



Volume 2
Issue : 2 (May - Aug 2025)

WE CONNECT

The Official Publication of WPF

Theme : Me Day

You cannot pour from an empty cup

Taking care of yourself is a form of strength!



F. L. Y

First Love Yourself

From Decoding DNA to Decoding Life: Life Lessons from a Senior Medical Geneticist



Dr. Shubha Phadke

M D [Pediatrics], DM [Medical Genetics]

Formerly Professor & Head, Department of Medical Genetics Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India

Current affiliation: Consultant, Jupiter Hospital, Baner, Pune

Her extensive research and clinical work focus on genetic disorders, counseling, and promoting genetic literacy in India.

- shubharaophadke@gmail.com | Location : Lucknow

I superannuated recently and wanted to address my past students who had gathered to relive & cherish memories of the student times and wish me for the future.

All of them have contributed so much to my growth and my department, the first department of medical genetics in India, that the words to thank them are not enough. Similarly, no words to thank the almighty for the wonderful life journey I got to live!

At that juncture, I wished to share my life experiences and what I learned through them. I am sharing the same string of thoughts in this article. I would not like to use the word 'advice', though age has probably given me the right to advise (which no one likes).

The first thought that came to my mind is the struggle of living multiple lives; life of a woman (a mother, a wife and more) and a doctor with a full time job and a lot of responsibilities.

This makes all women's lives different from those of men. There are two issues as a working woman; the first being issues related to sexual discrimination or harassment and the other is the constant conflict between home and family vs work.

Luckily, I did not have any experiences of suffering at workplace due to being woman but got glimpses of such issues while working as a chairman of the internal complaint committee of the institute.

Sometimes, not the boss, but subordinate male staff cause problems by showing resistance to a female boss. In both the situations, being tough and taking support of authorities is needed; especially in case of any harassment at work; make noise, register complaint.

The second struggle is balancing work and family by prioritising appropriately as the situation demands. This means sometimes gap in the career while children are young or pregnancy is difficult. Who can be a better mother than a pediatrician! Believe me, one- or two-years gaps once or twice in the career did me no harm.

I tell the resident doctors that after children grow up, there are decades for the career. There are no definite right ways of balancing family and work and one has to make their own decisions based on situation. The basis of decision should be responsibility to the family.

This is not a burden but the power of being woman. I still have not understood what gender equality really means but to me, it is the right to decide for yourself though sometimes the priority is not yourself. Secondly, being woman means strength to trust yourself.

Some more lessons from my experiences, I am sharing below. I knew these words of wisdom theoretically but they were not the reasons for my decisions. But as I look back they fit into my decisions and here, I endorse them.

I landed in medical genetics 35 years ago by chance. At that time the speciality was non-existent and future was fogged. But my decision to chose genetics to pediatrics did not need logical thinking.

Message is, listen to your heart!

The struggle of establishing a new speciality, a complex of more than 6000 genetic disorders from head to toe, diagnostic challenges, lack of support for laboratory diagnostics, learning genetic counseling, prenatal diagnosis was a taxing journey. Now after three and half decades while looking back I can laugh at failures and frustrations.

Message is finding strength in the failures. Challenges and setbacks are hidden opportunities & steps for upward growth.

A yearly short term introductory course in Medical Genetics & Genetic Counseling for clinicians which we started in the year 2000 was also a great challenge. There were hardly any takers. But the planning of the program, passionate teaching and commitment to provide an exciting learning experience by way of innovative interactive methods which will enthuse clinicians for this novel specialty made it a popular course and it could help medical genetics reach all corners of the country through the emissaries who attended the 21 courses held till date. The students in the department became teachers. They were taught to teach. Later they became excellent teachers and esteemed faculty for the courses held over years.

Similarly, my entry into the fetal medicine was by compulsion as the most important help needed for the families with genetic disorders, especially neural tube defects, thalassemia and Duchenne muscular dystrophy, was prevention by prenatal diagnosis. During 1990s, the community of fetal medicine specialists was non-existent. It was a matter of necessity to learn non-invasive and invasive prenatal diagnosis on my own. But in the long run, evaluation of a fetus for birth defects by a medical geneticist with pediatric background turned out to be a boon as I was evaluating children with malformations and was empowered with the knowledge of syndromes. It was a blessing in disguise for me as well as I liked the language of shapes and sizes and deciphering two dimensional images to three dimensional shapes in mind.

Message is to try to grow laterally, beyond your domain. The time needs more interdisciplinary activities.

An opportunity to be in a genetics lab in the University of Washington opened my eyes to how the labs in developed countries work. The system, meticulousness and high-end research were somethings which I was not exposed to, in India.

I listened to Prof Francis Collins, the key figure of Human Genome Project, explaining how he coined the word, 'Positional cloning' for directly identifying a causative gene for a disease and replaced the term, 'Reverse genetics'. Such opportunities to pursue science across the oceans fed the travel bug inside me.

Message is seeing the world to gain knowledge & wisdom. Short term fellowships and observer-ships make a world of difference for a prepared mind. It helps personal growth and the growth of the department back in India.

Three decades ago, the gap in what we were reading in journals and what we could do was enormous. But perseverance could take me from karyotype to cytogenetic microarray and exome sequencing. While working through these frustrating initial years, I continued to work in laboratory (a difficult task in the beginning) in addition to clinical responsibilities.

Message is, work till you the work is done, you succeed! Things work! After getting degrees, every-day is an examination! Learn to nag yourself.

The persistence and patience were also very essential to get the things done by navigating the complex maze of government functioning. Of course, the advantage of working in a government institute was ability to do what is right and what you like without the money matters. The job became a hobby!

During the journey, the most beautiful thing happened was getting intelligent, unusually bright, students who wanted to do different and joined me on the unchartered path. These young differently wired students cum friends got connected to my heart and the department. Teaching and learning are bi-directional. You learn more if you want to be a good teacher.

The message is if you are focussed and passionate about your work, like minded persons will join your uphill walk. Also, one has to keep learning and respecting the teacher in the student, especially postgraduate students. Good students are up-to-date, teach you and keep you on toes!

I had my plan of having a small clinic for children but, destiny made me professor of medical genetics. Hard work and determination were the keys to open the doors. I did what I thought was good, right and necessary without realising that those were high aims for the department.

So, the message is, NOT failure but low aim is crime! If you don't go ahead, the world goes ahead of you. "Effort is the greatest strength, there is nothing greater than effort"

"उत्साहो बलवानार्य नास्त्युत्साहात् परं बलम्" Lucky once in a while.... Don't wait for luck ... Work hard! In addition to hard work and perseverance, patience, positive thinking, planning and proactive actions help for success in the committed goals.

At the beginning of my career, the main objective was to establish prenatal diagnosis for beta thalassemia, and eradicate beta thalassemia by population-based screening as was done in the Mediterranean countries.

Over years, the outcomes of thalassemia major have improved so much; thanks to the development of oral iron chelating agents, safe hematopoietic stem cell transplantation and government funding for treatment that one wonders if population-based prevention of thalassemia is necessary or not.

Message is that the change needed in our perspective, guidelines, policies as situation changes.

As genetics evolved, the genomic testing has made it possible to screen everyone for thousands of disorders. This has presented many dilemmas and ethical questions to the laypersons who are lured with the possibility of screening the baby in the womb for all disorders without understanding limitations of such tests. On one hand the genomic medicine has opened up many diagnostic and therapeutic options but it also is posing many ethics dilemmas.

The message is that as the leaders of society, educated, knowledgeable doctors, medical geneticists we need to channelize the society in right direction, guide them to approach the fetal diagnosis with ethical angles.

Patients contribute a lot to our learning. They teach us medicine, so document each rare case or medical experience. Learn to ask why, if you are not reaching the diagnosis or something is different or cannot be explained.

My obsession with reporting every novel case or findings, lead to more than 300 publications in my career. Ten of these are new syndromes and in seven out of the ten, the causative genes could be identified with next generation sequencing; in some cases a decade after reporting the new syndrome.

Message is that we learn from the patient. Listening patiently may give you an important diagnostic clue. Research and patient care are interwound, each enhancing the other. One should be perturbed if diagnosis is not reached. Unbiased approach towards patient diagnosis, application of latest knowledge and appropriate interpretation of results of investigations are basic requisites for a logical clinician.

Patients and their families teach us about life as well. Hundreds of thalassemia and hemophilia patients' strengths to fight with the disease, endurance, grit, resilience and success in their lives makes us humble.

Families with children with disabilities, childhood deaths teach us coping strategies. We celebrated Down syndrome day, hemophilia day, thalassemia day and rare disease day every year.

The happiness and inner peace brought by their dances, hugs and smiles on parents faces cannot be described. The experience is an eye opener for new students in the department.

Message is to give eyes, ears and heart to the patients sitting in front of you. Work with patient support groups. Advocacy brings change in the society and policies. We can give and get strength from them.

Genetic counseling is an integral part of the management of genetic disorders. Talking to the patients about scientific aspect of the disease, genetic aspects and implications for the family, available treatment, reproductive options improves communication, command over language and ability to simplify the information.

Message is that do not consider talking to the patient as the easiest part, easier than diagnosis or a surgery. These are pillars of rapport with the patient on which the bridges of communication are built. Words can be healers; words can have adverse effects, hurt and block the communication.

Communication skills have to be improved by listening and practice. Talk to the patient in the language which he or she understands. Do not mix English to the local language. Experiences of genetic counseling help to improve communication with friends and family as well.

Physical exercises and games were my weakest points. Somewhere around forty, I started exercises, swimming, jogging and kept on improving.

Of course still I can not play any game, but can compete with my self to improve stamina and endurance.

Message is that it is never late to start physical exercises. What life journey is, nothing but to challenge oneself and stretch the limits!

I still want the window seat in the flight to look at the clouds, rivers, dams, mountains, railway tracks, if lucky, the Lotus temple from the sky.

Message is to keep the child in you alive! Also, it helps to remind oneself how miniscule dot one is on the earth!

Look at bright and beautiful. Teaching job gave me an opportunity to be with young students. They kept me updated about science and fashion. I could get to peep in their minds, to know how the young generation thinks.

I loved genetics, lived genetics, breathed genetics – will continue to do so! But I love to paint sarees, draw a rangoli, design mementos for conferences, cook for family, friends and students. Remember, unlike research manuscript, a failed recipe does not come back from the reviewer.

Message is, love your profession but don't forget your hobbies. Hobbies are stress relievers. Use your talent to keep your mind fresh and enhance the profession! Make your students and colleagues comfortable and friendly.

If we stood still in time & we would not have aged but then we would have stuck still at karyotype and would not have reached genome sequencing!

Time changes, accept the change.

**Look for Art in the Science
Don't forget the Science of Medicine
Care for Human in the Patient
& Never Ignore Your Conscience**