Pitt researchers help out at a camp for those with a rare genetic disorder that hampers the DNA-repair pathway. At Camp Sundown, kids get to stay up all night.
IT'S almost 9 o'clock on a warm, humid July evening in the woods in eastern New York State. The sun, as it dips beneath the horizon, sets wispy clouds ablaze with a crimson glow.

At the far side of a large clearing, next to a silent forest, you can just make out the silhouette of a long, low-slung building that looks like a military barracks. No lights can be seen from its windows. It looks uninhabited.

Just 10 minutes later, the sky turns ultramarine and floodlights illuminate the entire exterior of the building. The double doors that bisect the structure burst open. From them pour a couple dozen people who have been living inside in a self-imposed exile from sunlight for the past few days.

This is Camp Sundown, in Craryville, N.Y. The camp brings together a group of children, teenagers, and adults who suffer from xeroderma pigmentosum, or XP, a rare genetic disorder that renders people unable to tolerate ultraviolet light. Because their skin cells cannot repair the damage caused by UV rays, people with XP are thousands of times more likely to develop skin cancers as other people. So, they and their families live for the night.

Camp Sundown is run by Caren Mahar, a 46-year-old mother of five whose 15-year-old daughter, Katie, was diagnosed with the condition as a toddler. Mahar and her husband, Dan Mahar, established the XP Society along with the camp in the mid-1990s. They eventually raised almost a million dollars to build the barracks, which looks more like a dimly lit Holiday Inn on the inside than a military abode.

On this night, the XP campers and their families, who number 41 in all, will travel to the town of Campbell Hall, N.Y., which is about an hour-and-a-half bus ride away. They'll attend a festival and play baseball against a minor league team.

While the group waits for the bus to arrive, Mahar, who describes herself as "a sliver under five feet tall" rattles off orders to the almost dozen assembled volunteers with the authority of a drill sergeant:

"Every family has to be escorted at all times no matter what happens. Do you understand?"

Among the volunteers are four members of the laboratory of Laura Niedernhofer, an MD/PhD assistant professor of molecular genetics and biochemistry in the University of Pittsburgh School of Medicine. At Pitt, Niedernhofer studies the molecular and cellular processes involved in aging. Her group and others discovered that DNA damage, when not repaired, promotes aging. Therefore, a defective DNA-repair mechanism in early life can lead to premature aging. People with XP have an accelerated aging process, says Niedernhofer.

Sensitivity to the sun and a dramatically heightened risk of skin cancer are the main features of XP, notes Niedernhofer. "But as more and more of these individuals are being protected from the sun and are living longer, other types of symptoms consistent with premature aging are being discovered," she adds.

"The most predominant feature is a decline in their motor and mental skills, or neurodegeneration. They also develop problems with their hearing and vision, just like you would see in someone who is in their 70s or 80s. But, in XPers, these things can begin showing up as early as their 20s and 30s."

Among the XPers at Camp Sundown this year are children, teenagers, and adults with a range of symptoms, some mild and some very severe, depending on which genetic mutation they have and where it occurs in the DNA-repair pathway. Niedernhofer notes that there are seven "complementation groups" for this disease, meaning seven different genes can be affected. If the mutation is upstream in the DNA-repair pathway, XPers tend to have milder symptoms. If it is downstream, they tend to have more severe disease.

These families may live in the dark, but they're illuminating science.

"Much of what we have learned about DNA damage recognition and repair comes from studies on XP patients, like the [Camp Sundown] campers," notes Arthur S. Levine, dean of the medical school, senior vice chancellor for the health sciences, and a DNA-repair researcher himself.

Niedernhofer has been bringing members of her laboratory staff to Camp Sundown for the past several years. This year she's brought seven members of her lab at her own expense. Among them is Siobhan Gregg, one...
of Niedernhofer’s graduate students who is getting her first exposure to people with XP. Until now she has only worked with XP mice, which Niedernhofer helped to develop.

“It was a bit intimidating when I first arrived,” says Gregg. “But the families and kids really appreciate our being here and make us feel right at home.”

Gregg and her labmates play games with the campers and help out with events. Because the camp includes family members who do not have XP and can be up and around during the day, someone needs to be available around the clock to help out. Niedernhofer and half of her lab took the day shift; she has left the night shift to the other half of her lab group, which includes Gregg.

Earlier in the day, Niedernhofer delivered a short lecture on her research to the families attending the camp. Although there are no currently available treatments for XP other than avoiding the sun, her lab is experimenting on mice with compounds that may be able to alleviate the damage to skin cells caused by UV light.

“What the families really ask for is a bit of hope,” says Niedernhofer. “We are at a very early point in our research for treatments. ... We anticipate that someday we’ll recruit patients from this group for more detailed analyses of the disease in humans and clinical trials.”

The school bus arrives at 9:30 p.m., and campers immediately begin boarding for the long drive. The teenagers migrate toward the back, while the adults with smaller children sit in the middle. Several campers who are in wheelchairs are put in the front. As instructed, Gregg and the other volunteers post themselves throughout the bus. As the bus heads off into the starlit night, some of the teenagers nod in time to music only they can hear through their ear buds. Others tell each other good-naturedly or tell bad jokes evoking a smattering of groans and giggles. Adults sit in silence or make small talk. Some of the younger children begin to doze off.

About 45 minutes into the trip, as the bus is motoring down the interstate, the passengers are startled out of their seats with a kaboom! The bus has a flat, and the driver immediately begins easing the vehicle onto the shoulder of the highway.

Mahar, who has been following in her Hyundai Santa Fe, boards the bus and tells everyone to stay put. Within a couple of minutes, a highway patrol officer pulls up and, after a short consultation, asks the driver if she can make it to a nearby rest area, where it is safer to make repairs. The bus has double tires on the back, so she is able to drive it, albeit with a loud thumping from the flat tire, slowly to the rest area, which is about 5 miles further down the interstate.

It’s after midnight by the time the bus makes it to the rest stop. Mahar makes calls to Campbell Hall organizers, apprising them of the situation. They are still willing to hold the baseball game, but Mahar is growing increasingly doubtful that it can take place.

In between calls, she recounts the story of a nighttime cruise around the Statue of Liberty. On the ride home, the bus driver took a wrong turn and got lost in Manhattan. Because it was starting to get late, campers with the most UV sensitivity were loaded into a van that had been trailing them.

The van was able to get them back before dawn. The rest of the group, however, got back almost an hour later and watched in horror as the sun began to rise. Although everyone arrived safely back at camp, Mahar said it forced her to take more precautions. Now she always follows the bus in another vehicle with specially tinted windows that is large enough to hold several campers. “We are always racing against the sun,” she says.

It is well after 1 a.m. when the repairman arrives to fix the flat tire. By now, Mahar has called off the baseball game because there is not enough time. No one complains. Then Ronald McDonald arrives from the festival with a van full of gifts and refreshments for the kids. The clown lifts spirits, notes Gregg later: “The kids and families went from feeling nervous about the bus situation to enjoying themselves and laughing.”

At almost 2:30, there is no time to waste. The campers get back on the bus. Mahar loads several into her car and goes on ahead. By the time the bus rolls back into camp it is nearly 4:30 a.m. The campers file off the bus and immediately head inside in silence to the safety of their rooms. Within minutes, a glow begins emanating from the eastern sky.

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