

# The Heart Is a Muscle

Why the Gimbel brothers aren't waiting around for a muscular dystrophy cure.

BY JAMIE TALAN

**P**eter Gimbel was eight years old when he learned that the heart is a muscle. For Peter, this fact was downright frightening. Three years earlier, he and his younger brother Tommy were told they had a disease that was weakening their leg muscles. If the heart is a muscle, couldn't it give out, just like his legs? There's no wheelchair for that, he remembers thinking.

Duchenne muscular dystrophy (DMD) is a fatal genetic condition that causes muscle in every organ of the body, including the heart and lungs, to weaken. More than 40 different forms of muscular dystrophy exist, but DMD is the most common and one of the most severe. By age 12, virtually all children with DMD are in wheelchairs. While patients are now living longer—life expectancy is about 30 years—even common respiratory infections can prove fatal. Progressive damage to the heart is the typical cause of death.

But statistics have never stopped Peter or Tommy from living their dreams. Both young men left their childhood home on Long Island and went off to Brown University. Peter, now 28, is a social worker, husband, and father in Jersey City, NJ. Tommy, 26, is a religious studies scholar completing a doctoral program at New York University. They each have home health aides.

Peter and Tommy believe every day is an opportunity to teach others compassion—for all people with disabilities. “Everyone is disabled in some way,” says Tommy. “My disability is something that everybody can see.”

When Lesley (Lel) and Tom Gimbel Sr. heard that their first two children had DMD, they thought hard about how to raise the boys. Lel had just given birth to their third child when Peter and Tommy were diagnosed. The couple completed

of the disease; others have mutations that link back a few generations. One in every 5,000 women is a carrier.

Lel didn't know she was a carrier of the DMD mutation. Females can carry the genetic mutation even if they have a normal gene that makes dystrophin because they have two copies of the gene. Boys only have one copy, so if they have the mutated form, they get DMD.

Peter says that he grew up thinking a cure was going to arrive before the major milestones of his life—before the wheelchair was parked in his room; then before his graduation from high school, college, and graduate school; before he married his college sweetheart; before his daughter was born. The field has seen ambitious discoveries in the last three decades, including the identification of the DMD gene

and the many mutations, but no medicines have been developed that significantly protect the weakening muscle.

But no matter the odds, the promise of interesting things to do has always kept the Gimbel brothers looking forward to tomorrow. “We never told the boys what they couldn't do,” says Lel. “All opportunities were open for them.”

Lel and Tom Sr. love to exercise, and all of their children were encouraged to participate in sports. Tommy hoped that exercise would be the key to staying healthy. “I always hoped that exercise would keep my muscles strong,” Tommy says. Some experts believe that too much exercise can worsen weakened muscle, but Tommy enjoyed pushing himself. He went off to college with his daily and weekly exercise routines, which included swimming. Then, at the end of his fresh-



**LOOKING FORWARD** From left: Lesley Gimbel, Tommy Gimbel, Hope Gimbel, Lel Gimbel, Tom Gimbel Sr., Jessica Resnick-Ault (Peter Gimbel's wife), Peter Gimbel.

their family a few years later with another daughter.

“Independence was the greatest gift we could give our children,” Lel says.

## LOSING MUSCLE, GAINING STRENGTH

DMD is caused by mutation of a gene that makes dystrophin, a protein critical in keeping muscle fibers strong. Dystrophin is made up of 3,800 amino acids, and any damage to the gene can disrupt the way it works. Because the gene sits on the X chromosome, it is handed down from the mother. In about 40 percent of cases, DMD results from a spontaneous mutation. (New mutations are always appearing in our DNA.) One-third of every 600 boys born with DMD in the United States have mothers who have passed on the defect—some have no family history

man year at Brown, he found a community of people who got together to talk about the teachings of the Bible. He'd never even read the Bible, but the discussions captured his interest.

"I knew I couldn't rely on my body to see me through," he says. "My life needed a deeper dimension." He began taking courses in philosophy, ethics, and Judaism. After graduation, he went to Harvard Divinity School for two years for a master's degree before enrolling in the Ph.D. program at NYU, where he is president of the Graduate Christian Fellowship.

"I want to live longer than anyone else with DMD," says Tommy, who intends to teach religion and philosophy. "My parents always encouraged me to pursue the same activities as other kids and to never let my disability slow me down." Tommy says that he lives in the here-and-now and accepts that "if I want to be me, this body

comes with it. People don't need a disability to understand these basic things."

Peter also feels that he has a responsibility to give back to the world. He works for a nonprofit organization called DAWN Center for Independent Living in New Jersey, where he teaches a weekly seminar called "Living Well with a Disability." Patients with traumatic brain injuries, strokes, and spinal cord injuries learn how to cope with the challenges that people with disabilities face and have an opportunity to share their experiences. "The main benefit is giving people an outlet to talk about their problems," Peter says.

#### SCIENTISTS UNRAVEL DMD

In 1986, two years before Peter and Tommy were diagnosed with DMD, a

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researcher interested in the genetics of the disease was closing in on the genetic address. By 1988, when the Gimbel boys were diagnosed with the

condition, Louis Kunkel, Ph.D.—now a professor of pediatrics and genetics at Children's Hospital in Boston and Harvard Medical School—had identified the first genetic mutation that causes the condition.

Tom Sr. and LeI contacted Dr. Kunkel, who has been following the brothers ever since. The genetic discovery led immediately to a diagnostic test for the disease. Dr. Kunkel also set out to develop a gene therapy for DMD. The hope in gene therapy is to deliver a normal copy of the gene into muscle and have the gene make healthy and functional protein. There were—and continue to be—many challenges. For one thing, the gene is too large to package and deliver to muscle. Scientists spent years trying to miniaturize the protein. They finally did—and in 2006, the first U.S. human gene therapy trial for DMD began at Columbus Children's Hospital in Ohio. Another vexing problem: how to get the healthy gene working in muscles everywhere in the body.

The Gimbel brothers know they can get sick anytime. Last August, Tommy's heart started beating faster, a problem related to DMD, and in February, his right lung collapsed. When he went under anesthesia during a recent surgery to repair the punctured lung, he took the hand of the doctor and his last words before sleep were: "I have a blessed life." The surgery was successful. Tommy says he has too much living to do. NN

## Living Well with Spinal Muscular Atrophy

Jay Cohen and the Gimbel brothers are kindred spirits. Cohen was diagnosed with spinal muscular atrophy when he was nine months old. He barely crawled and never walked. Now 35 years old, his spine is so curved by the weight of his condition that he spends most of his days off his feet. But even from his bed, which is equipped with a computer powered with voice-recognition software, he has made a name for himself. In 2004, Jay developed a Web site for people with disabilities. It started off with a linked directory of places that people could obtain services; today, it has grown to include a large social networking portal with games, blogs, and information on a range of assistive technologies. Every member of the site has an opportunity to host his or her own blog. The site now logs about 30,000 visits a month. "There is a need for people with disabilities to stay connected," says Cohen, who spends a good part of his day administering the site and corresponding with patients all over the world.

When he does get out, he likes visiting Las Vegas. "So many places accommodate my wheelchair," he laughs. He lives with his mother and stepfather in Glenview, IL. When asked whether he ever wanted to live on his own, he was quick to reply: "Why ruin perfection?" Jay's website is [disabledonline.com](http://disabledonline.com).



For more information on muscular dystrophy, see RESOURCE CENTRAL on page 36.