

City checks its genes for healthier babies

Neha Bhayana

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MUMBAI: Like most two-year-olds, Shriya (name changed) is a little ball of energy, zipping around her house, chattering endlessly and keeping her parents on their toes. It's difficult to imagine that this energetic toddler narrowly escaped being permanently disabled — by a genetic disorder.

When Shriya was nine months old, her mother Preeti Acharya (32) noticed she would not “blabber, toss around or be inquisitive about toys”. When medical tests failed to find the cause, the family paediatrician recommended a genetic test.

Shriya was diagnosed with Methylmalonic Aciduria, a disorder caused by a defective gene, treatable by vitamin B12 therapy. After a course of 18 injections, Shriya got better. “Her development is delayed by three to four months and she has to take a pill daily but she is fine now,” said Acharya.

Many children like Shriya are benefiting from genetic testing as more and more Mumbaiites opt for them. While some test newborns, others undergo tests to determine if their to-be-born babies are healthy. And many take tests to ascertain their own susceptibility to diseases like cancer and diabetes.

Genetic testing can't prevent

TEST GUIDE



WHAT IS A GENETIC TEST?

A study of genes to assess one's vulnerabilities to hereditary diseases.

WHO SHOULD GO FOR IT?

Couples planning a baby if there's a family history of specific disorders or if they are related to each other.
Newborns, if there's a family history of disorders.

diseases, but it can improve the quality of life with treatment.

Malad-based PreventiNe Life Care has seen a big rise in the demand for tests. “We conducted 20 to 50 tests a month for newborns when we started 2.5 years ago, now it's 600 tests,” said CEO Abhimanyu Kumar.

Reliance Life Sciences, which introduced tests for predictive diagnosis for ovarian, breast and cervical cancer in 2004-05, has seen a “500 per cent increase” in testing between 2005 and 2009, said CEO K.V. Subramaniam.

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Genetic testing can save your newborn

GET YOUR baby's genes tested within a week after birth for effective treatment of disorders

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MUMBAI: A newborn in the United States does not leave the hospital till his/her heel is pricked for a few drops of blood required for a genetic test. The same happens in 52 countries across the world.

In January 2007, the Chandigarh government started funding mass genetic screening of newborns. A year later, Goa became the first state to make it mandatory. But the concept is yet to take off in the rest of India.

An estimated 6.2 lakh of the 2.6 crore babies that are born in India every year suffer from genetic disorders.

Experts feel that it is time we put a mandatory newborn genetic screening programme in place. "If the conditions are detected at birth, one can start treatment and help the child lead a normal life," said obstetrician Dr Saurabh Dani, who is attached with Bangalore-based Neo Gen Labs.

"So if a baby is diagnosed with Glucose-6-phosphate dehydrogenase deficiency, which is fairly common among Indian children, it can be managed easily if the child avoids eating kidney beans and taking sulphur-based drugs throughout his life," he said.

Dr Murlidhar Mahajan, a Kandivli-based paediatrician, said that genetic testing is even more important in India as many communities including Parsis, Muslims and South Indians have consanguineous marriages (between relatives).

Bhopal-based Manjeet Singh's niece, who lost her baby to a genetic disorder, agreed. "My niece is married to her cousin. Her baby looked perfectly healthy on delivery but refused to accept feeds and went into coma and died within a fortnight," Singh said.

Experts also pointed out that it is best to do the genetic test

within a week after birth.

"If the test is conducted when the child is a few months old and has already developed symptoms, then the damage is already done as over 90 per cent of disorders are present at birth and manifest at some point or the other in life," said Vrushali Joshi, senior research associate at Preventine Life Care.

Jaslok Hospital's neo-natologist Dr Meena Malkani agreed. "We conduct ultrasound and other tests to ensure the baby's development is normal. Genetic screening is another way of doing that," she said.

Jaslok Hospital has tied up with Metropolis laboratories to offer a test for seven common metabolic errors to all new parents.

Over 50 families have gone for the test in the last six months. A section of doctors, however, feel that making genetic testing mandatory is not feasible in a developing country.

"Genetic tests are too expensive to be effective as a mass screening tool. We need to ensure that every child gets immunized and has access to clean drinking water first," said Dr Soonu Udani, consultant paediatrician with PD Hinduja Hospital.

If the conditions are detected at birth, one can start treatment and help the child lead a normal life.

DR SAURABH DANI,
Obstetrician attached with Bangalore-based Neo Gen Labs



IMAGING: SURAJIT

CITY CHECKS ITS GENES...

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Subramaniam added that over 700 women, aged 25 to 50 years, have undergone tests at Reliance since 2005 to know of their probability of developing breast cancer.

Most of them had a family history of the disease.

The Metropolis' laboratory in Worli, which started offering genetic tests for pregnant women, newborns and infertile couples in December 2006, has also registered a rise in demand. While most tests are done on doctors' referrals, there is a perceptible growth in the number of people voluntarily opting for them.

Take the case of Chembur resident Nima Harris. The 29-year-old architect has no family history of any disease but she has registered her soon-to-be-born baby for a genetic test.

"I read about genetic tests in international pregnancy books and my cousins in the US have done it so I decided to get it done as a precaution," she said.

The laboratories say the tests are 98 percent accurate.

ON THE HEALTH FRONT

26 MILLION children born in India every year

6.2 LAKH have genetic disorders

1.6 MILLION born birth defects

1 IN 540

A 2004 study of hospitals in Hyderabad showed that one in every 540 children born has an Inborn Error of Metabolism.

IN US

1 IN 1,350

children has an Inborn Error of Metabolism.

Genetic testing guide

What are Inborn Errors of Metabolism?
These are disorders caused by the accumulation of chemicals produced naturally in the body to abnormal levels. They can cause slow physical development, mental retardation or even death.

When should genetic testing be done?
Genetic screening can be done during the first trimester of pregnancy, but this can only diagnose Down's syndrome. Doctors recommend genetic tests for a newborn, ideally when he/she is 24 to 72-hours-old.

How much does it cost?
A single blood/urine sample can be tested for up to 100 disorders. Most laboratories charge between Rs 4000-5000 for a package of 50 disorders.

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