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

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