

Institute of Medicine Review of the Cardiovascular  
Listing of Impairments for the  
Social Security Administration

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December 14, 2009

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 you today and I appreciate the opportunity to bring attention to our concerns. I want to thank the Social Security Administration for its responsiveness to several of our concerns over the past few years. Examples of these improvements include the production of an informational video for adjudicators on this rare disorder and better clarification of Marfan syndrome in the guidance to help adjudicators better evaluate claims.

Marfan syndrome is a life threatening disorder due to the potential for dissection of the aorta. Currently, Marfan syndrome is listed by SSA as a cardiovascular impairment specific to patients with dissections that are not controlled by prescribed treatment. However, these “uncontrolled dissections” are the most severe of circumstances which usually require hospitalization and unfortunately lead to loss of life in many instances in a very quick and abrupt manner. Most dissections, although severe and disabling can be controlled with medical therapies. However, people with a history of dissection are at a 70% greater risk of having further dissections, therefore, modification of the lifestyle is crucial to protect these individuals. The fact that cardiovascular disability determination is based on whether the dissection was uncontrolled misses the mark as a true indicator of disability in our opinion.

Recent studies of 192 patients have shown that 70% of patients undergoing elective repair of the aortic root have subsequent aneurysms or dissections at other sites of their vascular system and approximately 11% of those require additional surgeries (1). For those individuals that required emergency surgery because of a dissection, approximately 73% of the patients required additional surgeries (1). In several cases, patients may have required up to five additional high risk surgeries including repair of the mitral valve. Therefore, it needs to be noted that surgery and controlled dissections for Marfan patients is not a guarantee against further degradation of the aorta and other blood vessels. In addition, with the increase in surgical events, additional cardiovascular complications ranging from endocarditis, congestive heart failure, cerebrovascular accidents, arrhythmias and neurologic damage can occur and may result in cumulative detrimental effects.

It is this group of Marfan patients, who have had multiple surgeries and subsequent complications that should be clearly classified in the cardiovascular listing of impairments as disabled because of limited residual functional capacity or because of deterioration of one or more body systems resulting in chronic and debilitating functional capacity. The complexity of cardiovascular impairment, in addition to impairments in other body systems for those with Marfan syndrome, should be clearly defined under this listing. Currently, there are no functional

scales that cardiologists rely upon with regards to following patients with Marfan syndrome and aortic disease or dissection.

Marfan syndrome is rare medical condition effecting approximately 40,000 people in the United States. Because Marfan syndrome is a connective tissue disorder it manifests itself in several body systems ocular, musculoskeletal, pulmonary, and nervous systems not only the cardiovascular system. The diagnosis of Marfan syndrome requires a multidisciplinary evaluation by medical professionals knowledgeable and experienced in the evaluation and management of Marfan syndrome and related disorders of connective tissues. The age of onset of the cardiovascular manifestations in MFS is highly variable and has to be individualized. The evaluation includes a history and physical examination, imaging of the aorta by echocardiography and in some instances CT scanning or MR imaging, slit lamp eye examination performed by a knowledgeable ocular specialist, orthopedic evaluation and in some instances molecular genetic mutation analysis. To determine the severity of illness in various systems, imaging tests (such as echocardiography, CT, MRI) of the heart and skeleton and eye are performed. There are different degrees of involvement in all the organ systems involved in MFS. Marfan syndrome is a progressive disorder with virtually no prophylactic therapies available to modulate the deterioration of lungs, bones and joints, eyes and blood vessels. Life expectancy of Marfan patients has increased from 45 to 70 years of age due primarily to elective surgical repair of the aortic root. However, the aging Marfan population is a new entity and the natural history of this older generation is showing complex disability issues resulting from multiple aortic surgeries; degeneration of the bones and joints; visual problems associated with dislocation of lenses; dural ectasia resulting in back pain, headaches and difficulty moving the lower extremities; pulmonary problems; hernias; and early onset of arthritis. This is evident to us with the dramatic increase in inquiries to our office during the past 10 years. The chronic debilitating effects of this syndrome in this population have become the leading factors of disability. Individuals are 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 aneurysm or dissection. Even in closely monitored patients, instability of the aorta remains problematic and is a fact of Marfan syndrome probably underappreciated by many doctors who do not see many Marfan patients.

Functional impairment from the cardiovascular standpoint usually hinges on making the diagnosis of Marfan syndrome or related disorder. With these conditions, there are absolute and relative contraindications and restrictions placed on certain physical activities and workloads, because of the risk of undue stress on the aorta. After aortic dissection occurs, the restrictions are often even more severe with regard to functional limitations.

Processing of claims for patients with Marfan syndrome take a long time to be processed. Most people are denied on first application and must appeal the initial decision, often multiple times. For those denied multiple times, we believe this is due the severity of the standards listed in the cardiovascular impairments for Marfan syndrome. Marfan syndrome is a rare disease and it is not well known. The effect of Marfan syndrome on the whole person and not just the cardiovascular system is not understood by the adjudicators or reflected in the listings. Many appeals go all the way to the final judicial review. In most cases, Marfan patients have to be evaluated for effects in multiple body systems. They may not meet the severity requirements of any one body system in other listings, but when multiple, less severe impairments occur, required restriction on workload and lifestyle, a disabled state can occur as a result of cumulative effects across body systems. Adjudicators, and in many 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 problem is that physicians are unfamiliar with the social security application process and do not include the important facts in letters of support for patients. In addition, published medical evidence for many of the functional limitations is lacking. Therefore physician reports and requests for disability should be weighted more heavily by adjudicators in the process if an appropriate decision is to be made.

Marfan syndrome cannot be expedited under terminal illnesses because it is not considered terminal. However, we are very aware of cases where people have had problematic descending aortic dissections, were denied disability, struggled to continue working and died before ever receiving disability. Unfortunately, it is the lack of appreciation of the instability of the Marfan aorta that appears to create this devastating result.

In order to improve the current system, our recommendations are as follows:

- Develop functional scales of cardiovascular risk associated with dissection, multiple surgeries and family history.
- Develop groups of adjudicators that are dedicated to reviewing specific diseases, so they become experts in that disease state.
- Develop a listing of rare disorders and their disabling symptoms and impact on each body system for adjudicators to use would be extremely helpful.
- Multi-system disorders such as Marfan syndrome should be recognized as such so that impact of multiple disabilities can be better recognized and evaluated.
- Guidelines to evaluate multiple factors which do not have concrete medical evidence such as fatigue need to be developed.
- Guidelines for doctors that explain the types of information/medical records needed to document disability and helpful content for letters of support 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 should therefore be more willing to assist patients with the application and support documents.
- 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.
- 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.
- A survey of how many people have been denied disability the first time around and then received it after the second or third round would be a good indicator of how successful the initial process is. Further evaluation of why those applications were denied initially would provide insight into what is missing in the initial application process or what clarification needs to be made to the cardiovascular listing of impairments for Marfan syndrome so that these claims could have been awarded during the initial application process.

Thank you again for this opportunity for me to advocate on behalf of the Marfan community.