Sharmila Nikapota must bandage her daughter every day. Sohana, 9, has the genetic disease RDEB, causing irreversible damage to her skin

THE POSSIBILITY OF A MIRACLE

Sohana suffers from a rare, severely painful skin condition that could kill her. Can doctors find a cure? By Caroline Phillips. Photographs by Anastasia Taylor-Lind

NOTE OF OPTIMISM Clockwise, Sohana plays the piano wearing protective gloves; being cared for by her father, James; with the model Kate Moss

The couple were cuddled up on a sofa, talking about their good fortune. "We lead such a charmed life," James Collins told his wife, Sharmila Nikapota. The Cambridge-educated professionals enjoyed the theatre, dining out, romantic holidays. They had a lovely home and planned a family. "The world was our oyster," Sharmila, now 43, says. James is a commercial barrister, recently made a QC. Sharmila used to be a vet. On July 15, 2002, their perfect world was shattered. Their first child, Sohana, was born with the genetic skin condition recessive dystrophic epidermolysis bullosa (RDEB), one of the worst types of epidermolysis bullosa (EB). A lack of collagen VII protein in the skin causes it to blister and fall off at the slightest touch. Little can be done to alleviate the symptoms. The affliction is progressive, incurable, often fatal.

Sohana, now nine, is in constant and often diabolical pain. The level at which the shearing happens is so deep that it's equivalent to a third-degree burn. Every day she's bandaged afresh from shoulder to toe, like a walking mummy. Repeated skin damage leads to scarring, contractures and severe disability: all irreversible damage. At best, she may end up in a wheelchair, but most RDEB sufferers get a malignant skin cancer before the age of 35.

We're in the garden of the Collins's elegant north London house. Sohana's three siblings rough and tumble while she watches wistfully. It's rare to meet a child as captivating. She's impishly humorous, plays the piano and has a boundless imagination. She writes poetry ("I should like to dive into the stars' glow and shine/Keep the light of the moon..."), sings beautifully and has a remarkable *joie de vivre*.

At night Sohana has vivid dreams of running at school sports day or riding a horse across fields. She can do neither in reality: "On sports day I'll be watching, looking happy, but inside wishing I didn't have EB. I look on the bright side but can't help feeling left out. I've never had a day with no pain. It's almost torture to watch people do things I know I can't do."

A few days ago, she asked: "When there is a cure, Mummy, can I have a bike? Can I play tennis? Can I eat toast and Marmite?" "To which I obviously replied, 'Yes,'" says Sharmila. "I think she was testing my positivity." On another occasion, in a tremulous voice, Sohana asked: "Mummy, am I going to die from my illness?"

The answer may lie in the hands of John McGrath, professor of molecular dermatology at King's College Hospital, who works with Osaka University's Professor Katsuto Tamai. "We're developing gene, protein, cell and drug therapies to strengthen the skin," McGrath tells me. "Clinical trials are also in progress to assess cell therapies such as fibroblast injections into the skin [using normal skin cells from donors] or bone-marrow transplants."

"Five years ago," says Sharmila, "Prof [McGrath] couldn't envisage finding any kind of treatment. Now we could have one within two years." It's estimated that there are more than 5,000 victims of EB in Britain. One in 227 people carries the defective gene that causes EB, and one in 400 the faulty gene that can result in RDEB — and if that person's partner has a defect in the same gene, there's a one-in-four chance of their having a baby with RDEB. EB is an expensive condition to manage — dressings and specialist nurse care can cost the NHS up to £100,000 a year per sufferer. The current management of chronic wounds in British patients — including those with EB — costs more than £1 billion a year. McGrath's work could lead to significant savings. "It's time to flood rather than drip-drip money into research," says Sharmila.

Sohana was born four weeks prematurely at London's University College Hospital. She looked perfect, but "by the next morning, the skin had come off her feet. I couldn't hug her because her skin peeled off", Sharmila recalls.

'I COULDN'T HUG HER WHEN SHE WAS BORN BECAUSE HER SKIN PEELED OFF'

"I couldn't breastfeed her, probably because her mouth was sore." In RDEB, the internal mucous linings of the mouth, throat, eyes and anus may also blister. "Just taking her blood for investigation caused enormous trauma. She got blisters that kept getting bigger and bigger. Then the skin came off her legs."

The moment they received the diagnosis, James and Sharmila knew their lives would change irrevocably. "We were shell-shocked," says Sharmila. "I cried myself to sleep every night for a month." She gave up work. For six months they carried Sohana on a pillow, to avoid friction. "People say, 'How do you cope?' The answer is, 'You do because you have to.'"

Sharmila dedicates herself to Sohana, getting up at 6am every day to spend three hours changing Sohana's dressings while her daughter loses herself in a book — unless the pain is too excruciating. To prevent damage to the eyes, Sharmila administers eye drops every half-hour. "If she touches them at night, she suffers intense pain." Sharmila cradles her warm coffee cup as she talks. "Even rubbing her eyes gently can scratch off the protective cornea — leading to temporary blindness. In her first year at school, she had eye abrasions that took two months to heal. I'd just sit in a dark room and read to her while she kept her eyes closed." Then there's the continual round of hospital and dental visits.

Sharmila is honest about the daily strain. "Occasionally, I think, 'Escape, escape, escape...' I don't want to end my life. I just want to get away from the worry of it all." She rejected counselling. "Somebody giving me tea and saying, 'There, there dear'... how's that going to help?" She pauses. "Of course, sometimes I feel down." Does she feel guilty that her body is somehow defective? "Yes, I feel an irrational guilt. But I was relieved that James and I are equally culpable — it wasn't a dominant mutation. Both of us have the recessive gene."

When Sharmila got pregnant again, it was a time of huge anguish. "We did prenatal tests at 11 weeks and had three terrible days of waiting for the results." Would she have terminated her second pregnancy? "Yes," she says immediately. "We'd never want another child going through what Sohana has." Would she have had a selective termination in her third pregnancy if one of the twins had been affected? "Certainly. With the twins, I had high blood pressure after they were born and I also had a C-section. James used to bring Sohana into hospital and I'd do her dressings from my bed." The scarring and muscle shortening can lead to fingers and toes fusing — causing "mitten" deformities — and there's a risk of microstomia, in which a person's mouth decreases in size. So far, Sohana has avoided both. Has Sharmila ever taken time off sick? "No. If I get flu or high blood pressure, I just carry on. I've never taken to my bed."

It must put a huge stress on her marriage.

"We were very good friends before we got married," she explains. "I think that has ⋙→

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FAMILY MOMENT
Sohana (front) with her mother, twin siblings Zuleikha and Akhaila, and sister Jacinda

'I DON'T BELIEVE A LOVING GOD COULD INFLICT THIS ON A CHILD'

helped." But they cannot go away as a couple, even for a night: Sohana may need eye ointment and her dressings must be changed early in the morning. "We've joked about checking into the hotel down the road and coming back before breakfast, just to pretend to ourselves that we've been away." James does whatever he can to help. "He's squeamish, so has only changed her dressings once. But he busts a gut at work to support us. It impacts hugely on the family, particularly on the younger ones."

Jacinda is now eight, and the twins — Akhaila and Zuleikha — are five years old. "But they're constantly being told I can't do things with them." She is on the verge of tears. "The kids have to accept that Sohana's needs come first. I've hardly ever bathed the twins in the evening and I don't get to read them a bedtime story." Swallowing can be painful for Sohana due to scarring of the oesophagus. "So even a quick meal can take an hour, while the little ones wait. It's very tough on Jacinda because she's between Sohana and the twins, both needing a lot of attention." Sharmila adds that the family is "very fortunate" to have a nanny.

Do they manage to get away as a family? "We go on holiday, it's just that we have to take a lot of stuff with us, dressings, drugs..." They cannot do anything spontaneous. When they do go out, often they get stared at. "Children will gather around her, pointing. Or a child will stand in front of Sohana with its mouth open. I've taught her to say, 'Are you a goldfish?'"

The national charity for EB is called DebRA. It receives no state funding. Sharmila embarked on a mission to raise the £10m needed to allow a potential cure to reach the licensing phases, establishing a research fund, masterminding fundraising dinners and persuading high-profile celebrities, such as Kate Moss, to become involved.

Sohana is at mainstream school, where she has a full-time carer who helps her perform basic functions and administers medicines. "But we'd worked so hard at trying to normalise Sohana's condition," says Sharmila, "parents had no idea that, for example, I spend hours every day changing dressings."

The possibility of a cure has been in sight since 2007. That's when the respected surgeon John E Wagner conducted a revolutionary trial in Minnesota: he used bone-marrow transplants to treat children with RDEB. That procedure was unacceptably risky, leading to two deaths out of the seven children. But the trial write-up (in late 2010) emphasised how the cells within the bone marrow can repair skin and even make the collagen VII protein that RDEB sufferers lack.

McGrath and experts around the world had already identified 15 different genes in various forms of EB. They had used the data to develop prenatal tests, including preimplantation diagnostics. But the bone-marrow transplant trial was a catalyst towards the development of new treatments for EB. Now McGrath and his colleagues at Osaka University have had another breakthrough — in identifying which bone-marrow cells play a role in skin-wound healing and pinpointing the signalling mechanism that directs cells to the wounds. The research shows that damaged skin releases a distress protein called HMGB1 that, in turn, summons a key fraction of the bone-marrow cells to repair the skin. This has implications not only for repairing chronic tissue injury in skin, but also perhaps for that in other organs.

McGrath and his clinical colleagues are set to launch trials this year of bone marrow cells called mesenchymal stromal cells (which include the skin-repair cells). The plan is for 10 to 20 children with RDEB to receive intravenous preparations of mesenchymal stromal cells. Further trials are expected to follow using more selective cells that have the potential to restore healthy skin. McGrath is looking for a safer method for EB victims than full transplants.

What if the research money or clinical targets are not met, and an effective treatment or cure is not found? Would the couple ever consider euthanasia for their daughter? "If Sohana got to a point where she was suffering horribly and said to me, 'Please, I've had enough, I don't want any more of this,' then I would try to respect her wishes," says Sharmila, very quietly. "But she has a beautiful spirit. She is a sunny little soul and is a delight. The last thing we would ever want to do would be to go down that path." Does Sharmila have a faith to carry her through such endless trauma? "I don't believe a loving God could possibly inflict on a child the horrors of EB."

She is determined to stay focused on a positive outcome, but can't keep back the tears as she tells me, "how blood-curdling it is when Sohana is screaming in agony and I almost have to hold her down to do her dressings" ■

Sharmila has raised £1,070,000 so far, but it's a race against time. To donate to the Sohana Research Fund, visit www.justgiving.com/sohanaresearchfund or call 01344 771961