# ROAR FOR RARE: THE UNHEARD VOICES

sanofi

DISEASES THAT ARE RARE
**NEED MUCH MORE CARE.**

**COMMEMORATING**

# 25

**YEARS**

**OF**

sanofi

**TREATING GAUCHER IN INDIA**

**2024**

# INTRODUCTION

Rare diseases are serious, chronic, debilitating and life-threatening diseases that require long-term and specialized management. Their health expenditure is so high, that it would eventually impoverish the affected patients and their families. The fundamental 'Right to Health' rests on the foundation of equal rights and opportunities for all, where *no one* gets left behind.

Sanofi has been working towards enhancing awareness of rare diseases and capacity building of clinicians through innovative programs like Fellowships in Clinical Genetics; in collaboration with Society of Indian Academy of Medical Genetics and CSIR - Institute of Genomics and Integrative Biology.

Since 2013, a free diagnostic support program has been ongoing, to aid timely diagnosis of patients with rare diseases.

Sanofi India's Charitable Access Program (InCAP) is part of Sanofi's global humanitarian program [illegible] provide free treatment to patients [illegible] affliction with Lysosomal Storage Disorders (LSD) [illegible] Pompe, Fabry and MPS type one humanitarian program in India began in 1999, even though Sanofi Specialty Care did not have a local presence, then.

Globally, more than 700 people with RDs across 60 countries are in the Humanitarian Program, out of which 38 belong to India.

With this collection of real people stories who are on our humanitarian program, as well as those supported by government/institutional funding; we have attempted a glimpse into how the lives of people suffering from such debilitating disorders, have *transformed*.

This [illegible] the support from [illegible] like the States of Karnataka and Kerala, the Employee State Insurance Corporation (ESIC), the Indian Army and many such, who rose to the occasion and supported the treatment for a few of the children living with these rare diseases.

Sanofi is committed to stand strong as a health journey partner – Bringing 'hope' to those who think they have none.

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# THE UNHEARD VOICES
## STORIES OF GAUCHER FIGHTERS

**BRINGING HOPE | TRANSFORMING LIVES**

*#EveryLifeIsPrecious*

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Sanofi India's Charitable Access Program (InCAP) is part of Sanofi's global humanitarian program, through which we provide free treatment to patients in India afflicted with Lysosomal Storage Disorders (LSDs) – Gaucher, Pompe, Fabry and MPS Type I. The humanitarian program in India began in 1999, even though Sanofi Specialty Care did not have a local presence, then.

Globally, more than 700 people with RDs across 60 countries are in the Humanitarian Program, out of which ~138 beneficiaries are from India.

With this collection of real people stories who are on our humanitarian program, as well as those supported by government/institutional funding; we have attempted a glimpse into how the lives of people suffering from such debilitating disorders, have *transformed*.

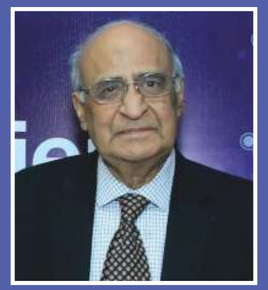
This would not have been possible without the partnership and alliance of other partners like the States of Karnataka and Kerala, the Employee State Insurance Corporation (ESIC), the Indian Army and many such, who rose to the occasion and supported the treatment for a few of the children living with these rare diseases.

Sanofi is committed to stand strong as a health journey partner – Bringing 'hope' to those who think they have none.

**BRINGING HOPE | TRANSFORMING LIVES**

*#EveryLifeIsPrecious*

# FOREWORD BY
# *Late Dr. I. C. Verma*



Professor and Senior Consultant
Advisor, Institute of Medical Genetics and Genomics
Sir Ganga Ram Hospital, New Delhi, India

I feel honored and privileged to be asked to write the foreword for this delightful book, sensitively describing the improved life of those patients of Lysosomal Storage Disorders, lucky enough to fall under Sanofi's India Humanitarian Program. The Enzyme Replacement Therapy (ERT) they received transformed their lives, enabling them to fulfil their desires and ambitions. These stories are not only inspiring, but a testimony to what modern science and technology has made possible for those who have the will to succeed. Sanofi in India deserves accolades for their generous support to this program to help patients suffering from LSDs, who otherwise cannot afford this expensive therapy.

Of course, patients with Gaucher disease achieved the best results, growing from children with bloated tummies to smart adults, to becoming engineers and scientists. Patients with Mucopolysaccharidosis Type 1 were able to flex their joints and muscles and benefitted from increased movement. Those with Fabry disease kept their hearts and kidneys strong and avoided dialysis and kidney transplantation, while those with Pompe disease strengthened their hearts and the older ones increased their mobility. Overall, each one improved the quality of his/her life.

The stories of these patients also remind us of their brethren who were not able to obtain enzyme therapy. This starkly points out the intransigence of the authorities in denying therapy to patients of LSDs, because of cost. Admittedly the enzymes are expensive, but these are not beyond the resources of the Government of India. These case histories are a remarkable demonstration of what appropriate therapies can do, and hopefully will make the authorities realize their duties towards the Constitution and towards the Father of the Nation, Mahatma Gandhi, who famously remarked that the state of our society will be judged by how we treat the poorest of the poor who lack the means of supporting themselves.

The other lesson we learn from these case histories – is that the earlier the diagnosis is made and earlier the treatment with ERT is started – best are the results. Once the physicians know that treatment would be available for those who need it, they would surely make an early diagnosis for early start of therapy, to ensure best results for their patients. They have the necessary clinical skills, but they have not felt any compelling reason to exercise it.

In the end, I congratulate Sanofi for initiating and continuing the Indian Humanitarian Program and showcasing these stories of hope and success.

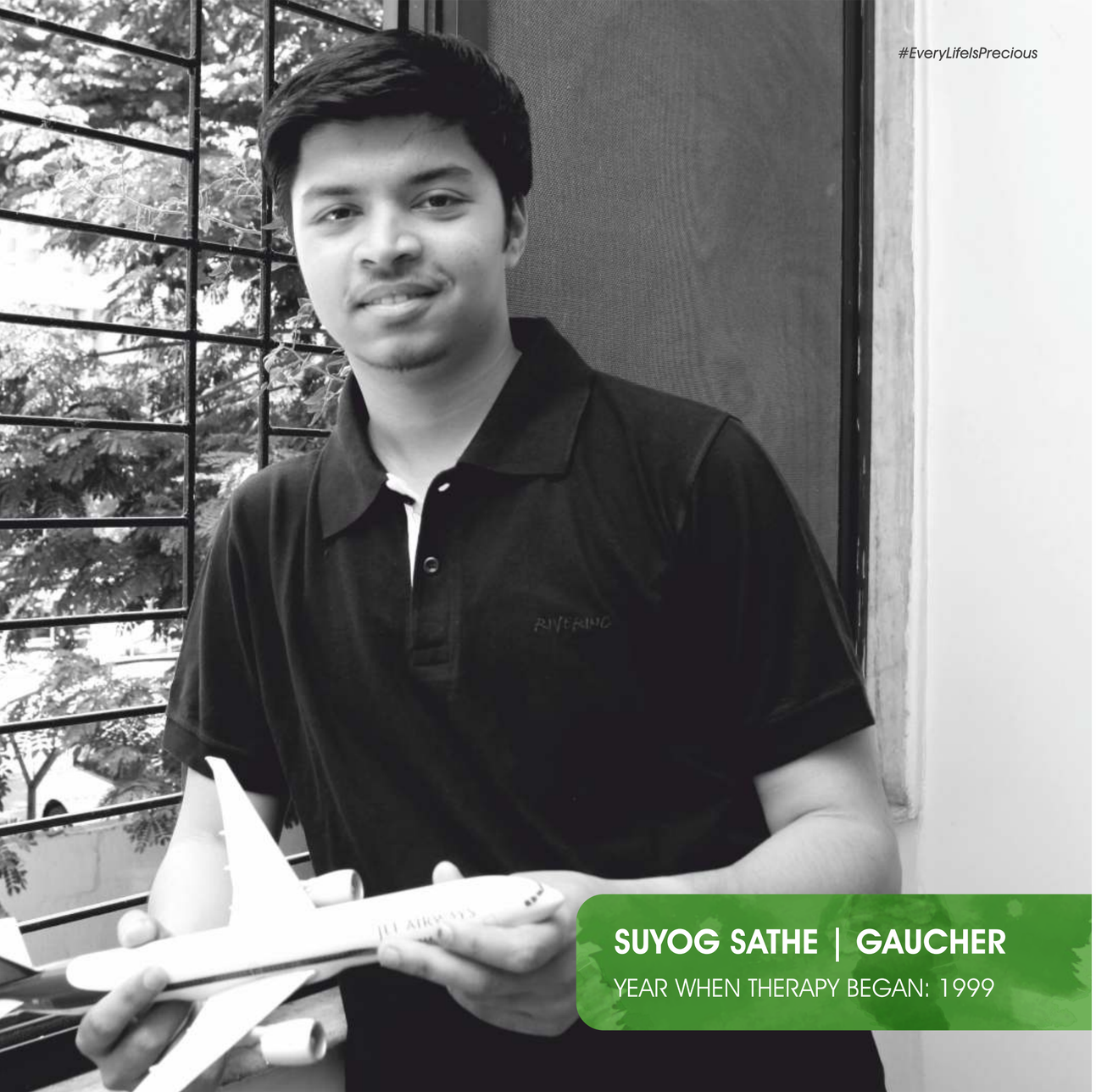
**Ishwar Verma**
Chairman, Indian Medical Advisory Board,
India Charitable Access Program (INCAP), Sanofi Specialty Care

# OUR
# GAUCHER FIGHTERS

Standing Together for a Healthier Tomorrow

*#EveryLifeIsPrecious*

*#EveryLifeIsPrecious*



**SUYOG SATHE | GAUCHER**

YEAR WHEN THERAPY BEGAN: 1999

# SUYOG SATHE

Two-and-half-year-old Suyog was severely anemic and abnormally pale. Nothing improved for him, even after a blood transfusion. It was only in 1997, when his blood sample was sent to Manchester, that he was diagnosed with Gaucher disease. By this point, his liver and spleen had swollen considerably, and he had difficulty in breathing.

Many internet searches later, his parents heard of and wrote to Genzyme USA pleading for their son's life to be saved through the enzyme replacement therapy. They were delighted to hear back that their son would not only receive treatment assistance, but that it would be completely free.

By early 1999, Suyog's therapy began and he was the first recipient of this treatment from Sanofi Specialty Care in India.

Today, Suyog leads a healthy life. He is an electronic engineering graduate and works at an IT firm in Mumbai. He also works as a Regional Manager – South Asia, for the International Gaucher Alliance.

He aspires to climb the corporate ladder, while he fuels his hobbies of travelling, music and photography.

# VILOL MALAVDE

At the age of three, when treating Vilol for high fever, doctors also detected a swelling on his liver, by chance. After many more tests and examinations, he was diagnosed with Gaucher disease.

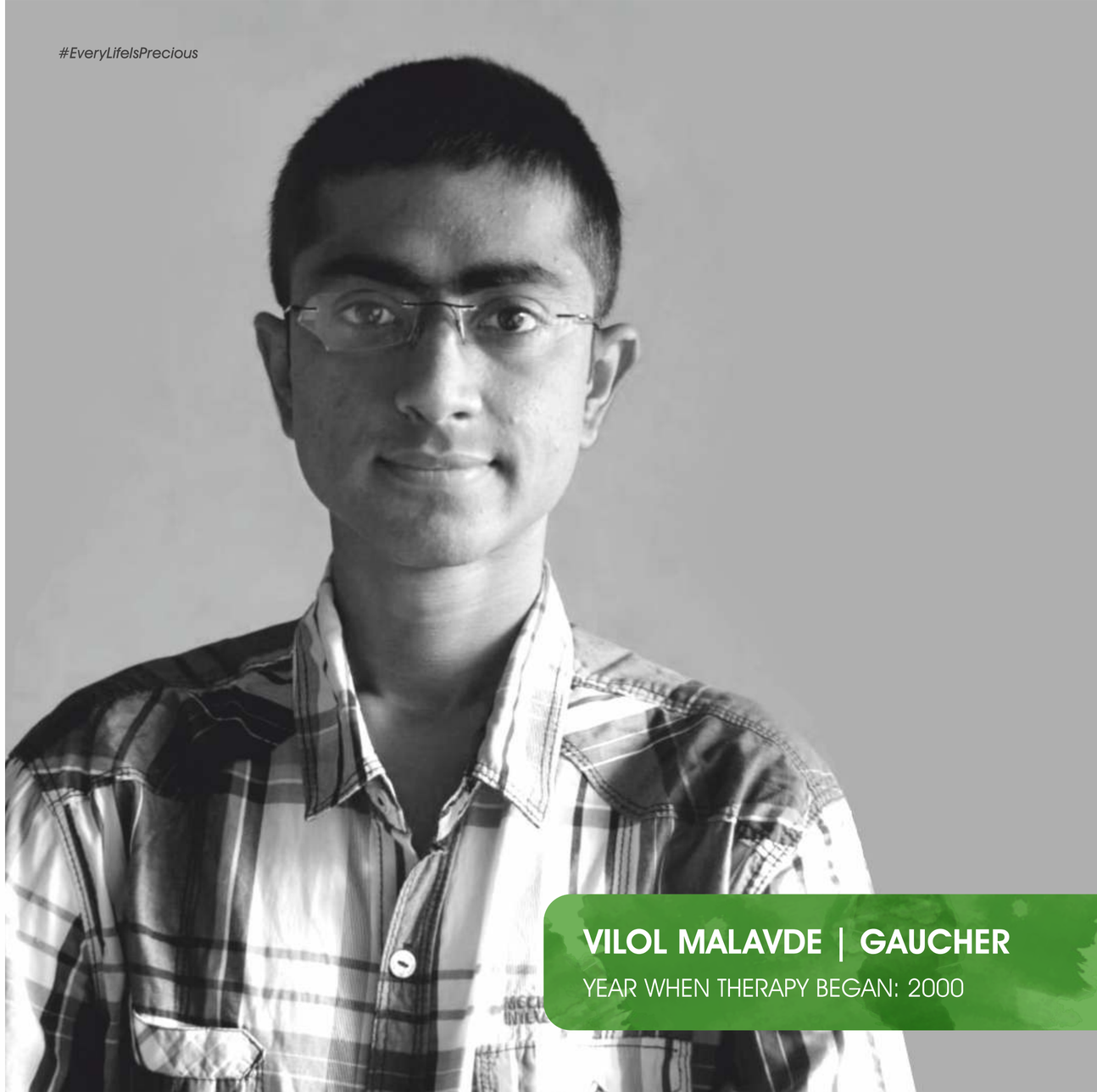
As Vilol grew older, his condition worsened. Following a severe pain in his legs and joints, bleeding from the nose, and vomiting blood, he had a brain hemorrhage. This left him in a coma for three days, and doctors feared that he would not survive beyond three months.

Lying on a hospital bed, when young Vilol asked his father if he would survive, his father knew he was going to do everything in his capacity to save his child.

Fortunately, Vilol recovered from coma, but his health kept deteriorating. The struggle continued until he received an infusion at the age of 13. Since then, Vilol has lead a near-normal life. He completed his education on time and currently works in Mumbai.

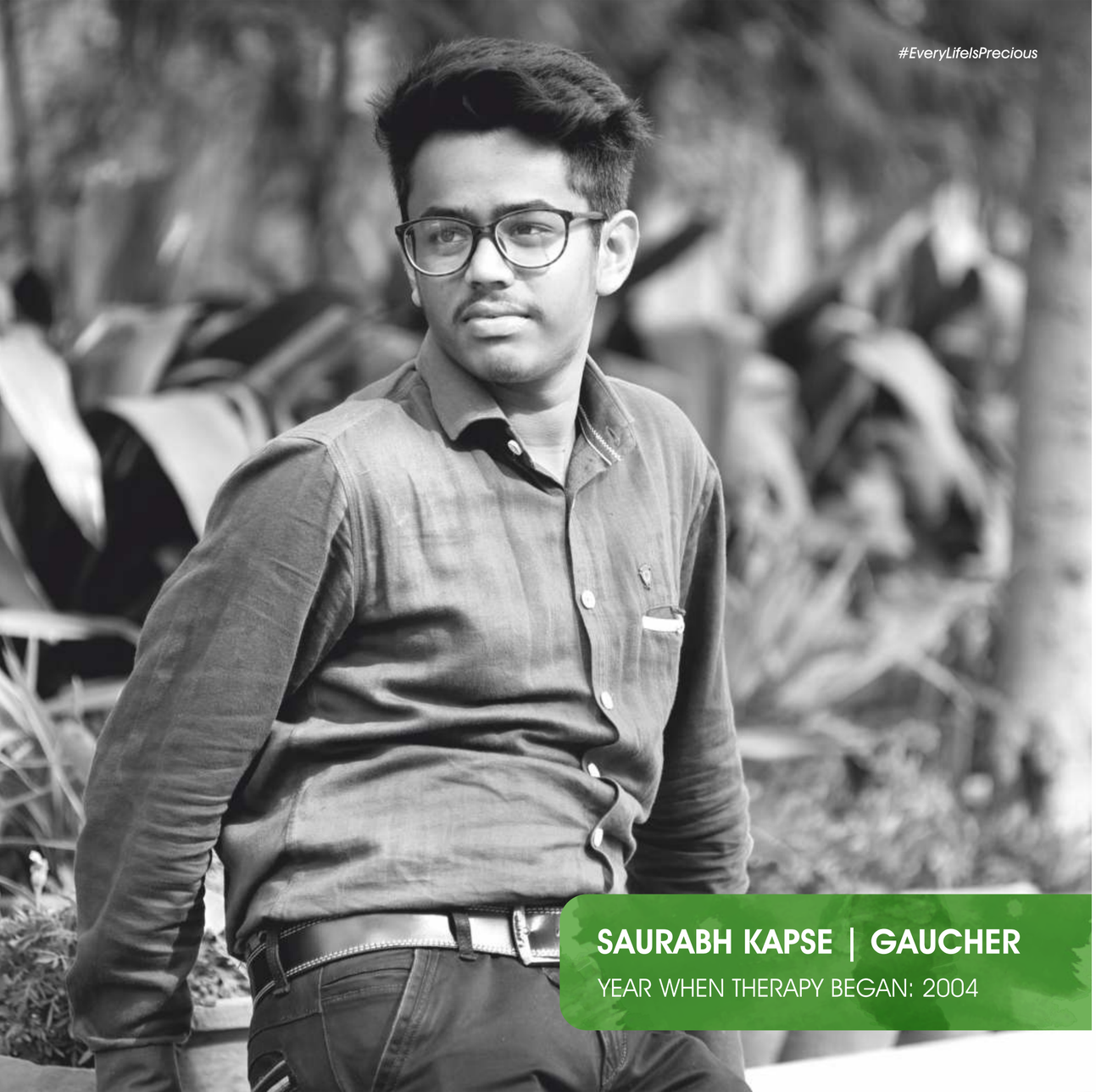
He is happily married and wishes to lead a fulfilling life with his wife and family. He also hopes to help other Gaucher patients through counseling, to ease their anxiety and pain.

*#EveryLifeIsPrecious*



**VILOL MALAVDE | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2000




**SAURABH KAPSE | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2004



# SAURABH KAPSE

Three-year-old Saurabh had an abnormally sized stomach due to liver and spleen enlargements. His blood tests showed low counts of hemoglobin and platelets. He frequently experienced nosebleeds and developed an aversion to food, due to indigestion.

His family doctor noticed soon that both his sister Aishwarya and Saurabh were showing similar symptoms. Due to poor bone health, their height wasn't increasing, either. He referred them to a pediatrician.

The pediatrician suspected his condition to be a rare genetic disease and referred both siblings to the Ganga Ram Hospital in Delhi. After thorough medical tests, they were diagnosed with Gaucher disease in the year 2000.

Since no treatment was available in India then, they underwent splenectomy and were on an antibiotic regime to avoid any susceptibility to infection. Finally, in 2004 they started infusion therapy. He has been on treatment, ever since.

Saurav recently graduated as a Mechanical Engineer. He loves the outdoors, plays cricket and football and works out at the gym to keep fit. He has won numerous awards as an athlete in college and is an avid science-fiction reader.

# AISHWARYA KAPSE

Aishwarya was all of one-year old when she started showing initial symptoms such as an enlarged stomach, weakness, loose motions, indigestion and low appetite for food.

Till 2000, her treating physicians were clueless about the reasons behind her worsening health condition. She was on symptomatic treatment based on recommendations of various specialists. She underwent splenectomy for interim relief, but it was all in vain. Her condition continued to worsen until she, along with her brother who showed similar symptoms, were referred to the Ganga Ram Hospital in Delhi. There, both siblings were diagnosed with Gaucher disease.

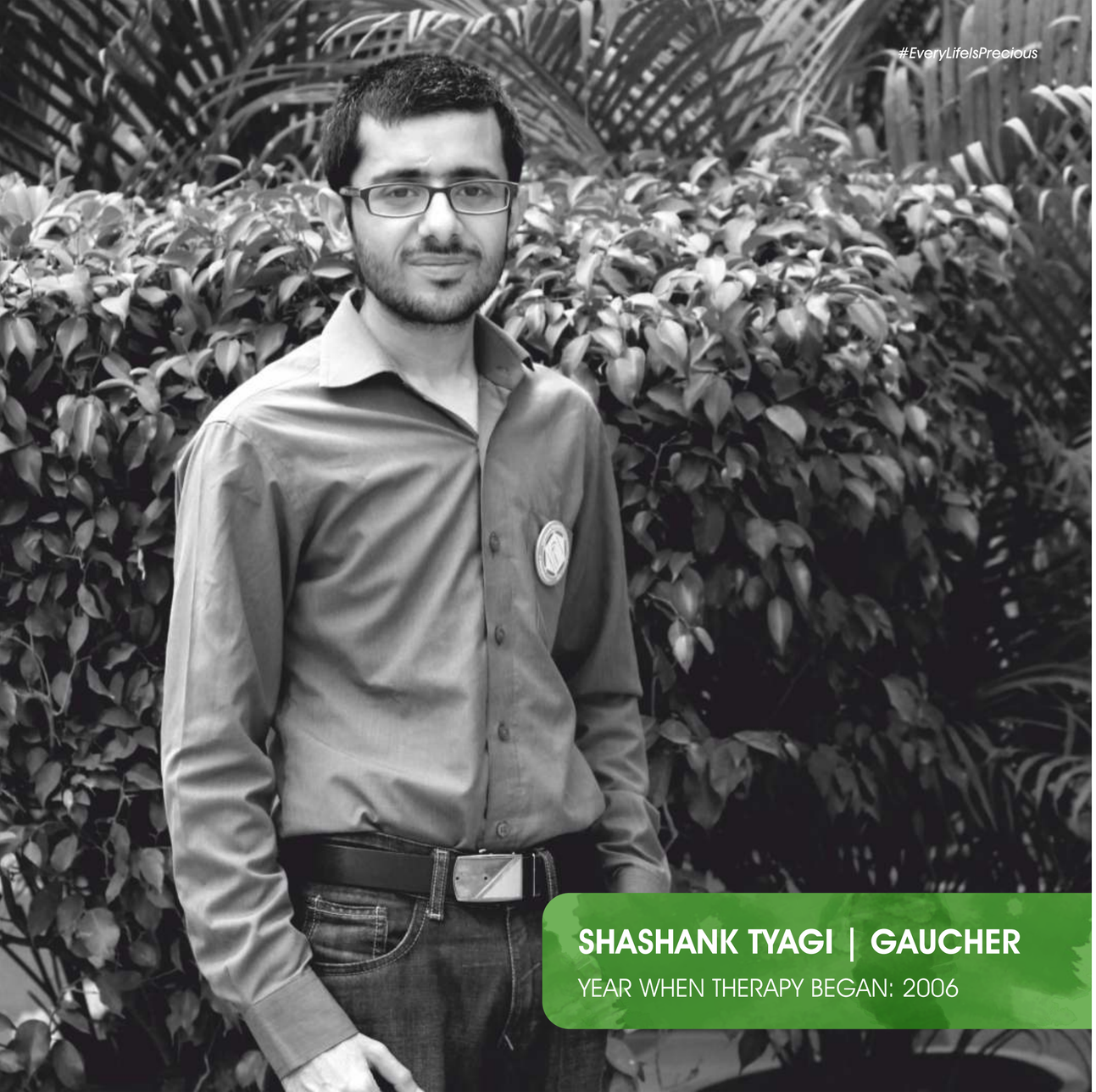
Aishwarya's treatment began in 2004, along with her brother Saurabh's. Looking back now, no one can tell she is the same child that suffered such a traumatic journey of misdiagnosis and delayed treatment.

Currently, an Electronics and Telecommunications Engineer, Aishwarya continues her search for work opportunities in the Information and Communications Technology space. She swims and cycles to stay fit, enjoys reading novels and continues to keep her passion for baking and cooking alive.

*#EveryLifeIsPrecious*

**AISHWARYA KAPSE | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2004




**SHASHANK TYAGI | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2006



# SHASHANK TYAGI

Shashank recalls his journey of a decade full of misdiagnosis through never-ending medical screenings and tests. It all started when he was four years old. He used to throw up whatever little he ate, could not digest any solid foods and was growing lower on energy and immunity with each passing day.

He was always tired, physically drained and on a constant liquid diet, due to which, his life was very inactive when compared to other young, bubbly, energetic children of his age. And that he wasn't growing tall as per his age, made him sadder.

Through these ten tumultuous years, his family and he went through immense emotional ordeal, fear and hopelessness. Shashank was even put on various alternative therapies, but nothing improved his condition. His health was worsening, and his liver and spleen were enlarging.

Finally, in 2004, he was diagnosed with Gaucher at AIIMS, Delhi. Two years later, he received his first infusion through CMC, Vellore. His body started responding to the therapy. He started eating better and his body started regaining the strength it needed. He could run, cycle, play football and go to school like all other children of his age.

There has been no looking back, ever since. Currently, Shashank lives a near-normal life and is now an MBA graduate.

He's a Pan India coordinator of the Lysosomal Storage Disorder Support Society and is raising awareness on rare diseases and also works as a Regional Manager - South Asia for the International Gaucher Alliance since 2018. Shashank aspires to work at a corporate firm soon and wants to live his dream of bungee jumping and paragliding.

# SHREYA SETHI

Shreya's birth was like a dream come true for her parents. The couple had a boy earlier, and with a baby girl, their family was 'complete'. But their happiness seemed very short-lived, when doctors reported major health concerns for Shreya.

Her spleen was enlarged and her hemoglobin kept dropping, persistently. She was admitted to the ICU for blood transfusion, twice. It was only when her blood samples were tested for rare genetic disorders, that the family found out she had Gaucher disease.

Her health kept deteriorating for a year after her birth, constantly indicating near-nil chances of survival. The family was heart-broken but determined. Various internet searches later, they approached Sanofi Specialty Care in 2011, requesting any possible assistance with their daughter's treatment.

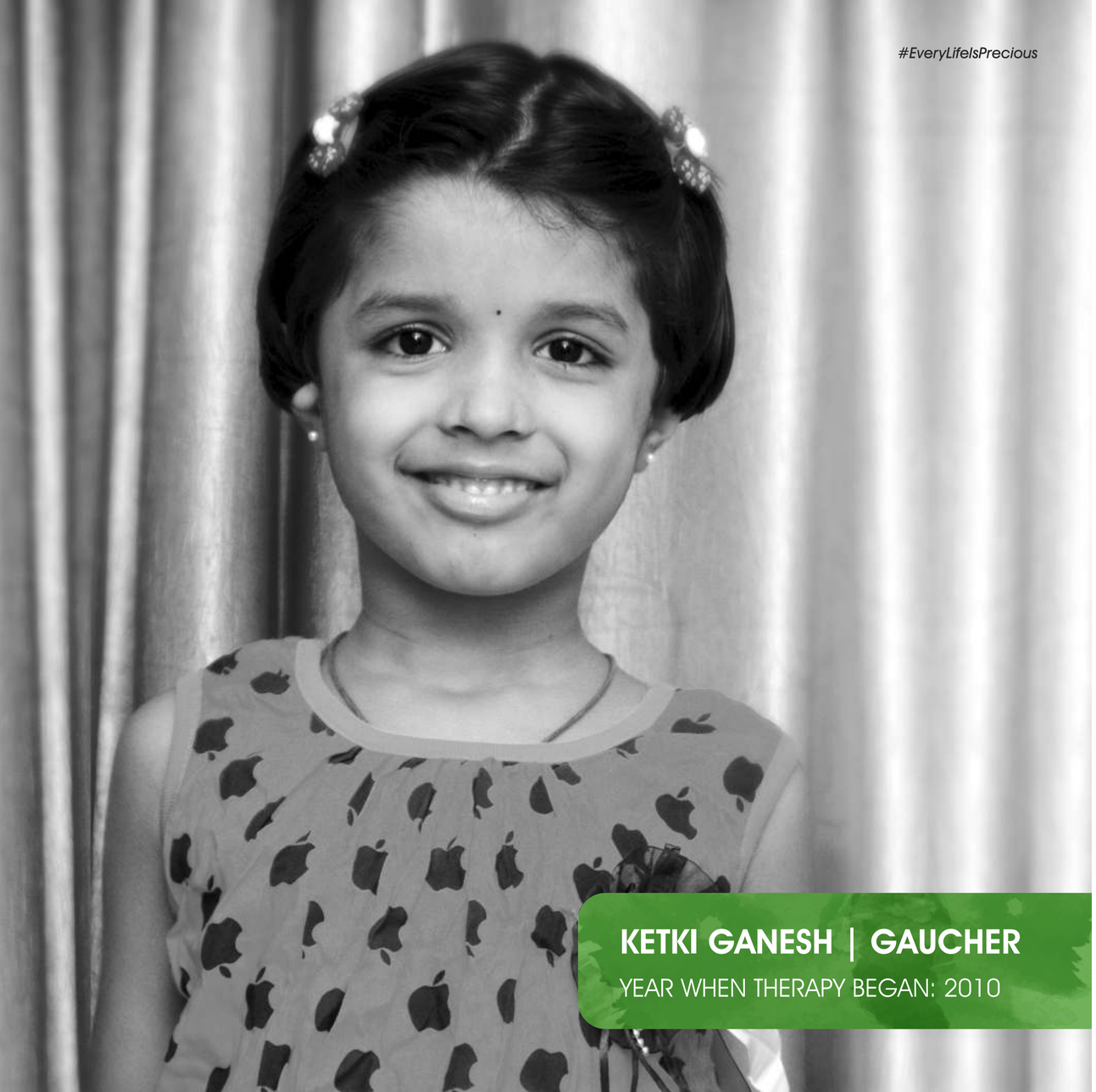
A response in the affirmative ensured their joy knew no bounds. As soon as Shreya started receiving therapy, her condition stabilized – her blood count improved, and her spleen and liver reduced to normal size within two months.

Now Shreya attends school regularly and is academically sound. She loves watching movies and spends her time cycling, creating craft and, at times, grooving to her favourite Bollywood music.

*#EveryLifeIsPrecious*

**SHREYA SETHI | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2012



**KETKI GANESH | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2010



# KETKI GANESH

When 2-years old, Ketki's family noticed that she had an abnormally hard and strangely large abdomen for an infant. The puzzled but suspicious family immediately visited a specialized hospital for children in Pune to get her diagnosed.

Fortunately, Ketki's pediatrician correctly diagnosed it as a form of rare disease. After her bone marrow and blood tests, it was confirmed that she was suffering from Gaucher. The classic symptoms of a large spleen and low hemoglobin count reaffirmed their diagnosis.

Her pediatrician immediately connected the family to Sanofi Specialty Care, seeking treatment and humanitarian assistance. Within six months of her diagnosis, Ketki received her first infusion.

Now in school, Ketki is a diligent student. Her brother and she are inseparable, always together, curating new child-like mischief. She aspires to be a teacher. Her parents commend her patience, bravery and smiling spirit all throughout her therapy infusions.

# GIRIJA KUMARI

Girija's parents lost their older son Ganesh to a condition they were unaware of. When Girija was born, she started displaying the same symptoms that Ganesh had too – an enlarged stomach and low hemoglobin.

Girija's brother passed away even before his condition was diagnosed, but her doctor and her parents were not going to allow history to repeat itself.

They ensured all necessary tests were undertaken in time and as expected, Girija too, was diagnosed with Gaucher within six months of birth – a disease her parents later realized was the cause of death for Ganesh, too.

In 2011, Girija's doctor facilitated the process for her first infusion within six months of her diagnosis.

Girija attends school regularly now. She is an avid watcher of dance shows on TV and loves making new friends. The little girl is a bundle of joy who aspires to become a doctor someday and heal the world.


**GIRIJA KUMARI | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2011

**DEEPESH KUMAR | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2015

# OUR GAUCHER FIGHTERS NOT GOING DOWN WITHOUT A FIGHT

At the tender age of 12, when most teenagers are out and about, making new friends, cultivating a hobby, exploring relationships and savoring the joys of finishing childhood, Deepesh started falling progressively ill. What began as a mild fever, soon turned chronic.

Over time, his condition worsened. Various doctor visits and multiple medical tests later, he was diagnosed with a potentially debilitating condition – Gaucher.

Luckily, Deepesh and his family did not suffer too long. In 2015, his treatment began soon after a year of diagnosis. He feels fortunate that he did not experience the suffering of either spleen enlargement, or other physical deformities, thanks to the timely availability and accessibility of treatment.

Today, Deepesh leads an almost-normal life. He enjoys regular schooling and is extremely passionate about dance.

# OUR GAUCHER FIGHTERS NOT GOING DOWN WITHOUT A FIGHT

It was strange and perplexing for seventeen-year-old Sangeetha and her parents to notice that she was finding it difficult to gulp down food. Never had she faced a problem like this. It took the family a few more complications to realize that there was something more serious than what met the eye.

Sangeetha's spleen started swelling up and as a result, her stomach began protruding. In addition, she had loose motions, headaches and low hemoglobin levels. It took two years of several consultations and a series of medical tests, for doctors to diagnose her with Gaucher disease; this was in 2016. The ordeal didn't end there for Sangeetha & her family. They struggled for two more years to get treatment, even post diagnosis.

Finally, her treatment began in 2018. With regular infusions, her health started to gradually stabilize and even improve.

While due to her failing health Sangeetha had to discontinue her studies from the first semester of B.Sc, she now hopes to pick it back up and complete her degree.

**SANGEETHA M. | GAUCHER**

YEAR WHEN THERAPY BEGAN: 2018

PATIENTS ARE PEOPLE, FIRST.

WE ALL LAUGH AND CRY.

WE HOPE AND DREAM.

WE HAVE FAMILY AND FRIENDS WHO LOVE US.

WE WANT TO LEAD NORMAL LIVES.

THIS IS WHY WE DO WHAT WE DO.

sanofi

**BRINGING HOPE | TRANSFORMING LIVES**

*#EveryLifeIsPrecious*

**Sanofi Healthcare India Pvt. Ltd.**

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