

Written Statement for the
Social Security Administration for
Compassionate Allowances

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Commissioner and Members of the Panel

My name is Dr. Josephine Grima and I am the Vice President of Research and Legislative Affairs at the National Marfan Foundation. I am honored to appear before this panel today. I would like to extend a special thank you to Diane Dorman from NORD for her support and attention to our concerns. I would also like to acknowledge Dr. Stephen Groft who has been a valued advisor for the National Marfan Foundation for many years. And lastly, I want to thank the Social Security Administration for their help during the past few years addressing our concerns that more information about Marfan syndrome and rare disorders is required and for producing an informational video on Marfan syndrome for their adjudicators, in addition to providing more guidance for Marfan syndrome under the cardiovascular impairment listing.

Marfan syndrome is a rare medical condition effecting approximately 40,000 people and is classified as a heritable disorder of connective tissue. Connective tissue provides structure and support for the body. In addition, it provides many regulatory functions which control development before birth, growth after birth, cushioning of joints, and enabling passage of light through the eye. All organs contain connective tissue, and therefore the manifestations of the Marfan syndrome appear in many parts of the body, especially in the bones and ligaments (the skeletal system), the eyes (the ocular system), the heart and blood vessels (the cardiovascular system), the lungs (the pulmonary system), and the fibrous membrane covering the brain and spinal cord (the nervous system).

The diagnosis of the Marfan syndrome may be difficult because there is no specific laboratory test for the condition. Instead, it is a composite of clinical findings that are required to make the diagnosis of Marfan syndrome in an individual. The diagnostic evaluation for the Marfan syndrome should be performed by physicians experienced with the condition and should include the following:

- Detailed medical and family history.
- Complete physical examination.
- Thorough eye examination by an ophthalmologist who uses a slit lamp to look for lens dislocation after fully dilating the pupil.
- Echocardiogram (a sound-wave picture of the heart) looking for involvement of the cardiovascular system that is often not evident from the physical examination.

People with Marfan syndrome are at risk due to the weakness of the aorta and can result in a possible tear or rupture which can lead to sudden death. Therefore, Marfan syndrome is currently listed under the cardiovascular impairment listing. Patients with a chronic uncontrolled dissection are in need of urgent medical attention. In many cases, the dissection or tear can be controlled

with medical therapies. Therefore, in most cases, this is not the characteristic that renders them disabled, however it is the measure used to determine disability under the current system. Instead, it is the chronic debilitating effects of this syndrome on Marfan patients which are slowly becoming known and when in combination become the factors leading to disability. People with the Marfan syndrome may have to endure multiple cardiovascular surgeries to repair several portions of the aorta. Repair of descending aortic aneurysms has a higher risk of serious complications and the neurological damage that occurs with these more difficult surgeries and increases the length of time required to recover and in some cases, patients may not fully regain the level of energy required to maintain a job. Difficulty breathing which can be a result of impaired lung and/or heart function or medications, combined with premature onslaught of musculoskeletal and joint deterioration and pain results in continual fatigue. In addition, they can develop related chronic illness problems such as edema, varicose veins, digestive system problems, incisional hernias, cardiac arrhythmias, extremely poor eyesight and dural ectasia resulting in headaches and numbness and pain in the legs. Individuals are therefore unable to sit or stand for long periods of time to perform basic sedentary work activities. In addition, jobs with more physical requirements are not advised for people with Marfan syndrome because any strenuous activity can cause a dissection of the aorta or aggravate an existing dissection.

The psychological, social and financial consequences of multiple body system involvement in the Marfan syndrome is devastating, especially in the aging population. The effect of the death of multiple family members - parents/siblings/children within the primary family and throughout the extended genetically affected family - weakens the patient's already strained critical support system. Each major "incident" translates into non-productive years, disrupted or discarded careers, lost wages and assets, and an emotional and psychological disconnect and alienation from society. Couple this with the specter of early death and the resulting angst and pervasive depression that ensues further debilitates the patient.

Since the Marfan syndrome is a condition that compromises the integrity of the body's connective tissue, and connective tissue is everywhere in the body, many body systems can be affected. The Marfan syndrome can present itself with a host of medically determinable physical and mental impairments, and exertional and non-exertional limitations relating to any or all of the affected body systems. Any combination of adverse involvement of these body systems can result in a lifetime of chronic pain, the inability to perform even non-exertional work, unmanageable expense, and resulting disability.

In the case of Marfan syndrome, it may be difficult to make a case for disability based on the listing of impairments that currently exists. Not all Marfan patients will require disability as they get older. However, there are a few which really need to be considered even though they do not meet the current strict eligibility

requirements. The diagnosis of Marfan syndrome obviously has to be established and there is well documented information on diagnosis. Once diagnosis is confirmed, the symptoms which are serious enough to interfere with the ability to obtain gainful employment need to be evaluated. Usually, these will consist of orthopedic pain which impedes mobility, dural ectasia which causes back pain and leg pain, loss of vision due to dislocation of lenses, loss of pulmonary function, muscle pain, fatigue and most important cardiovascular problems such as existing dissections. People with a family history of dissection are more likely to suffer from more dissections. Individuals that have had 2 or more dissections, might very well be a good candidate for disability since the physical and emotional stress as well as other cumulative effects of other symptoms may impede a person to obtain gainful employment. Recently, unpublished data from a reputable laboratory from Johns Hopkins has shown considerable muscle degradation and inability to recover from injury in Marfan mouse models which may be a significant cause of fatigue and muscle pain in the human population

The NMF would like to see the Social Security Administration take a more serious look at the functional limitations of a person with Marfan syndrome and other rare disorders. The only criteria for Marfan syndrome that is currently in place is a dissection that is not medically managed which in many cases means that death is imminent if surgery is not performed which indicates that the criteria is much too strict. In addition, the instability of the aorta is probably underappreciated by many doctors who do not see many Marfan patients.

With regard to the processing of claims for patients with Marfan syndrome who meet the criteria, claims take a long time to be processed and it usually takes longer to get approved due to Marfan syndrome being a rare disease and not being well known. In addition, the effect of Marfan syndrome on the whole person and not just the cardiovascular system is not understood.

Most people must appeal the initial decision, often multiple times. For those that get denied multiple times, we believe this is due the severity of the standards listed in the cardiovascular impairments for Marfan syndrome. Many appeals go all the way to the final judicial review. In many cases, Marfan patients have to be evaluated for effects in multiple body systems and again many may not meet the severity of any one of the body systems in other listings, but when multiple less severe impairments occur, a disabled state can also occur. Adjudicators and in some cases physicians are unfamiliar with the multiple system effect of Marfan syndrome and its impact on the day to day functional abilities of many people. Another major roadblock is that physicians are unfamiliar with the social security application process and don't include the important facts in letters of support for patients causing patients to appeal the initial decision. In addition, medical evidence for many of the functional limitations is lacking and therefore we believe it is necessary for adjudicators to evaluate physician requests for disability. When appropriate physician reports are obtained, they may require a greater weight in the evaluation so an appropriate decision can be made. Finally,

SSA staff are often unfamiliar with rare diseases and don't take the time with patients to listen and learn in order to better understand the patients disability and consider the application or help the applicant to get better or proper documentation of their claim.

The supplemental Security Income program (SSI) is definitely a good program for those who do not qualify for disability but it again has to have more information on rare disorders such as Marfan syndrome.

In order to improve the current system, a listing of rare disorders and their debilitating symptoms and impact on each bodily system for adjudicators to utilize would be extremely helpful. Multi-system disorders such as Marfan should be recognized as such so that degree of multiple disabilities can be better recognized and evaluated. Guidelines to outline the need to evaluate multiple factors which do not have concrete medical evidence such as fatigue need to be developed. Guidelines for doctors explaining the types of information they should include in letters of support and medical records for patients applying for social security benefits would also be helpful. Adjudicators should have a better understanding of how difficult it may be for an individual with a rare disorder to obtain documents to support their application and therefore be more willing to assist patients to fill out the application and obtain support documents rather than simply turning them away.

Several improvements could help people with rare disorders. In most cases, rare disorder often result in a multitude of chronic symptoms and disabilities.

1. Providing information in the listing of impairments for rare disorders such as Marfan syndrome to include more aspects of the disorder than just the most severe. There are many debilitating symptoms with Marfan syndrome such as ocular issues, orthopedic issues, dural ectasia and the effect of multiple complex and risky surgeries with lasting effects. The listing for Marfan syndrome should recognize that most people with Marfan syndrome have overlapping issues that prevent them from working. For example, an orthopedic issue alone may not prevent an individual from working but severe back issues in addition to extreme ocular issues and the potential for dissection could prevent an individual from working.
2. SSA workers need to be more familiar with rare disorders and should have a reference they can use when unfamiliar with a particular illness.
3. Guidelines needs to emphasize that for people with rare disorders it can be much more difficult to obtain diagnosis and proper medical evidence and guidance on how to obtain appropriate letters of support and other documents as well as greater emphasis on these documents may be needed when applying for social security benefits.
4. When people are denied disability, many give up and do not understand the role of an appeal. This right needs to be clearly stated in plain language in a prominent place. The reasons for the denial need to be

specific and clear. If an application lacks important information, this needs to be stated. The denial letter should explain what additional information might make for a different decision.

5. In many cases, people applying for disability have no income, they lack medical insurance to receive medical care that would address their medical needs and document their disability. When applying for disability many need to see SSA doctors for information to substantiate their claim but many of these doctors do not know Marfan syndrome. Having physicians that know about rare diseases would be a benefit.

Thank you again for this opportunity.