WRITTEN TESTIMONY OF:

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TO

UNITED STATES HOUSE OF REPRESENTATIVES APPROPRIATIONS
SUBCOMMITTEE ON LABOR, HEALTH AND HUMAN SERVICES,
EDUCATION AND RELATED AGENCIES

BEFORE

THE HONORABLE RALPH REGULA, CHAIRMAN

PUBLIC WITNESS HEARING
WEDNESDAY, MARCH 29, 2006
2:00 p.m.

CONCERNING FY2007 APPROPRIATIONS FOR THE
NATIONAL INSTITUTES OF HEALTH

Chairman Regula and members of the Subcommittee, the members of the National Marfan Foundation (NMF) thank you for the opportunity to provide written testimony in support of the FY2007 budget of the National Institutes of Health (NIH), the National Institute of Arthritis, Musculoskeletal and Skin Diseases (NIAMS), the National Heart Lung and Blood Institute (NHLBI) and the National Eye Institute. We are grateful for the Committee's on-going support of funding for NIH research.

It is estimated that 200,000 people in the United States are affected by the Marfan syndrome or a related disorder. Marfan syndrome is a genetic disorder of the connective tissue. Because the connective tissue is essentially "the glue" of the body, this disorder manifests itself in many systems such as the heart, eyes, skeleton, lungs and blood vessels. It is a progressive disorder that causes deterioration of the connective tissue and blood vessels in the body. The life-threatening aspect of this disorder is the weakening of the aorta. The aorta is the largest artery that supplies blood to the heart. The eventual weakening of the aorta causes it to dissect causing large tears in this vessel that will cause death if surgical intervention is not sought. There is no simple diagnosis and no cure. **But there is finally hope for an effective non-evasive therapy for the Marfan syndrome due to the research that has been done by NIH supported scientists over the past 15 years. We are now on the brink of a clinical trial with the Pediatric Heart Network of the National Heart Lung and Blood Institute. These clinical trials are essential to prove the safety and efficacy of these therapies in humans.**

The National Marfan Foundation (NMF) represents people affected with the Marfan syndrome and related disorders which includes familial aortic aneurysms which result in aortic dissection. Aortic dissection is a leading killer in the United States and approximately 20% of the people it affects are young people who have a genetic predisposition to the development of aneurysms. A very deadly and silent killer, with no outward physical signs. However, those with Marfan syndrome usually have outward physical characteristics that only some astute doctors recognize. People are usually very tall, slender and loose jointed. They have long arms, legs, fingers, a chestbone that is either protruding or indented, scoliosis, and flat feet. The diagnosed are able to monitor their aortas for aneurysms and hopefully get elective surgery to replace damaged portions prior to a deadly dissection. However, surgery is not the cure for these disorders. Unfortunately, these disorders are progressive and degenerative in which the rest of the aorta is prone to develop aneurysms also. Therefore, people need to be monitored throughout their lives to detect changes in the size of the different portions of the aorta. In addition, those with Marfan syndrome also experience deterioration of their muscles, joints and bones and are also prone to have dislocated lenses in the eye that can be serious enough to cause blindness in some instances. The cumulative effect of this degenerative progression of the disease results in severe disability, debilitating pain, consequently limiting the individual's lifetime work experience and earnings. **Voluntary health organizations such as ours consistently hear the frustrations, confusion and despair of people who deal with the daily medical issues.**

The NMF would like to share with you one of those stories. Jessica Falco is a member of the National Marfan Foundation and a patient who has courageously battled Marfan syndrome for 18 years. Jessica is a freshman at Loyola University, and the daughter of Susan and Randy Falco. Susan is a member of the National Marfan Foundation's Board of Directors, and Randy is the President of NBC Television. Together, the Falco family has been a tremendous champion for

NMF and the entire the Marfan syndrome community. Here is Jessica's story, who spoke in front of the committee today.

> My name is Jessica Falco and on behalf of the National Marfan Foundation I am honored to appear before the Subcommittee. I would like to extend a special thank you to Congressman Fitzpatrick for his help and attention to our concerns.
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> Obviously, I didn't choose to have Marfan syndrome. It chose me. But it affects the body, not the spirit. My goal has always been to do my best to achieve my dreams and not let this disorder get in the way of my life. That said, there are obvious physical challenges that I and others with Marfan syndrome face every day.
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> I was diagnosed at the age of two. A year later, I was put into a body cast to correct the scoliosis. This was a heavy, hard plastic shell that covered my torso from chin to tailbone. I wore it around the clock, except for when I took a bath or shower, and I didn't take it off for more than eight years.
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> When it finally did come off, I traded the cast for the operating table. I had two difficult operations on my spine at ages eleven and thirteen. The operations lasted nine hours each and were followed months of physical therapy.
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> Fortunately, the surgery worked, and I was able to walk without wearing a heavy plastic jacket. That was a big accomplishment in my life.
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> My other major hospitalization came when I was fifteen and my lung suddenly collapsed. This time, I was in intensive care for a month, basically tied to a hospital bed while we all waited for my lung to reinflate.
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> Physical play is a part of every childhood, except with Marfan syndrome it could be fatal. It was a tough lesson to learn as a young child. How do you tell your friends in second grade that an accidental bump in the chest could cause an aortic aneurysm?
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> But, as I said, this disease may limit the body … but not the mind or soul. It is something I live with. It is <u>part</u> of my life … but it <u>isn't</u> my life. I am currently a freshman at Loyola University. I have hopes and dreams for the future just like every other nineteen-year-old.
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> Mr. Chairman, we are at an unprecedented time of hope in our battle against this disorder. The future holds great promise for patients and families and we look forward to working with you to create a better life for our community. Once again, thank you for the opportunity to testify. I am grateful to my parents and my brothers for all their love and support.

Currently NHLBI and NIAMS are helping to support several programs for Marfan syndrome such as a PHN clinical trial, a national registry for genetically triggered thoracic aortic aneurysms, program projects and many investigator initiated grants. This is an exciting time in research for Marfan syndrome which provides hope for patients and families with this disorder and we need the funding support to keep the movement in these areas.

The cause of the Marfan syndrome is an alteration of the gene that encodes for the protein, fibrillin-1. This major research advance was discovered in 1991. Basic research in molecular biology has helped us to investigate the interaction of the fibrillin-1 gene and its regulatory functions in the extracellular matrix to better understand how the Marfan syndrome comes about. Development of a mouse model has brought insight that Marfan syndrome is due to an increased activity of a family of growth factors that are normally regulated by fibrillin-1. More importantly, it has been found certain drugs can block these growth factors and can prevent the manifestations of this disorder in the Marfan mouse model. An FDA approved drug has shown significant improvements not only in the cardiovascular system but also in the pulmonary and musculoskeletal systems in the mouse model. This is truly an incredible discovery. We may truly be on the verge of a drug therapy to help prevent the development deadly aneurysms. It has taken years of research funded by NIH to bring us to this day and therefore we are truly grateful for the research dollars that were given to both NIAMS and NHLBI that helped support these research efforts. We look now to the government to continue the much needed support of research dollars to NIH. We look to the Pediatric Heart Network of the NHLBI to provide support for a clinical trial to test a new drug in the Marfan population which will begin in September 2006. This potential treatment represents the first opportunity to prevent aortic aneurysm and therefore eliminate the need for risky heart surgery in addition to show beneficial affects in the lung and muscle. It is the hope and the closest thing to a cure that we have today. We need the continued support in order to make this a reality.

In addition to the clinical trial, quality of life issues such as back, joint and muscle pain must be addressed. NIAMS has been instrumental providing mechanisms for Marfan research and should expand its resources to help investigate the musculoskeletal issues of Marfan syndrome. Funds used to study this rare disease to uncover the reasons for premature osteoporosis in the Marfan population can lead to therapies to help millions. Furthermore, chronic pain is so often a part of their daily lives, and much more effort needs to be put forth to help develop treatments for this pain relief. We believe that development of specialized centers of excellence in which researchers and clinicians can partner together and develop multi-prong research programs as well as medical care in multiple areas will allow patients to go to experienced clinics and have all their needs attended to. These centers can also study a group of related disorders such as familiar aortic aneurysms and the newly identified syndrome known as Loeys-Dietz which like Marfan syndrome can have devastating cardiovascular effects at even younger ages. This environment will also provide a network for future clinical trials and provide for a place to cultivate young researchers in this field. It is our hope that both NHLBI and NIAMS look forward to accomplish these goals through all available mechanisms possible. The tools and technology are there to be exploited; we just need the funding to continue the growth in this area.

The National Marfan Foundation would ask members of the subcommittee to expand research not only for Marfan syndrome but for research in general to safeguard the scientific and medical

productivity throughout the country. With possible decreases in funding levels of current R-01 grants, each currently funded investigator may have to release one employee. In addition, the funding levels and rates of approval are likely to favor the more senior experienced investigator and deplete the scientific community of the younger people, to say nothing about discouraging this group and pushing them toward moving out of research altogether. This will likely affect the middle tier institutions and have serious economic effects.

Hope for a better quality of life for patients with Marfan syndrome lies in advancements made through NIH supported biomedical research. For FY07, NMF encourages the Subcommittee to provide a 5% increase for each NIH institute and center. We are particularly focused on research being conducted on Marfan syndrome through the National Institute of Arthritis and Musculoskeletal and Skin Diseases, the National Heart, Lung and Blood Institute, and the National Eye Institute.

In the coming years our scientists are anxious to establish a long-term, multifaceted research program that brings together each relevant NIH institute for the purpose of addressing all manifestations of Marfan syndrome. We look forward to working with our friends at NIH to create a cross-cutting research initiative that will enable investigators to tackle the many different aspects of Marfan syndrome.

Finally the National Marfan Foundation is working hard to increase awareness of the disorder given the tragic consequences that result in patients who are not properly diagnosed. Early detection and treatment of the Syndrome are essential if we are to prevent deadly aortic aneurysms. NMF is anxious to partner with the Centers for Disease Control and Prevention as part of this educational effort. We look forward to working with the Subcommittee to help facilitate a formal partnership with CDC in FY07.

The NMF hopes the Congress will work with us and be dedicated to improving the treatment and quality of life of all patients with the Marfan syndrome. We look forward to no child having to deal with the challenges of this disorder, and no loved one will suffer the loss of a life cut short by Marfan syndrome.

The NMF as a member organization supports the Ad Hoc Group for Medical Research Funding, the NIAMS Coalition and the NHLBI Constituency Group in their request to sustain the current momentum of research which will benefit all Americans. NIAMS and NHLBI provide a primary resource for rare, genetic, multi-system disorders – such as the Marfan syndrome. Funding biomedical research through the NIH is today's investment in America's future. The technology and the science *are* available to understand and ultimately cure or eradicate many of these devastating genetic disorders.

The National Marfan Foundation is a non-profit voluntary health organization dedicated to saving lives and improving the quality of life for individuals and families affected by the Marfan syndrome and related disorders.

The Foundation, which was founded in 1981, accomplishes these goals by:

- Educating patients, family members and the health care community about the syndrome.

- Advocating and funding basic and clinical research into the syndrome's detection and treatment.
- Providing national support services and a network of local and special interest support groups for patients and relatives to share experiences and improve their medical care.

The NMF works to achieve its mission through a comprehensive resource center that includes a toll-free number and personalized email/phone communication; an annual conference for affected people, their families and health care providers; informative publications targeted to both the public and medical community; a coast-to-coast support network; national and international scientific symposia; public awareness campaigns; medical education initiatives at hospitals and national professional meetings; and an aggressive research program that funds meritorious projects in basic, clinical and translational research.

For questions or additional copies of this testimony, please call

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Biography

I was born in Westchester County and grew up in Pound Ridge, New York. At a very young age I began taking piano lessons, which I really enjoy. I attended St. Patrick's Elementary School until 7th grade when I was accepted into The Convent of the Sacred Heart. There I was the head of the community service club, played in the Instrumental club and wrote for the school newspaper. I graduated in June of 2005 and I am presently a freshman at Loyola College in Maryland considering a major in fashion design.

DISCLOSURE STATEMENT

The public witness, Jessica Falco and the National Marfan Foundation did not receive any federal grant in the last three years.