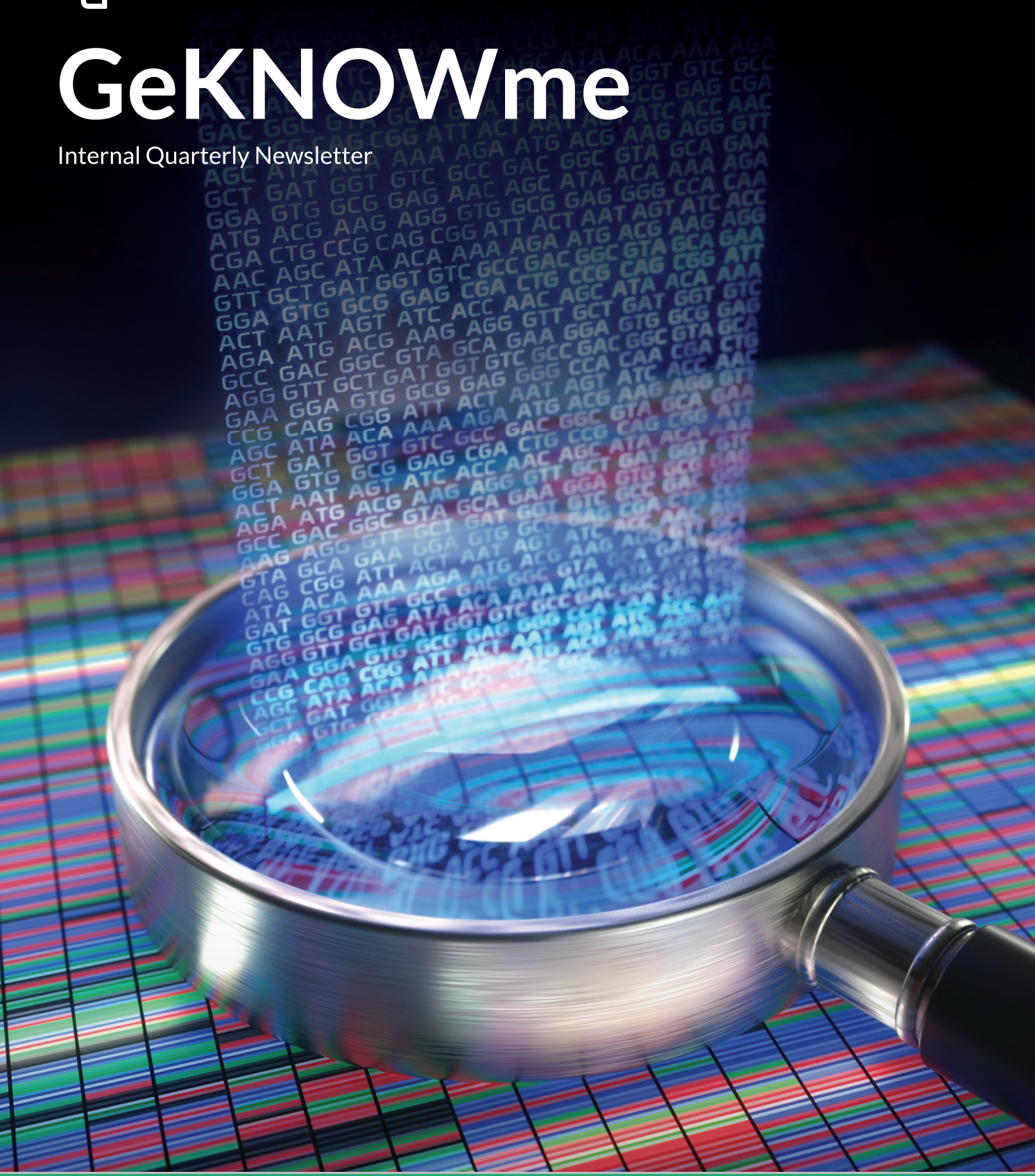
Vol 14 | July 2020

MEDGENOME

GeKNOWme

Internal Quarterly Newsletter



WORDS FROM THE FRONTLINE

Hiranjith G H

Senior Director, Corporate Marketing & Business Operations, USA

I am happy to contribute to this section of our quarterly newsletter, which I believe is a good medium to communicate few opinions and thoughts to each and everyone in the organization.

My team's current focus is to continue to develop the research services business and establish MedGenome as a preferred Next-Generation Sequencing (NGS) and Informatics service/solution provider in the US and Europe. We have been successful in engaging big pharma clients with our Transcriptome sequencing, Single Cell sequencing and TCR sequencing solutions, leading to multi-year master service or laboratory service agreements with these organizations. Our ability to provide end-to-end (hypothesis generation, sample/library preparation, sequencing optimization, analysis result) consultative solutions has allowed us to partner with biotech CSO's and research heads, which has developed into an effective word-of-mouth branding for MedGenome. Our streamlined operations enabling quality deliverables and fast turn-around-times for sequencing have allowed us to support projects for large academic institutions, even if they have access to internal genomic core facilities. Being a reliable service provider, we were able to strike co-marketing partnerships with multiple platform and kit manufacturers in the region. It is great to build a team and work alongside the team to plan and deliver on these objectives.

With increased adoption of next-generation sequencing in biomarker and translational research, our continued growth will be supported by our ability to:

1. Optimize new assays, workflows and solutions that will be relevant to our customer's research, driven by our experienced application and lab scientists
2. Offer custom, advanced and proprietary analyses solutions on NGS data, enabled by our skillful bioinformaticians and subject matter experts
3. Build a stronger brand across our target geographies through strong sales and marketing efforts by the team
4. Leverage systems and processes to derive operational efficiencies from sample receipt to invoicing the customers for the projects delivered. Extending this to the customer, appropriately, enables us to offer a window to manage our customers - be it to deliver data/results/analyses, to reduce iterations in sample shipment logistics, to run targeted campaigns, to experiment cross-selling opportunities and much more
5. Focus and assign resources to the impactful initiatives driving business and cross-functional collaboration with agility

With the right effort and the right team, the above objectives can be achieved.

I have been fortunate to work alongside brilliant and passionate colleagues since joining MedGenome, back in March 2014, when it was incorporated as an independent company in Bangalore. Along the way, I got to learn at close quarters from the founders of MedGenome and imbibe from experts who advised me at various junctures, across a variety of roles, responsibilities and teams. I have benefited immensely from the various technical, process and commercial insights offered by my team members over the years.

As we try to overcome the current crisis situation brought about by the coronavirus (COVID-19) spread, starting early this year and potentially extending for few months, it is time to fall back on the trust that we have on our team, our customers and our collaborators to sustain the momentum that we have gained. I look forward to collaborating with each one of you and contributing towards the continued growth of MedGenome.

Contents

Most Talked About

The News

EDGENOME NEWS | March to june 2020 | MEDGENOME NEWS

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

1

Science
THE WIRE

From Shed Hair, Scientists Obtain Whole Genomes of Wild Tigers

Featured image: A tiger in Ranthambore National Park. Photo: Flickr/Tausif. CC BY ND 2.0

Wild tigers are hard to study. That's why India's tiger census uses camera traps to take photographs of tigers as they walk unsuspectingly by; geneticists pick up tiger poop or hair from forest trails used by the elusive big cats, to obtain DNA.

Tiger poop, or scat, is the most accessible source of tiger DNA in the wild. Even the degraded genetic material in it can give scientists a small peek into how the tiger and its population are doing. However, there's a move common and better source of tiger DNA in the wild, scientists have now found.

Tiger hair – shed when the big cats exuberantly mark their territories at scratch posts – are more common in the wild than scat. Hair can even provide whole-genome sequences (the entire genetic information of an organism) if the right laboratory methods are used, according to a study published in the journal *Ecology and Evolution* on May 18.

The approach, if replicated and extended, could "persuade us all that hair sampling is a trustworthy second option in assessing genetic data from free-ranging species, which is valuable for management concerns", commented conservation geneticist Stephen O'Brien, founder of Russia's Theodosius Dobzhansky Center for Genome Bioinformatics, who was not involved with the work.

What tiger genes tell

Managing the last wild tiger populations – most of which are in India – is a

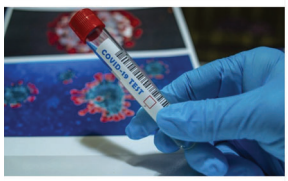
Trending | Transasia Bio-Medicals Partners with Go to Deploy Mobile Covid-19 RT- P...

2

AMEDTODAY
REIMAGINING INDIAN HEALTHCARE

MedGenome Labs gets ICMR Approval for RT-PCR Testing for COVID-19

MedGenome Labs strengthens its presence in infectious disease testing



Bengaluru: MedGenome Labs, a genetic diagnostics lab collaborates with Dr. Iravatham's clinical laboratory in Hyderabad, a niche testing lab in the field of Tuberculosis diagnostics strengthening its presence in infectious diseases testing. MedGenome's partner Xcyton Diagnostics is also awarded the ICMR approval to undertake COVID-19 testing at its lab in Bengaluru.

Citing their strengths, Dr. Vedam Ramprasad, CEO, MedGenome Labs, said "Our collaboration with Dr. Iravatham's lab is a milestone in TB testing ecosystem and we are committed to take more steps to integrate SPIT-SEQ, our proprietary whole genome sequencing based test for diagnosis and drug resistance detection directly from sputum samples, into TB testing ecosystem".

The partnership with Iravatham's lab will help integrate MedGenome's flagship innovation SPIT-SEQ, with various tests offered by Dr. Iravatham for Tuberculosis and also set up a state of the art BSL2/3 infectious testing lab in Hyderabad. "With India in the limelight as far as global TB burden is concerned, diagnosis and drug resistance detection take the centre stage. Offering a complete range of tests, right from smear microscopy to a sequencing-based test, will help shorten the time to diagnose, and lead to timely intervention.", said, Dr Chitra Iravatham, Dr Iravatham's Clinical Laboratory (DICL).

3

healthworld.com

World Meningitis Day : Tie to defeat Meningitis by rapid diagnosis

According to Lancet 2018 report, there were 2.8 million cases of meningitis globally in 2016. In India, there were around 5 lakh cases with a high mortality rate of 12% in 2016.

by Dr Gunisha Pasricha, Principal Scientist- Infectious Diseases at MedGenome Labs Ltd

Meningitis is a devastating disease and is major public health challenge. It is caused by the inflammation of the protective coverings of the brain and spinal cord known as meninges. The infection of the fluids surrounding the brain and spinal cord leads to inflammation of meninges. Globally, meningitis is a significant cause of morbidity and mortality in the pediatric population. According to Lancet 2018 report, there were 2.8 million cases of meningitis globally in 2016. In India, there were around 5 lakh cases with a high mortality rate of 12% in 2016.

Meningitis can be caused by many different microbes including bacteria, viruses, and fungi but the highest global burden is seen with bacterial meningitis. The currently widespread approach to identify infection causing microorganism is through a culture test (a test to grow these foreign invaders like bacteria and fungi, from the patient samples). Newer gene-based technologies have also enabled identification of infection-causing-microorganism directly from the patient sample for meningitis. There are a few niche diagnostic companies (such as Xcyton) in India that offer the test using Syndrome Evaluation System (SES), which is one such example of newer molecular techniques available to the doctors and patients for enhanced outcome. Once the causative microorganism is identified, it can help in faster and informed course of treatment for Meningitis and other deadly infectious diseases like Sepsis or TB.

Advantages of the newer gene-based technology over the conventional techniques include; rapidity, higher accuracy since it based on gene targeting, its ability to detect any of the microbes in a single test; higher sensitivity wherein the newer test can detect microbe in the patients sample even if it is present in lower quantity or loads. The newer techniques can also identify drug resistance and virulence factors which are important to cause the disease.

As soon as we talk about molecular or gene-based testing, the first thing that comes to our mind is 'cost of the test'. However, understanding the core of cause helps the clinician decide precise course of treatment. This involves removing unwanted antibiotics based on identification of exact causative organism, which otherwise would have been continued suspecting the chances of a particular microbe causing the disease. This phenomenon is referred to as

4

...Linc
News

...scientists unravel why some people are ...more susceptible to Covid-19

Findings may help make a drug to fight the deadly infection

A team of researchers from India and the US, led by a scientist at the Chennai-headquartered private genome research foundation, has found why ...me individuals are more susceptible to Covid-19 ...fection than others.

...esearchers from MedGenome Inc and SciGenom ...esearch Foundation (SGRF) jointly analysed the DNA ...quence and variation data from over 300,000 ...dividuals belonging to 400 different populations to ...redict susceptibility to the Covid-19 disease caused by ...e SARS-CoV-2 virus. Nearly 1 per cent of the samples ...re from people of Indian origin.

...he team led by Sekhar Seshagiri, President of Chennai-...ased SGRF India, identified variations in the ...giotensin-converting enzyme 2 (ACE-2) protein gene ...at are predicted to make individuals more susceptible ...the virus. Their findings are published online on the

5

BioSpectrum
ASIA EDITION

MedGenome, SGRF predict COVID-19 susceptibility

Image credit- shutterstock.com

A group of Indian and US researchers from MedGenome and SciGenom Research Foundation (SGRF) collaborated to analyze DNA sequence and variation data from over 300,000 individuals to predict susceptibility to the COVID-19 disease caused by the SARS-CoV-2 virus. The group identified variations in the ACE-2 protein gene that are predicted to make individuals more susceptible to the virus – their findings are published online at https://biorxiv.org/cgi/content/short/2020.04.07.029470v1. They also report variants in the ACE2 gene that will protect individuals carrying some of those variants.

SARS-CoV-2 is a highly contagious virus and is the cause of the current COVID-19 pandemic. Symptoms caused by the virus include fever, chills, cough, diarrhea and pneumonia. While some infected individuals are asymptomatic, about 10% need hospitalization and ~1-5% of the cases are fatal. Understanding why some individuals are more severely affected than others by the virus is important for managing at risk individuals. In this study posted in bioRxiv, the research team has attempted to understand how genetic factors affect susceptibility to the virus.

SARS-CoV-2 enters human cells by binding to ACE2, a cell surface protein that functions like a door to the cells for the virus. ACE2, an enzyme created by the ACE2 protein, usually plays a role in the maintenance of blood pressure but is tricked by the virus into becoming an entrance. One of the reasons the SARS-CoV-2 virus is highly infectious is because it can bind to the ACE2 cell receptor with higher affinity compared to the previous SARS-CoV virus which caused the disease called SARS which was less contagious, even though it caused an epidemic in 2003. Natural human ACE2 protein variation encoded in an individual's DNA can alter the virus to bind to ACE2 either strongly or weakly and hence alter their susceptibility to the disease which has caused the current pandemic.

"We analyzed ACE2 protein altering variants from over 300,000 individuals variant data from across the world and have found variants that are predicted to bind to the virus less tightly and hence make individuals less susceptible – giving these individuals natural protection against the invading virus' said Dr. Sekar Seshagiri, President, SciGenom Research Foundation, India and lead study author. "The converse is also true; in this data set, we found ACE2 variants that will render individuals more susceptible to the virus" said Dr. Eric Stawiski, Vice President Bioinformatics, MedGenome.

The team used its world class bioinformatics capabilities to accelerate the analysis and complete this study in very short order. The variants in ACE2 identified are not common, but this work sets the stage for larger studies that can focus on understanding the role and impact of these variants in patients. by studying variations in clinical outcomes and data in patients with the different genetic variants identified. The team is continuing to conduct more research to build on these findings to help fight COVID-19 and is also supporting Pharma and Biotech companies to help find solutions for COVID-19 through its research support services teams.

"Our finding on ACE2 variants can help enable the development of a rationally engineered soluble ACE2 that can quickly be developed as a therapeutic against this deadly virus" added Dr. Sekar Seshagiri. "We are also in talks with Pharma partners to advance their drug development work with genomics insights and ...particular in the context of this COVID-19 crisis. I think we have a potential drug solution ...and can be developed quickly with the right Pharma/Biotech partner," said Sam Santhosh, ...ma.

6

EXPRESS HEALTHCARE

MedGenome Labs collaborates with Dr Iravathams Clinical Laboratory Hyderabad

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The partnership with Dr Iravathams lab will help integrate MedGenome's flagship innovation SPIT-SEQ, with various tests offered by Dr Iravathams for tuberculosis, also set up a state-of-the-art BSL2 / 3 infectious testing lab in Hyderabad

MedGenome Labs collaborates with Dr Iravathams Clinical Laboratory at Hyderabad to enhance its tuberculosis diagnostics offering and gets ICMR approval for RT-PCR testing for COVID-19 with its current partner Xcyton Diagnostics.

One of the biggest problems that India has faced in the field of healthcare is in the area of infectious diseases. Before COVID-19 was recognised as a global pandemic, India was at the centre of the discussion on reducing global tuberculosis burden. With an immensely high prevalence of tuberculosis in India, the world is looking towards India to control the same. Indian government has set a deadline of ending TB by 2025. While India is one of the highest contributors to the global TB burden, the same nation, on the other hand, is looked upon as a proactive decision maker as far as controlling COVID-19 spread is concerned.

However, There are reports that show how the COVID-19 lockdown is taking us back to 2013 as far as our fight against TB is concerned. The lockdown may aggravate the existing TB burden as more TB patients may go undiagnosed and hence untreated.

With both these public health issues in focus, MedGenome Labs, India's biggest genetic diagnostics lab collaborates with Dr Iravatham's clinical laboratory in Hyderabad, a niche testing lab in the field of tuberculosis diagnostics strengthening its presence in infectious diseases testing. MedGenome's partner Xcyton Diagnostics is also awarded the ICMR approval to undertake COVID-19 testing at its lab in Bengaluru.

Citing their strengths, Dr Vedam Ramprasad, CEO, MedGenome Labs, said "Our collaboration with Dr Iravatham Lab is a milestone in TB testing ecosystem and we are committed to take more steps to integrate SPIT SEQ, our proprietary whole genome sequencing based test for diagnosis and drug resistance detection directly from sputum samples, into TB testing ecosystem."

The partnership with Dr Iravathams lab will help integrate MedGenome's flagship innovation SPIT-SEQ, with various tests offered by Dr Iravathams for tuberculosis and also set up a state-of-the-art BSL 2 / 3 infectious testing lab in Hyderabad. "With India in the limelight as far as global TB burden is concerned, diagnosis and drug resistance detection take the centre stage. Offering a complete range of tests, right from smear microscopy to a sequencing-based test, will help shorten the time to diagnose, and lead to timely intervention," said Dr Chitra Iravatham, Director, Dr Iravathams Clinical Laboratory (DICL).

Dr BV Ravikumar, Founder and Managing Director, XCyton Diagnostics said, "We are equipped to undertake a minimum of 1000 tests per day, with an ability to scale up the capacity manifolds in a matter of a few days. We are

7

EXPRESS HEALTHCARE

HOME | BLOGS | INTERVIEWS | FEATURES | CEOS CORNER | DEBATES | CASE STUDIES | EVENTS | VIDEOS

Home » Blogs » How non-invasive prenatal testing (NIPT) is helping during the COVID crisis

How non-invasive prenatal testing (NIPT) is helping during the COVID crisis

By EH News Bureau — On Jun 8, 2020 — BLOGS | GUEST BLOGS

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Read Article

***Dr Priya Kadam**, Associate Director- Reproductive Genomics, MedGenome Labs shares insights on the importance of non-invasive prenatal testing during pregnancy to determine chromosomal abnormalities irrespective of any prior risk, screening should be advised as a part of first trimester screening in pregnant women*

An unexpected outcome, as the world battles COVID-19, is the sudden focus on modern molecular testing techniques. These techniques have been around for a long time but are now coming into the forefront. Tests such as a RT- PCR have been around for a long time but are only now coming into the limelight. It's safe to say that large portion of Indians now know what an RT-PCR test is. Molecular diagnostic is not only more accurate, but also non-invasive in some cases.

One such modern test is non-invasive prenatal screening test (NIPT). This is a test that can be used to detect common chromosomal abnormalities in unborn babies from as early as the 10th week of pregnancy. The technique is based on next generation sequencing. This technique is used to sequence the entire genetic code in humans to find genetic mutations that are causing disease and disorders. The benefits of the test are convenient sample collection and high screening accuracy of over 99 per cent. NIPT also offers increased safety compared to invasive procedures such as Amniocentesis and Chorionic Villus Sampling (CVS).

Experts advice universal prenatal screening for common chromosomal disorders as part of first trimester screening in pregnant women. Take the case of a couple from Mumbai already evaluated and found harbouring an existing genetic condition which can cause a serious disease in their offspring. The couple opted for an assisted pregnancy through a young egg donor to eliminate the risk to the offspring being born with the disorder. The donor was evaluated for the same condition and found normal. This ensured her egg could be used to conceive a child without the genetic condition the couple harboured. Since it was a precious pregnancy the couple further decided to undergo a non-invasive prenatal testing to assess for any other common conditions that may occur spontaneously. Unfortunately, the foetus was found to be at high risk for Trisomy 21 (Down Syndrome). This highlights the fact that though women with advanced maternal age (35 years and above) are more prone to common chromosomal abnormalities, the condition should also be evaluated for in younger women. Both the donor and the woman carrying the pregnancy were less than 35 years of age.

Hence, screening for chromosomal abnormalities irrespective of a prior risk information is important and should be advised screening irrespective of age, other genetic conditions and backgrounds. In addition to testing, the couple should undergo genetic counselling to ensure that they are aware of the risks involved and understand the test results. A comprehensive discussion should be had with the family doctor or a health advisor about the tests available and the way forward after the tests results are available.

The COVID lockdown brought NIPT into the forefront because it offers high accuracy screening with low risk of infection compared to invasive procedures such as Amniocentesis and CVS both of which may require hospital admission which can expose both doctor and patient to the risk of COVID infection. There has been increase in prescription of NIPT's due to the COVID lockdown. The reasons could be multifactorial, number of elective procedures in hospital had reduced during lockdown and there might have been patient reluctance as well for fear of COVID infection. NIPT is seen as a good alternative due to higher accuracy and minimum discomfort to the patient. Th... trend is likely to continue as reliability of NIPT is well established and more doctors are becoming aware of th... of NIPT in the current circumstances.

1 2 3 4 5 6 7

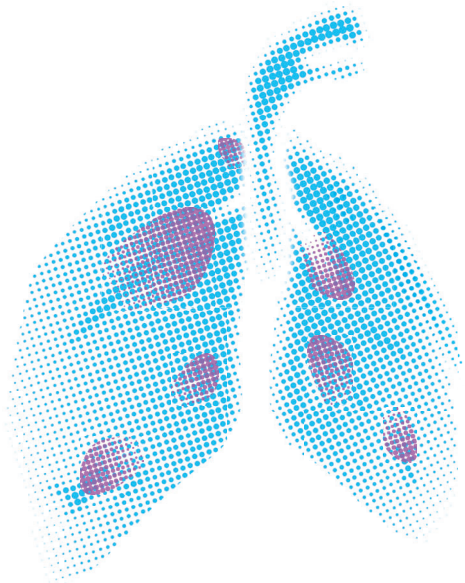
For press articles, please click - https://diagnostics.medgenome.com/press/

MEDGENOME LABS STRENGTHENS ITS PRESENCE IN INFECTIOUS DISEASE TESTING

MedGenome received the ICMR approval for RT-PCR testing for COVID-19

In partnership with XCyton Diagnostics and Narayana Nethralaya, Bengaluru

We are offering RT-PCR based COVID-19 test which detects and amplifies one or more loci of the RNA virus. The RT-PCR test is regarded as the current gold standard for molecular diagnosis with high sensitivity. RT-PCR kit is validated by ICMR. With over 20,000 (approx.) samples tested, we are looking at making a strong impact and help India fight COVID-19.



Collaborated with

Dr Iravatham's Clinical Laboratory, Hyderabad to enhance the Tuberculosis diagnostics offering

Our collaboration with Dr. Iravatham's lab is a milestone in TB testing ecosystem and we are committed to take more steps to integrate SPIT SEQ, our proprietary whole genome sequencing based test for diagnosis and drug resistance detection directly from sputum samples, into TB testing ecosystem.

MedGenome's Proud Moment

MedGenome has been awarded the "Outstanding Research in Diagnostics (National)"

by ET Healthworld at the ET Virtual Diagnostic Summit

ETHealthworld.com India Diagnostics Awards 2020, is an initiative aimed at recognizing the continued efforts put towards encouraging preventive care by players in the diagnostics industry during these testing times caused by the coronavirus pandemic by rewarding private diagnostic laboratories across India for their outstanding contributions at national and regional levels, towards providing high-quality diagnostic services to patients.

MedGenome won the award for Outstanding Research in Diagnostics (National) due to several publications across areas such as reproductive health, infectious diseases, inherited conditions and Oncology.

For more info: please click **https://health.economictimes.indiatimes.com/microsite/india-diagnostics-award**

MedGenome connect

Claria
Reproductive Genetics

Oh, how life has changed!! Without a doubt, the last 3 months have affected all our lives in one way or another. We lost our momentum, thanks to a lowly virus that continues to play havoc in our lives.

But all is not lost, the past three months have changed the way we function, hopefully for the better. As expected, all live events had to be cancelled due to the lockdown, but this period also unlocked digital avenues for us. We revived the MedGenome webinar series, and what a hit it was among the clinician community. We conducted two Claria webinars and these were on Recurrent Pregnancy Loss by Dr. Meenakshi Lallar and Utility & Best Practices in NIPT by Dr. Priya Kadam and Dr. E Venkatswamy. The response was tremendous with one of the highest registrations and attendance recorded for any of our webinars. The webinar series has helped build a lot of awareness about MedGenome and we hope to regain and accelerate our momentum over the next few months.

ACTIA
Inherited Genetics

Actia also witnessed more emphasis on webinars as a means to engage physicians. There was a total of four webinars done under the Actia banner. We hosted our first key opinion leader in the form of Dr. Sunita Bijarnia, Senior Consultant Geneticist from Sir Ganga Ram Hospital, Delhi who gave a two-part talk on Inborn Errors of Metabolism. This series received one of the highest registrations among our other webinars conducted in this segment. Dr. Sheetal Sharda gave another two-part webinar on Demystifying NGS, and hopefully, this was an eye-opener for the people who attended it. The quality of the interactive sessions after both the webinars was stellar showing that doctors lapped up the digital content with delight.

Also, April was Autism Awareness Month and we highlighted the need for genetic testing to aid in the diagnosis of Autism through various mailers and social media platforms.

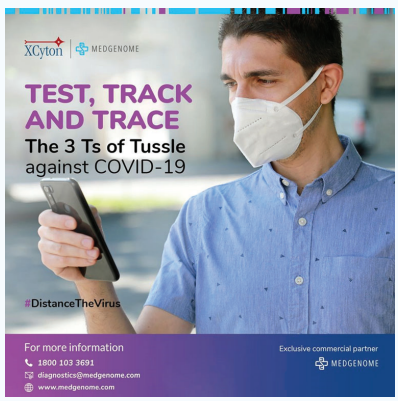
MedGenome connect

Prima
Cancer Genetics

With our lives transitioning from physical to digital lately, physician events were no different. As far as Prima is concerned, we conducted four webinars on various topics in last 3 months. The webinars were focused on cancer types (myeloproliferative neoplasms and acute myeloid leukemia) as well as specific markers (*ASXL1* and *TP53* gene mutations in various hematological malignancies), conducted by Dr Arun Kumar and Dr Vidya Veldore. All these webinars were moderated by Dr Rahul Bhargava, HOD, Haematology, Haemato-Oncology and BMT, Fortis Memorial Research Institute, Gurugram. The webinars were well received and witnessed a decent attendance. Over and above the live attendance, even the post webinar recordings had a large number of views by the physicians.

mocra
Infectious Disease Genetics

As 24-April was Meningitis awareness day, we did our part of creating awareness through emailer, social media as well as press articles for genetic testing for early and accurate diagnosis. Followed by this, we launched the COVID-19 RT-PCR test in the third week of May, which was followed by an extensive email campaign, digital campaign as well as a press release. Once we started sample collection from air passengers arriving at KIAL, Bengaluru, the focus shifted on a rigorous feedback mechanism and subsequent process improvement.



From our US office

In continuation with our previous newsletter update on our Research Services Blog, we are happy to share with you that the blog features some more interesting and cutting-edge scientific articles covering a wide range of topics including: Cancer, GenomeAsia, Covid-19, Biomarkers etc. We hope you have enjoyed reading them and any insightful comments are welcome.

MedGenome colleagues are encouraged to take initiative and contribute towards the blog. You can share your blog articles with Vinay and Hiran @ mgus-blog@medgenome.com

Further to our current research efforts in US, we have added new services to our existing portfolio - the Epigenomic Profiling and BCR Repertoire Analysis Services. More details can be seen on our website:

Epigenomic Profiling: **https://research.medgenome.com/ngs-services/epigenomic-profiling/**

BCR Repertoire Analysis Services:
https://research.medgenome.com/ngs-services/bcr-repertoire-analysis/

Whitepapers can be downloaded from our resources section here:
https://research.medgenome.com/white-papers/

In the last quarter, we ran marketing campaigns on:

1. FAST Turn-Around on pre-made library sequencing
2. MedGenome's COVID-19 research solutions
3. Single Cell Gene Expression Assays
4. CUT&TAG Epigenetics assay
5. TCR Sequencing
6. BCR Sequencing

Making a difference

NIPT Detects Chromosomal Abnormalities in Pregnancy Classified as Low Risk

Case Discussion

Monosomy X is characterised by short stature, problems with the heart and kidneys, absence or poor development of sexual characteristics, learning difficulties and sometimes mild intellectual disability. The symptoms range from mild to severe.

There is a small chance of false positives with NIPT. Therefore, and according to guidelines, invasive confirmatory testing was recommended, but due to maternal anxiety, they decided to terminate the pregnancy. The couple decided to test the products of conception by further confirmatory testing.

A chromosomal microarray (CMA) was performed on the fetal tissue sample and it was found out that approximately 30% of the foetal cells had only one X chromosome, confirming the NIPT results. Though it is hard to predict antenatally, even in such a case, the abnormality would have manifested. Since all other tests including the ultrasound scans were normal, the condition could have been missed out, if not for NIPT.

As the couple is at increased risk for having another pregnancy with an abnormality, they were advised to go for genetic testing early in the next pregnancy

Patient information

A young couple visited their obstetrician for the routine check-up during their pregnancy, they were suggested Non-Invasive Prenatal Testing (NIPT) to rule out chromosomal abnormalities in their pregnancy

Genetic History

Family history was negative. The couple were considered in the low risk category, but they went ahead with NIPT to rule out the population risk.

Genetic testing at MedGenome

Non Invasive Prenatal testing was done at MedGenome

Results

The NIPT result came as a high risk for Monosomy X. The couple was counselled about the condition, advised diagnostic testing and explained its consequences.

Deciphering Tiger genome from shed hair

By : Anup Chughani
Senior Scientist, MedGenome Labs Ltd., India

Tigers are an elusive species which were on the brink of extinction a few decades ago. It is estimated that 95% of the tiger habitat has been lost and remaining is fragmented due to extensive human activities like - clearing of forests for agriculture, building of road networks and other development activities. Since the tigers are highly territorial and typically prefer large territories, many may not survive in scattered islands of habitats. This islanding leads to higher risk of inbreeding and poaching as they venture out to establish territories.

Thanks to extensive conservation efforts by various governments and NGOs, their population globally is growing back at a steady rate (Figure 1). In spite of these efforts, there is a deeper need to understand how these species have diverged and isolated in patches of forest cover across Asia. One way to achieve this is to look at their genomes.

Genome sequencing data is critical to estimate genetic relatedness in an elusive species like the tiger. Typically, studies like this need sampling the animals in the wild which may involve capturing, tagging and blood/tissue collection. Although this can be easily done with the herbivores or other small animals, for an elusive large carnivore such as the tiger, it is a challenge or rather dangerous to immobilize the animal. For such species, samples such as scat, urine, saliva, skin, hair or other environmental DNA is more feasible but is not without its own set of challenges. For example, scat samples may be dominated by microbial and prey DNA while the urine sample may be contaminated with environmental DNA. Saliva samples at the kill site may belong to more than one individual and may have prey DNA. In contrast, shed hair samples are expected to be cleaner than the above non-invasive sources.

Figure 1

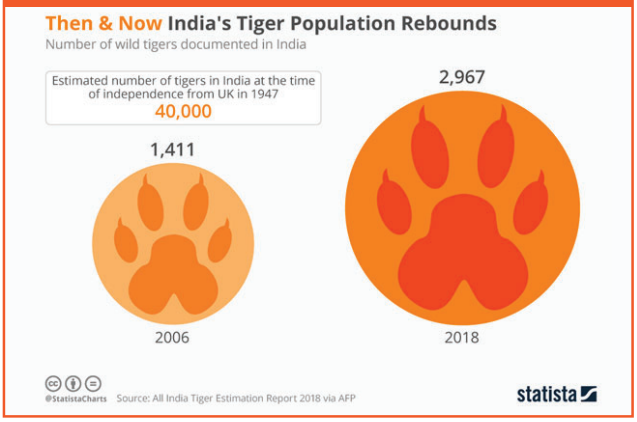
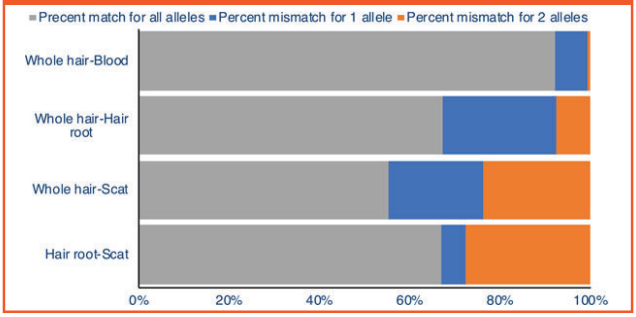
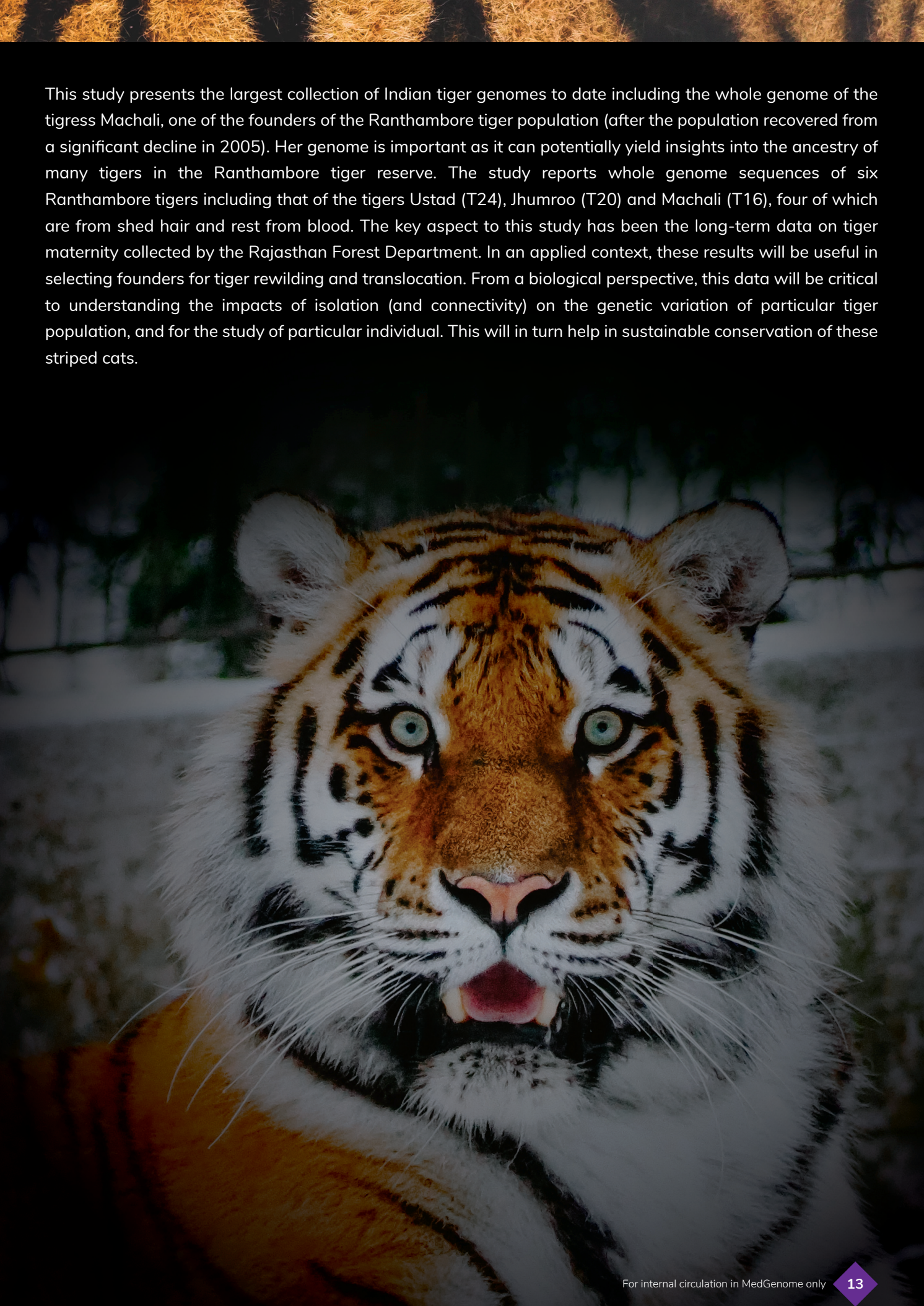


Figure 2



In the last couple of years, we have been working with Dr. Uma Ramakrishnan and her team at NCBS, Bangalore to generate sequencing data for different tiger populations. We have been using DNA extracted from different sources of samples viz., blood, tissue, scat, hair, etc. for library preparation, sequencing and analysis. In this study, during the field work in Ranthambore Tiger Reserve, individual tigers were followed (at safe distances) and the hair samples were collected wherever the tigers rested or groomed. Later on, these samples were processed further for DNA extractions and sequencing. It turned out that DNA content from shed hair was better than that from scat samples (which was traditionally collected). It was also found that DNA sequences from shed hair were concurrent with the blood samples and yielded identical results on population connectivity (Figure 2). Using the sequences from shed hair, two new matrilines in Ranthambore Tiger Reserve were discovered. It was also discovered that the tiger T47 might belong to the same matriline as the famous tigress Machali. T47's ancestry was previously unknown. Hence, shed hair from field-observed tigers could help sequence the genetic material and establish previously unknown genetic links. The results of this work was published in the journal Ecology and Evolution (DOI: 10.1002/ece3.6157)

This study presents the largest collection of Indian tiger genomes to date including the whole genome of the tigress Machali, one of the founders of the Ranthambore tiger population (after the population recovered from a significant decline in 2005). Her genome is important as it can potentially yield insights into the ancestry of many tigers in the Ranthambore tiger reserve. The study reports whole genome sequences of six Ranthambore tigers including that of the tigers Ustad (T24), Jhumroo (T20) and Machali (T16), four of which are from shed hair and rest from blood. The key aspect to this study has been the long-term data on tiger maternity collected by the Rajasthan Forest Department. In an applied context, these results will be useful in selecting founders for tiger rewilding and translocation. From a biological perspective, this data will be critical to understanding the impacts of isolation (and connectivity) on the genetic variation of particular tiger population, and for the study of particular individual. This will in turn help in sustainable conservation of these striped cats.



Artificial Intelligence and Machine learning in healthcare

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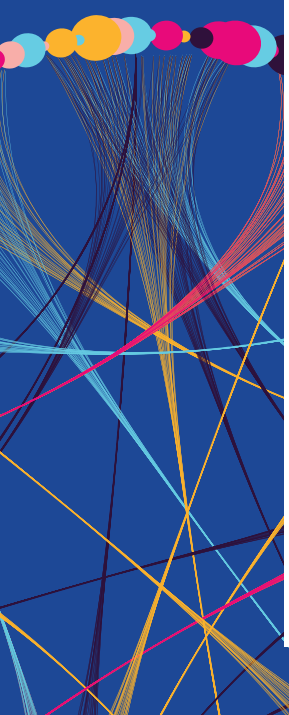
Artificial intelligence (AI) and **Machine learning (ML)**, which represent simulation of human intelligence for decision making is the current buzzword across many industries and are often used interchangeably. According to computer scientist and machine learning pioneer Tom M. Mitchell, ML is the study of computer algorithms that allow computer programs to automatically improve through experience. According to the U.S. Food and Drug Administration (FDA), AI is "the science and engineering of making intelligent machines", whereas ML is "an AI tool that can be used to design and train software algorithms to learn from and act on data " (https://www.fda.gov/medical-devices/software-medicaldevice-samd/artificial-intelligence-and-machine-learning-software-medicaldevice#whatis). So ML is one of the enablers for achieving AI. ML algorithms can help in understanding complex data patterns present in input training data. Machine learning algorithms can be grouped into the following categories: (a) supervised learning, (b) unsupervised learning, or (c) reinforcement learning. In supervised learning, the algorithm is given an input data along with corresponding target label/group. The algorithm tries to learn from input features and target groups. The classification and regression tasks are named as supervised learning since then they learn from the labelled training data. In unsupervised learning the input data is provided without target group. The algorithm's task is to identify the underlying structure in the data and then assign group label. Unsupervised learning is also called clustering. In the case of reinforcement learning, the algorithm's aim is to find the most suitable action that will maximize the reward. Like clustering, reinforcement learning does not need the target label for training. Another group of algorithm that is becoming popular is the deep learning (DL) framework. Generally deep learning methods require much larger amount of data to train efficiently compared to machine learning, but once trained properly, their efficiency is usually better than machine learning frameworks. Usually interpretability of machine learning concepts is much more clearer compared to deep learning. Deep learning architectures have better efficiency in areas where it is impractical to design features/concepts from input data. So, where ML requires domain knowledge and expertise, DL can be applied more or less in a much wider arena, given large amount of data to train the systems efficiently.

The most popular AI method that we currently use are Human-AI interaction gadgets by Apple Siri, Google Home and Amazon Alexa. The video prediction systems used by Netflix, Amazon and YouTube are powered by ML algorithms that help in identifying the right content. The shopping cart items shown by FlipKart and Amazon are driven by ML programs that look up customer's past searches both on their website and internet and then suggest them the most suitable items. AI/ML methods are increasingly becoming essential in our daily lives e.g. Google Maps, where it processes real-time information about traffic and commuter routes quickly on the user's mobile phones and provides the best route to take. Healthcare is another field that is thought to be highly suitable for the application of AI tools and techniques. These techniques will enhance the quality of automation and make decision making in primary/tertiary patient care in public domain much more robust. **AI in healthcare has the potential to transform the quality of life for billions of people worldwide. It is predicted that in near future every clinician whether a specialty doctor or a general physician will be using AI/ML for making clinical decisions. They will use these techniques to interpret medical scans, pathology slides, skin lesions, retinal images, electrocardiograms, endoscopy, genetic diseases, faces, vital signs and many more.**

Accurate interpretation of radiology images plays an important role in clinical diagnosis and treatment planning. In the last few years there have been many studies that show how AI can outperform humans in the interpretation of medical images from various diseases. Chest X-rays with over 2 billion scans worldwide every year are one of the most commonly used medical scans for diagnosis of several thoracic diseases (chest X-ray database- ChestX-ray8; Wang, X. et al. 2017). **A deep learning program viz., CheXNet developed by Rajpurkar et al. in 2017 uses a 121-layer Convolutional Neural Network (CNN) trained on large publicly available chest X-ray dataset with over 100,000 records from 14 diseases, can detect pneumonia from chest X-rays better than trained radiologists.** Later on a team from Google (Li et al., 2017) analysed the same dataset using residual neural network (ResNet) architecture and performed better than the reference baseline for diagnosis of pneumonia, heart enlargement and collapsed lung. The sensitivity of manual identification of cancerous pulmonary nodule by clinical community has not been satisfactory and ranges from 36% to 84% depending upon tumor size and cohort. Recently a deep neural network (DNN) was applied to detect cancerous pulmonary nodule from X-ray of chest using DNN (Nam et al., 2018). This method yielded much better results compared to manual identification methods and its overall performance was better in 16 out of 18 clinicians. The clinicians who performed better than the AI method had over 13 years of experience. Another interesting application of AI has been in identification of bone fracture from images as compared to human interpretation e.g. using a DNN method on wrist fractures detection increased accuracy from 81% to 92% and reduced the misinterpretation by 47% (Lindsey et al., 2018).



AI and ML methods are now applied in various problems of genomics including variant calling, variant classification, detection of functional and regulatory elements in the human genome.

Particularly in genomics data, it is impractical to use hand crafted statistical rules for the task of interpretation; e.g. let us take the example of generic variant-calling tools, most of which are prone to systematic errors that are biased due to dependency on sample preparation, sequencing platform and technology, sequence context, and inherent biology of the sample in question. Recently published DeepVariant (Poplin et al., 2018), a CNN (Convolutional Neural Network)-based variant caller trained directly on read alignments without any specialized knowledge about genomics or sequencing platforms, has shown significantly higher performance than the popular variant callers including GATK. Similarly, Illumina's

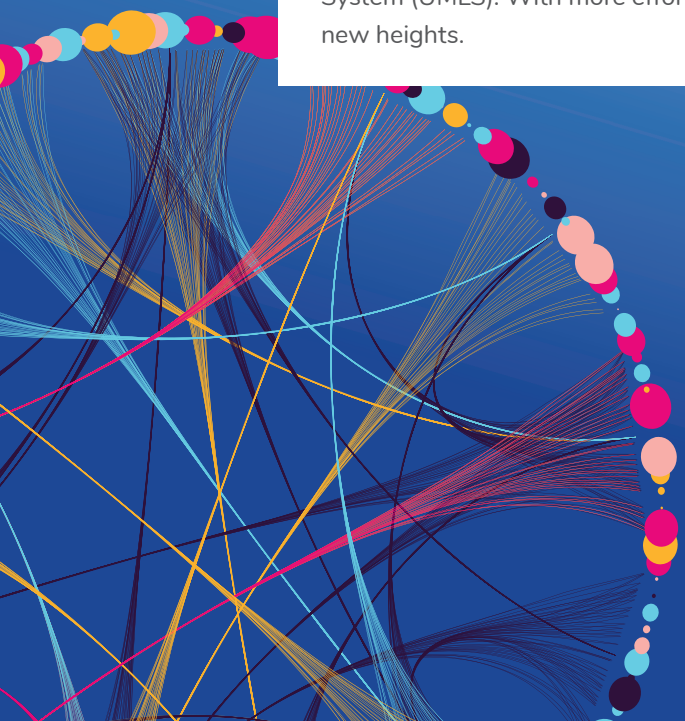
SpliceAI (Jaganathan et al., 2019) uses deep learning for predicting intronic variants that can lead to cryptic splicing. **Cryptic splicing is enriched in autism and intellectual disability patients as compared to healthy individuals, therefore understanding the impact of variants in intronic is an important step in clinical diagnostics.** Many AI tools that use CNNs and RNNs (Recurrent Neural Networks) are also being used for predicting cis and trans regulatory binding sites. DeepSEA (Lyu et al., 2018), another multitask CNN trained on large-scale functional genomics data learns sequence dependencies at multiple scales and predicts DNase hypersensitive sites, transcription factor binding sites, histone marks, and the influence of genetic variation on those regulatory elements, with accuracy superior to tools for prioritizing non-coding functional variants. AlphaFold from Google Deepmind is another application of AI where Levinthal's paradox (protein folding problem) has been addressed with the aid of BlueGene supercomputing from IBM and is able to predict protein structure large genomic datasets (Senior et al., 2019). Protein structure prediction is an important step in understanding protein function and is difficult to obtain experimentally. AlphaFold is neural network-based program that infer protein structure by analyzing covariation in homologous sequence. AlphaFold generated highly accurate structures for 24 out of 43 free modelling domains.

EHR (Electronic Health Record) analysis is another field that is catching lot of action from deep learning.

Patient clinical data is a very rich source of healthcare data that can impact clinical decision making in heavy manner; but it exists in multitude of formats ranging from radiology data to clinical notes. A flexible data structure Fast Healthcare Interoperability Resources (FHIR) format is being encouraged to represent clinical data in a consistent, hierarchical, and extensible container format, regardless of the health system, which simplifies data interchange between sites. EHR data can be converted to features or concepts of an RNN (there's a reason to call them Recurrent Neural Networks), that can identify patterns of patient characteristics, diagnoses, demography, medications, and other events capable of predicting patient mortality or hospital readmission. NLP techniques can capture unstructured data, analyze the grammatical structure, determine the meaning of the information, summarize it and thus can reduce cost and extract the information for deep analytics. **Combined with genomic data, NLP-based methods have boosted phenotype-informed genetic analysis, resulting in automated genetic diagnoses and is now been used to diagnose rare diseases.**

Compared to standard statistical approaches that use direct feature learning, deep learning has been shown to simultaneously harmonize inputs including free-text notes, to produce predictions for a wide range of clinical problems and outcomes that outperform state-of-the-art traditional predictive models. Automatic Mendelian Literature Evaluation (AMELIE) (Birgmeier et al., 2020) is one such example of a text-mining tool that parses 29 million PubMed abstracts as well as several full text article to associate the causal variants with their phenotypes. In diagnosis of rare Mendelian disorder, singleton patient analysis (without relatives' exomes) is the most time-consuming scenario. By connecting the phenotype of patients with literature AMELIE ranked the causal genes at top for 66% of 215 diagnosed singleton cases from Deciphering Developmental Disorders (DDD) project. In MedGenome we have implemented random forest based ML tool (VaRTK – Variant Ranking ToolKit) to prioritize variants in our diagnostics samples. VaRTK provides pathogenicity score to each coding variants and is trained on 33 features generated from variants and genes. These features are derived from our clinical reports by genome analysts. In a recent testing (June 2020) on 184 cases where a variant has been reported by genome analyst VaRTK ranks ~90% of pathogenic cases in top 20. By incorporating our latest advancement in VaRTK where we include phenotype score, we are able to rank 93% of the pathogenic cases in top 20.

AI features or concepts are not generally explainable in human comprehension and is often referred to as a black box. Since huge fraction of the audience to claimed AI products do not understand the black box many companies are misusing the AI term for their various product and services. In a report from Verge, 40% of European startups that claimed to use AI don't use the technology. In clinical decision making, which is a high-risk situation, acceptance of AI predictions is subject to acceptance both by clinician and patient community. The genomic and healthcare data suffer from multilayer substructure; with data being prone to biasing arising out of confounding factors such as socioeconomic status, cultural practices, unequal representation, and other non-causal factors that relate to the delivery and accessibility of medicine and clinical tests rather than to their efficacy (Topol, 2019). AI systems must be carefully applied to differentiate between these types of non-causal bias. However, the road is not as dark as it seems. FDA is currently recognizing many AI-driven health analytics in medical diagnostics. One such example is the recent FDA approval of X-ray imaging technique for detection of lung diseases by GE Healthcare. More research is underway to bring confidence to AI systems in high risk areas like clinical diagnostics. This goes hand in hand with development of more cluster computing as well as more clinical data synchronizing efforts like FHIR and standardization of medical terminology like Unified Medical Language System (UMLS). With more efforts in these areas, acceptance of AI in clinical set ups will gain new heights.



The Medici Effect

What Leonardo da Vinci and Michelangelo can teach us about DTC Genomics?

By: Dr Vidyadhar Karmakar,
Product Head, ApnaGenome

The House of Medici was an Italian banking family and political dynasty that came to power in Florence, Italy in the 14th century. The Medici family funded contemporary artists, whose work eventually led to The Renaissance, which is best known for the artistic development and contributions of Leonardo da Vinci and Michelangelo. Since the Medici family did not intend the Renaissance to happen but their financial support contributed to it, Frank Johannsson in his book on innovation calls it "The Medici Effect". The title of my article is inspired by this book, which argues that innovation occurs at the intersection of ideas from diverse industries, cultures, and disciplines. And we are witnessing such massive inter-disciplinary innovation in Genomics that is revolutionizing healthcare. The power dynamic is evolving as "patients" are now becoming "consumers" and companies that innovate and treat them as such are getting rewarded. However, the potential of genomics is much larger because it can serve the "patient" consumer as well as the "healthy" one.

Hence, to harness the true potential of DTC genomics, one needs to think "out-of-the-clinical-box". In my opinion, this paradigm shift in thought is the secret of success in DTC Genomics. In this article, I will share some of my experience and practical insights on setting, scaling and stabilizing DTC Genomics business.

To harness the true potential of DTC genomics, one needs to think **"out-of-the-clinical-box"**

Setting the Stage:
Direct-to-Consumer (DTC) Healthcare

It is a
$700
Billion- Dollar
global
industry

Direct-to-consumer healthcare bypasses a clinician recommendation and directly engages with the consumer. This absence of a clinician recommendation is the defining feature of direct-to-consumer (DTC) healthcare. However, DTC healthcare is not a new industry. It is a USD 700 billion-dollar global industry that includes diverse products e.g. over-the-counter drugs, hearing aids, glasses, contact lenses, nutraceuticals, oral/skin health and other categories[1] (Figure 1). In India, DTC healthcare is a USD 3.5 billion market with CAGR of 10-11% (Euromonitor).

The biggest driver of DTC healthcare industry is self-initiation/selection in which people, as consumers, are proactive about managing their health. Consumers, given their increasing level of awareness and education, are now savvier than before on their healthcare needs and are now more comfortable and confident of their decisions. DTC companies seek to address the needs of such proactive customers and hence consumer-centricity is critical for success in DTC healthcare business.

> "Consumer-centricity is critical for success in DTC healthcare business"

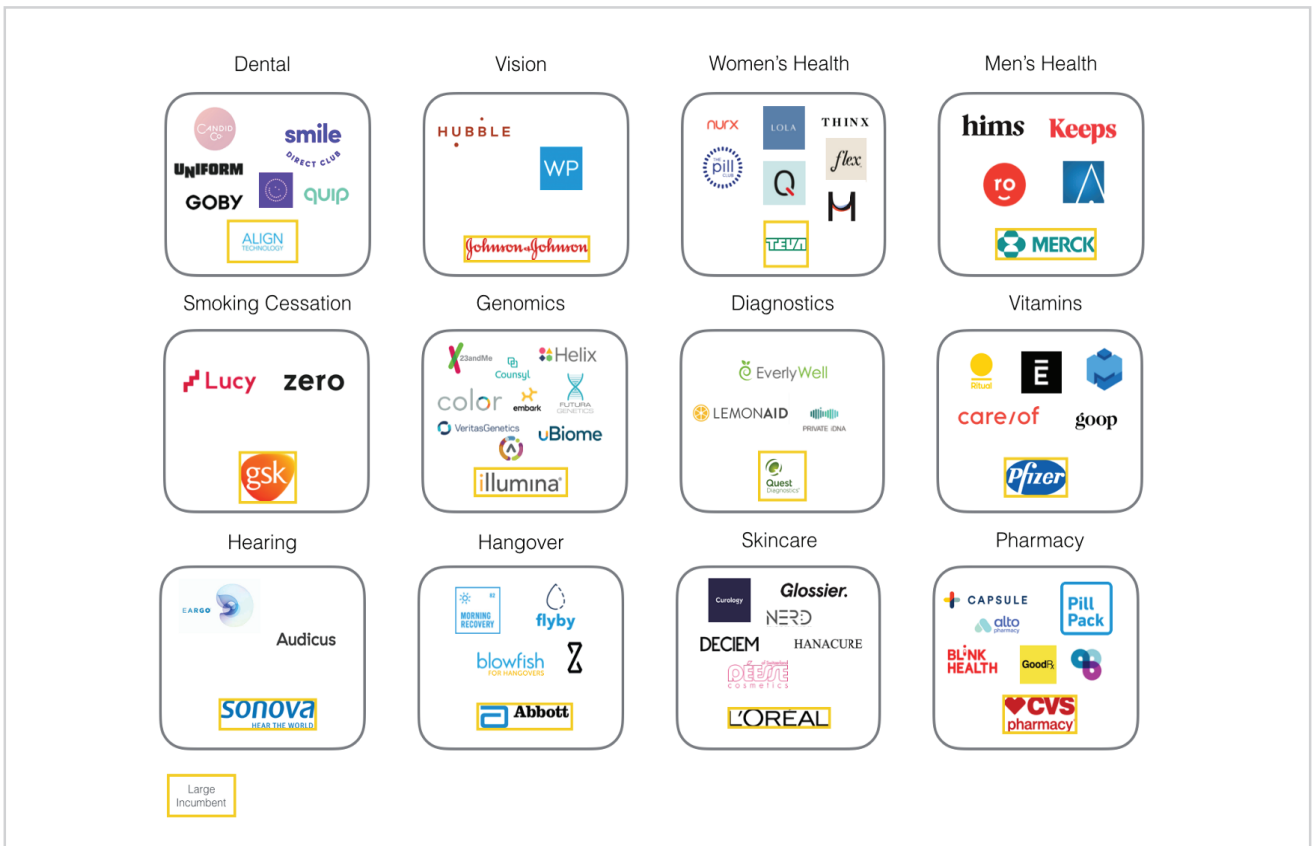


Figure 1 DTC Healthcare Companies[2]

Although the DTC industry is attractive for consumers and providers, the very "direct-to-consumer" feature that bypasses the typical filters and safeguards of health-care systems makes it vulnerable to risks and exploitation by unscrupulous companies. The risk is that low value, or even harmful, products often inundate the commercial health-care market. The regulatory concerns stem from DTC products making explicit claims about wellness and lifestyle but implicitly mean health benefits for serious medical conditions. Such products and solutions are apparently innocuous and could fall under the regulatory radar. Nevertheless, as the DTC industry diversifies and grows in new verticals to bring science and technological development for benefit of the people, the regulators, clinicians, and researchers should assess the inherent risks and how to minimize them while bringing new technology to maximize benefits to individual and public health.

DTC Healthcare 2.0: **Consumerization of Genomics**

Given the backdrop of DTC Healthcare, it is now easier to visualize DTC genomics as a natural fit in the industry. Notwithstanding the critics, with more than 30 million DNA tests sold globally, DTC Genomics is now a serious category of DTC healthcare. While these numbers are impressive, in my opinion, we have just tickled a demand for genomics-based consumer industry. It is an evidence that with the right strategy and marketing mix for the respective geography, DTC genomics can also be a multi-billion-dollar industry. However, given the diverse areas that DTC Genomics can impact, estimating DTC Genomics market size is a bit difficult. The diversity of impact areas of DTC Genomics is in its conceptual promise - our DNA codes the blueprint of our life and a genome scan can reveal information for healthcare, wellness/lifestyle and recreational uses. DTC Genomic companies have positioned themselves somewhere along this spectrum of recreational, health and wellness or in a hybrid category thereof.

> "Ancestry testing that drove the DTC Genomics sales allowed 'productization' of genomics"

With the bulk of the global DTC Genomics sales coming from the US, the key question is how were companies able to achieve such high volumes? In my opinion, ancestry testing that drove the DTC Genomics sales allowed "productization" of genomics and helped it get a strong foothold of consumer's mind. Companies like 23andme, Ancestry and MyHeritage addressed an unmet market need or a question that people had about their origins. Record keeping and discussion about ancestry was common over dinners and coffees. Thus, curiosity about their ancestry was already a part of their environment. DNA based ancestry testing just provided answers with a new and exciting flavor especially in discovering unknown relatives. An affordable pricing of $49-$99, ease of saliva-based testing process allowed DNA Ancestry Testing product to be made available and accessible across multiple sales channels. In addition, despite the concerns on data, the scientific participatory research communities e.g. on depression, Parkinson's etc created by 23andme helped in user engagement and "crowd-source" research to be part of the solution and not the problem. This sense of belonging to the company through an altruistic purpose also created brand loyalty.

From an operational viewpoint, ancestry testing was also a perfect use-case for productization of genomics. In a clinical setting, pre- and post- genetic counseling is integral to genetic testing. However, counseling is a service and hence labor and time intensive, adds significant costs, reduces throughput and slows down product delivery. To fasten product delivery, companies simplified the content and user interface (UI), enriched user experience (UX) and strategically removed counseling service as integral to the product journey. Thus, DTC Genomics companies solved a major operational hurdle to achieve logistics and economies of scale. However, that did leave a gap in the market since consumers still wanted to speak to someone to understand the report. This allowed other start-ups to come up with counseling services.

Setting the Stage:
Lifestylization of Genomics

There is a huge difference in what a product does versus what the product does for a consumer. While DNA ancestry is an interesting value proposition for a genetically heterogeneous consumer, it is of limited appeal to consumers whose ancestors have not migrated much over the course of history. Thus, lifestyle was considered as a possible value proposition in this case to bring genomics into their environment. Based on the assumption that people have power to choose healthy and unhealthy lifestyles and are at least partly accountable for their health, several companies globally launched lifestyle and wellness genomics products and met variable degrees of success and showcased willingness of the consumers to buy these products. If we take this evidence in the context of DTC Healthcare, USD 700 billion industry, we realize that DTC Genomics is a huge untapped opportunity.

> *"There is a huge difference in what a product does versus what the product does for a consumer"*

Like DTC Healthcare products, a consumer can self-initiate the purchase of DTC Genomic products on lifestyle, health and wellness. From a product management perspective, this is a huge innovation in product positioning and disrupts the market dynamics in various ways. For instance, this impacts clinicians from being gatekeepers of health-information to a situation where their involvement is marginal, at the best. This is because DTC Genomic positions itself in a regulatory category alternative to medical tests and hence bypasses the need of clinician prescription. The lifestyle and wellness label on the product, while legitimizing the seriousness of the test, negotiates a position to serve the customer that is hybrid between patient and consumer. This positioning between tightly regulated medical tests and less regulated consumer market signifies a blurring distinction between established normative categories. From a product marketing perspective, this democratizes personal health information, reduces the regulatory hurdles to catalyze faster speed to market, reduces consumer's decision-making time to purchase a product and sets the platform to boost sales.

From an analytical validity standpoint, there is no debate that even well-funded and established companies have rushed to market with DTC Genomic products supported by scattered peer-reviewed research studies. For actionable products, companies will have to invest in properly researched products that create value for consumers. The companies willing to invest in research for consumer-centric product development would raise a barrier of entry for new entrants and gain a long-term sustainable competitive advantage.

DTC Genomics in India

DTC Genomics products have been sold in India for a decade. But let the truth be told, despite 1.3 billion population, all Indian companies put together could not sell more than 20K tests in 10 years. In this section, I will share my views of the drivers and challenges of DTC Genomics products and industry in India:

Challenges:

1. Information asymmetry

The customer, the clinician and the company that sells such tests are the three primary stakeholders of a DTC Genomic test (Figure 2). It is important to note that, "consumer" (or influencer) of the test information is different from the customer (i.e. one who pays). A PGx report would be consumed by a clinician to advise a personalized prescription to a patient or to personalize preventive health checkups based on genetic predispositions. Likewise, a dietician would consume the genetic report to plan a personalized diet recommendation based on genetics. For product information uptake and actionability, consumers and customer both need to be educated about pros and cons of genetic testing. The ability of consumer to provide valuable guidance to the customer is hampered by lack of awareness and education about the genomic testing. Hence, awareness and education becomes the key to reduce information asymmetry of various stakeholders. Importantly, with ~10% of Indians comfortable with English and language preference changing with city tier, choice of language makes a huge difference in creating education/awareness and subsequent impact thereof.

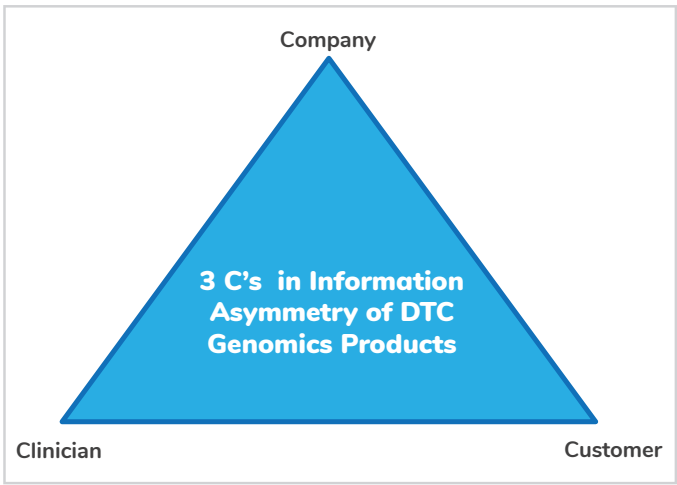
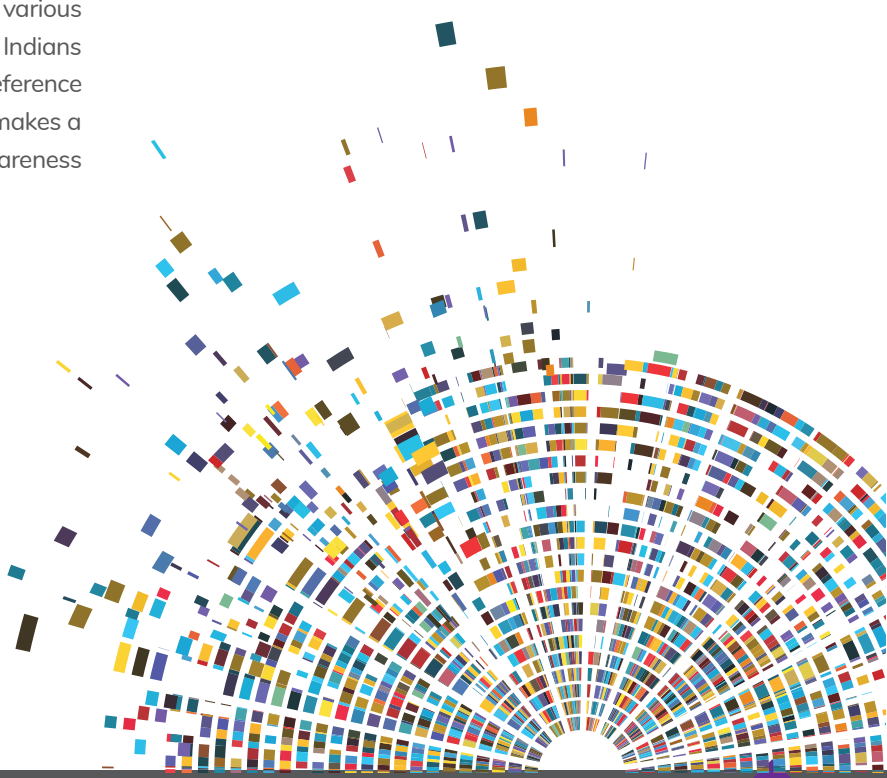


Figure 2 Information Asymmetry in DTC Genomics Stakeholders



2. Product Poverty

A customer's product journey begins much before product purchase. It includes - awareness, interest, consideration, intent, evaluation, purchase, use, feedback, continued engagement and customer-referrals (Figure 3). A company has to cover this product journey to delight the customer with an awesome UX and UI. Ironically, in the product journey, a successful product sells the problem it solves and not the product itself.

> "A successful product sells the problem it solves and not the product itself"

Figure 3 Customer's Product Journey

The conceptual promise of DTC Genomics products on health, wellness and lifestyle is to empower an individual for proactive and personalized healthcare and lifestyle. The entire product journey of the customer has to be designed around delivering the promise. Likewise, after the purchase and delivery of the report, counseling is a critical feature in the genomic wellness product journey. A counselor should be able to bring out the "why" of the report by connecting health, nutrition and fitness sections of a wellness report to offer personalized advice. Thus genetic counselors need to be trained and the companies should focus on platforms or KPIs to measure counselor performance vis-à-vis product experience.

UI of the report decides how the customer will access his/her genetic information. Reports should be brief and language should be simplified for customers to understand. A strategy focused only on selling the test report coupled with a poor UX/UI of a product results in a poor customer engagement, less customer referrals and directly hits the cost of customer-acquisition.

Another aspect of product poverty is the technology and genomic data used in generating a report. While this does not directly hit the sales or cost of customer acquisition, technologically poor products spoil the market. Without proper discovery and validation in target ethnicity, the sensitivity, specificity and predictive power of such reports is questionable.

3. Product Affordability

As per PWC report[3], ~900 million Indians earn less than USD 150 per month and share of wallet of medical and health expenses is about ~7% of total household expenditure. Indians spend more on curative care as compared to preventive care (Figure 4). Given this economic context and health priorities, the price of DTC Genomic products that currently range from USD 70 to USD 275 (Exchange rate INR 73) are not affordable to most of the Indians. In addition, Indians are price-sensitive and seek value-for-money. Hence DTC Genomic companies, to make the product affordable, often tie-up with banks for the consumer to pay for the product with EMIs.

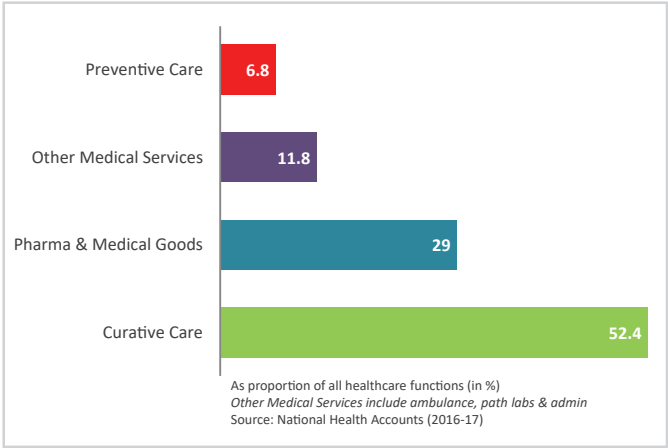


Figure 4 How Indians spend on Healthcare

Drivers:

For India, on one hand we have data on healthcare challenges e.g. adverse drug reactions and non-communicable diseases, which have a genetic component to it. On the other hand, we also have data on increasing interest in proactive health e.g. willingness to spend on nutrition and fitness, health check-ups etc. Hence, one might anticipate that a combination of the healthcare problems along with consumer behavior could have translated into a significant interest in DTC Genomics.

> *"People would buy products that work and DTC companies need to demonstrate it to the consumers"*

However, when we look at the insignificant sales, we clearly see that is not case. Thus, I am not sure if there are any clear drivers for DTC Genomics thus far. However, as awareness and education about consumer genomics is increasing, it is reflecting in increasing inbound queries directly from consumers, key opinion leaders, doctors, dieticians and wellness coaches trying to understand more about genomics products to help health and wellness goals. Thus in my opinion, key future drivers for DTC Genomics in India would be: (i) good products that have evidence-based actionable insights for customer engagement and benefit, (ii) awareness and education about product evidence by clever use of various media for communication. In summary, people would buy products that work and DTC companies need to demonstrate it to the consumers.

ApnaGenome -
Cracking the Indian Consumer Genomics

Indian consumer is highly heterogeneous. Hence, to crack the Indian market, there may not be single blockbuster product like ancestry-testing. In my opinion, a strategy of "one-app fits all" may not work for India. Hence, keeping in mind the various challenges and drivers of the industry, we need a consumer-centric approach for products, services and solutions that are developed based on market research and identification of consumer need and pain points. Our products, services and solutions will be built on a 5A ideological framework (Figure 5): Analytically Valid, Actionable, Affordable, Accessible and Available.

Figure 5 Customer's Product Journey

> "our aspiration for ApnaGenome is to create loved and trusted brands of consumer-centric products, services and solutions that are underpinned by the science of the genome"

This ideological framework will empower our mission, which is to empower people by making actionable benefits of personal genomic information affordable, accessible and available for guiding healthcare and lifestyle decisions. To accomplish our mission, we would face various challenges discussed in this article. In such situation of spoiled markets, rampant misinformation and poor products, these challenges are major threats to our business and long-term growth of industry. However, our core values of basing our decisions on data, respecting privacy of each consumer and focus on utility and impact would guide us in our mission. Also, we need to be cognizant about the fast-paced iterative nature of science and technology with deliberative and cautious nature of medicine. Our competitors provide us valuable learning to plan strategic and tactical details to execute our mission. We would learn from the competitors, not copy them by developing me-too products that are current norm in DTC Genomic industry. Our philosophy is to create products, services and platforms that allow continued customer engagement and not a one-time sale/report delivery. This would allow us to setup necessary frameworks, processes and systems to acquire genotype-phenotype data, which will be valuable for further research and development. In summary, our aspiration for ApnaGenome is to create loved and trusted brands of consumer-centric products, services and solutions that are underpinned by the science of the genome.

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PANDEMIC POSITIVE

Vachana Jayaram,
Associate Director-HR

For someone in the Human Resources industry for over 15 years, I have come to love everything about "HR". The most important aspect being the human connect and the relationship I have built with my team, my stakeholders and best of all, my co-workers.

However, the last 90 days have been quite challenging, discomforting or least to say, thought provoking. Imagine this - for someone who has always advocated the importance of physical presence of an HR for all official events, big or small - the birthday parties, the fun Fridays, the rewards and recognition, the welcome handshakes and the farewell hugs – I neither got a hug from my previous team, a 'will miss you' cake nor did I get that warm 'welcome aboard' handshake from the new one. All thanks to this pandemic!

While people are dealing with greater challenges, these do come across as first world problems. But what makes me wonder is that - **How are we reinventing and adapting ourselves to such situations?**

Did I miss the "normal" send-off? Of course, I did! Was I happy that my friends and colleagues ensured a virtual, equally engaging, tear-inducing-good-bye? Hell, Yeah!

That is exactly why the current pandemic situation, barring the pain the world is going through, is appealing to me. While the times are scary and fear-inflicting, everything about this is new, unprecedented. Something that none of us alive have seen or experienced in our lifetime.

Should we fear? I say we need not. Instead, let us look at this as an opportunity.

> **"An opportunity to experience, challenge, change, innovate, transform, adapt and most importantly be more positive and more receptive."**

The best lesson nature has taught us is that in any adversity, it is the fittest who survives. In the current scenario, it may not necessarily mean the body strength. It means that we make best use of the available resources to keep our minds healthy and reinvent ourselves to be relevant to the environment we are part of.

How do we do this? Learn a new life skill, engage with family, debate with anyone who stimulates a positive thought process, set some goals, go back to your hobbies, do whatever brings a smile on your face, sleep for 8 hours, de-clutter, teach, reengineer what you do and how you do. Resources required for these are easily available.

> **“All we need for a good start is to ‘think’”**

Thinking generates ideas, ideas can bring in the required innovation, and controlled innovation can transform the entire world.

If one pandemic can change the social, behavioural and economic landscape, imagine the infinite changes every day thinking can bring. We do not need world class brains to achieve this. Just someone who is willing to think, to solve a problem in hand.

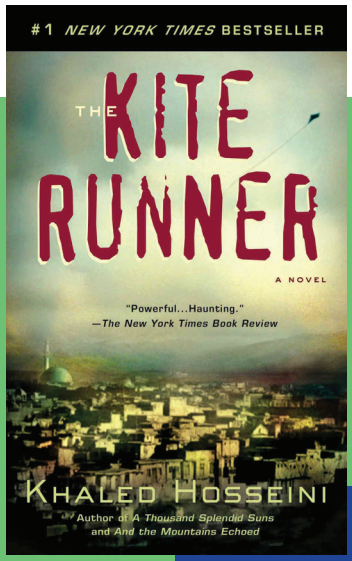
This is essential mainly because I see two groups of people thriving through this season. One group, who sees this as an opportunity to transform the way we are doing things and the other, who quickly adapts to these transformations. Just these two. The ones who are hating the change, either make the dreadful pandemic magically go away or join one of these two surviving groups.

At the risk of sounding philosophical, I believe the purpose of humans is to make the world a better place for all living organisms. For this to happen, each of us must think what positive changes we can bring in our respective areas. I also believe that we, the **‘MedGenomians’**, being the frontline warriors in this situation, have a crucial role to play in making this world a better place.

Let us innovate. Let us adapt. Let us be more positive about this pandemic.

From our Colleagues

Verbum



Book:

The Kite Runner by Khaled Hosseini

Book review by :

Dr Sanghamitra Mishra,
Senior Scientist, Operations Dept.

How often do we hear this word, ‘refugees’? Brushing it aside by turning the newspaper page, mildly worrying while looking at the gruesome picture war paints, sometimes grateful for our better luck. In a world striving for peace yet soiled by unrest, thousands of people get dislodged every day from their comfort and seek a safer shelter, but most importantly seek ‘freedom’. Often, the word ‘refugees’ is referred to as a collective noun like the whole word is not made of individual, pained, tattered souls.

The Kite runner is a story of how beauty turns to ugly and gets a chance to turn back to beautiful. Set in the backdrop of Afghanistan in its process of destruction, the story narrates of relationships, father and son, the changing friendship of two boys and the many others that the protagonist encounters in his journey to redemption. It speaks of betrayal and reclamation of peace.

Amir: "It's a sad story."

"Sad stories make good books," she said.

"They do."

Khaled Hosseini's first novel, a story about changing lives in a turbulent country, was triggered by a news report that the Taliban had banned kite flying in Afghanistan. Having grown up in his native country with the sport, the writer found this cruel and was inspired to pen one of the most devastating stories of two little boys and how political unrest changed their world. Sad or not, the 'Kite Runner' is a powerful story. It indulges between the innocent friendship, hatred, and other relationships in a changing landscape. Hassan is a pure soul who is devoted to his best friend, Amir. He is a follower, Amir's kite runner. Amir, also the narrator, is the son of Hassan's father's benefactor. He is a privileged lad who is later unfolded to be weak in friendship. The plot is gripping, nails till the end. The story and the narration speak for itself as a first round bestseller.

Hosseini's characters are simple with visible layers, if any. However, he makes up by adding characters as the story unveils, who exit from the scene and stay back in the narrator's memories and reappear at times. The writer paints the story with hurt, hatred, guilt and conscience. There are some white characters like Soraya, Hassan and Sohrab and some dark like Aseef and his likes and all others are shades between these. The hatred in the neighbourhood bully Aseef who grows up to be a Taliban and the outcome of his ingrained scruples of supremacy of his race is bone chilling, a reminder that hate sinks in early and deep.

"They called him "flat-nosed" because of Ali and Hassan's characteristic Hazara Mongoloid features. For years, that was all I knew about the Hazaras.

Then one day, I was in Baba's study, looking through his stuff, when I found one of my mother's old history books. It said the Hazaras had tried to rise against the Pashtuns in the nineteenth century, but the Pashtuns had "quelled them with unspeakable violence."

Amir is privileged, lonely and seeks validation. He finds a friend and soulmate in Hassan. Hassan on the other hand is dedicated to his Ali Agha. Although shown to be faltering in his ideals, sometimes selfish, Amir is a constantly evolving character in the novel. He has a craving for discovery. That is why he does not always accept what is around him but goes to find his own truth.

Hosseini reminds us of our own mild racisms, of where it begins, the hate for someone's food, clothes, or having a typical physical appearance like a flat nose. The hatred and the abhorrence to 'hazaras' shown by the neighbourhood kids or the indignation with which Amir's teacher dismisses a book because it doesn't furnish his views on the Hazaras are reflections of all around the world.

"They do nothing but thumb their prayer beads and recite a book written in a tongue they don't even understand." He took a sip. "God help us all if Afghanistan ever falls into their hands."

Two little boys dreading about the inevitable. Hosseini plays with words and gives contrasting epithets to the same league in a single conversation. While the Taliban use prayers to enforce their supremacy, an innocent child turns to another God for help.

I dropped next to him, lay on a thin patch of snow, wheezing. "You're wasting our time. It was going the other way, didn't you see?" Hassan popped a mulberry in his mouth. "It's coming," he said. I could hardly breathe, and he didn't even sound tired. "How do you know?" I said. "I know."

Hassan is an exceptional kite runner. Hosseini also subtly shows the pride and confidence of the simple Hazara boy, unaccepted except by his benefactors.

.... I find it hard to gaze directly at people like Hassan, people who mean every word they say.

Amir may be a coward at friendship, but he is honest with himself. He admits his hypocrisy. He is a contrast to Hassan's sincerity. They both have the dynamics of a relationship of innocence and need.

Hassan returned the smile. Except his didn't look forced. "I know," he said. And that's the thing about people who mean everything they say. They think everyone else does too.

Hassan is an epitome of innocence and sincerity, a true friend. He believes his friend is the same, unsuspecting of what was coming. Amir on the other hand

I was glad I didn't have to return his gaze. Did he know I knew? And if he knew, then what would I see if I did look in his eyes? Blame? Indignation? Or, God forbid, what I feared most: guileless devotion? That, most of all, I could not bear to see.

Amir later faces a hard journey towards freedom and learns to value and savour when freedom is reached, still haunted by the innocent guilt. The book gives an intense picture on child abuse, another social burner. Hassan and Sohrab or both had the ill fates of being hated so wildly in their innocence. A relief to the protagonist' guilt comes as a chance. **There is a way to be good again, Rahim Khan had said on the phone just before hanging up. Said it in passing, almost as an afterthought. A way to be good again.** Rahim is Amir's uncle, his father's business partner. He is Amir's friend and conscience and helps him find redemption from the guilt by introducing him to Sohrab, Hassan's lost son.

'People' is another collective made of individuals with a story, mostly different. Amir is introduced as a coward but makes a choice of changing his truth, of facing his guilt. He is one with multiple stories like many.

"Do you want me to run that kite for you?"

His Adam's apple rose and fell as he swallowed.

"For you, a thousand times over," I heard myself say.

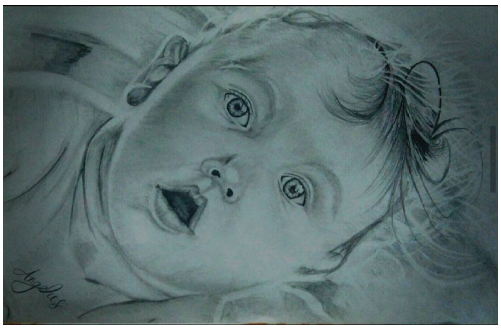
And finally, when Amir runs for Hassan's son, he partly pays back to his beloved friend.

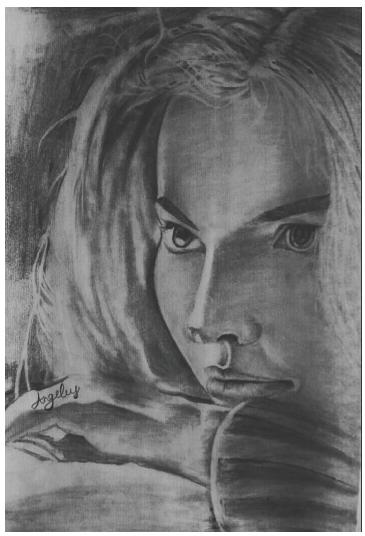
From our Colleagues

Art meets Science

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

By: Anjali M. Patil
Research Associate Trainee





By: Peri Shiva Prasad
Genome Analyst trainee

From our Colleagues

Our employee's little Picasso :)

By: Mridha Baskar (6 years)
DNA of Malini Manoharan, *Bioinformatics Scientist*

By: Smera Ponnamma (14 years)
DNA of Survesh Chinappa,
Asso. Director, Procurement & Logistics

By: Mridha Baskar (6 years)
DNA of Malini Manoharan, *Bioinformatics Scientist*

By: Shivani Muthamma (10 years)
DNA of Survesh Chinappa
Asso. Director, Procurement & Logistics

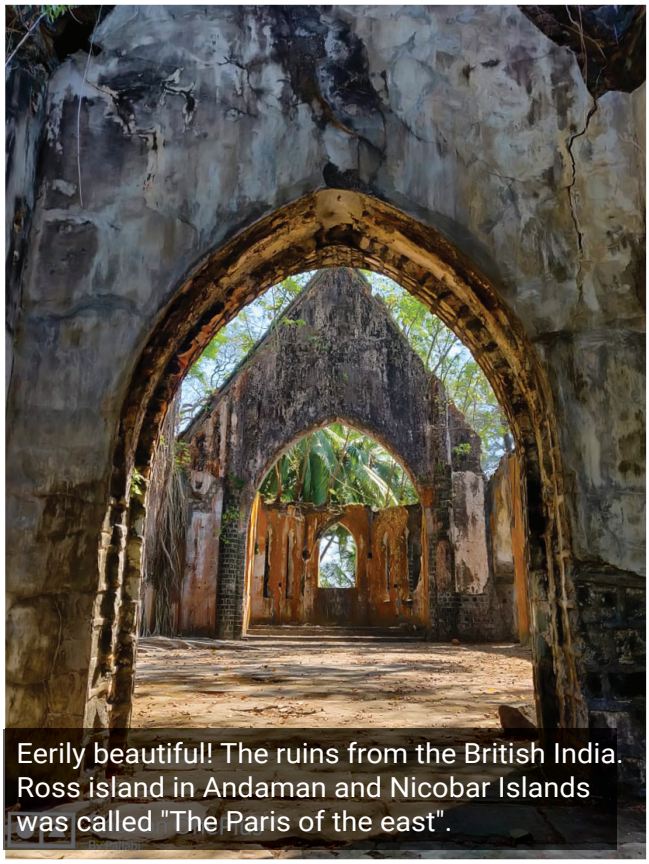
By: Mridha Baskar (6 years)
DNA of Malini Manoharan, *Bioinformatics Scientist*

From our Colleagues

By : Pallabi Dasgupta, *Project Manager*

The sky is pink, Pattaya.

Vanilla sky melting over the concrete jungle, Bangkok



Eerily beautiful! The ruins from the British India. Ross island in Andaman and Nicobar Islands was called "The Paris of the east".

A Paradise to Behold at Tiger Nest Monastery, Bhutan.

Work-life during lockdown

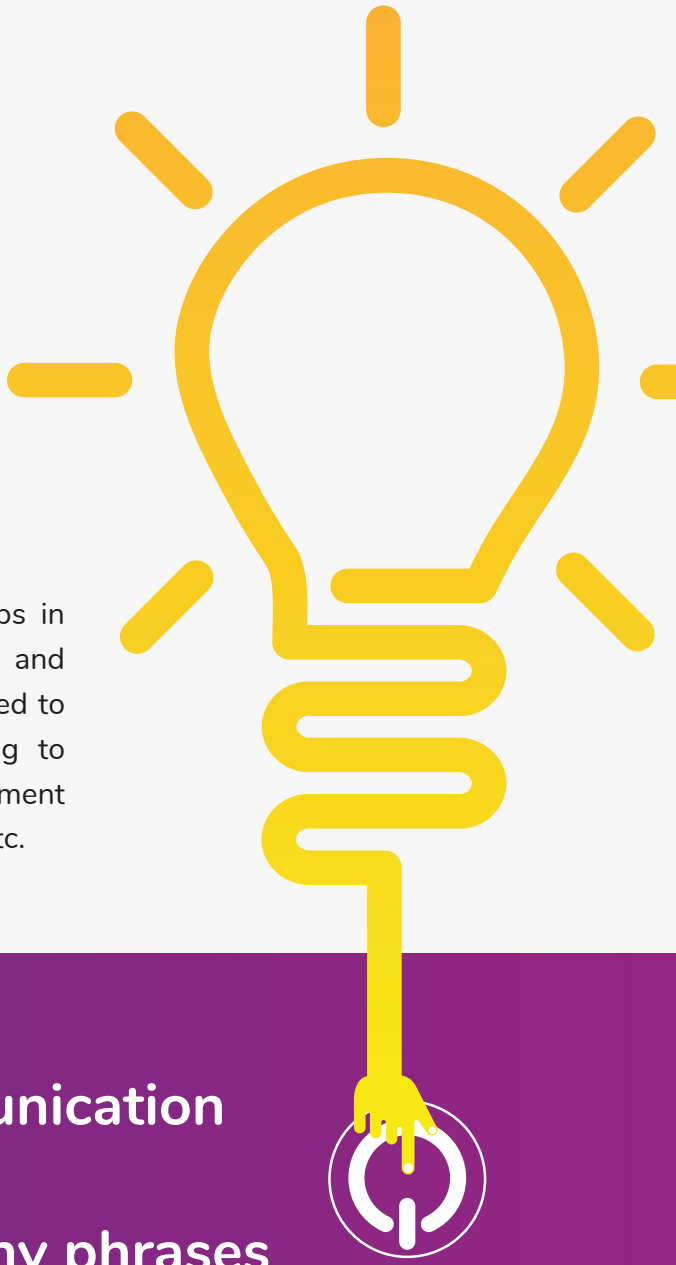
> “Tough times go away, tough people do not”
> — by Walter Payton

Glimpse of our team during lockdown. While some are working from office and others from home, but we are united by a common goal i.e. to deliver our best efforts amidst these tough time.

Naming contest

> “Great communication begins with connection”
> — Oprah Winfrey

A good communication chain at the workplace helps in increasing the productivity, performance, and self-confidence. Keeping this in mind, we have decided to create a new channel of communication pertaining to announcements, newsletter, and employee engagement activities such as events, competitions, celebrations, etc.



We are planning to create a
new email id for this communication
and welcome all of you to
suggest some smart & catchy phrases to name it!!

Exercise your grey cells to help us in giving this initiative a new name. The best idea will be chosen to create the official email id for this communication.

(For example, if the name selected is MedCom, then the email id will be “medcom@medgenome.com”)

WINNER

will receive due recognition for his/her effort

Kindly mail your suggestions by 15th July 2020 to editor@medgenome.com

Note: All HR related announcements and information regarding pay, tax, policies, important notices, etc will be circulated via hr@medgenome.com

Employee connect

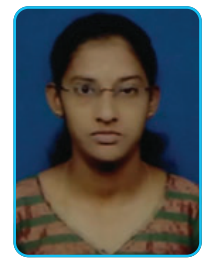
WELCOME

Our New-Joiners


Ajay Katoch


Akram Husain R S


Ayam Gupta


D Sashi


Dikshaya Prabhakara


Kaisar Gani Shaikh


Kodali Priscilla


Madhu S


Mohd Arif Khan


Mridula K P


Mukesh Prajapati


Nithil P U


Preethi Elangovan


Rajesh Suresh Sonawane


Rajesh V S


Richu Rachel Yeldos


Sajitha Suresh


Sandeep Ruhela


Sanjay Nayak


SanojK S


Trupti Suresh Inamdar

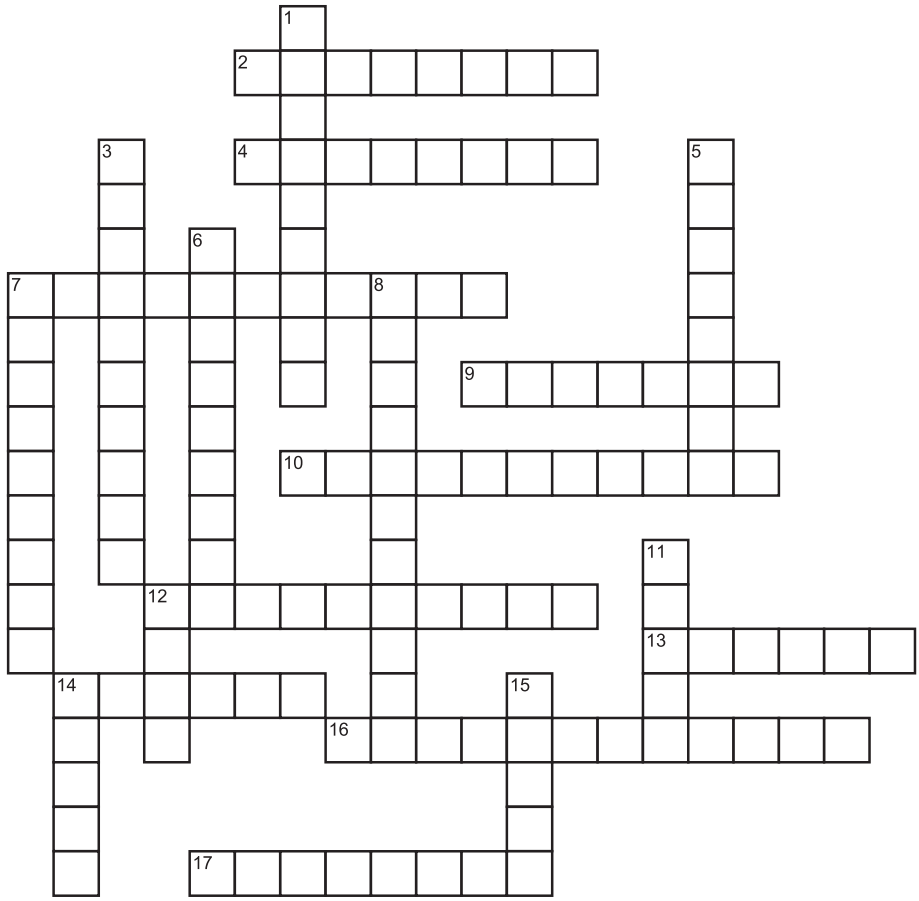

V Hamsagiri


Vachana Jayaram


Vidhyadhar Karmarkar


Vithya Natarajan

Employee connect



Down

1. The trait that is hidden when other traits are preset.
3. Two copies of the same allele.
5. Separate units.
6. An image of chromosomes.
7. The physical appearance of a living thing.
8. Genetic traits are _____ from a parent.
11. Stores female reproductive cells.
12. Region of DNA where instructions for one trait are kept.
14. Paired male and female cells for reproductive purposes.
15. Characteristic like hair freckles or dimples.

Down

2. The genetic makup of a living thing.
4. The field of biology that studies how genes control appearance.
7. The likelihood that an event will happen.
9. Different versions of a gene.
10. Long molecules made of DNA that hold genes.
12. All the individuals born at the same time.
13. The part of the flower that creates pollen.
14. A monk who experimented with pea plants.
16. Two different alleles for a trait.
17. The trait that is visible when other traits are present.

Previous Winner

Himadri Baul, *GA Trainee*

Kindly mail your answers by 31st July 2020 to editor@medgenome.com. The first two people to answer the puzzles correct will be featured in the next edition of our newsletter.

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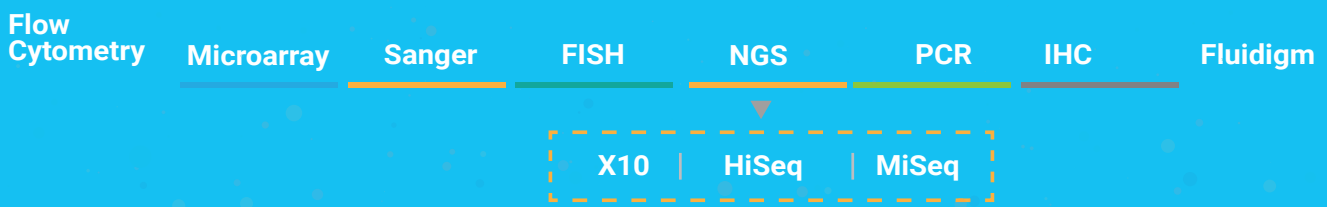

INDIA

Bangalore
Chennai
Delhi
Kochi
Mumbai

US

Foster City

SINGAPORE