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HT MIND&BODY — GLYCOGEN DISORDER

Afflicted by genetic disorder, Afghan family finds hope in India

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NEW DELHI: Mosam Khan had lost two of his children to glycogen storage disease (GSD). He has three more children affected by the same disease. Determined to not let the rest suffer the same fate, the Afghani family arrived at Delhi from Jalalabad hoping to find a cure.

GSD is a genetic disorder due to which there is a defect in the enzyme that affects glycogen breakdown to glucose, forcing it to accumulate in the liver.

Children with GSD are usually short, have round faces, develop progressive severe liver enlargement that causes abdominal swelling and have fainting spells due to a sudden drop in blood sugar.

A few may also have weakened muscles but what eventually proves fatal is organ failure. The only definitive cure is liver transplant.

There is a 25% chance of every child born to couples having the carrier gene being affected.

Sana, 7, had multiple episodes of early-morning fits during the first 2 years of her life. She had developed poor night vision due to severe Vitamin A deficiency, threatening her with the risk of permanent blindness. She was also short and underweight due to growth failure.

As her liver started failing, and doctors back home were not able to treat the condition, Mosam brought her and her two younger siblings to Delhi in June and rented a room in south Delhi's Bhogal area.

Mosam donated a part of his liver to her two months ago.

Sana's elder sibling, 16-year-old Subhanallah, who himself is awaiting a donor match, became her caregiver as their father recuperated.

"My wife couldn't come because she had to look after our two other children, who are healthy," says Mosam.

Ahmed, 5, was the next from the family to receive a transplant a month and a half ago. Their uncle donated his liver, who had a smooth recovery post surgery.

"This is the first time we have done multiple transplants within a family as it can get really taxing for a family to take care of more than one person having undergone a transplant. One has to be extra careful in terms of hygiene, medicine compliance and follow ups, but the parents were really committed and requested us to go ahead with the surgeries," said Dr Anupam Sibal, senior consultant paediatrics hepatology, Apollo Hospital.

Talking about the challenges during surgery, Dr Subhash Gupta, chief transplant surgeon, says, "In metabolic diseases, especially in children, the vessels are extremely tiny, stiff and not dilated, so joining them is difficult. We had to be very cautious."

The family has flown home, and is now waiting for a matching donor for their eldest son. His mother wanted to donate a part of her liver but as she had already undergone an abdominal surgery a few years ago she was not found fit to be a donor.

"The risk would be too high in her case as there is already a lot of scarring and lesions due to the previous surgery," said Dr Gupta.

The family, however, is hopeful that they will be back soon for the surgery.

"As he is the eldest of the siblings, he understands things better. He keeps asking when he is going to get a match. We tell him it is just a matter of time," says Mosam.

THERE IS A 25% CHANCE OF EVERY CHILD BORN TO COUPLES HAVING THE CARRIER GENE BEING AFFECTED WITH GSD

IT'S ALL IN THE GENES

An abnormality in the genome can have serious repercussions and treatment is difficult.

FAQS

What is Glycogen?
Glucose is a significant source of energy for the body. It is stored by the body in the form of glycogen and later released into the body with the help of special proteins called enzymes.

What is Glycogen Storage Disease (GSD)?
People are born with the disease. In GSD, an abnormal amount of glycogen is stored in the liver, and the liver cannot control the use of glycogen and glucose. Certain enzymes are missing that control the change of sugar (glucose) into its storage form (glycogen) or release of glucose from glycogen.

How many types of GSDs are known?
There are at least 10 different types of GSDs. The types are put into groups based on the enzyme that is missing. The most common forms of GSD are types I (one), III (three) and IV (four).

How common is the disease?
About one in 20,000 people can have a type of GSD.

Is there a cure for the disease?
There is no definitive cure as such but children are treated symptomatically.

■ Ahmed with his doctor, Dr Anupam Sibal (L) and older brother, Subhanallah. The siblings are being treated for GSD. SUSHIL KUMAR / HT PHOTO

GSD: SIGNS AND SYMPTOMS

- Poor growth
- Muscle cramps
- Low blood sugar
- Significantly enlarged liver and in some cases, heart and kidney
- Swollen belly
- Nausea
- Fits
- Dizziness
- Abnormal haemoglobin and iron levels
- Anaemia
- Osteoporosis