

"We try to send patients home with hope and the feeling that this isn't the end," says Dr. William Gahl (fourth from left, with Jill, in bed, and his team).

SHE WAS HEALTHY ONE DAY, GRAVELY ILL THE NEXT—
AND NO ONE KNEW WHY. ENTER A REAL-LIFE DR. HOUSE
AND HIS TEAM OF DISEASE DETECTIVES BY KRISTEN MASCIA

WHAT'S WRONG WITH JILL DUFOUR?

PHOTOGRAPHS BY EUGENE RICHARDS

HEALTHIER DAYS



Jill (with her German shepherd Digger) in 2000.



"I used to be a real fun hog," says Jill (in 1998 on vacation near Newport, Ore.).

As she jogged through the woods during her wild-fire-fighter fitness tests in April 2002, Oregon forest ranger Jill Dufour heaved for air. Normally a pro at the annual exam—a 3-mile hump with a 45-lb. pack strapped to her back—Jill felt herself dragging. Chalking it up to the flu, she trudged on.

But with every passing day, her symptoms grew weirder and worse. Her ankles swelled mysteriously, strange red bumps spread across her body, and she could hardly muster the energy to get out of bed. "I was so scared," Jill, then 39, recalls. "I had no idea what was going on." Then doctors made a startling discovery: Her kidneys were failing—and no one knew why.

Thus began a six-year odyssey that led Jill and her longtime partner, Anne Boeder, 48, to specialists from Oregon to Toronto, Boston, Paris and Minnesota's Mayo Clinic, in search of an answer. Doctors ruled out a dozen diseases, from lupus to HIV to Lyme, but they still hadn't nailed it in June 2008, when a deadly infection put Jill in crit-

ical condition. Anne summoned Jill's sister Judy, her only living relative, from Detroit, and the couple started planning their estate. Says Anne: "We didn't know if we'd have another 10 years, another day, another hour."

But just when they'd run out of places to turn, a phone call offered one last hope. It was Dr. William Gahl, director of the world's only research program dedicated to the study of mystery diseases. Gahl, 60, heads the Undiagnosed Diseases Program at the National Institutes of Health in Bethesda, Md., running a team of real-life Dr. Houses—ambitious, tenacious, top-of-their-field specialists whose mission is to discover and cure diseases so new they don't

have names. "We call them 'fascinomas,'" says Gahl, referring to the fascination these mystifying, bizarre cases hold for his 50-member team. Open since 2008, the program has seen some 200 patients, most of whom it has helped, and has discovered a brand-new genetic disease. "We get desperate letters—'You're my last chance, that sort of thing,'" says Gahl, who saw Jill in May 2009 after learning of her case from her personal physician. He rolls his eyes when compared to Hugh Laurie's Dr. House, a television character who solves the most complex cases within an hour-long episode, but acknowledges, "We're treating things no one has ever seen before."

"I'M FEELING OPTIMISTIC, AND I'M GETTING MY LIFE BACK"

—JILL DUFOUR



"Anne," says Jill (with her awaiting test results at the NIH Clinical Center), "is my rock."

Sitting up in her hospital bed at 9:45 a.m. on the first morning of her five-day visit, Jill braced herself for a dizzying array of exams. Seated by her side, Anne gently rubbed her arm, while Jill's physician and friend Mary Meyer fiddled on a laptop across the room. After the first of what would be 30 blood draws, Jill looked exhausted. "I've prepared," she said, her blue eyes tearing up, "for bad news."

But news of any kind would have to wait—on yet more tests. Every morning Jill was visited by one or more specialists—including a nephrologist, oncologist, cardiologist and immunologist—and spent afternoons undergoing MRIs, CT and PET scans and pain-management treatments ranging from IV drips to acupuncture. On the last day, Jill and Anne anxiously awaited Gahl's final visit. The team's announcement: They believed a protein in Jill's body called VEGF wasn't working correctly, causing her kidneys to fail. If doctors could just get that protein to do its job, they might be able to save those vital organs. "The battle isn't over yet," Gahl promised. Jill's eyes filled with tears. "I don't know how to thank you," she said

Over the next months, as Jill rested at home in Portland, Gahl often thought back to the promise he'd made to her: "We'll never stop working." In their lab the doctors analyzed and reanalyzed Jill's samples. Then, in August, a breakthrough: With the help of a Boston VEGF expert, they found an antibody in Jill's blood that was impeding the VEGF protein. They could now try to eliminate that antibody, saving Jill's kidneys—and perhaps sparing her from a lifetime of dialysis.

Meanwhile, on new medications, Jill started feeling better. She shed 40 lbs. she'd gained from steroids, returned to her environmental-policy post with the forest service full-time, went back to cooking dinner for friends and recently started taking short bike rides with Anne and their German shepherd Digger. In December the couple took a trip to San Diego—their first vacation in four years—and are talking about an

DISEASE DETECTIVES: CRACKING JILL'S CASE

In May 2009 at the NIH Undiagnosed Diseases Program in Bethesda, Md., a team of medical sleuths brought their specialized skills together to try to discover the cause of Jill's kidney failure. "None of us had ever seen what she has," says team leader Dr. William Gahl. Here's how they did it:



SPECIALIST VISIT

During a May 13 exam by Dr. Anthony Ippolito, a rheumatologist, Jill named the diseases—like scleroderma and Wegener's granulomatosis—that doctors had already ruled out as causes of her kidney problems.

BRAIN IMAGING

A technician checks images of Jill's brain during an MRI May 13. The results helped the team rule out a tumor in her pituitary gland—a possibility doctors had considered after an abnormal finding from one of her blood tests.



FINDING ANSWERS

Members of Dr. Gahl's team (clockwise from left, Drs. Galina Nesterova and Fred Gill, with nurse practitioners Michelle Braun and Colleen Wahl) pore over the results of Jill's nephrology tests May 13.

adventure in Spain. "We can go out and have dinner or go to the movies like normal people—all those little things people take for granted," says Anne. "We have hope again." Which is medicine in itself. "I'm working on

getting my life back," says Jill. "And when I get my feet back under me, I'm going to be full of hell."

For more information on the NIH Undiagnosed Diseases Program, log on to rarediseases.info.nih.gov/undiagnosed