Rebuilding blood

Why are some sickle cell disease patients protected from the disease? Learn about our biomedical research in the field.

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It was a medical mystery. The patients should be showing symptoms of sickle cell anemia. They harbored dangerous mutations in the gene for hemoglobin, the protein in red blood cells that carries oxygen. But they were relatively healthy. Why were they protected from the devastating disease?

Academic groups around the world launched an investigation and made an interesting discovery. Most of us stop producing the fetal form of hemoglobin—to which oxygen is highly attracted—soon after birth. The unexpectedly healthy patients, however, keep making fetal hemoglobin, thereby compensating for the defective adult form of the protein.

Researchers at Novartis and the biotech company Intellia Therapeutics would like to switch the gene for fetal hemoglobin back on in those suffering from sickle cell anemia. The team hopes to extract red blood cells from patients, edit their DNA—using technology called CRISPR—to flip the switch, and then inject the revised cells back into the body. At this point, the project is in its infancy.

“Human data are driving our therapeutic strategy for hematological disorders,” says Lloyd Klickstein, head of translational medicine for the New Indications Discovery Unit at the Novartis Institutes for BioMedical Research. “We know that the fetal form of hemoglobin modifies the course of sickle cell anemia, so we’re optimistic about this potential gene therapy.”

The team also is looking beyond sickle cell anemia to other hematological applications of gene editing. An experimental treatment by Novartis called HSC835 potentially expands the possibilities.

In some cases, it will be difficult to safely obtain enough blood cells from a patient to make editing feasible, especially if he or she needs frequent blood transfusions to stay alive. HSC835 is designed to multiply blood stem cells, which give rise to all the other cells of the blood. It might be possible to combine gene editing with HSC835 outside the body when the supply of blood cells from a particular patient—or group of patients—is limited.

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