THE SUNDAY READ

Pune-based Nalini Nivangune (seated, right) and her family say that genetic profiling has helped them to prepare for any eventuality

Tracing their fear

Mirror meets families who have been part of genetic tests for cancer to find stories of hope, anxiety and stigma

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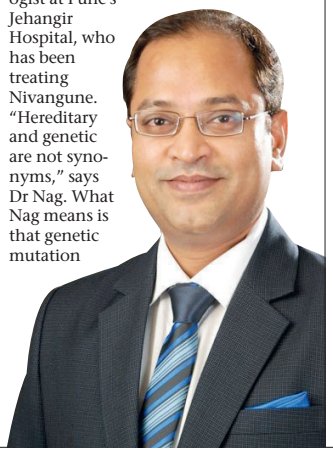
Two years ago, Pune-based Nalini Nivangune, who is one of six sisters, was diagnosed with breast cancer. In 2002, the 62-year-old's younger sister, Alka Pasalkar, succumbed to the disease at 45.

"My mother and father lived up to 80 and were able-bodied until the end," says Nivangune, "This was the first time someone in the family got cancer, and none of us knew anything about the disease then." Two months after Nivangune's diagnosis, her younger sister Savita Sharma, 56, complained of severe stomach ache. One doctor dismissed it as fear psychosis as a result of her sister having been recently diagnosed with cancer. But an alert Sharma went to another physician who advised a sonography and scan that revealed that she too had cancer, of the ovary. The female members of the family are a classic case of the Hereditary Breast and Ovarian Cancer (HBOC) genetic condition.

"Tests should only be conducted in families that have a history of a particular disease. At the same time, emotional counselling is required to ensure that patients are able to cope with stark diagnoses
–Dr Shishir Shetty

Nivangune's family, including her sisters, son, nephews, nieces and eight-year-old granddaughter, have taken the Germ Line Cancer Test, usually performed on a blood sample, to determine which of them were carriers of the mutated gene. Only two of her siblings have tested negative for carrying the mutated BRCA1 gene that causes cancer. Her son Nandan, 38, and sister Asha Kadu, 50, have both tested positive. This puts Nandan at the risk of male breast and prostate cancer and Kadu at the risk of breast and ovarian cancer, but the family that has lived with the disease says that there is no more fear, just a desperate need for more information. "No one knows how cancer can be cured unfortunately," says Nandan, who works in an electronic leasing and financing company in Pune.

But there is an important distinction to be made when it comes to cancer, says Dr Shona Nag, oncologist at Pune's Jehangir Hospital, who has been treating Nivangune. "Hereditary and genetic are not synonyms," says Dr Nag. What Nag means is that genetic mutation need not necessarily be hereditary. "Only 5-10 per cent of cancers are hereditary. Hereditary means when it is passed on from one generation to the next and at least one of the offspring is carrying that trait and is passing it on to the next generation."



Sharma says that the bond between her family members has grown stronger ever since they were diagnosed. "Can you tell we have cancer?" she smiles. Her thinning scalp, a result of chemotherapy, is indeed the only giveaway. "If you have family support, nothing can be daunting." Her 29-year-old son, an IT professional, does not want to disclose the results of the test, or even his name for fear of being stigmatised at his workplace. His sister, who is based in Aurangabad, has tested positive. Kadu, who runs a provision store, also lives in fear of customers boycotting her store because cancer runs in the family. The gene has become an identity marker for the family. "There has been so much stigma that those who have been close to us have distanced themselves," says Nandan, "Some of them act as if cancer is contagious." His cousin, who wishes to remain anonymous, adds, "I don't want my actions to be judged in the office because someone in my family has cancer."

Nag says that the tests can help guide further treatments. "Preventive surgeries for breast or ovarian cancer

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can provide 95 per cent protection," she adds. Treatment, however, can be expensive. Sharma has spent close to eight to 10 lakh on treatment and her older sister, Nivangune, who has undergone 18 rounds of chemotherapy besides mastectomy, has spent approximately 25 lakh, and is still being treated for cancer. Nandan says that the disease has made them put away money in a more systematic manner. "My wife is a nurse so she understands the situation. We are saving so that our two-year-old daughter's future is secured," he says, "It has also made us realise how important medical insurance is. It is why we could afford all of these expenses."

"In a village called Gudran, some 100km from Bhavnagar in Gujarat, Anvarali Manjayani is awaiting the results of a test that, he says, is extremely vital for the future of his children

Genetic testing, or an awareness of the insights that genetic material (chromosomes) can provide into our biological destinies, so to speak, has been around for longer than you think. In fact, inherited diseases were linked to chromosomes as far back as the early 1900s, and by the 1950s scientists had developed tests for Down syndrome, among others. Genetic testing is now available for more than 2,000 conditions, and the tests available include, among others, diagnostic testing, pre-natal testing and even pharmacogenetic testing, which focus on matching medicines with a person's genetic make-up. Hollywood actor Angelina Jolie's pre-emptive double mastectomy in 2013 acted as a catalyst, especially among women across the world, in furthering genetic diagnoses, and today, awareness about it has also percolated to smaller towns in the country.

Earlier this month, again in Pune, Ashok Gholap, 66, invited his extended family over to his home. His relatives came from across Maharashtra. Gholap told a newspaper that he wanted his relatives to undergo a Germ Line Cancer test that would help flag vulnerabilities. And, in a village called Gudran, some 100 km from Bhavnagar, Anvarali Manjayani is awaiting the results of a test that, he says, is extremely vital for the future of his children. Two months ago, the cloth trader's wife, Yasmin, succumbed to breast cancer. Yasmin's sister, too, was afflicted with breast cancer – as well as ovarian cancer – but she survived, says Manjayani. He is now waiting for the results of the genetic tests conducted on his wife when she was alive. These, he says, will determine whether he needs to get his three daughters tested.

"We got to know about the tests only after my wife fell ill," says Manjayani. "We were advised that on the basis of Yasminben's test reports, doctors will suggest whether I should get my daughters tested or not.

Gudran-based cloth trader Anvarali Manjayani is getting his daughters tested after his wife Yasmin succumbed to cancer two months ago

Bengaluru-based MedGenome is a market leader in genomic diagnostics

About a week ago, Dr Shishir Shetty, director of oncology at Apollo Hospitals, performed an operation on a senior citizen, a mother of two sons. She was diagnosed with colon cancer about two years ago, and a gene test confirmed that she was suffering from Familial adenomatous polyposis (FAP), an inherited disorder characterised by cancer of the large intestine (colon) and rectum. In March 2015, the lady underwent large intestine removal surgery, and the operation performed last week by Shetty was involved tackling a cancer relapse she suffered earlier this year. The FAP had also affected her liver. Dr Shetty's patient is convalescing at the moment, but she is also worried about her sons. "She has already lost her two elder sisters and a younger brother to colon cancer, and the chances of other younger members in the family too being afflicted with it is very high. So, the lady's extended family is now undergoing tests to determine whether they have the genetic condition," says Shetty. The woman's younger son has been diagnosed with the same gene, and has been advised by Shetty to get his large intestine removed. The sons, who are 32 and 30, are wary of what the future holds for them. "I'm not sure if I want to go in for the surgery, but I'm periodically undergoing tests to rule out colon cancer," says the younger son. His elder brother would rather focus on his career at the moment. "I'm a bit unsettled by my brother's diagnosis, but at this moment, I have a career to think of," he told Mirror.

Over the years several private laboratories offering personalised individual DNA readings and genetic tests have come up in India. ACTREC, the R&D arm of the Tata Memorial Hospital, has been conducting genetic analysis of hereditary cancer since 2007, a project funded by the Indian Council of Medical Research. Yet others include Chennai-based XCODE, which offers predictive genetic tests for obesity, stroke, diabetes and cardiovascular diseases; Goregaon-based Super Religare Laboratories and Hyderabad-based Map My Genome. But Dr Shetty says that while genetic testing is now more accessible, there are still concerns that need to be addressed. "First of all, tests should only be conducted in families that have a history of a particular disease, and more often than not, it is cancer. At the same time, emotional counselling is required to ensure that patients are able to cope with stark diagnoses. " Other experts echo his concerns.

"We have discussed genetic testing with many families, but these pertain to only hereditary cancers, to test whether the family has a high risk or not. These cancers mostly include breast, ovarian, and gastro-intestinal," says Dr Prasad Narayanan, chief medical oncologist at Bengaluru-based Cytecare hospitals. But one needs specific indications as to why one want to get tested, he clarifies. "When you have a first degree relative who has either one of these cancers, or if there is a male patient who has breast cancer, it makes sense to get the test done to understand whether you have a high risk of passing on the disease genetically," he adds.

"When it comes to Germline Genetic Testing, it is necessary only if someone within the family – a blood relation has already been diagnosed with any of the cancers. However, what is most important is testing and analysing the patient's blood first. If it's a somatic or acquired disease, then there's almost no chance that the person will pass on the cancer to the family. However, if the germ line test reveals that the person's cancer was inherited (for example BRCA gene mutation for breast cancer), it is important for the family to get tested to see if they are carriers as well, or not," says Sam Santhosh, founder and chairman of MedGenome, a market leader for genomic diagnostics in South Asia and a leading provider of genomics research services globally. Santosh does not recommend recreational genetic testing. "Recreational genetic testing – called so because it's not necessarily a test prescribed by a physician - is where a family goes in for an ad hoc genetic screening for diseases, and it is unlikely to be fruitful because we don't possess enough technology or data as yet to come to conclusions whether a person is genetically pre-disposed to complex diseases such as cancer."

— With inputs from Lata Mishra, Alok Brahmbhatt and Priyadarshini Nandy