

POLICY PLAN 2009/2010

MSS RESEARCH FOUNDATION



Foreword

This is the policy plan 2009-2010 of the MSS (Marshall Smith Syndrome) Research Foundation.

Following a successful first year, the board went 'on retreat' for a day to discuss policy which laid the basis for this policy plan. The policy is grouped around five main themes. These can be summarized as follows:

- 1) **Research:** with the support of a medical advisory board, substantial effort will be put into further research on MSS with the breathing difficulties as the first priority;
- 2) **MSS Website 2.0:** the website will be further developed as a network and knowledge-sharing platform for MSS, based on the latest developments in Internet 2.0;
- 3) **International Care Standard:** an international care standard will be developed to provide medical guidelines, treatment recommendations and best practices, etc;
- 4) **Fundraising:** efforts will be made to increase the group of "Friends of the MSS Foundation" and personal sponsorship campaigns under the slogan of "Breathtaking campaigns for MSS";
- 5) **International outreach:** MSS has no boundaries; the aim of the organization is to be there for all MSS patients worldwide.

Together with the children, parents, doctors, paramedics, donors, sponsors and everyone else involved, we are also looking forward to taking further steps in 2009 and 2010 towards improving the position of people with MSS, increasing awareness among the general public and promoting further scientific research.

The Board of the MSS (Marshall Smith Syndrome)
Research Foundation,
The Hague, July 2009

Froukelien Schiebaan-van der Mooren (Chairperson)

Henk-Willem Laan (Secretary/Treasurer)

Liaan Jansen

Wout Koelewijn

Sonja Bracke

What is MSS?

Marshall-Smith Syndrome is a rare disorder which affects growth and development. It is characterized by abnormal development of the skeletal system in childhood, breathing difficulties due to underdevelopment of the throat and delayed neurological development, among other things. Its cause is still unknown.

What is the objective of the Foundation?

- to strengthen the position in society of people with the Marshall-Smith Syndrome (hereafter referred to as MSS);
- to provide the general public with information on MSS;
- to finance and foster scientific research into MSS;
- to carry out any further activities connected with the foregoing in the broadest sense or which may be beneficial for this purpose.

Among other things the organization seeks to realize its goals by promoting contact among fellow-sufferers, informing the general public about MSS, working to promote the interests of people with MSS and encouraging research into the cause and treatment of MSS, as well as enabling the funds raised by the organization and provided by other bodies to be used for research projects at Universities and Research Institutes in the Netherlands and elsewhere.

(Articles of Association, Article 2).

Theme 1: Research

Research into the cause of MSS has begun at the Institute of Child Health of University College London. The MSS Research Foundation intends to play an active role in this scientific research. From the patients' perspective it is very important to prevent the condition worsening and improve the quality of life. A better understanding of the pathophysiology of the syndrome could also be very important in this context. The first research priority however will be focused on the parents' request to understand the cause and treatment of the breathing difficulties, because this is the most life-threatening aspect of MSS.

The board will provide further support to the research group in London in terms of finance and information. New patients will be made aware of the research in progress and the assistance that they could possibly give. The research group will report its findings on an annual basis.

The board would very much like to set up a medical advisory body. The role of this advisory body has to be further defined. Input and criteria for this will first be gathered through other patient groups and research organizations. Representation will be sought with regard to various medical specialisms as well as in the paramedical field. These could include pediatrics; clinical genetics; ENT medicine; physiotherapy and occupational therapy. Grant terms also need to be drawn up for research institutes to apply for grants for research into MSS.

The board will provide further support to the research group in London in terms of finance and information.

Contact will be made with at least two patient associations to discuss their research policies. Further to this a plan will be developed to provide a qualitative framework for present and future research. The terms which apply to subsidy requests or requests for assistance will be amended on this basis.

No	Objective	Desired result	Action to be taken by	Accomplished
1.1	Fostering scientific research by the ICH UCL and other research institutions	A better understanding of the cause and treatment of MSS, based on research	Froukelien	Anytime
1.2	Criteria for medical advisory body and identifying potential members	Criteria and candidates for medical advisory body	Froukelien	Nov 2009
1.3	Formation of a medical advisory body	Proper assessment of current research and new research topics	Froukelien	Jan 2010
1.4	Terms for grant applications made by research institutes	Clear standards and evaluation criteria for new research applications	Wout	Dec 2009
1.5	Developing a scientific research plan	Scientific research plan developed	Froukelien	March 2010
1.6	Improving grant terms based on recommendations of advisory body	Better assessment and evaluation of research applications	Wout	June 2010

Theme 2: MSS Website 2.0

Over the next two years the board wishes to further enhance the website. The ultimate aim is to improve the quality of life of young MSS patients worldwide. New contacts made through the internet could be vital to them. Information on the syndrome can also be made available worldwide and further developed. The current developments in Internet 2.0 offer enormous potential for this. The board intends to apply the following three principles* in developing the **MSS website 2.0**:

- 1) **‘Wisdom of crowds’**: Medical knowledge is no longer the exclusive domain of medical experts. Knowledge is collectively built up through the internet. Based on the collective wisdom of the MSS community, much more information can be exchanged and knowledge generated.
- 2) **‘Social technologies’**: The website should make use of the powerful modern tools of social innovation or ‘user generated content’ (e.g., Hyves, Facebook, Twitter, Wikipedia, etc.) to create a real MSS community.
- 3) **‘Protecting the commons’**: The knowledge generated is very valuable. Not directly in terms of money, but more specifically in helping parents and, through research, in saving lives. Therefore the rights (and also licences) and intellectual property will need to be properly protected.

The biggest challenge lies in making the website available in many different languages. Every effort will be made to make important information on MSS available in as many languages as possible. This also means that translators must be able to easily convert content into other language versions. It will be attempted to find a local contact in as many countries or languages as possible.

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The primary goal is to create a Wikipedia-like environment in which worldwide knowledge on MSS can be built up. The aim is also to greatly improve the design of the website. In this way, through the use of all the available ‘social technology tools’, the website can become a real online MSS community. This will lay the foundations for the website to be used for the purposes of research, by conducting online surveys and thus creating information in databases for current and future research.

**) Thanks to David Golub, Vice-President Mark Krueger & Associates, Inc., New York, USA*

No	Objective	Desired result	Action to be taken by	Accomplished
2.1	Creating a Wikipedia-like environment for knowledge generation	Increasing and sharing information on MSS	Henk-Willem	Dec 2009
2.2	Improving website design	Better accessibility and more attractive website	Henk-Willem	Dec 2009
2.3	Utilizing the available social technologies on the website	Enhancing the online community	Henk-Willem	March 2010
2.4	Looking for translators	Making the website available in as many languages as possible	Bestuur	Anytime
2.5	Finding contacts to be included on the website	Local contact person in country / for that language	Sonja	Anytime
2.6	Developing a tool for online questionnaires on the website	Facilitating online research	Henk-Willem	June 2010

Theme 3: International Care Standard

Because most medical practitioners are unfamiliar with MSS, it often takes a long time to arrive at a correct diagnosis, care is uncoordinated and inadequate, and there is insufficient information available. Through the development of a care standard for MSS it will be possible to set a standard which the requirements for good care must meet when dealing with children with MSS. A care standard will make it possible for the parents of a child with MSS to approach care more from their own perspective.

A professional care standard will incorporate detailed medical information together with the knowledge and experience of the parents of children with MSS.

The aim over the next few years is to work towards a detailed international care standard which provides medical guidelines, treatment recommendations and best practices, etc.

A care standard will make it possible for the parents of a child with MSS to approach care more from their own perspective.

A working group will be set up to draw up a care standard. This group will comprise parents and professionals involved with MSS. The working group will examine what criteria the care standard should meet. Essentially the guideline on standards of care for rare diseases of the Dutch Genetic

Alliance (VSOP) will be followed. Existing care standards (e.g., for Schisis and Duchenne patients) will also be considered. The aim is to learn from their experience. This will be done systematically on the basis of questions and criteria drawn up in advance.

Work towards a preliminary version of the care standard will take place on the basis of the medical articles on MSS and an overview of known treatments of the various symptoms. If possible, online questionnaires sent to all parents and those providing treatment will be used for this purpose.

The aim is that an initial version of the international care standard will be ready by 2011. As soon as the first version of the international care standard is completed this should also be made available online via the internet.



Professor Hennekam (above) and Dr. Shaw of the Institute of Child Health of University College London are undertaking research on MSS.

No	Objective	Desired result	Action to be taken by	Accomplished
3.1	Setting up Care Standard Working Group on the basis of expertise	Care Standard Working Group	Sonja	Sep 2009
3.2	Examination of existing care standards	Criteria for care standard	Sonja	Jan 2009
3.3	Online questionnaires sent to doctors, paramedics and others involved in treatment, as well as parents	Input for the care standard	Sonja	June 2010
3.4	First version of care standard to be made available online with editing/translation tool	First version of care standard available online	Henk-Willem	Dec 2010
3.5	Care standard to be regularly updated with new information	Latest version of care standard available online	Sonja	after 2010

Theme 4: Fundraising

Perhaps the most important objective is to raise funds for research. The MSS Research Foundation would very much like to be a charitable organization which can play an active role in facilitating groundbreaking research into MSS. A great deal of money will have to be collected for this over the coming years.

The MSS Research Foundation will work to raise funds mainly through two types of fundraising activities:

- 1) **Breathtaking campaigns:** By organizing “Breathtaking campaigns” money can be collected for the charity in ways which are fun. These breathtaking campaigns are a good way of encouraging people to organize personal sponsorship activities within their own networks to raise money for MSS research. This might include taking part in a sponsored New Year’s Day Dip, the Nijmegen Four Day Marches, etc.
- 2) **Finding friends:** The Foundation offers the option of becoming a Friend for a fixed regular donation. This group of friends will ensure a certain level of income every year. The goal is that this group of friends will also grow over the next couple of years. The board has set itself the goal of achieving 100 friends by 1 January 2010 and 200 friends by 1 January 2011.

In 2010 the board will organize an “MSS Family and Friends Day” as a way of involving the group of donors in all the activities. This event can be organized in combination with a breathtaking campaign in order to raise money at the same time. To make the event attractive to people, fun activities can be organized such as a wagon race, bath duck race, etc. These activities can also be used to raise money.

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With new studies it will generally always be determined whether or not co-financing can be found or whether there are any open national or European ‘calls’ for research funding.

No	Objective	Desired result	Action to be taken by	Accomplished
4.1	Finding friends	At least 100 “Friends of”	Board	Dec 2009
4.2	Breathtaking campaigns	Money for research	Board	Anytime
4.3	Organization of Family and Friends day with breathtaking activity	More involvement of friends and financial campaign	Wout	May 2010
4.4	Finding friends	At least 200 “Friends of”	Board	Dec 2010

Theme 5: International outreach

MSS has no boundaries. International outreach offers opportunities but also requires time and effort. The potential includes access to and exchange of information, support and resources. To try to achieve this the board has formulated the following goals:

- All relevant documentation (policy plan, budget and minutes of meetings) to be translated into English and made available on the website;
- The use of teleconferencing to enable parents to share knowledge internationally;
- Parents of children with MSS will automatically be granted special membership of the MSS Research Foundation, (still to be further defined);
- The organization of an MSS Family Event abroad to support the realization of the Foundation's objectives on other continents.

A particular goal, the feasibility of which will have to be investigated, is making aids and resources available to families in difficult circumstances. This could be for parents of a child with MSS in a third world country, or countries where certain aids are not available. This could eventually become one of the main goals of the organization.

The aim of the Foundation's board is not only to take useful steps, but also to take them in the right way. In the allocation of jobs and responsibilities to board members careful consideration is also given to each board member's affinity and professional background. When vacancies are to be filled, the necessary complementary experience and competences are also taken into account.

MSS has no boundaries.

No	Objective	Desired result	Action to be taken by	Accomplished
5.1	Accessibility of documents	Important documents translated and published on website	Wout	dec 2009
5.2	Teleconferencing	All parents in contact with one another and the board once a year	Board	Anytime
5.3	Special membership for parents	All parents special members	Sonja	jan 2010
5.4	Policy surrounding campaign for aids for young MSS patients in need	Helping families in difficult circumstances by providing aids	Wout	mei 2010
5.5	MSS Family Event abroad	Contact with other sufferers worldwide	Board	feb 2011

Contact details

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Lindsay from USA



Mattéo from France



'New Year's Dive in Scheveningen



Nina from
The Netherlands



Adriana from
The Netherlands



Ella from UK



Matthew from UK



Rafaëla from Brazil



Joas from
The Netherlands



Adam from UK