MEDGENOME

Vol 06 | July 2018
www.MedGenome.com

GeKNOWme

Internal Quarterly Newsletter

Management speaks

DR. AMITABHA CHAUDHURI
VP, R&D

Dear MedGenome Colleagues,

This year MedGenome will be five years old (officially incorporated in November 2013). Being the first MedGenome employee in the US, I feel proud of what we have achieved so far, both in the US and in India. Let me start with the US lab, which has grown from a three-member team working in a shared incubator space in San Francisco, to a 22-member team occupying an independent building in Foster City, California.

A separate immuno-oncology team works side-by-side with the Next-generation sequencing lab leveraging NGS to make inroads into the complex mechanism of immune-mediated tumor clearance. We have added a 10X Genomics single-cell sequencing platform beginning of this year to help ease out interesting biology. The clinical lab in Foster City has put up a CLIA-certified WES assay by working closely with many groups in MedGenome Labs in India. Many of you may not realize the herculean effort and the teamwork required to get one test certified and validated. It is a tremendous achievement by the MedGenome Labs, Bangalore to bring close to 300 tests accredited by CAP.

Having built a strong foundation in leveraging NGS for diagnostics and research, supported by a very talented team of bench scientists, bioinformaticians and software engineers, where do we see ourselves in the next five years? The vision of MedGenome, as laid out by our Founder and Chairman, Mr. Sam Santhosh is to leverage the unique genetic structure of India to investigate the biology of complex diseases. The mandate is to identify and understand the global needs in discovering novel disease biology and then support global organizations with data to enrich their drug discovery and development pipelines. Many novel therapies have come from studying isolated disease populations.

We have taken up this challenge and forged many clinical collaborations and partnerships, set up genetic centers to capture rich clinical data and through GenomeAsia initiative have started investigating the molecular genetic landscape of the Indian population and their impact on disease prevalence. We have also set up initiatives in few globally relevant disease segments led by experienced Disease Heads and given them the freedom to build the disease programs keeping aligned with the vision of the company. The Ophthalmology disease program has launched Ophthatome last month at the ARVO conference in the US. This is our third knowledge base after OncoMD (cancer) and Diabetome (diabetes).

Finally, the success of a science-driven company is not measured by revenue alone, but also by its ability to innovate, build intellectual property, publish original papers in peer-reviewed journals and present novel findings in scientific meetings. We have some measure of success in all the above areas, which need to be boosted and bolstered further as we explore the uncharted territories of human diseases and disorders. We have successfully nurtured three critical foundations to deliver good science – advanced technological platforms, innovative and passionate scientists to take on challenging problems, and the ability to work together as a team to become more efficient and productive. Let us all work together to solve challenging problems in human health and have fun doing it!

Highlights

Most talked about

MedGenome in news

1

Cheaper, faster DNA tests revolutionising diagnoses

Charges Come Down By More Than Half

Genomics is causing a revolution in medicine and healthcare globally "and it's happening now", said an Australian research paper published on Friday

Malathy.Iyer@timesgroup.com

Mumbai: Delhi IT professional Salim Khan's considers his brush with genomics at KEM Hospital in Parel a medical wonder. He had painfully accepted that medicines wouldn't work for his incurable neurological condition, myasthenia gravis. But seven years into his suffering, a subsidized genetic test costing just Rs 20,000 revealed that the doctors in Delhi hadn't diagnosed his disease hasn't had an attack in a year, and he has gained weight.

Affordable, rapid DNA sequencing has not only changed Khan's life, but that of eight other patients at KEM's neurology department over the last year. "Previously, we didn't have such an easy and affordable access to genetic tests. It would take months for an accurate diagnosis," said Dr Ravat.

Genomics is causing a revolution in medicine and healthcare—their genome—has plummeted from hundreds of millions of dollars to around the cost of a shoulder MRI," the report said.

V P Ramprasad of Medgenome Labs in Bengaluru pointed out that 15 years ago, genetic tests from India were sent abroad, cost about Rs 1 lakh, and the results would take four to six months. Today, better technology and infrastructure means tests are done locally, take two to three years. And the benefits are being felt all around.

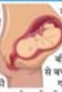
Seven-year-old Renu suffered over 30 epileptic fits every day and took five different anti-epileptic pills. The daughter of a farmer from Shegaon in Maharashtra's Buldana district, she would close her eyes and appear to fall asleep every few minutes. "If this happened when she was standing or sitting, she would fall and hurt herself. Her school asked us to keep her back at home as she would keep falling," said her father, P Shelke.

When the Shelkes were referred to KEM Hospital seven months back, Dr Ravat's team immediately sought a genetic test. Three weeks later, the test results showed that Renu had protein deficiency called

2

पैदा होने से पहले ही पता चलेगी बच्चे की बीमारी

गर्भावस्था के नौवें हफ्ते में ही संतान में संभावित असामान्यता का पता लगाना अब संभव



[illegible]

इन अस्पतालों को अध्ययन में किया गया शामिल

[illegible]

2

HEALTH ZONE

Dr Tulika Tayal, Dr Priya Kadam and Dr V Madhavi

Prenatal screening essential for pregnant women: Experts

More than 38,000 babies are born in India each year with the Down Syndrome

OUR BUREAU

A group of medical experts have strongly stressed the need for prenatal screening of pregnant women to identify chromosomal disorders in babies before birth. Doctors from various hospitals of the city attended in an event organised by MedGenome Diagnostics Company and opined their views on the same.

The doctors informed that the majority of genetic disorders are untreatable and impose a huge emotional and financial burden on the family. Pre-natal genetic screening can identify such chromosomal abnormalities in the foetus. While all couples who have conceived should ideally opt for pre-natal genetic screening, it is strongly recommended for those with advanced maternal age, previous positive family history and other high-risk factors for early detection of potential disorders, said the doctors.

The incidence of chromosomal disorders in India is 1:166 per live births. India being one of the highest birthing countries in the world, there is greater incidence of Down Syndrome, which occurs in approximately 1 out of 830 live births.

"More than 38,000 babies are born in India each year with the Down Syndrome. Yet, only 50 per cent of pregnant Indian women undergo full antenatal check-up. Prenatal screening will let them take informed decision in case of seriously affected pregnancy. Even if they decide to continue with the pregnancy, the prenatal test gives them an opportunity to adjust to this information before the baby is born, and plan the treatment, in case of some disorders, well before birth. Also, if the test is normal, it reassures the mother about the health of the baby. Prenatal screening has a major role to play in bringing down the incidence of chromosomal disorders," said Vasikarla Madhavi, Geneticist, Fernandez Hospital.

On the other hand, Noninvasive Prenatal Test is a high-performance screening test and is highly accurate and can be done as early as the 10th week of gestation and carries no risk of miscarriage. Although it is highly accurate, but it's not a diagnostic test. NIPT tests are performed with a simple draw of blood from the mother.

Dr Priya Kadam added, "Government needs to take an active part in spreading awareness about the importance of genetic testing, provide funds for R and D and cover genetic diagnostic tests under health insurance and by engaging private and government hospitals."

, 22 June 2018
per.thehansindia.com/c/29725338

3

Battling thalassaemia

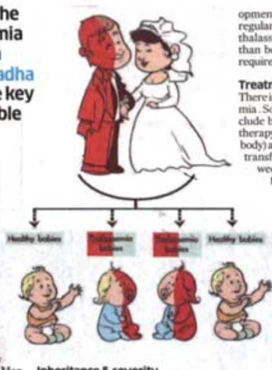
BLOOD BASICS With the number of thalassaemia cases on the rise each year, Dr Sheetal Sharadha says awareness is the key to prevent the incurable disorder

India has one of the largest numbers of people suffering from thalassaemia. There are over 40 million carriers and over 100,000 thalassaemia majors undergo blood transfusion every month. Keeping this in mind, it is important to create awareness about the disease.

Thalassaemia is an inherited blood disorder in which the body makes a decreased quantity of haemoglobin. In India, thalassaemia is one of the causes for haemolytic anaemia amongst infants. This form of anaemia is associated with an excessive number of red blood cells being destroyed by the body (due to defective haemoglobin). Marriages with close members of the family or with members amongst the local communities result in at least 3-17% of thalassaemia cases in India.

The types

Normal adult haemoglobin consists of



Inheritance & severity

Thalassaemia major is inherited in an autosomal recessive manner. Being an autosomal recessive condition means that two copies of the defective gene are required for the disorder to occur in a severe form. If only one copy of the gene is defective, it is known as thalassaemia minor.

opment. Affected children will require regular lifelong blood transfusions. Beta thalassaemia intermedia is less severe than beta-thalassaemia major and may require episodic blood transfusions.

Treatment

There is no permanent cure for thalassaemia . Some of the available treatments include blood transfusions, iron chelation therapy (removal of excess iron from the body) and bone marrow transplant. Blood transfusions at intervals of two to three weeks are prescribed to patients affected with thalassaemia major. However, with consistent blood transfusions, the iron content of the body increases and there is a likelihood of the excess iron causing problems to vital organs like the heart and the liver. Under such conditions, experts suggest iron chelation therapy to remove the excess iron from the body.

Bone marrow transplants can be curative for some children with beta-thalassaemia major. It is an expensive therapy and involves matching a compatible bone marrow donor with the patient.

Prevention

Couples planning to get married should undergo carrier testing along with genetic counselling. This is critical in case of thalassaemia, as carriers are at a greater risk of having a child with thalassaemia major especially if they are married to another

1. **Dr Ram quoted in The Times of India on how genetic tests are aiding in better diagnosis**
   https://timesofindia.indiatimes.com/city/mumbai/cheaper-faster-dna-tests-make-diagnosis-better/articleshow/64056698.cms

2. **Story on NIPT research study in Navodaya Times and Hans India basis the Press conference held in Delhi and Hyderabad**
   https://diagnostics.MedGenome.com/pdf/2018/HansIndiaJune22Hyderabad.pdf

3. **Authored article on Thalassemia by Dr Sheetal in Deccan Herald**
   https://www.deccanherald.com/living/health-and-well-being/battling-thalassaemia-671668.html

'Genetic testing can help improve the survival rate'

ST CORRESPONDENT
reporters@sakaaltimes.com

Pune: "Genetic testing and targeted therapy drugs can significantly increase the survival rate of lung cancer patients. Cases of lung cancer are rapidly increasing in India due to rampant smoking, including active and passive smoking," said doctors.

LUNG AND OTHER CANCERS

Dr Vidya Veldore, Principal Scientist at Oncology with MedGenome, said most cancers, including lung cancer, arise due to genetic mutations.

"Once the cancer-causing the side-effects of chemotherapy," said Dr Veldore.

"So there is an urgent need to create awareness about the importance of genetic testing for not only lung cancer patients but also in other cancers," Dr Veldore further added.

She added that genetic testing has resulted in a paradigm shift in the treatment and management of cancer.

Speaking about the threat to public health posed by tobacco consumption, which further causes lung cancer, Dr Shailesh Bondarde, Medical Oncologist, Apex Wellness Rishikesh Hospital, Nashik, said India accounts for one-sixth of the 6 million tofor 42 per cent of all male deaths due to cancer and 18.3 per cent of all female deaths. Two most common cancers caused by tobacco are mouth cancer and lung cancer. Tobacco does not harm the individual alone. It also increases health care costs and decreases productivity," said Dr Bondarde.

He added that lung cancer is now treated in a personalised way, unlike earlier when the same line of treatment was used in all cases.

"Lung cancer does not show symptoms till it has reached an advanced stage when it becomes difficult to treat or manage. Symptoms such as a persistent cough, chest pain and ...ness

Decoding MedGenome's Genomics and Fundraising in India

Interaction with Sam Santhosh of MedGenome regarding the genomics of venture capital fund raising

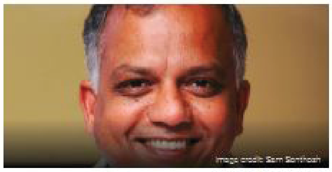


Image credit: Sam Santhosh

Rahul R
Senior Correspondent

April 10 · 5 min read

Opinions expressed by Entrepreneur contributors are their own.

Has anyone heard of the business of deciphering your DNA ?,You heard it right, your Genes.That's the talk of the town and people are investing in that, not just in the US but in India as well.

In March 2018, San Francisco-based Genomics venture MedGenome (with labs in Bangalore, and Singapore) raised USD 10 Million in a Series C round with investment from HDFC Ltd., HDFC Life and HDFC Asset Management to complete Series C funding of USD 40 million. Now, it is worth noting that MedGenome has been in existence from 2013 and has completed Series A and Series B in addition to two Series C rounds relatively rapidly by wooing investors through intuitive business models.

In a bid to understand MedGenome's business model, and to analyse the startup's fundraising mechanism, Entrepreneur India interacted with Sam Santhosh who is

weekend **Wellness**

SUNDAY | MAY 20, 2018

THE FREE PRESS JOURNAL

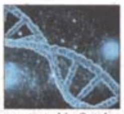
Decoding diabetes...

Maturity Onset Diabetes of the Young (MODY) is the commonest form of monogenic diabetes, and a breakthrough study leads to the discovery of a novel MODY gene, the NKX6-1. SHILLPI A SINGH finds more about this new entrant

WHAT IS MATURITY ONSET DIABETES OF THE YOUNG

Mutation causing pre-cancerous condition found in Guj family

Times News Network



The research said in people with the gene mutation, the condition could gradually turn malignant, leading to colorectal cancer by the age of 40-50

Bengaluru: Researchers from a Bengaluru company and a Vadodara hospital have discovered a genetic mutation responsible for a rare pre-cancerous inherited condition in six members of a family.

Considered a breakthrough in cancer research, the study showed that six members of a Gujarati family were suffering from Familial Adenomatous Polyposis (FAP), a rare pre-cancerous condition which accounts for 1%-3% of colorectal cancer cases. The research began when a 52-year-old patient, Paresh (name changed), complained of weight loss and change in bowel movement in 2014. Diagnosis showed an abnormal growth in his colon, which was a benign polyp which could turn cancerous.

The research, conducted by Bengaluru-based MedGenome Labs and Vadodara-based Kailash Cancer Hospital and Research Centre, was possible because Paresh's family members agreed to share his clinical history and blood samples. A genetic analysis of 25 people from his family aged 6-60 was done. While the first patient was tested in October 2015, the entire family was tested in July 2016. MedGenome took about four weeks to identify the mutation in one female and five male members.

The analysis revealed mutation of Adenomatous Polyposis Coli (APC) gene present in six people who were diagnosed with FAP. It was also found in four other family members who didn't have FAP. However, no one in the family suffered from cancer, including Paresh who was screened first, but members were prone to it given their genetic disposition.

The research said in people with the gene mutation, 100-1000 benign growths appear in the colon and rectum in teenage years, and the condition could gradually turn malignant, leading to colorectal cancer by the age of 40-50.

Dr Rakshit Shah, surgical oncologist, KCHRC, Vadodara, said, "Detection of this genetic mutation in a patient will enable us to identify individuals most vulnerable to FAP and colorectal cancer. Preventive measures can be taken so that the incidence can be brought down," he said. According to him, every individual in the family of a patient suffering from colorectal cancer must undergo genetic analysis.

Under normal conditions, the APC gene is a tumour-suppressor one which prevents uncontrolled growth of cells. But mutations in same gene lead to malfunction, the scientists said.

"Our analysis revealed the presence of a mutation in the APC gene which hasn't been identified before. It underscored the pow...

4 Story in Sakal Times on how genetic testing can help lung cancer patients

5 Mr Sam Santhosh interview in Entrepreneur magazine
https://www.entrepreneur.com/article/311664

6 Story in The Free Press Journal on MODY research paper
http://www.freepressjournal.in/health/decoding-diabetes-mody-is-the-commonest-form-of-monogenic-diabetes-read-to-find-out-more/1279612

7 Story on our Goraj case study in the Times of India
https://timesofindia.indiatimes.com/city/bengaluru/bluru-lab-discovers-gene-mutation-causing-pre-cancerous-condition-in-gujarati-family/articleshow/64375366.cmsl

MedGenome connect

ACTIA

Claria and Actia business team meeting at NN office

Between March and May 2018, Actia organized six CMEs/RTMs (primarily on Neurology & Nephrology) and gained nationwide visibility by participating in 2 conferences (NeuroXchange and PIDCON), in Delhi, Bangalore, Chennai, Kolkata, Hyderabad, Trivandrum and Jaipur. With this, we could reach out to nearly 750 clinicians across India with an intent to drive awareness on the usage of genetic testing in their clinical practice.

With an endeavor to strengthen our dominance in the therapy areas we operate in and to kick-start venture into new therapy areas like Ophthalmology and Endocrinology, the Budget meeting for FY 2018-19 was organized at Bangalore office from 24th - 26th April. Both Actia and Claria teams came together on one forum and demonstrated their intent of synergy and commitment to make this financial year a memorable one.

Coming 90 days promise to be exciting as Actia will venture aggressively into Tier 2 towns to spread awareness on the importance of genetic testing among clinicians. The concerted effort of Actia and Claria will pave way for this. With the launch of new tests (KT & FISH), enhanced coverage of clinicians in new therapy areas like Endocrinology and Ophthalmology, inclusion of new tests in our services and path-breaking KOL engagement programs, the Actians will have fire in their bellies to define new paradigms of success!

PRIMA

The quarter April - June was quite exciting and full of events for Team Prima. We participated in some important national and international conferences across India, while the prominent one being, ONCOPATH at Aluva, Kerala, where we had our stall. The visibility and awareness on Prima and its offerings was boosted with our presence in this events. The major therapies touch-based through these engagement programs were Solid tumours and haematological malignancies. We could reach out to more than 100 clinicians through our participation.

MedGenome Sales team along with Dr Boben Thomas at ONCOPATH, Kerala

CLARIA

L to R: Dr. Priya & Dr. Hema Purandarey at MOGS conference held in Mumbai, June, 2018 and Dr. Priya speaking at MOGS

The April to June period was a busy one for Claria with close to 15 events happening during this period. We started with a round table meeting on PGS/PGD by Dr. Sam Balu for doctors at Gunjan IVF in Ghaziabad, which was leveraged in the press through local media. Dr Balu was also the speaker at the Dialogue conference organised by Chennai Women's Clinic and Scan Center. While, Dr. Priya Kadam was invited to give a talk on Genetic testing at the CME Rajarajeshwari Medical College and to Cloud Nine Hospital, Pune and the Kovai Medical Center, Coimbatore respectively for a talk on NIPT.

On the conference circuit we made our presence felt at The Federation of Obstetric and Gynaecological Societies of India (FOGSI) event at PGI Chandigarh and at the Mumbai ObGyn society conference where Dr. Priya Kadam and Dr. Hema Purandarey gave talks on NIPT and Genetic screening respectively.

In addition to our regular events we also had a series of press conferences in Delhi and Hyderabad respectively, to highlight the findings of our NIPT paper that was published earlier this year. Dr. Priya along with the local collaborators addressed the conference. Further, we also organised a Webinar on the Screening for Twin pregnancies using NIPT addressed by Dr Priya and a separate Q&A session with Dr. Priya and Dr. Sam, where we invited questions from doctors on their doubts in the area of genetic diagnostics.

Making a difference

Genetic testing aids in treatment of a seven-year-old suffering from Fanconi Anemia

Master John (name changed), a seven-year-old boy was referred to Dr. Sachin S Jadhav, a leading Haematologist and Bone Marrow Transplant (BMT) physician, with severe deficiency of red blood cells, white blood cells, and platelets. Clinical examination was done, and his bone marrow was tested immediately. It was found that there was a failure of bone marrow development leading to the deficiency of all types of blood cells in John. As there are several reasons for bone marrow failure, a Cytogenetic stress test was done which revealed that the bone marrow failure is actually due to DNA damage. So Dr. Jadhav decided to opt for genetic testing to understand the problem.

Genetic testing at MedGenome Labs in Bangalore revealed a mutation (a disease causing change) in a gene called *FANCG* that is responsible for DNA repair. It was also observed that in John, both the copies of this gene had the mutation. He had no normal copy of *FANCG* gene to repair his damaged DNA. He was diagnosed with Fanconi Anemia.

Fanconi anemia is a rare genetic disease that mainly affects the bone marrow resulting in decreased production of all types of blood cells. The condition is most often diagnosed in children between 2 and 15 years old. Due to the abnormal gene the cells cannot repair their damaged DNA. It is an inherited disorder, where in both the copies of the gene need to be faulty for disease to manifest itself. It is known as an autosomal recessive genetic disorder with 25% risk of recurrence in every pregnancy. Thus to inherit Fanconi anemia, a person must get 1 copy of the abnormal gene from each parent.

When his parents were tested, it was found that they were carriers for this disease that is, one copy of *FANCG* gene was mutated. A carrier is a normal healthy individual who has one copy of a disease causing mutation. His maternal grandfather, paternal grandmother and paternal aunt were also found to be carriers on further investigation.

In the United States the carrier frequency or heterozygote frequency for Fanconi anemia is 1:181 whereas in Israel it is 1:93. Though there is no such data available in the Indian population, the number of patients or families with Fanconi anemia in India is rather high. This could be due to a relatively high degree of consanguineous marriages, especially in South India. One study from Mumbai revealed a significantly high frequency (36.4%) of parental consanguinity in Fanconi anemia patients compared to controls (3.33%) in our country.

Once Dr. Jadhav came to know that an abnormal gene was the cause of Master John's condition, he realised that Anti-Thymocyte Globulin (ATG) therapy will not help this boy. The only cure then is a bone marrow transplant, and that too with a reduced intensity conditioning chemotherapy regimen, since patients with Fanconi anaemia cannot tolerate the usual doses of chemotherapy. Also, by testing the family members, he realized that almost everyone in the family was a carrier, and hence he needs to look for an unrelated donor. Master John is currently stable and an allogeneic Bone Marrow Transplant is being planned.

From our US office

We are happy to share with you that our in-house developed proprietary platform OncoPept™ has been awarded the winner of the 2018 MedTech Breakthrough Awards for 'Best Overall Genomics Solution.'

With over 3,000 global nominations this year for the MedTech Breakthrough Awards, the competition was incredibly fierce. Notable winners from other categories were GE, WebMD, FitBit, Philips, 23andMe, Humana, Abbott, Apple and an impressive list of top companies and startups in the MedTech industry.

Our OncoPept™ platform is used by researchers to help identify biomarkers aimed at delivering personalized therapies including more durable, improved and responsive cancer immunotherapy treatments.

We presented our cancer immunotherapy solutions capabilities at the Next Gen Immuno-Oncology Congress Conference in Boston and discussed our Diabetome knowledge base at ADA in Orlando. MedGenome also hosted a symposium in Foster City office in May 2018, where Dr Henry Lynch talked about "Lynch Syndrome: Diagnosis, treatment and management", Dr Kishore Guda presented on "*In silico to In vivo*: Systems Approaches in decoding complex Gastrointestinal malignancies" and Dr Aju Mathew presented on "Applications of Genomics in Breast Cancer Clinic".

Sneak peek into the world of science

Ophthatome – A knowledgebase for ophthalmic disease research

by Dr. N. Soumittra, Disease Head - Ophthalmology

Medical Big Data

Big data and big data analytics are the buzz words that we have been hearing for the past few years, which have relevance in all fields and specialities. In the field of medicine, the process of clinical documentation and analysis have been very meticulous and exhaustive in the past contributing to major discoveries in associating diseases with genes, understanding disease epidemiology and in generating and testing hypothesis. The advances in computational science and data processing have streamlined the management of medical big data creating opportunities to impact the health care system with accurate prognostication and disease management. The sources of medical big data are electronic medical records, medical imaging, clinical registries, large clinical trials, large epidemiological studies and administrative claim records. Medical big data applications include rational clinical decision, predictive or prognostic modelling of disease progression, disease surveillance, public health and research.

MedGenome has taken a lead to create disease-specific knowledgebases using large unstructured clinical datasets to initiate hypothesis-driven research in complex diseases.

Ophthatome

Ophthatome – a knowledgebase for ophthalmic disease research was launched in the Association of Research in Vision and Ophthalmology (ARVO) annual meeting, held at Honolulu, USA in April 2018. This knowledgebase of ocular diseases is a comprehensive collection of clinical, phenotype and biochemical data providing researchers and clinicians with a platform to design studies that address critical unmet needs in eye disorders. Ophthatome currently contains curated clinical and phenotype data of 581,466 cases that includes 524 disease types and 1800 disease subtypes, covering 35 different eye parts and more than 40 clinical variables. Nearly half of the total cohort have longitudinal data with a maximum of five-year follow-up.

The searchable interface enables performing complex queries to select specific disease cohorts based on demographics, disease types and subtypes, disease course or severity, specific tissues affected by the disease, drug response and many other clinical and phenotypic parameters.

The knowledgebase provides options to select cohorts with specific well-defined quantitative and qualitative traits apart from disease types and subtypes. The availability of clinically well-defined disease cohorts facilitates powerful genomic, pharmacogenomic and clinical research to discover novel biology in ocular diseases.

The database is continuously updated with new data - ~100,000 additional cases will be added in the next 2-3 months along with further enhancement of features, pedigrees and genetic data where available.

Ophthatome Team

Ophthatome is developed by MedGenome in collaboration with Narayana Nethralaya, a tertiary eye care hospital and research institute, Bengaluru. The ophthatome knowledgebase is built on the electronic medical record data. We thank Drs. Arkasubhra Ghosh, P. Narendra, Sushma Tejwani, Mr. Sankar Das and Ajith Shetty from Narayana Nethralaya for sharing the data and helping us on data mapping and clarifications on data clean up. A big appreciation to my ophthatome team members, Mr B. Muthu Narayanan, Mr. S. Praveen Raj, Mr. J. Somasekhar, Mr. T. Chandrasekhar, Mr. Durgesh Kumar, Mr.G. Raghunathan, Mr. Paul George, Mr. Laj Mathew, for the development and Mr. Hiranjith, Mr. Michael Nemzek, Dr. Ankita Das, Ms. Angelica LaVallee, Dr. Amit Chaudhuri for the launch at ARVO.

Familial Studies in MedGenome

by Dr. Sameer Phalke, Senior Scientist

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes (changes in the number or structure of entire chromosomes, the structures that carry genes).

As we unlock the secrets of the human genome (the complete set of human genes), we are learning that nearly all diseases have a genetic component. Some diseases are caused by mutations that are inherited from the parents and are present in an individual at birth, like sickle cell disease, cystic fibrosis, Tay-Sachs disease etc. Other diseases are caused by acquired mutations in a gene or group of genes that occur during a person's life. Such mutations are not inherited from a parent but occur either randomly or due to some environmental exposure (such as cigarette smoke). These include many cancers, as well as some forms of neurofibromatosis.

Over last decades next generation sequencing has revolutionized the human genomics. With reducing cost and increasing throughputs of Next generation sequencing (NGS) technologies one can obtain a whole genome or exome (coding part of the genomes) sequence under USD 1000 within a week. This dramatic progress in sequencing technologies has enabled opportunities to design and address questions to identify gene mutations responsible for different rare and neglected familial genetic disorders etc., which will be useful in managing human health and disease [1,2,3].

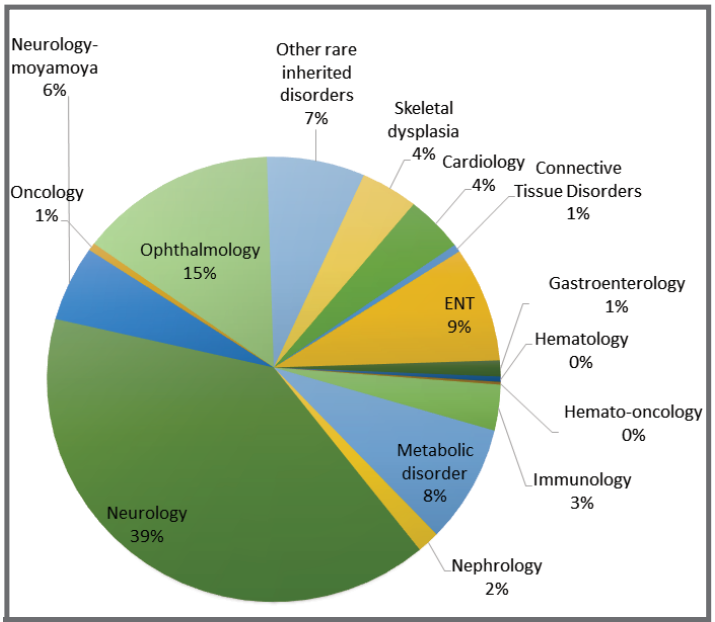


Figure 1

Despite the rich patient resource for Mendelian diseases available in the country, discovery of disease causal variant(s) in different genes have also been suboptimal. This has contributed to our limited understanding of disease biology in general which in turn has hampered novel drug development. To address these issues MedGenome undertook a project on Familial Genetic Disorders Study (FGDS) across Indian families. The focus of this study is to identify novel genetic alterations which leads to several rare mendelian genetic disorder using whole genome and whole exome sequencing. For this project we collaborated with several clinicians and hospitals across India and based on some inclusion/exclusion criteria we have already been able to recruit more than thousand families. Diseases covered in this study range from Neurology, Ophthalmology, Cardiology and many other disease areas (Figure 1). One small cohort in Ophthalmology disease category consists of families which were diagnosed with Autosomal Recessive Bestrophinopathy (ARB).

ARB (MIM 611809) is an ocular condition that usually manifests in the first two decades of life, but may become symptomatic as late as the fifth decade[4, 5, 6]. Clinical manifestation of ARB is characterized by central visual loss with typical hyperopic conditions, sub-retinal lipofuscin deposits that are predominantly outside the macula, absence of light rise in EOG, reduced ERG, accumulation of fluid within and/or beneath the neurosensory retina and development of angle-closure glaucoma[7, 8].

Mutations in *BEST1* gene have been described in a variety of ocular disease phenotypes including *ARB4*, Best vitelliform macular dystrophy *(BVMD)*[9], autosomal dominant vitreoretinochoroidopathy[10], autosomal dominant microcornea, rod-cone dystrophy, early-onset cataract posterior staphyloma syndrome and retinitis pigmentosa[11, 12].

The *BEST1* (bestrophin-1) protein is expressed in the basolateral plasma membrane of the retinal pigment epithelium (RPE) where it regulates multiple functions essential for normal vision. It primarily functions as a calcium-activated chloride channel.

ARB may manifest as the result of a total absence (null phenotype) of functional bestrophin-1 protein in the RPE, improper localization to the cell membrane with intact anion channel activity or lack of channel activity specifically. Among the roughly 270 mutations reported in *BEST1* thus far, only about 40 compound heterozygous and homozygous mutations have been associated with ARB.

As part of our familial genetic disorder studies (FGDS) effort we studied four unrelated families in which more than one individual was clinically diagnosed with Bestrophinopathy. Figure 2 exemplifies one of family in this cohort. Overall 20 individuals with 8 affected and 12 unaffected members were analysed using whole exome sequencing (Agilent SSV5 exome panel).

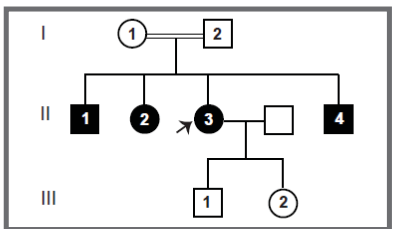


Figure 2

Exome sequence analysis identified five mutations in *BEST1* (p.Tyr131Cys, p.Arg150Pro, p.Arg47His, p.Val216Ile and p.Thr91Ile) in the affected individuals. Three of these five mutations (p.Tyr131Cys, p.Arg150Pro and p.Val216Ile) have not been previously reported. Inheritance pattern of *BEST1* mutations confirmed the diagnosis of ARB in probands in 3 of the 4 families, while the inheritance of heterozygous *BEST1* mutation in one of the families was suggestive of BVMD. In coherence with recessive phenotype, interestingly, 1 of the 4 ARB families was a compound heterozygote carrying a mutation in canonical isoform and the other mutation in an alternate *BEST1* transcript isoform, highlighting, for the first time, a role for alternate *BEST1* transcripts in bestrophinopathies. Our report expands the list of pathogenic *BEST1* genotypes and the associated clinical diagnosis. Findings of this study have been published in Scientific Reports recently[13].

MedGenome's familial genetic disorder study has several such small cohorts with rare genetic conditions. Several such small cohort analysis are being done currently. Our sequencing efforts will definitely bring in new insights into our current understanding of disease biology, diagnosis and potential gene therapies for many of the genetic disorders in Indian context.

References

1. Genomic approaches for understanding the genetics of complex disease. Lowe WL Jr, Reddy TE.Genome Res.2015 Oct;25(10):1432-41. doi: 10.1101/gr.190603.115.
2. Exome sequencing as a tool for Mendelian disease gene discovery. Bamshad*et. al., Nature Reviews Genetics*12, 745-755 (November 2011) | doi:10.1038/nrg3031
3. Unlocking Mendelian disease using exome sequencing. Gilissen*et al. Genome Biology*2011, 12:228
4. A Novel BEST1 Mutation in Autosomal Recessive Bestrophinopathy. Lee CS,*et al. Invest Ophthalmol Vis Sci*56, 8141-8150 (2015):2
5. Biallelic mutation of BEST1 causes a distinct retinopathy in humans. Burgess R,*et al. The American Journal of Human Genetics*82, 19-31 (2008).15
6. Childhood-onset autosomal recessive bestrophinopathy. Borman AD,*et al Archives of Ophthalmology*129, 1088-1093 (2011).16
7. Autosomal recessive bestrophinopathy associated with angle-closure glaucoma. Crowley C, *et al. Documenta Ophthalmologica 129, 57-63 (2014). 17*
8. Autosomal recessive vitelliform macular dystrophy in a large cohort of vitelliform macular dystrophy patients. Kinnick TR, *et al. Retina* 31, 581-595 (2011).18
9. Identification of the gene responsible for Best macular dystrophy. Petrukhin K, *et al. Nature genetics* 19, 241-247 (1998). 3
10. Mutations of VMD2 splicing regulators cause nanophthalmos and autosomal dominant vitreoretinochoroidopathy (ADVIRC). Yardley J, et al. Invest Ophthalmol Vis Sci 45, 3683-3689 (2004). 5
11. The spectrum of ocular phenotypes caused by mutations in the BEST1 gene. Boon CJ, Klevering BJ, Leroy BP, Hoyng CB, Keunen JE, den Hollander AI. Progress in retinal and eye research 28, 187-205 (2009).
12. Missense mutations in a retinal pigment epithelium protein, bestrophin-1, cause retinitis pigmentosa. Davidson AE, et al. The American Journal of Human Genetics 85, 581-592 (2009).
13. Next generation sequencing identifies novel disease-associated BEST1 mutations in Bestrophinopathy patients. *Thong et. al., Scientific Reports, in press* (2018)

Sneak peek into the world of science

Autoimmune Diseases: The Known and the Unknown

by Dr. Sudhanshu Shukla, Scientist (R&D) Autoimmune Diseases & Dr. Lakshmi Mahadevan, Principal Scientist

Autoimmune pathologies occur when immune cells lose their ability to distinguish between self and non-self antigens and attack single or multiple organs resulting in compromised functions of the targeted organs in affected individuals. More than 100 autoimmune diseases have been identified till date. Some autoimmune diseases target specific organs, for example, muscle in Myasthenia Gravis, pancreas in Type 1 Diabetes, the thyroid gland in Grave's disease and gastro-intestinal tract in Ulcerative Colitis. Few autoimmune pathologies like Systemic Lupus Erythematosus may affect different parts of the body including joints, blood vessels, kidneys, lungs and nervous system. In general, the exact molecular events leading to pathogenesis of autoimmune diseases are complex and remain largely unknown till date.

Autoimmune diseases (ADs) have been reported to affect 5 -10 % of the world population, with the prevalence being up to 75% more in females than males. Globally, the estimated total incidence across all autoimmune diseases is 0.09% per year. Incidences and prevalence have increased significantly over the last 30 years. The age of onset varies in different autoimmune diseases, with some like type 1 diabetes developing primarily in childhood and adolescence, whereas others like myasthenia gravis and multiple sclerosis occurring in mid-adult years and few others like rheumatoid arthritis seen more among older adults.

Majority of autoimmune diseases are predicted to be the result of interplay between polygenic risk factors (involvement of multiple genes) and environmental factors making it difficult to identify the key genetic determinants of the disease and understand its pathogenesis. The contribution of genetics in autoimmune diseases has come from multiple lines of evidence. First, existence of few rare monogenic autoimmune diseases such as Autoimmune Polyendocrinopathy, Autoimmune Lymphoproliferative Syndrome and Immunodysregulation Polyendocrinopathy X Linked Syndrome has increased our knowledge about the disease biology to some extent. Second, many studies have shown that the risk of developing autoimmune diseases is increased in first degree family members, with the highest concordance (24-50%) between monozygotic twins than in dizygotic twins. Third, evidences such as the co-existence of different ADs within family members indicate that common genes and similar pathogenic pathways underlie multiple autoimmune diseases.

The current treatment methods used in ADs focus on controlling the overactive immune response and in ameliorating inflammation but fail to cure. Clinicians face many challenges related to timely diagnosis and treatment of patients suffering from autoimmune pathologies. This is attributed to insufficient knowledge about the disease mechanism and non-uniform patient responses to the same treatment regimen. Most AD treatments are symptoms-based and lack of unreliable diagnostic parameters to distinguish disease-specific symptoms at early stages of the disease or during disease relapse, reduce treatment efficacy in most autoimmune diseases. Research to treat and manage ADs has focused on inhibiting inflammatory cytokine production or blocking their functions, depletion of B-cells, inhibition of B-cell functions including their activation and proliferation, blocking autoantibody production and blocking T-cell activation. These studies have led to the use of therapies for disease management, such as Methotrexate, an inhibitor of folic acid metabolism or Azathioprine, an inhibitor of DNA synthesis to inhibit cell proliferation; Belimumab, which binds to BAFF ligand and inhibits proliferation of B cells; Anti CD-20 antibody, which depletes matured B cells; CTLA4-immunoglobulin (Abatacept), which binds with CD80 & 86 receptors on Antigen Presenting Cells thereby blocking the interaction of these receptors with CD28 on T cells and leading to inhibition of T-cell activation and B-cell immune responses; neutralizing antibodies to TNF (Remicade,

Enbrel, Humira and Simponi) and IL-6 (Tocilizumab and Siltuximab); and Ustekinumab, which blocks IL-12 pathway. Few studies using gene therapy approaches in animal models are also underway for treating autoimmune diseases. However, the vast knowledge gap in our understanding of autoimmune diseases hampers designing novel strategies for effective treatment.

Previous genome-wide linkage studies such as the "Human Genome Diversity Project", "HapMap Project", "HUGO Pan-Asian Project" and "1000 Genomes Project" conducted in highly heterogeneous ethnic populations to analyze human genetic variations have helped in the identification of new genetic risk variants in many autoimmune disorders. Paradoxically, India which constitutes almost one fifth of the world population has been under represented in these studies. A large and ethnically diverse population like India's, with an estimated 5-10% people suffering from common and rare autoimmune pathologies, which is predicted to increase due to changing life style and environmental factors, harbors several advantages to study genetics and etiology of autoimmune diseases. At the same time, AD research can truly leverage the existence of a large number of ethnic groups (population isolates) in India who have maintained genetic homogeneity through generations by endogamy. The stratified Indian population therefore provides a unique opportunity to investigate the underlying molecular mechanisms of familial autoimmune diseases, which in turn will provide a framework for understanding & dissecting the undiscovered aspects of autoimmune disease biology in detail.

The study of complex diseases has been revolutionized with the cost-effectiveness, high throughput, accuracy and large-scale data output offered by Next Generation Sequencing (NGS). GWAS studies, with a minimal representation of the Indian population, identified autoimmune diseases associated nucleotide changes but could not be easily linked to molecular & cellular causal down-stream events. Sequencing based fine mapping studies conducted so far have revealed that almost 90% of the autoimmune associated SNPs reside in non-coding regions and ~ 40% of this affects cis-eQTLs (expressed Quantitative Traits Loci) and on few occasions trans-eQTLs. Additionally, eQTL studies have been directed towards mainly protein coding genes ignoring non-coding RNAs, which constitutes almost 65% of transcribed RNA and is predicted to impact gene expression. These large genomic datasets can be reanalyzed with available information from other NGS studies to generate testable hypothesis to develop novel treatment strategies or finding specific drug targets for autoimmune diseases.

MedGenome Labs with its state-of-the-art NGS sequencing and data analysis platforms is uniquely poised to investigate some of the pressing questions in the AD field to fill the knowledge gap and to support next generation autoimmune disease research. Using NGS based technologies such as whole genome, whole exome, whole transcriptome and whole epigenome sequencing, we plan to understand the molecular mechanisms that lead to dysregulation of the immune response in the context of autoimmune diseases. This is expected to be helpful in identifying and prioritizing drug targets for autoimmune diseases by re-analyzing the findings from GWAS studies and focusing on variants in the non-coding regions of the genome. We plan to also select autoimmune diseases prevalent in isolated populations to discover novel genetic determinants of the disease. Further, we plan to leverage the power of single cell sequencing to characterize the phenotype of immune cell subtypes causal to the disease. Taken together, MedGenome's autoimmune disease program is looking ahead to leverage the unique population structure of India along with advanced technological potential to unravel the complex interplay between genetics and environment causing autoimmune diseases.

From our Colleague

Yoga - the Science vs. the Myth

by Vinay CG, Manager - Content and Communications, US Presales Support

More than any subject Yoga has been shrouded in mystery for centuries. From levitating yogis to himalayan saints with unexplained supernatural powers have captured the attention of curious minds not only in India but also in the West. The quest to attain pure and supreme consciousness has always been the goal for these yogis – thus making them to endure extreme physical pain and harsh environments. Often the success attributed to their physical prowess and concentrated mind is the intense practice of Yoga they undertake for years together. Yoga has always been their tool to reach this state which they call the "Samadhi" – the final layer of yogic practice. It is said the person who achieves such a state goes beyond physical dimensions into the realm of the unknown, where boundaries and limitations vanish and what remains is just supreme bliss. Different forms and traditions have been around and many have contributed to its evolution and what we see as a modern version of poses today is a kind of import from the west. Yes, India exported its Gurus to the west – such as BKS Iyengar, Pattabhi Jois, TKV Deshikachar, Swami Rama, Bikram and many more who made it so famous that today Yoga is considered as a form of beneficial exercise in many Western Universities. The west adapted to this new form of stretching, bending and inverting exercises so fast that they created a cult of yogis. The first generation started to travel to India to study deeper and carried it to the west thus making it a popular exercise form. Thirumalai Krishnamacharya was a pivotal figure in rejuvenating the lost science of yoga and perfected it so well so that it could reach a common man. The Mysore Maharajas encouraged Yoga and due to their constant support Thirumalai Krishnamacharya established the first ever studio of yoga in the Mysore Palace **(Figure 1)**. His disciples like BKS Iyengar, Pattabhi Jois and TKV Desikachar could further spread yoga to all corners of the world and the rest as it is said is History.

Figure 1: Yoga display at Mysore Yoga Studio – Students of Thirumalai Krisnamacharya

Interestingly, we don't know how yoga was born, but the first glimpses of the asana – famously known as the Bhadrasana – one among the most difficult poses in yoga is found in one of the oldest artefacts excavated from the Indus Valley. This particular figure as shown in the image here is called "Pashupathi" – the king of animals/beasts (Figure 2). He sits in the majestic Bhadrasana. This earlier relic and many more similar to these being dated more than 3000-4000 years is a testament to their evolved consciousness and the ability to observe their own body in all its minuteness.

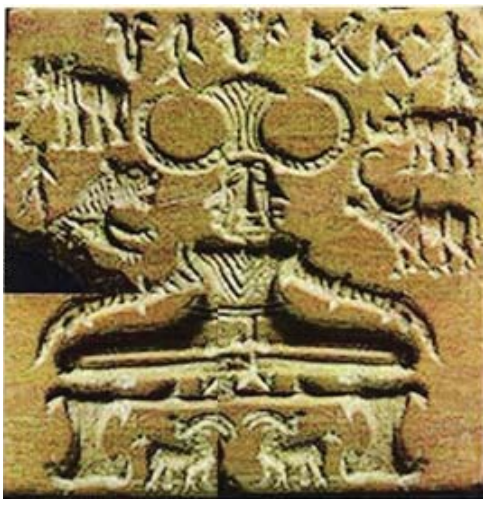


Figure 2: Pashupathi in Bhadrasana.

It is said Kapila – a sage was the first to recognise and systemise yoga, however, the credit of recording and scripting goes to "Patanjali" – the man who gave a form and shape to this ancient knowledge.

From our Colleague

In his yogasutras the first line says "Now begins yoga". He never said anything more than that, thus preventing one to assume that he will offer something sublime and uniquely unfathomable space. Patanjali further says Yoga aids in "Chittha Vritti Nirodah" – meaning "Yoga is the cessation of the mind. Then the witness is established in itself". The tradition of yoga has always been an individual abstraction rather than a collective movement. Patanjali belonged to the school of Sankhya – one among the 6 dharshan philosophies that has been handed over to us by our ancestors.

Patanjali's 8 limbs of Yoga

The Sankhyans were skeptics and were thoroughly scientific in their approach in understanding the Nature. They observed that knowledge can be gained by "Prathyaksha" or direct perception, "anuman" or inference and by "Shabda" or word. Grounded in the notion that there is dualistic reality one that of "Purush"(self) and the other "Prakriti" (matter) the Sankhyans went on to divide the matter into what we now usually use in our modern day jargons as "Sattvic", "Rajasic" and "Tamasic". They are bound to be present in both animate and inanimate.

(The deeper philosphies and the extensive literature related to yoga's evolution is beyond the scope of this article and hence a superficial overview is provided for the reader to know the lineage and history.)

Good to know. But, how does it matter to me and you is the obvious question one may ask in the era where modern science and technology surrounds us everywhere. The modern day reader's questions are more existential like: Why do the Himalayan yogis live longer and if they do live long also why should I bother, does the arousal of Kundalini bestow super human powers? Does practicing Tantra yoga increase virility in humans? And finally how does it matter to me whether India celebrates an international day of yoga or not.

May be the first answers to these questions comes from the methodical studies conducted both in India and abroad on the effects of yoga on human body. Though the data obtained so far lacks the kind of systematic study done in clinical trials – but they talk more and provide us a starting point to discover what has been neglected so far domestically and elsewhere.

Delinking the myths and the perversions such as levitating yoga (Figure 3) has been their foremost concern and a systematic scientific inquiry into yoga was much needed.

The year 1851 marks the first systematic study by Nobin Chander Paul – a young medical student who was intrigued by a Yogi's capacity to go into a deep burial for 40 days under the observance of the then Maharaja of Punjab Ranjith Singh. This particular wandering yogi caught the attention of many including NC Paul. He recorded his observations in a book titled "A Treatise on the yoga Philosophy". The challenging poses and purifications in yoga were least of his concerns but he was more focused on something that was not observed and recorded so far. The ability of a yogi to reach the state similar to animal hibernation by manipulating the breath. Yes, by practicing intense Pranayama a yogi could bottle up CO_2 inside the body – which helps in lowering the metabolism much like animals who seal off their burrows in their period of hibernation. This also showed the kind of trance like state experienced by yogis inside the cave. This simplest explanation provided the much needed relief from the existing myth of yogic supernatural powers.


Figure 3: People jumping from spring mattresses – mimicking levitation

Later on in 1926, Jagannath Gune reported the headstand (Figure 4) and shoulder stand (Figure 5) promoted blood circulation but not high pressure – thus making yoga a

form of exercise that can promote physical vitality. Debunking the stories of many such as the much acclaimed "Autobiography of an Yogi" - the long-time friend of Swami Yogananda – Basu Kumar Bagchi alias Dhirananda – who became a scientist at University of Michingan (1957) later leaving Yogananda's ashram conducted several studies on the effects of yoga and pranayama on human respiratory system – reporting yogis can slowdown the life basics such as respiration and heart rate but they couldn't stop the heart from beating. A finding which contradicted ages of miraculous claims that yogis can stop their heart.

Figure 4:
Headstand (without support)

Figure 5:
Shoulder stand displayed by BKS Iyengar.

The year 1975 onwards saw studies conducted at Harvard by a certain doctor by name Herbert Benson who reported that the meditators could lower their respiratory rate, blood pressure and oxygen consumption.

Over the years it has been reported in many scientific studies that yoga does have a significant effect on the human body and mind. One such significant study came from Duke University Medical Centre in 1980s where a control group with designated physical activities were compared with the experimental group who did yoga. The measurement was taken before and after the training period and they had found that the control group improved in terms of VO_2 max significantly while the yogis, despite their poor performance felt better about themselves than the control group. This was very intriguing to the scientists and the yogis even reported enhanced sleep, better health, endurance and flexibility. The duke findings were indeed interesting because just physical exercises were not enough to affect the human physiology but the feel good factor of an individual played a much better role in gaining physical health.

The NIH from 1998 started public funding for the study of Yoga and many new findings are being discovered on the effects of yoga on conditions such as diabetes, arthritis, insomnia, depression and chronic pain.

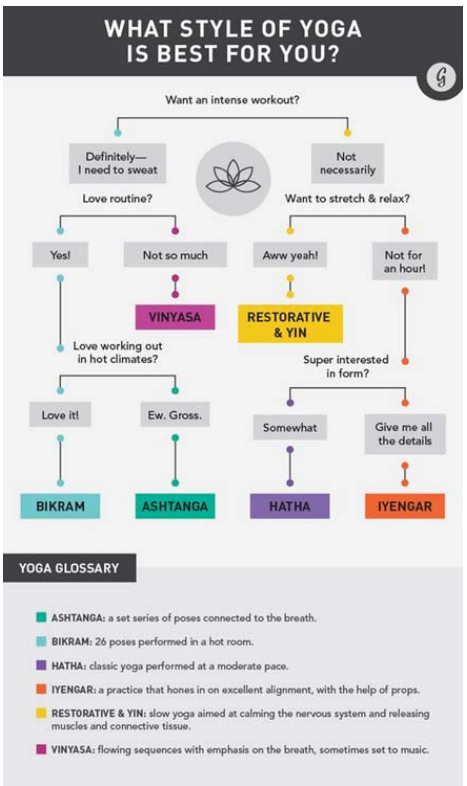
Overall, yoga has made significant strides in the west and today 20.4 million Americans practice yoga every day. Yoga combined with Pranayama has been profoundly helpful in addressing common issues such as back pain, digestive issues, muscular pain, fatigue, mood disorders and arthritis.

The modern definition for yoga is best explained in the quote: "If you want to define yoga in terms of adjustment, then it is not an adjustment with the society but an adjustment with the existence itself. It is an adjustment with the whole".

So, it can be concluded that yoga is nothing but a form of physical exercise which promotes overall well-being and deepens one's ability to look at his or her body and mind in a totally different way with no myths attached.

Yoga is existential, experimental. No belief is required, no faith is needed—only courage to experience.

Modern Styles of Yoga Forms – How do I choose?

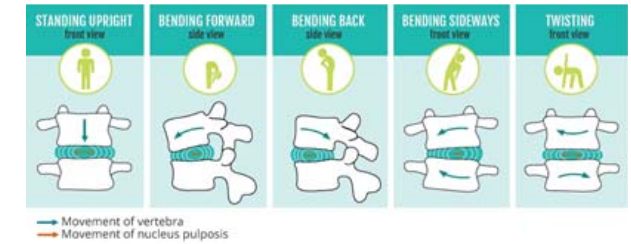


From our Colleague

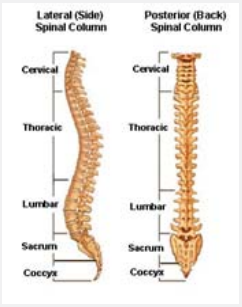
WEST VS EAST

Push Up	Chaturanga Dandasana
Plank	Dwipada Prasaranasana
Superman Flutter Kicks	Shalabasana
Lunge	Virabhadrasana
Crunch	Navasana

Before you do Yoga keep in mind how your spine works for you:



Spinal Massage through Yoga



Anatomy of the Spine

Yoga Poses for Back Pain *

Common Asanas to target different parts of the spine

Sitting Parvatasana
(Mountain Pose)

Bhujangasana
(Cobra Pose)

Marjariasana
(Cat pose)

Ustrasana
(Camel pose)

Supta Vajrasana
(reclining thunderbolt pose)

Purvotanasana
(intense eastward facing stretch)

Bharadvajasana
(named after a vrishi –
brings nourishment to your back)

Urdhva Mukha Svanasana
(Upward facing dog pose)

Shalbasana
(locust or grasshopper pose)

Dhanurasana
(Bow Pose)

Halasana (Plow Pose)

* *Please note these should be performed under the guidance of a professional yoga teacher.*

Consult a doctor to understand your spinal health before you want to pursue an intense yoga program.

From our Colleague

Employee Health Safety And Environment

by Fazal Mohammed Asif, Manager - Quality Assurance and
Srinivasan Rajendran, Senior Manager - Quality Assurance

We take pride of being associated with the second lab in India with CAP accreditation & the largest lab with CAP scope in ASIA for Genomics.

Safety is the state of being "Safe", the condition of being protected from harm or other non-desirable outcomes.

Employee Health Safety and Environment- It is a discipline and specialty that studies and implements practical aspects of environmental protection and safety at work. In simple terms it is what organizations must do to make sure that their activities do not cause harm to anyone. Regulatory requirement play a role & organisations must implement suitable safety measures.

From Health & Safety Stand point- Creating safety procedures for identifying workplace hazards and reducing accidents and exposure to harmful situations and substances. It also includes wearing of PPE (Personnel protective Equipment) like Apron, mask Goggles, Gloves and lab slippers while entering the Sample Analysis zone, and Functional Checks of every equipment's maintenance and calibration. Storage cabinet for Hazardous chemicals, Emergency Eye wash for any spillage, Fume hood and biosafety cabinet for analysis of samples. MedGenome is under 24 hours Surveillance CCTV with Access restriction.

Personnel Health Safety ensured through Annual Health Check-up, HBV Vaccination, Ergonomic evaluation & through employee health insurance.

From an environmental standpoint- It involves creating a systematic approach to complying with environmental regulations, such as handling waste management system, Daily Monitoring of temperature, Humidity and pressure of each Room, Noise checks & air quality checks for xylene are monitored on regular basis, Fire equipment are checked for its expiry.

From a Safety training Standpoint- Safety Training is made mandatory for all MedGenome Employees, includes Usage of PPE (Personnel protective Equipment), accident prevention & response, emergency preparedness, Handling of Hazardous chemicals, Spill Management, Hand wash procedure, Hand's on training and mock drill on Fire safety and lab tour to show all the safety equipment used in the lab at the time of emergency.

Scheduled Inspections are performed for every aspect of Quality and Safety, Corrective and Preventive action are taken in case of any Non-compliance. Successful **training programs & Audits** will lead the organisation towards achieving Safety goals.

It is important that 'safety' is disassociated from 'bureaucracy'.

Employee connect

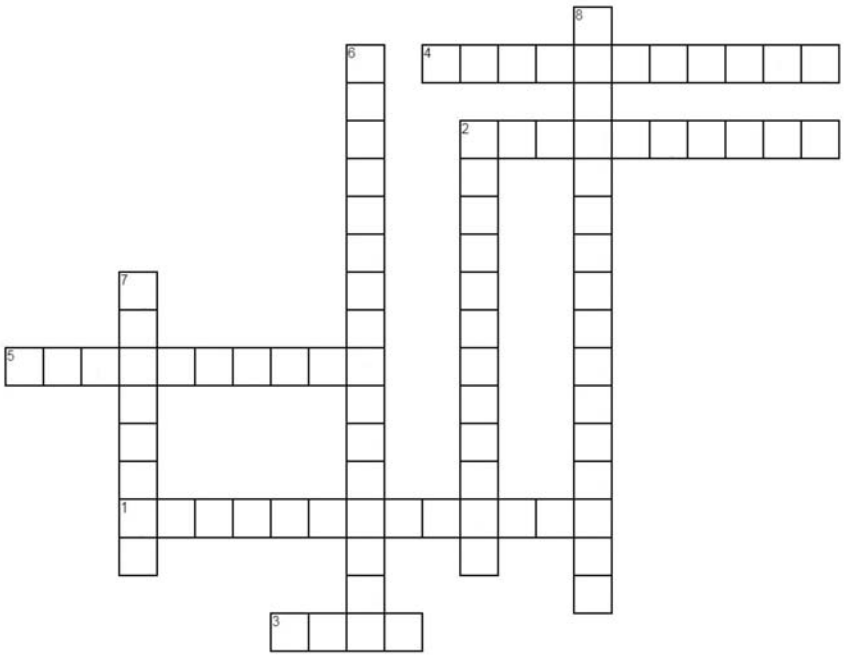
Our New-Joiners

Employee connect

Our New-Joiners

Employee connect

Cross word puzzle



Across

1 *Finding the expected proportions of possible genotypes in the offspring of a cross.*
2 *When there are two identical alleles for a trait*
3 *The original parents or the true breeding plants.*
4 *The likelihood of something happening or being the case.*
5 *Outward appearance of an organism regardless of its genes*

Down

2 *When there are two different alleles for a trait*
6 *Every individual has two alleles of each gene*
7 *Combination of genes in an organism*
8 *Genes of different traits are inherited independently*

Last Puzzle winners:

Crossword Puzzle winner

Pradeep kumar,
Manager-Sales Force Effectiveness

Spot the difference winners

Deepak Dadi,
Medical Lab Technician

Appikonda Sri Hari Chandan,
Research Associate

Kindly mail your answers to editor@MedGenome.com. The first two people to answer the puzzles correct will be featured in the next edition of our newsletter.

Employee connect

Congratulations to our recent Ph.D graduates

Savita Jayaram and Meeta Sunil

Accolades

One of our customer was touched by the act of our employee, Amalendu Deb, and since then has been recommending MedGenome to all their near and dear ones. Few days back, Amalendu had actually coordinated to collect 6 samples from one of the patient for checking for a donor match for his bone marrow transplant. However, after sending the sample the patient expired and on being informed of the same, Amalendu proactively got the patient's family a refund. The patient's family was moved by the gesture and was full of appreciation of him, who helped them in their testing times.

For being thoughtful, supportive and acting with integrity MedGenome appreciates Mr Amalendu Deb.

Photo feature

CELEBRATIONS

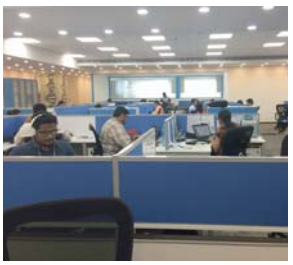
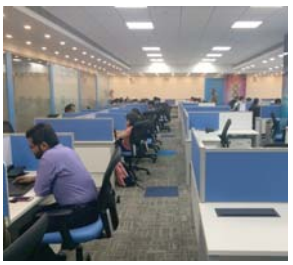
Birthday Celebrations




Environment Day




New Office - Bangalore

MEDGENOME

A market leader in Genomics-based Diagnostics and Research

One-stop solution for all your Diagnostics and Research needs

Flow Cytometry | Microarray | Sanger | FISH | NGS | PCR | IHC | Fluidigm

NGS: X10 | HiSeq | MiSeq

INDIA

Bangalore
Chennai
Delhi
Kochi

U.S

Foster City

SINGAPORE

Website: www.MedGenome.com

Email: diagnostics@MedGenome.com

Phone: 1800 103 3691