



International Federation of Marfan Syndrome Organizations

www.marfanworld.org

The International Federation of Marfan Syndrome Organizations (IFMSO) was formally founded in 1992 at the Second International Symposium on the Marfan syndrome in San Francisco, California, USA.

The purposes of IFMSO are to:

- Share current, accurate, information about the Marfan syndrome worldwide and facilitate international communication among medical professionals and the general public.
- Support and foster research throughout the world and facilitate communication with research centers and researchers worldwide.
- **Provide support to begin and develop a Marfan Syndrome Organization in different countries.**

Marfan Syndrome

The Marfan syndrome is a heritable disorder of the connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels. The condition affects both men and women of any race or ethnic group. It is estimated that at least one out of every 5,000 people in the world has the Marfan syndrome or a related connective tissue disorder.

Because connective tissue holds the body together, the Marfan syndrome manifests itself in many body systems, including the bones, ligaments, eyes, heart and blood vessels. Often the aorta, the large artery that carries blood away from the heart, is weakened. Many people with the Marfan syndrome experience an expansion of the aorta. Without proper monitoring and medications to reduce the stress on the aorta, the aorta could tear, resulting in sudden death.

The Marfan syndrome is difficult to diagnose because there is no specific laboratory test for the condition. In addition, characteristics of the disorder vary greatly among affected individuals. Most affected people do not have all the possible signs and complications of the syndrome.

An accurate diagnosis of the Marfan syndrome can be assessed by a complete physical examination that focuses on the systems affected by the disorder. This includes:

- Echocardiogram, a sound wave picture of the heart by a cardiologist.
- Slit-lamp eye examination by an ophthalmologist after fully dilating the pupil.
- Skeletal examination.
- Complete family history.

The recent identification of the chromosome, gene and component of connective tissue (fibrillin-1) in which the mutation for the Marfan syndrome is located offers great promise for the diagnosis of the condition. It is hoped that as better understanding of the fibrillin-1 is gained, earlier and more accurate diagnosis of the Marfan syndrome and related connective tissue disorders will be possible.

International Marfan Community Activity Highlights

International Symposia:

- Baltimore, Maryland, USA, 1988
- San Francisco, California, USA 1992
 - Formation of IFMSO
- Berlin, Germany, 1994
 - Marfan Syndrome Clinic Criteria
 - Marfan Syndrome Diagnostic Criteria
- Davos, Switzerland, 1996
 - Revised Marfan Syndrome Diagnostic Criteria Accepted
 - IFMSO Fact Sheet
- Helsinki, Finland, 1998
- Seattle, Washington, USA, 2001

1986: Diagnostic criteria for Marfan syndrome refined in the Berlin Nosology for Heritable Disorders of Connective Tissue. Beighton P, de Paepe A, Danks D, Finidori G, Gedde-dahl T, Goodman R, Hall JG, Holister DW, Horton W, McKusick VA. International nosology of heritable disorders of connective tissue, Berlin, 1986, *Am J Med Genet* 1988; 29:581-594.

1996: Diagnostic criteria for Marfan syndrome revised further in the Ghent Nosology. De Paepe A, Devereux RB, Deitz HC, Hennekam RCM, Pyeritz RE. Revised diagnostic criteria for the Marfan syndrome. *Am J Med Genet* 1996; 62:417-426.

For Additional Information About the Marfan Syndrome, Please Contact the Nearest Marfan Syndrome Organization at www.marfanworld.org or info@marfan.ca.

Current Members

Argentina
Queensland, Australia
Victoria, Australia
Belgium
Canada
Cuba

Denmark
Finland
France
Germany
Greece
Holland
Italy

Japan
Mexico
New Zealand
Norway
Poland
Singapore
Slovakia

South Africa
Spain
Sweden
Switzerland
United Kingdom
United States

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Treasurer – Cheryl Williams, United States: dwilli7171@aol.com