with a few aggregators & conversations were initiated with some of the major players in this space. We are also revamping our Genessense website with with new pages on individual tests, about us, partners, blogs along with scientific insights. We have worked on various tests brochures such as Genessense premium and Genessense advanced. We have also posted Kardiogen video for no smoking day on all social media platforms.

No Smoking Day - Kardiogen Social Media Post

As per WHO, Tobacco is responsible for 20% of fatalities from coronary heart disease. While tobacco smokers are at higher risk of heart diseases, quitting smoking can drastically reduce the risk.

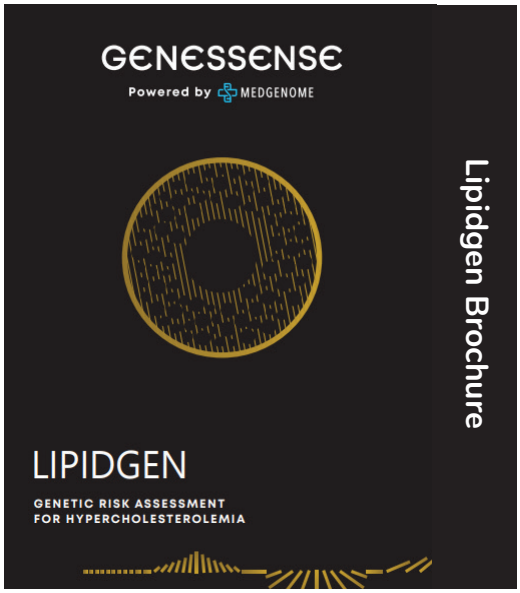
Empower yourself to take charge of your well-being and lower the risk of CAD (Coronary Artery Disease) by making the conscious decision to quit smoking.

Take the first step towards a healthier heart by booking a KardioGen Screening with us. This will assess your risk of heart disease, enabling you to make informed decisions for a healthier future.

#NoSmokingDay #QuitSmoking #HealthyHeart #KardioGenScreening #KardioGen #Genessense #Genetics #GeneticTesting #Genes

Genessense Premium Brochure

Genessense Advanced Brochure



Lipidgen Brochure

Lipidgen Report

What's new

Research publications

Optimization and Validation of a Harmonized Protocol for Generating Therapeutic-Grade Dendritic Cells in a Randomized Phase II Clinical Trial, Using Two Varied Antigenic Sources.

Journal : *Vaccines* Read more

Acute lymphoblastic leukemia with myeloid mutations is a high-risk disease associated with clonal hematopoiesis

Journal : *Blood Cancer Discovery* Read more

Diagnostic exome identifies a novel PRKG2 mutation in a proband with skeletal dysplasia

Journal : *Clinical Genetics* Read more

Comprehensive germline profiling of patients with breast cancer: initial experience from a Familial Cancer Clinic

Journal : *Ecancermedicalscience* Read more

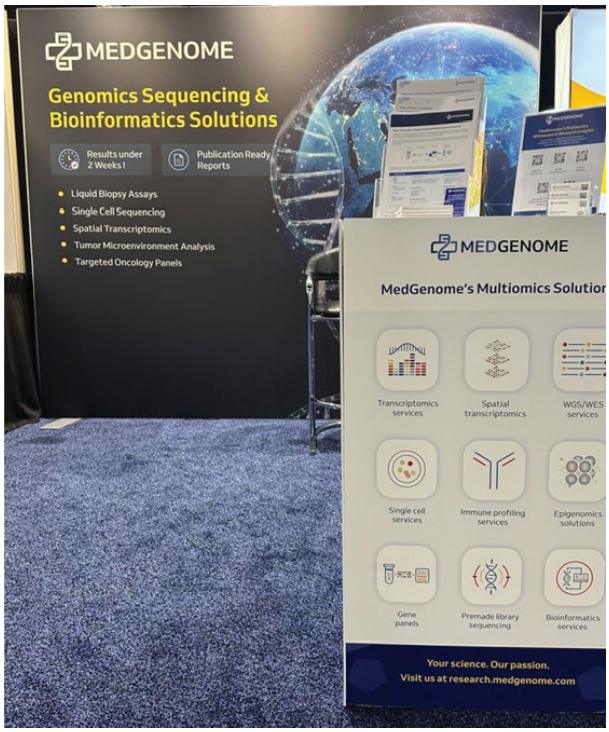
Tests launched

- LipidGen
- Pan-TRK IHC BY VENTANA
- Lymphoma Fish panel (*BCL2, BCL6, MYC -BA*)
- HLA Donor Specific Antibodies, IgG (Class- I & II)
- *GBA* gene analysis by amplicon sequencing
- Monogenic Parkinson Disease Panel by NGS & MLPA

Proud moment

Surajit Chakrabartty, CFO, MedGenome received the **CFO100 2024 Roll of Honour** under the category 'Winning Edge' in Strategy Execution at the 14th Annual CFO100 2024 event in Mumbai. Our sincere gratitude to CFO Collective and the jury for recognising our continued efforts in making a difference in the healthcare ecosystem.

From Our US Office



We started the new year by introducing new service lines like single - cell flex and spatial transcriptomics, demonstrating our ability to handle diverse sample types and design experiments to accelerate research. At prominent conferences such as AGBT and TAGC, we engaged with industry leaders, highlighting our novel solutions and MedGenome's long-read sequencing capabilities. The response to our *De novo* genome assembly and annotation grant program, in partnership with PacBio, was exceptionally positive. Our recent attendance was at the AACR conference where we showcased our new offering spatial transcriptomics, alongside other cutting-edge cancer genomics offerings: single cell, immune profiling, liquid biopsy service and more.

We're happy to share that MedGenome, Inc. has received the **Best of Foster City Award** for six consecutive years, earning us a spot in the 2024 Foster City Business Hall of Fame.

Our blog is constantly evolving with fresh and informative articles to keep you updated. Explore our latest blog articles at https://research.medgenome.com/blog/.

Our recent articles cover a range of topics including:

1.Transcriptome sequencing to uncover gene expression signatures and disease biomarkers
2. From Single Cells to Spatial Landscapes: Unraveling Gene Expression with 10x Flex and Visium
3. Empowering Prevention: Genomic Insights for National Cancer Prevention Month
4. From Sequences to Solutions: Exploring Colorectal Cancer Research & Treatment
5. Unveiling the Complexity of Head and Neck Squamous Cell Carcinoma: From Genes to Microenvironment

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

Rare Diseases – Genetic Diagnosis and Way Forward

Thenral SG, Ph.D
Principal Scientist

Rare diseases affect millions of people worldwide. The definition of rare diseases differ by country or region as does their policy landscape. Though individually rare, there is an estimated 7,000+ rare diseases which affects 3.5–5.9% of the world's population i.e., ~300 million people worldwide. The incidence varies by race, ethnicity, and geography; recessive disorders are higher in communities with consanguineous marriages.

Over the past decades, rare diseases have progressively been acknowledged as an important public health issue, greatly impacting the lives of people living with such conditions, their families and caregivers, healthcare systems and society. The Orphanet includes more than 7,800 disease-gene relationships. Data on how many people suffer from different diseases that are considered rare globally, is lacking in India. According to Indian Organization for Rare Diseases (IORD) so far only about 450 diseases have been recorded in India from tertiary care hospitals.

Many inherited rare diseases are caused due to an aberration in a single gene. The mutated gene perturbs the normal biological processes resulting in chronic or severe or sometimes life-threatening forms of the disease. The causes of several rare diseases yet remain unknown, with only symptomatic treatment available at times. The diagnostic odyssey for patients can vary from months to decades with an average time of 4-5 years, including visiting several specialists and undergoing extensive and often expensive work-ups.

Use of NGS methodologies in detection of rare diseases

Until 10 years ago, genetic testing was expensive and usually limited to a few genes at a time. The advent of next-generation sequencing (NGS) technology has had a dramatic effect on the cost, accuracy, and utility of genetic testing. Accurate diagnosis can help in better management of the disease. Tailored treatment options where possible may be offered avoiding unnecessary investigations. Identification of the causal variant also informs patients about the risk of passing the disease to future generations and to make informed reproductive decisions.

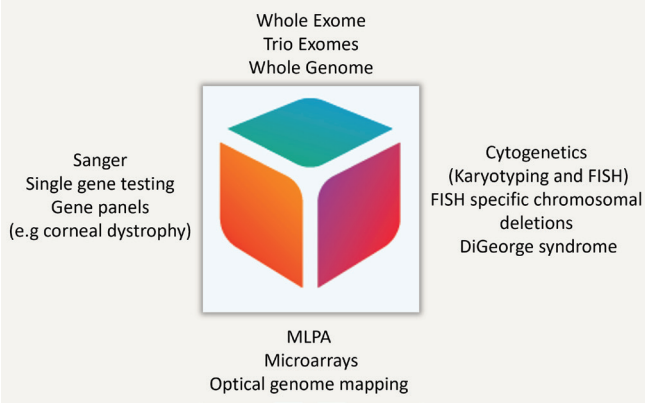
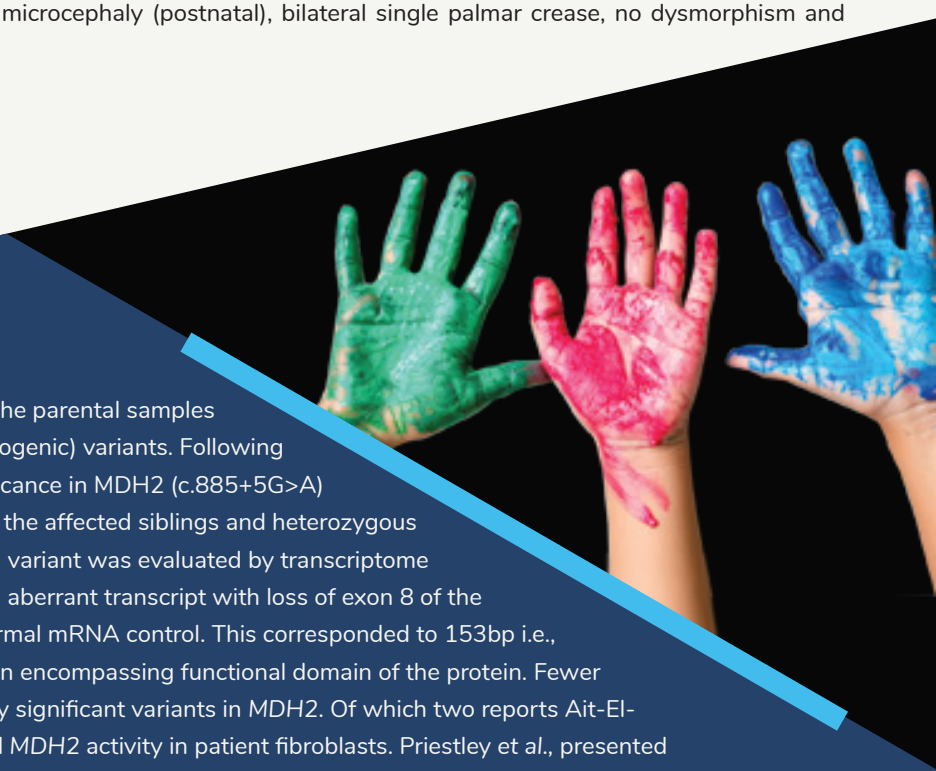


Figure 1: Genetic testing

Case study

First girl child of consanguineously (second degree) married couple was born at term of uncomplicated pregnancy with 3 kg birth weight, cried immediately after birth. She presented in the early neonatal period with seizures on fifth day of life requiring two antiseizure medications for control. At 4th month of age she started having epileptic spasms which were refractory to Steroids, Vigabatrin and Ketogenic diet. She had severe global developmental delay with refractory polymorphic epilepsy, pseudobulbar dysfunction, cortical visual impairment and microcephaly. Spasticity was noted in all four limbs without neck control. No obvious high-risk factors were noted in the mother or any adverse events during the delivery. MRI brain done at day 7 of life showed cystic changes in bilateral basal ganglia. Arterial lactate done later at 2 years was 3.5mmol/l Child died at age of 3 years due to refractory seizures and aspiration pneumonia.

The second, a male child was born at term of uncomplicated pregnancy with a birth weight of 2.7 kg, cried immediately after birth. During this pregnancy, the obstetrician had noticed abnormalities in the basal ganglia and had advised fetal MRI at 26 weeks. Fetal MRI brain demonstrated signal changes with early cystic changes in bilateral lentiform nucleus. On comparing with the elder sibling's (child 1) MRI, similar cystic changes were noted in lentiform nucleus along with extensive white matter cystic changes. Hence, autosomal recessive genetic disorder was considered. As the pregnancy was advanced, alternate options could not be considered. Child had seizures on first day of life. MRI brain (4 days of life) showed well defined cystic lesions. He had recurrence of seizures from 2nd month, requiring multiple anti-epileptics. At 3 months he developed epileptic spasms requiring multiple medications (Steroids, Vigabatrin, Lacosamide) without complete cessation of spasms. He also had severe global developmental delay like his elder sibling. On examination he had microcephaly (postnatal), bilateral single palmar crease, no dysmorphism and central hypotonia with dystonia..



The whole exome sequencing on both children and the parental samples did not detect any significant (pathogenic/likely pathogenic) variants. Following analysis, a splice proximal variant of uncertain significance in MDH2 (c.885+5G>A) was identified. The variant was homozygous in both the affected siblings and heterozygous in the unaffected parents. The effect of the observed variant was evaluated by transcriptome sequencing of the blood cells. The variant resulted in aberrant transcript with loss of exon 8 of the gene evidently not expressed when compared to normal mRNA control. This corresponded to 153bp i.e., deletion of 51 amino acids leading to inframe deletion encompassing functional domain of the protein. Fewer than 20 individuals have been reported with clinically significant variants in *MDH2*. Of which two reports Ait-El-Mkadem S et *al*., and Ticci et *al*. have shown reduced *MDH2* activity in patient fibroblasts. Priestley et *al*., presented clinical characteristics of seven patients with discernable biochemical signature including urinary excretion of malate and subependymal cysts similar to those observed in our proband causing early-onset encephalopathy. This is likely the first report from India for this rare form of epileptic encephalopathy. This case demonstrates the rarity of the disease gene, genetic and phenotypic heterogeneity and challenges faced in the confirmation of variants of uncertain significance.

Figure 2: Shows the comparative IGV of the RNA sequencing data, where the exon 8 of the *MDH2* gene is not expressed in the affected and is expressed in the normal asymptomatic individual

Epileptic encephalopathies (EIEE) are severe forms of childhood epilepsy characterized by refractory seizures and developmental delay accompanied by other comorbidities. EIEE has been associated with ~200 genes; identifying the cause with seizure control in the first 2 years of life is critical to manage the neurodevelopmental outcomes. Resistance to anti-seizure medications is one of the major concerns in the treatment of epilepsy. Thus, early genetic testing and identification of the disease causal gene with clear pathogenicity can inform the most appropriate treatment option/s. For example, patients with mutations in *KCNQ2*, *SCN2A*, and *SCN8A* can benefit from sodium channel blockers, while the same are avoided in those with *SCN1A* mutations; mTORopathies are managed using mTOR inhibitors etc.

Role in treatment and management

Genomic testing is now becoming a routine tool for diagnosing rare childhood genetic disease. Coupled with extensive knowledge gained from biomedical research, improved disease management strategies are now available for rare diseases, especially for paediatric patients in the neonatal or paediatric intensive care unit. There are ~251 rare diseases that have been suggested by Pierre *et al* wherein delayed diagnosis could be detrimental. Though only limited curative therapies are currently available for rare diseases, several treatment/management strategies based on the genetic diagnosis have become accessible.

A few use case scenarios are mentioned in the figure below:

Dietary management

~95 diseases

e.g. dietary galactose management is effective in galactose metabolism

Hematopoietic stem cell transplantation

~157 diseases

e.g. severe combined immunodeficiency, thalassemia, and congenital amegakaryocytic thrombocytopenia

Enzyme replacement therapy

~ 28 disorders

e.g. Phenylketonuria, Glycogen storage disease, Gaucher disease, etc.

Supplements

~175 disorders

Minerals (ZnSO4 NaCl, KCl, MgCl,CaCl, NaHCO3, Na3PO4, CuCl2, MnSO4), vitamins (vitamin K,vitamin D, vitamin A, vitamin E, pantothenic acid, pyridoxine, riboflavin, cobalamin, folate), sugars (galactose, fucose,mannose), and other natural products (creatine, phosphatidylcho-line, ornithine etc).

Immunoglobulin therapy

42 immunodeficiencies with agammaglobulinemia, hypogammaglobulinemia, or dysgammaglobulinemia

Gene therapy

Examples: beta-thalassemia, spinalmuscular atrophy, adenosine deaminase deficiency, and RPE65-related Leber congenital amaurosis

Solid organ transplantation

~18 disorders

Liver, kidney, Heart, Thymus transplantation

Medications, vaccinations

~ 328 disorders

For disease management, most are specific some are general like the antibiotics.

Reference: Content adapted from Bick D *et al.*, An online compendium of treatable genetic disorders. Am J Med Genet C Semin Med Genet. 2021 Mar;187(1):48-54.

Figure 3: Treatment and management of rare diseases.

Conclusion

Most rare diseases require lifetime treatment and support for the patient to maintain basic quality of life. The clinical diagnosis sometimes remains elusive even for experts, in spite of the increasing molecular technologies and scientific knowledge. Several reasons can be attributed for the same: identification of new gene-phenotypes, lack of awareness of the disease, limited access to genomics services, lack of resouces at medical centers etc. Despite these challenges, the emergence of new technologies and constant innovation in treatment and management modalities does hold promise and hope for patients with rare monogenic disorders.

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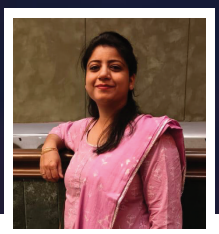
Acknowledgement: Thanks to Dr. Lokesh Lingappa for his critical inputs and for the detailed clinical discussion of the case.

NGS MRD in leukemias: Is it needed more than ever?



Ritika Chauhan, Ph.D
Lead Genome Analyst

With an evolving therapeutic treatment landscape for leukemias, such as Acute Myeloid Leukemia (AML), Acute Lymphoblastic Leukemia (ALL), and Chronic Myeloid Leukemia (CML), it is very crucial to monitor the treatment response for relapse as well as for progression free survival. The patients who achieve complete hematologic remission after the treatment of blood cancer often harbor residual leukemic cells in bone marrow or peripheral blood that can result in relapse. It is important to understand that these cells can be present at a very low level and cannot be detected by conventional methods. Both treatment decisions and prognosis are dependent on patient disease status which requires accurate assessment of these residual leukemic cells, which can be achieved by Minimal Residual Disease (MRD) assessment. MRD is considered as a prognostic and predictive approach to monitor disease progression and is used as an end point to monitor disease status and evaluate new treatments in leukemia.

What is MRD and how MRD testing can affect treatment?

Minimal residual disease (MRD) is the term that refers to describing the small number of cancer cells which remain in the body even after treatment. These small numbers of cells may not cause any physical signs or symptoms at the time of treatment but can remain dormant for a long time and can become active or multiply resulting in relapse of the disease. The remaining cancer cells being dormant could be due to the possibility of not all the cancer cells responsive to the therapy and the cancer cells becoming resistant to the drugs used in the treatment (Figure 1). An MRD positive test means that disease was still detected after the treatment, and an MRD negative test means that no disease was detected after the treatment.

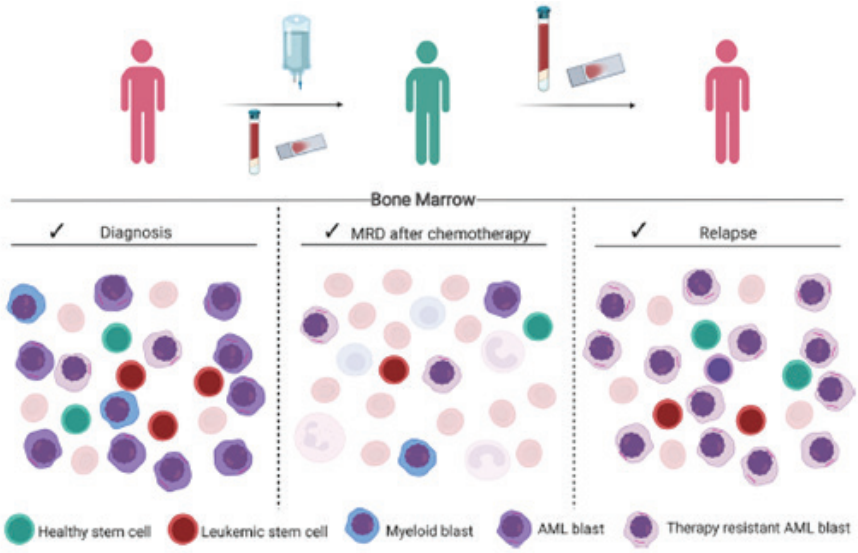


Figure 1. Residual leukemic cells observed after treatment in terms of relapse even after successful induction and complete remission. (Adapted from Ngai LL et. al., 2020)

The clinicians prefer MRD testing to measure the effectiveness of treatment for risk stratification which helps them to categorize their patients as high-risk patients or low-risk patients. MRD testing provides an insight for how well the cancer cells have responded to treatment, to confirm and monitor remissions, patients who may need to restart the treatment, and an early detection of relapse. It also helps them to take decisions on patients who may benefit from other treatments, such as stem cell transplantation or combination therapy.

Approaches for MRD testing

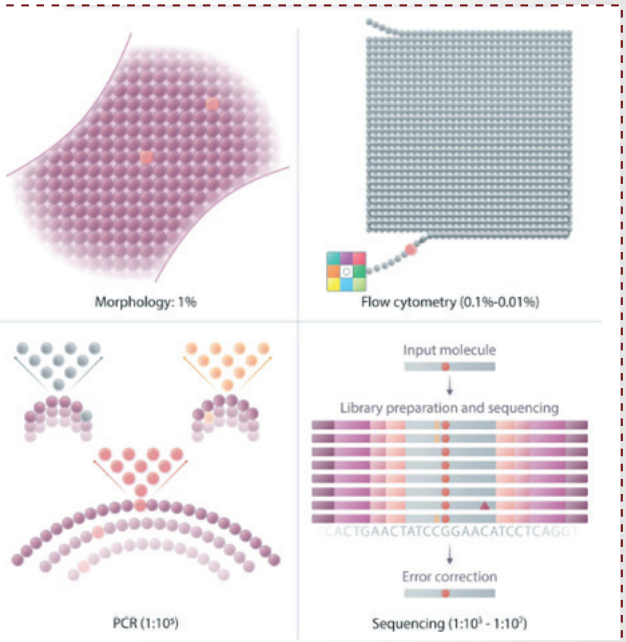


Figure 2. Sensitivity of different assays to test MRD based on available technology. (Adapted from James S Blachly et, al., 2022)

Currently, there are three methods to diagnose and monitor MRD through phenotypic marker patterns or differential gene patterns by multiparameter flow cytometry (FCM), quantitative polymerase chain reaction (PCR), Next generation sequencing (NGS) and their valuable points have been described in Table 1. MRD testing uses highly sensitive approaches, and the different MRD techniques can be expressed in several ways by reflecting the number of cancer cells that can be detected in a sample per number nucleated cells (Table 2).

The sensitivity of these assays plays an important role in measuring MRD for clinical management of patients. The morphological examination of the marrow metaphase karyotypes, or fluorescent *in-situ* hybridization is limited to a sensitivity of about 1%. The sensitivity of multiparameter flow cytometry with carefully chosen markers is 10-3 or acute myeloid leukemia constituting 0.1% of events. The molecular testing, such as PCR, whether normalized to reference transcript such as ABL1 or quantified absolutely with digital PCR can look deeper, between 10-4 to 10-5, while conventional NGS has an error rate of 0.1%, error corrected technology has improved limit of detection to be better than 10-7 as seen in Figure 2. There are different criteria for when to test for MRD which is based on the factors specific to patient disease, such as after bone marrow transplant, during treatment to confirm the depth of remission, after one year on maintenance therapy, at regular intervals after treatment is completed, or at other specific times. The MRD assessment for leukemia patients depend on the quality of sample, which is very important as at times there is a possibility of hemodilution. Both the sample types: fresh peripheral blood and bone marrow aspirate with 2-5 mL volume is recommended for MRD testing.

Table1. Techniques to detect Minimal Residual Disease (MRD)

Techniques	Flow Cytometry (FC)	Molecular Techniques		
	Multiparameter flow cytometry (MFC)	Polymerase Chain Reaction (PCR)		Next generation sequencing (NGS)
		Real-time quantitative PCR (qPCR)	Digital PCR (dPCR)	
Description	Evaluates individual cells by checking the presence of certain protein markers on cell surface.	Expands or amplifies the specific small amount of DNA or RNA to detect and identify malignant cells based on their characteristic genetic abnormalities- mutations or chromosomal changes.		Error corrected NGS with unique molecular identifiers.
Technique specificity	MFC relies on the detection of the expression of antigens on neoplastic cells compared to normal cells with fluoro-chrome-linked antibodies specific to cancer cell antigens.	PCR is widely used for patients harboring well-defined genetic aberrations- *BCR-ABL1*, *CBFB-MYH11*, *RUNX1-RUNX1T1* gene fusion.		NGS testing rapidly examines stretches of DNA/RNA and detects mutations and other genetic abnormalities.

Application	MFC is applicable to nearly 100% of patients with AML, ALL, CLL and MM.	For molecular MRD testing for hematological malignancies, we need to know that almost 98% of lymphoid malignancies contain clonally rearranged immunoglobulin (Ig) and/or T-cell receptor (TCR) genes (VDJ recombination). There are only 25-30% of cases which are well defined for chromosomal aberrations or mutations.	
Sensitivity	Sensitivity limit is 10-4	Sensitivity limit is 10-5	Sensitivity limit is 10-6
Sampling	Fresh bone marrow aspirate is required for reliable results.	• Fresh or frozen peripheral blood is needed for testing • Baseline sample with detectable disease is required to subsequently characterize the clones that will be analyzed	• Both fresh and frozen samples can be used for NGS MRD testing. • Baseline sample with molecular markers details is mandatory to perform NGS MRD testing
Test timeline	Results are available within a day.	Results are available in several weeks.	Results are available in 12-14 days. FDA has approved NGS MRD testing by Clono - SEQ® Assay for B-ALL, CLL and MM patients.
Limitations	• Technically heterogenous • Variable sensitivity • Difficult to standardize and interpret results	• Only be applied to the patient's harboring mutation • Labor intensive • Expertise for interpretation • Expensive technique	• Only be applied to the patient's harboring mutation . • Labor intensive • Expensive technique • Interpretation requires expertise

Table 2: Sensitivity thresholds for MRD testing

Maximum sensitivity (no. of cancer cells per no. nucleated cells)	Percentage (%)	Sensitivity threshold
1 in 20	5%	
1 in 1,000	0.1%	10-3
1 in 10,000	0.01%	10-4
1 in 100,000	0.001%	10-5
1 in 1,000,000	0.0001%	10-6

NGS MRD in Acute Myeloid Leukemia

Acute Myeloid Leukemia (AML) is an aggressive clonal hematopoietic stem cell malignancy with a 5-year overall survival of 30%. It is a disease characterized by heterogenous biology resulting in varying clinical outcomes including relapse. The high rate of relapse implies the post-treatment low level persistence of residual leukemia cells or clones that are not detected by routine disease monitoring methods. The detection of post-treatment MRD by several different laboratory methods has proven to be potent prognostic tool for long-term disease outcomes and early detection of relapse. Testing for MRD in patients with AML in remission can stratify them into groups with higher and lower risks of relapse and survival. The MRD assessment in AML can be used to monitor and identify impending relapse, a potential surrogate end point for overall survival in clinical trials to accelerate the development of novel treatment strategies, and a prognostic/predictive biomarker to refine risk assessment and inform treatment decision-making.

A diverse array of sensitive molecular methods has been used to detect MRD in AML such as RT-PCR and ddPCR, but NGS is a promising tool for sensitive MRD monitoring and has been used successfully to monitor mutations and chimeric gene fusions. The error-corrected sequencing involves the physical incorporation of random oligonucleotides or unique molecular identifiers (UMI) at library preparation stage prior to amplification of DNA. This helps to tag individual's DNA molecules with a unique molecular fingerprint. As per 2021 ELN guidelines update which states that AML patients who are not included in the molecularly defined subgroups should be monitored for MRD by MFC flowcytometry. The patients testing MRD negative by flowcytometry prior to alloHCT still have a 20% to 30% relapse rate, and deintensification of standard treatment based on an MRD test result should only be attempted cautiously as part of a clinical trial. The ELN recommends molecular MRD testing in AML based on certain criteria and Figure 3 provides an overview to have a better understanding before performing NGS MRD testing in AML patients, and Figure 4 represents the major checkpoints which need to be listed while putting up the results of this assay.

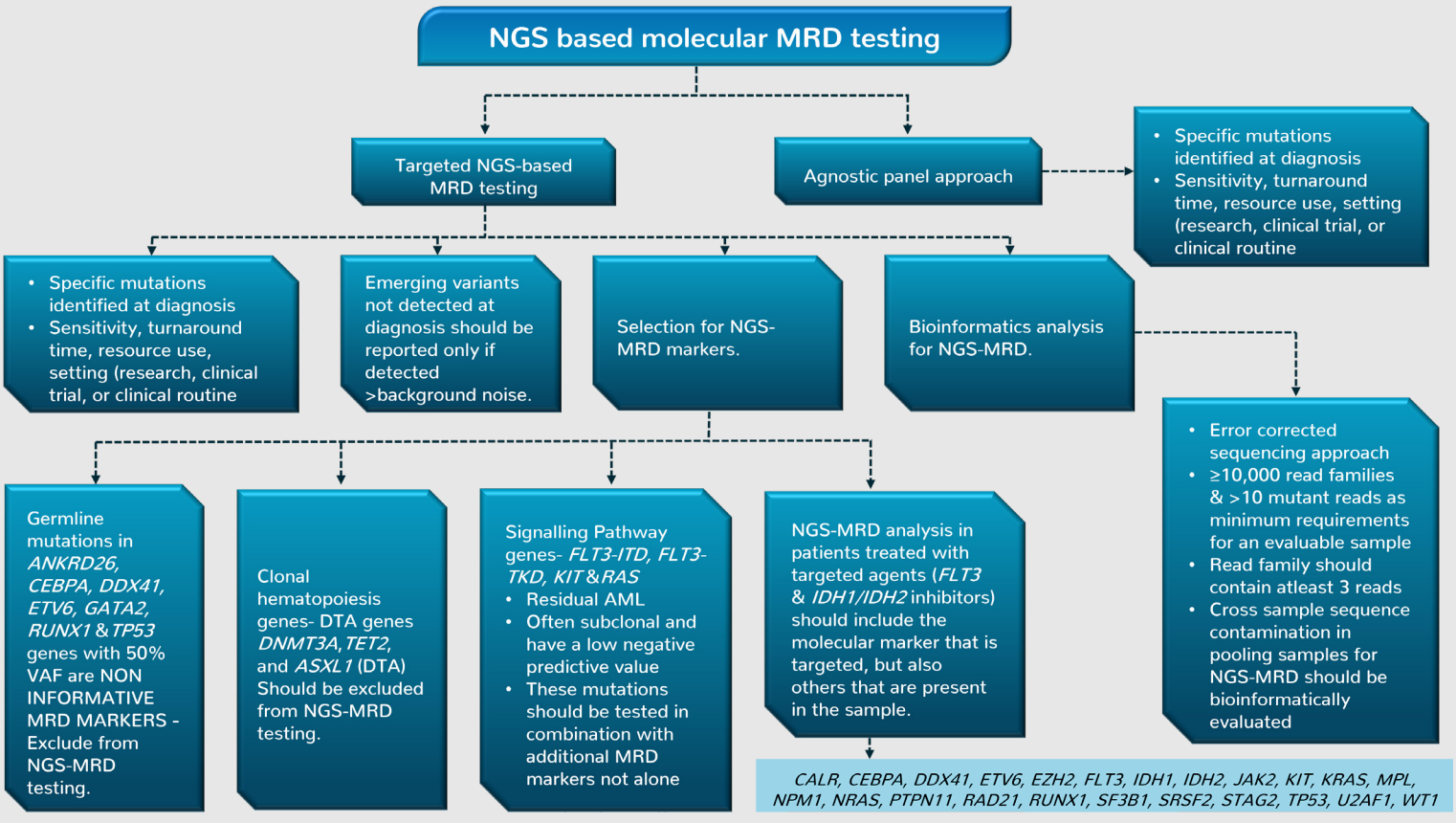


Figure 3. ELN recommendations for NGS based MRD testing in Acute Myeloid Leukemia

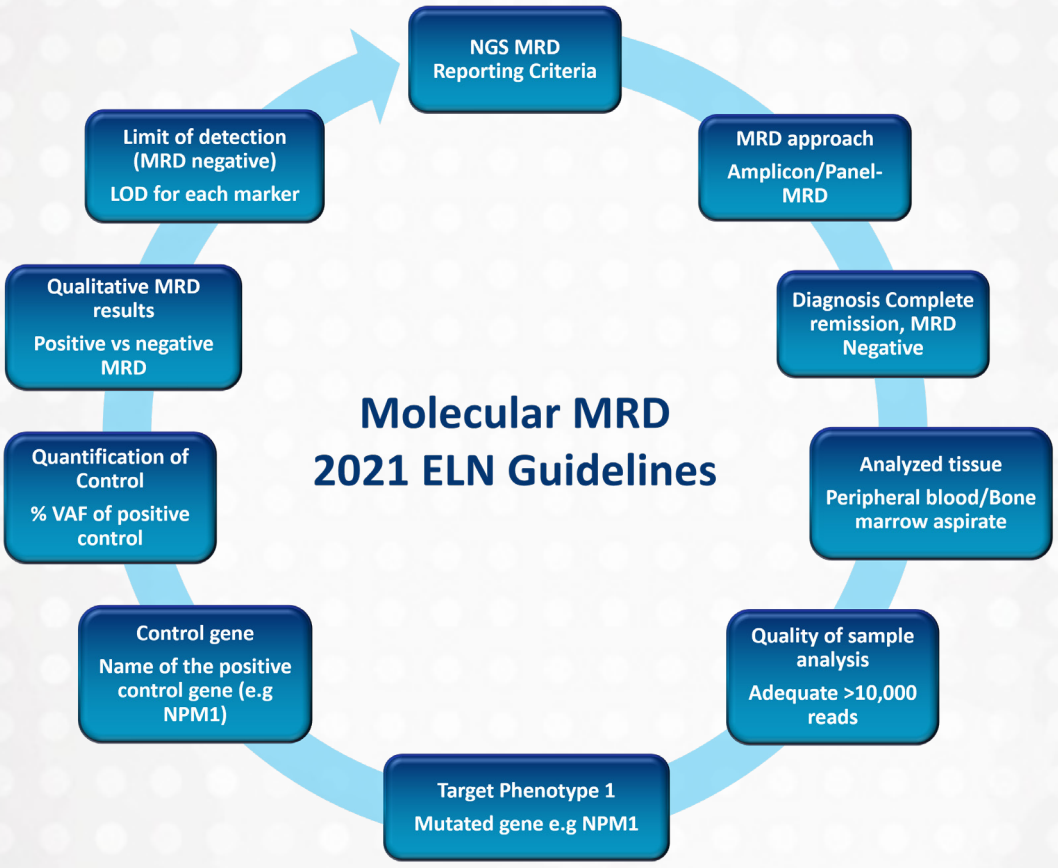
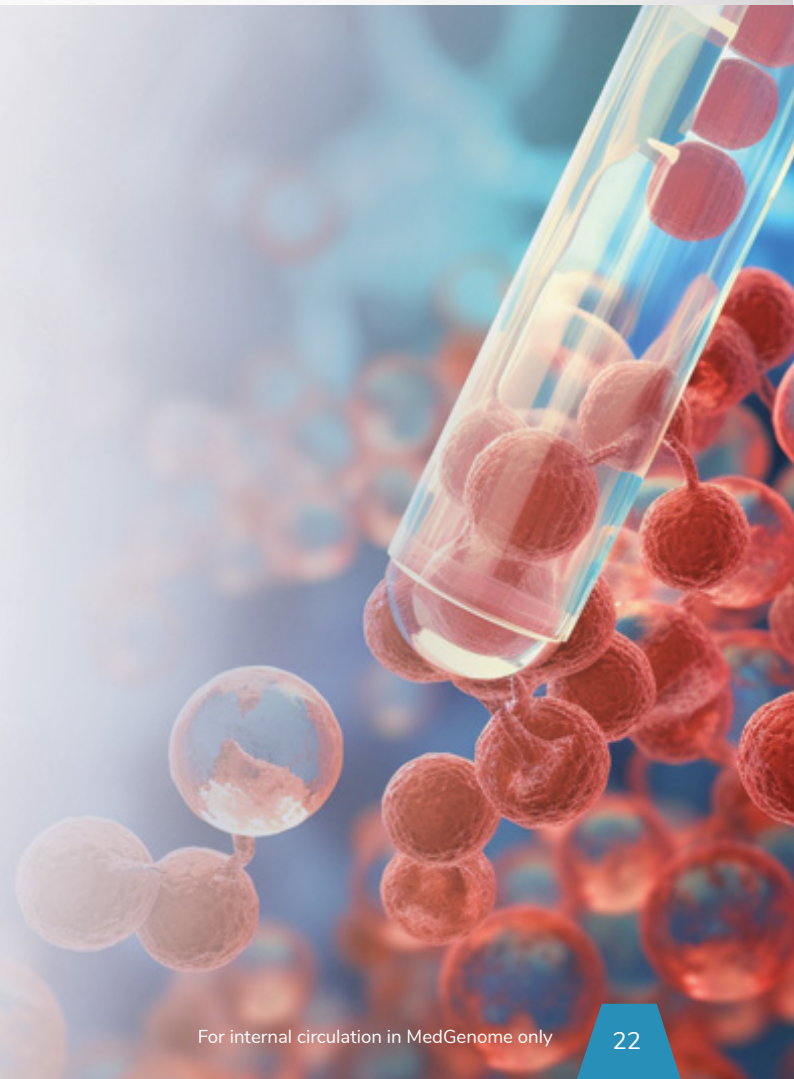


Figure 4. Flowchart explaining the list of elements to be reflected in results for NGS MRD in AML as per ELN guidelines.

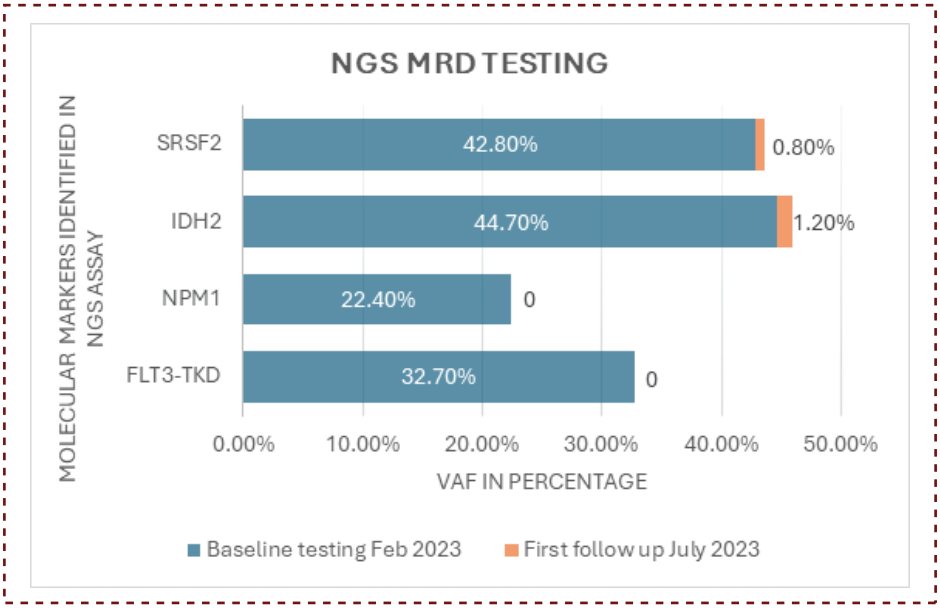
Conclusion

MRD is quickly evolving in terms of biological, technical, and clinical research fields. It is potentially relevant for several clinical decisions before and after the transplant. Among leukemias, AML has consistently been at the forefront of cancer genetics and genomics due to its molecular and immunophenotypic heterogeneity. NGS-based MRD assay could be practically and equivalently applied to all pre and post treatment AML samples. It is not only more analytically sensitive and prognostically relevant than the non-molecular assays which were traditionally used to access the disease burden, but also very practical approach to refine risk assessment and to make early treatment decisions for leukemia patients. Currently, the NGS-based MRD testing is an explorative field, with new and more sensitive assays being investigated.



Case scenario

A 59-year-old AML patient with post chemotherapy was tested in our laboratory for NGS MRD testing with UMI's and was identified to have molecular markers such as FLT3-TKD (D835Y), NPM1 (p.Trp288CysfsTer12), IDH2 (R140Q) and SRSF2 (P95L) in Feb 2023 with a variant allele fraction (VAF) of 32.7%, 22.4%, 44.7% and 42.8% respectively. However, after 5 months NGS MRD testing results indicated the presence of variants in IDH2 (p. Arg140Gln) and SRSF2 (p.Pro95Leu) genes at a variant allele fraction of 1.2% and 0.8% respectively, which helped in early treatment decisions.



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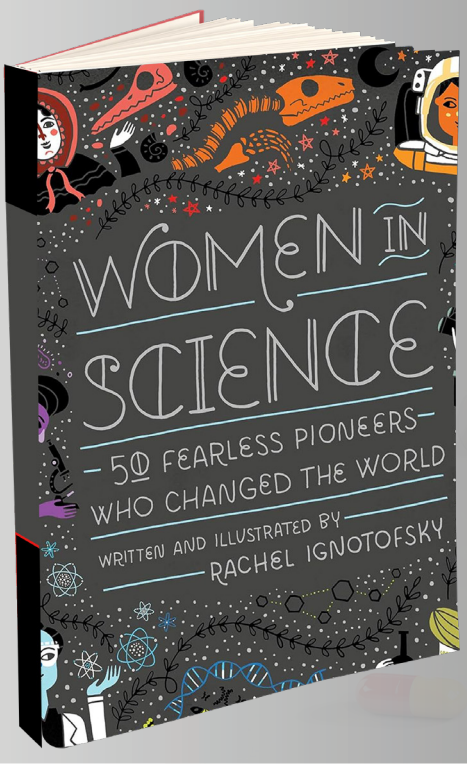

Book

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

Author - Rachel Ignotofsky

Book review by

Avinash Pradhan, Ph.D
Associate Scientist

THE UNSUNG WOMEN HEROES OF SCIENCE

This book celebrates the lives and accomplishments of women scientists throughout history, from ancient times to the present day. It covers a diverse range of scientific disciplines, including physics, chemistry, biology, astronomy, mathematics, and computer science. Each profile in the book provides a brief biography of the featured scientist, highlighting her background, contributions to science, and impact on the world. The biographies are accompanied by vibrant illustrations that capture the essence of each scientist's work and personality. In this book review, we will cover the extraordinary journey of Nettie Maria Stevens.

Nettie Maria Stevens was born on July 7, 1861, in Cavendish, Vermont, to Julia and Ephraim Stevens. After the death of her mother, her father remarried and the family moved to Westford, Massachusetts. She was graduated from Westford Academy in 1880. Stevens taught high school and was a librarian. Her teaching duties included courses in physiology and zoology, as well as mathematics, Latin, and English. Her interest in zoology may have been influenced by taking a teacher training course she attended on Martha's Vineyard in the 1890s. After graduation at the top in her class, she attended Stanford University, where she received her B.A. in 1899 and her M.A. in 1900. She also completed one year of graduate work in physiology under Professor Jenkins and histology and cytology under Professor McFarland. Stevens continued her studies in cytology at Bryn Mawr College, where she obtained her Ph.D. and was influenced by the work of the previous head of the biology department, Edmund Beecher Wilson, and by that of his successor, Thomas Hunt Morgan.

Her work documented processes that were not researched by Wilson and she used subjects that he later would adopt along with the results of her work. After receiving her Ph.D. from Bryn Mawr, Stevens was given an assistantship at the Carnegie Institute of Washington in the year 1904–1905. Several subsequent studies of germ cells in aphids appeared as a result. One paper (1905) won Stevens an award of $1,000 for the best scientific paper written by a woman. Another work, "Studies in Spermatogenesis," highlighted her entry into the increasingly promising focus of sex-determination studies and chromosomal inheritance. It was at this institute that Stevens had her sex determination work published as a report in 1905. At Bryn Mawr, Stevens focused on topics such as the regeneration in primitive multicellular organisms, the structure of single cell organisms, the development of sperm and eggs, germ cells of insects, and cell division in sea urchins and worms. In 1906, she discovered that male beetles produce two kinds of sperm, one with a large chromosome and one with a small chromosome. When the sperm with the large chromosome fertilized eggs, they produced female offspring, and when the sperm with the small chromosome fertilized eggs, they produced male offspring. This pattern was observed in other animals, including humans, and became known as the XY sex-determination system.

Stevens was one of the first American women to be recognized for her contribution to science. Her research was completed at Bryn Mawr College. Her highest rank attained was the associate in experimental morphology (1905–1912). Using observations of insect chromosomes she discovered that, in some species, chromosomes are different among the sexes. The discovery was the first time that observable differences of chromosomes could be linked to an observable difference in physical attributes (i.e., whether an individual is male or female). The experiments completed to determine this used a range of insects. She identified the Y chromosome in the mealworm, Tenebrio. She deduced that the chromosomal basis of sex depended on the presence or absence of the Y chromosome. She successfully expanded the fields of genetics, cytology, and embryology. Stevens failed to gain a full regular university position, however, she achieved a research career at leading marine stations and laboratories. Her record of 38 publications includes several major contributions which further the emergence of ideas of chromosomal heredity. As a result of her research, Stevens provided critical evidence for Mendelian and chromosomal theories of inheritance. Stevens worked to be able to become a full researcher at Bryn Mawr, however, before she could take the research professorship offered to her, she died on May 4, 1912, of breast cancer at Johns Hopkins Hospital. Following her death, Thomas Hunt Morgan wrote an extensive obituary for the journal Science. In an earlier letter of recommendation he wrote, "Of the graduate students that I have had during the last twelve years I have had no one that was as capable and independent in research as Miss Stevens." Studying egg tissue and fertilization process, Stevens was the first to recognize that females have two large sex chromosomes in the shape of Xs and that males have one of full size X and another that is missing a portion, making it resemble a Y. Wilson performed tests only on the testes as eggs were too fatty for his staining procedures. After her discoveries, Wilson reissued his original paper and acknowledged Stevens for this finding. Stevens at Bryn Mawr was breeding Drosophila melanogaster in the laboratory as subjects of her research some years before Morgan adopted it as his model organism. Her career span was short, but she published approximately 40 papers. Nettie Maria Stevens was buried in the Westford, Massachusetts, cemetery alongside the graves of her father, Ephraim, and her sister, Emma. Nettie Maria Stevens passed away on May 4, 1912, at the age of 50, but her legacy continues to inspire scientists and educators in the field of genetics.

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:
Pratiksha Shivaji Bhalerao
Bioinformatics Analyst

Poetry - Rhythmical beauty of words!

The Poem of the Ocean

The poem being recited about her by the ocean ;

Was she reciting the poem to the ocean or
the ocean was reciting a poem to her?
Because how can someone resist deciphering
the beauty of existence that her eyes and soul hold?

After looking at such a pristine and absolute soul
the ocean was mesmerized all at once!
Maybe he will tell the secret to the moon when she is not around
that he witnessed a girl with long dreams and unspoken words.

Moon was curious to know more and then he decribed
"She was sublime visage of tides which falls on rocks...
the shell with oyster which has pearl at heart."

" When the sun was falling on her face
her hair were shiny like golden threads,
eyes were dreamy and the smile was reflecting eternity
which resides in you ,me and in the glorious sun."

"The golden threads were playing with her face...
when the sun was gazing at her
trying to touch the dreams in her eyes....
that blush on the cheeks...and the smile
which was reflection of sanctity on me...
the threads were playing with her while she was just looking at me...
the beauty of her was a reflection on the surface of me...
I was so fortunate the moon to tell u that...
she was just sitting next to me...just sitting next to me..."

And she became a poem of the ocean...

By
Pratiksha Shivaji Bhalerao
Bioinformatics Analyst

Poetry - Rhythmical beauty of words!

If not adore women, respect them,
If not support, understand them,
If not salute, educate them,
If not prioritize, consider them,
Don't win over, but win with them,
It's the minimum one can do,
Never ever bully them, they know
to take a stand

By
Lekhashri Ranjitha
Bioinformatics Analyst

From our Colleagues

By:
Azmat Naseem
Research Associate -
Genomic Medicine

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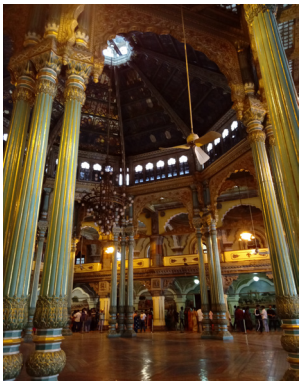
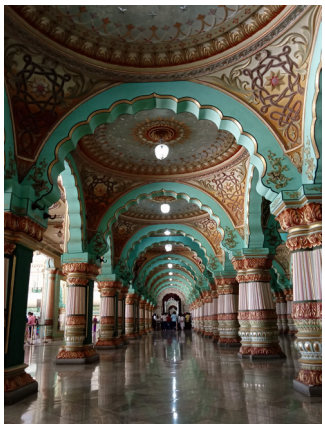
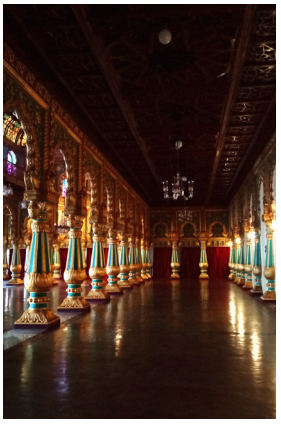




Employee Connect

Our New-Joiners


Manoj R B


Megha Manojan


Manish Tiwari


Bishrul Hafi


Shraddha Yadav


Srithika Mohanrengaraj


Ranjitha R


Sarah Andrea Wilson


Rachel S


M Agaath Hedina


Bharathi Kathirvel


Hemanth Kumar R


Rahul Ram


S R Manikandan


Ashish Joshi


Amit Sharma


Lavanya B


Sherin Raju


Shakib Farooq Dabir


Ashish Kumar


M Ruban

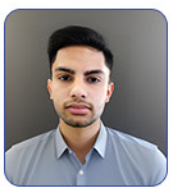

Himanshu Mehta


Rajan Kumar Jha


Usha H R


Soundarya K R


Kamlesh Gopal Mandal


Rohit Kundu


Parichay S R


Nithinson C Sunny


Nilesh Suresh Dhawale


Lenesh Karthikeyan


Palak Sahebrao Patil


Dhiraj Bharali


Neethu Franklin


Naveen P Raju


Akhil M M


Navin C V


Dakshinamoorthy S


Santhanakrishnan Sarathanathan


Ankit Mahajan


Suraj Tomar


Pradeep Kumar


Sabanna


Venkatesh Perumal R


Romina Raulo


Madhusudan B K


Abinaya Madhavan


Ajab Sing Rathor


M Bhanu Shankar


K Balaji


Kathiravan R


Arathi K


Girish S R


V R Hariharan


Srinidhi C


Shiva kumar N


Ashita Omprakash Awasthi


Vishal Kotwal


Moumita Ghosh


Mayank Mishra


Abhilash Nautiyal


Imran Bepari


Sushmakumari


Venkatavaradhan Chakravarthy


Ajay R


Neeraj Joshi


Pallavi Rohan Salvi


Tajamul Sidiq


Rahul Sharma


Sandhya R


Satyajeet Sarkar


Rajeev Ranjan

Tarique Anwar

Sunny Sunil Badgujar

Nilmani

Aswathy K P

Cormaty Shreya Sudarshini

Shubham Kumar

Panshu Dwivedi

Saheli Chakraborty

Saurabh Tiwari

Sachin Raina

G Anbudass

Photo Feature

Makar Sankranti / Women's day Celebrations

Embracing the spirit of Makar Sankranti, the MedGenome team decorated the office with traditional motifs and wore ethnic outfits, filling the environment with warmth and positive festive vibes.

International women's day was celebrated with a token of appreciation card and chocolate for all the lovely ladies of MedGenome. Along with the gift, a few fun filled games were organsied. It was a delight to watch everyone dressed up in the theme color 'Laddoo peela' (Yellow) representing joy and positivity.

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