

ANNUAL REPORT 2009



MSS



RESEARCH
FOUNDATION



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www.marshallsmith.org
 Stichting MSS Research Foundation
 Altingstraat 120
 2593 SZ Den Haag
 Kamer van Koophandel 273.090.21
 RABOBANK 1403.51.159



1. Foreword

The highpoint of 2009 was the MSS Family Event! Never before have so many families with a child with the Marshall-Smith syndrome come together. Nine families from all over the world met each other in Valkenburg aan de Geul in the Netherlands.

This is the second annual report of the MSS (Marshall-Smith Syndrome) Research Foundation. The Marshall-Smith Syndrome (MSS) is a rare disease 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.

Progress has been made in 2009 in several areas: the contact between fellow-suffers was further developed; scientific research yielded its first results; various Dutch media devoted attention to children with MSS and their parents; and the new website has become pivotal in providing information.

It is responses like this which make it clear just how important it is that there is research, as well as contact between doctors and between parents. We have now enabled 28 families to contact one another. Nevertheless, the specific problem of the rarity of the syndrome still means that children and their families still often find themselves in an isolated position.

When we were given our son to take home with us we were told that we were taking him home to die. They told us that they did not expect him to survive and if he did, then he would only be able to lie on his back in a vegetative state. We are so happy that our son has exceeded that expectation and has quality of life. (Nathan is now 8 months old)

We hope you enjoy reading this annual report. Together with the children, parents, doctors, paramedics, donors, sponsors and everyone else involved, we also look forward to taking further steps in 2010 towards improving the position of people with MSS, increasing awareness among the general public and promoting further scientific research.

This annual report further provides the figures for 2009. The annual report with the complete financial report can be found on www.marshallsmith.org.

On behalf of the Board of the Marshall-Smith Syndrome Research Foundation (Stichting MSS),

The Hague, April 2010
 Drs. Froukelien Schiebaan-van der Mooren,
 Chair



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2. Objectives

The Marshall-Smith Syndrome Research Foundation was set up on 15 December 2007 by Gerhard Post, notary at law based in Ommen. The MSS Stichting has its seat in The Hague. The organization is registered in the trade register of The Hague Chamber of Commerce under number 27309021. The objectives of the organization are as follows:

To strengthen the position in Dutch 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).

3. What is MSS?

The Marshall-Smith Syndrome (MSS) is a very rare condition. Worldwide there is only a handful of children who suffer from it. In the Netherlands there are three children with MSS. Because of the serious respiratory problems and associated complications many children die shortly after birth or in early infancy. Life can be prolonged through aggressive treatment of the respiratory problems. The cause of MSS is not yet understood. There is still little useful information available on this serious syndrome. It is not an easy diagnosis to make. The diagnosis has to be made by a clinical geneticist based on various tests. An X-ray of the hand from which the more advanced bone age can be seen, will often be a typical feature. Because there is no quick diagnosis, children with MSS are at risk of failing to receive adequate treatment and may suffer permanent damage to their health as a result..

Characteristics of Marshall-Smith syndrome

RESPIRATION & HEARING

*Significant respiratory problems resulting from narrowing in nose and throat region
Deafness, ear disorders*

ACCELERATED AGING OF BONES

*A higher age of the bones
Unexplained bone fractures*

GROWTH & NUTRITION

*Difficulties with growth and nutrition
Lagging mental and physical development*

FACIAL CHARACTERISTICS

Prominent forehead, shallow eye sockets, retracted chin, sunken nose bridge, the white of the eyes showing a bleu tint.

4. MSS Research

In taking part in scientific research the aims of the MSS Research Foundation are twofold: First: what is the development of the children and what problems do they have? Second: how does a child develop MSS and what causes the problems?

In 2009 the MSS Research Foundation and the Institute of Child Health of University College London worked together on a scientific study of MSS. The initial results of the study were presented at the Family Event. There is now more insight into the clinical presentation of MSS. Seven people were fully examined and data from five other children was also used for this. It would, of course, be

very helpful if more children could be examined. From the first DNA results it appears that there are several places where a genetic origin for the syndrome might be found. Further analysis of chromosome material, mainly from the parents, would be necessary for this. Indications of a collagen disorder were also found in the urine of children with MSS.

Further work was done in 2009 on investigating the chromosome abnormalities. The question is whether these chromosome abnormalities are inherited from one of the parents of the children or whether they occur sporadically (de novo). De novo chromosome changes are the most probable cause of the Marshall-Smith Syndrome. The results of this study are expected in 2010.



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

5. Contact between fellow-sufferers

5.1. Contact

What began in February 2007 in the Netherlands as contact between two Dutch parents with a child with Marshall-Smith Syndrome, grew in 2009 to become a real MSS community in which (as of March 2010) 28 families from all over the world have contact with one another. This is 10 more families than in 2007-2008.

Children with MSS are known of in the Netherlands (3), Belgium (1), Norway (1), Germany (1), Canada (1), USA (8), UK (4), Israel (1), France (3), Brazil (1), Spain (1), Mexico (1), Paraguay (1) and Croatia (1).

In most cases the contact was first made through the MSS website. The words of parents who find the website are still very moving; they are so happy to be able to make contact with other parents who also have a child with MSS. They are also very pleased to be able to find information on MSS and are more than willing to take part in the study by providing information.

5.2. MSS Family Event

The highpoint of 2009 was undoubtedly the MSS Family Event which was held in the Ronald McDonald children's holiday village (kindervallei) in Valkenburg aan de Geul. For four days the nine families enjoyed being together, a full program, eating together, the enchanting surroundings and the hospitality of the staff of the children's holiday village. Supported by volunteers who were there to lend a hand or help with translation, it was a wonderful gathering in which the weather was also kind to us.

Friday was devoted to arriving, meeting everyone and an enjoyable meal. On Saturday morning the fathers took part in an adventure hike and Rafaela from Brazil was given a new walking frame, which she has used this year to learn to walk.

Saturday afternoon was devoted to the study: Professor Raoul Hennekam, Dr. Adam Shaw and Dr. Inge van Balkom gave a detailed presentation on how the study is progressing. Among the audience were friends and family of the Dutch families and some of the event sponsors. Saturday evening we all watched the 'één vandaag' TV broadcast in which Henk-Willem Laan was followed during the preparations in the week just before the Family Event.



On Sunday we all enjoyed singing along with the Rainbow Tree and in the afternoon - while the mothers were relaxing at the beauty spa - the children could enjoy the mobile children's petting farm outside. On Monday everyone made their way home again, feeling happy; with some touching farewells.

One of the main aims of the event - promoting contact between families - was more than successfully achieved during the weekend. The families got on extremely well together and both the parents and the children enjoyed being together.

The parents were particularly amazed at the remarkable and sympathetic way in which the doctors who were present were able to add to their research into MSS during the few days of the family event. Between the various activities those children who had not yet been seen by the doctors were examined and the parents had time to ask all their questions. After the event every family was sent a video and a photo DVD so that they can look back on the weekend and enjoy the memories of the event whenever they want.

From the positive feedback we received from many of them, we can definitely say that the MSS Family Event was greatly appreciated by all the families. The MSS Research Foundation intends to organize another MSS Family Event in 2011 and hopes to be able to welcome even more families then.

5.3. Patient organizations

The MSS Research Foundation is a member of VSOP (an association of 57 patient organizations) and in December 2009 asked VSOP to start work on drawing up a care standard for MSS. The MSS Research Foundation is a member of the European organization for rare diseases, Eurordis. A board member attended the Membership Meeting in Athens in 2009 where, besides making many contacts, a great deal of information relating to the website was obtained.

6. Fundraising

Fundraising enables the Foundation to cover the cost of its activities. This task is therefore vital to achieving the goals of the Foundation. The following fundraising activities took place in 2009:



6.1. Letter via parent network

Potential donors are approached through networking. The existing network is specifically drawn on in the quest to find new donors. This has resulted in many single donations and 'friends' of the Foundation.

6.2. Rare Diseases Fund

The aim of the Rare Diseases Fund (ZZF) is to support and foster activities related to rare diseases. The MSS Research Foundation maintains contact with ZZF and was pleased to receive a grant of EUR 6,000 for the second part of the study into the cause of MSS.

6.3. Private fundraising activities

Private fundraising activities are particularly important to our organization. Private fundraising not only makes a financial contribution to the activities of the Foundation, but it also gives a huge boost to morale to see other people working to support the Foundation. An example of this in 2009 was the New Year's dip in the North Sea at Scheveningen in freezing conditions.

6.4. Friends of the MSS Research Foundation

Friends (donors) of the MSS Research Foundation are people who have committed to make an annual donation of at least EUR 25. The number of friends grew in 2009 from 19 to 51 people. The regular income generated by the group of friends is very important in financing recurring expenses. The board has set itself the goal of regularly informing the friends about the activities of the MSS Research Foundation and to keep them involved in the work of the Foundation.

6.5. PGO Fund grant

The PGO Fund extends grants to national patient organizations, organizations for the disabled and associations for the elderly in the Netherlands. The Fund makes an annual institutional grant to the MSS Research Foundation which constitutes a major support for the activities of the Foundation. This helps to cover the cost of activities in the areas of providing information, promoting the interests of patients, contact between fellow-sufferers and enhancing the professionalism of the organization.



7. Information

7.1. General

Providing information to the general public is one of the Foundation's core tasks. Given the limited financial resources available, choices have to be made. Activities organized by Foundation are used to inform the media who are then invited for an interview or a visit. This has raised the general level of awareness about MSS. The following took place in this context in 2009:

7.2 Website

A large part of the information for the general public is provided on the interactive website: www.marshall-smith.org. In 2009 we therefore devoted particular attention to making the MSS website more professional. For example, towards the end of 2009 a new design was implemented to bring the website into line with the rest of our communication and to make the website a recognizable part of the MSS information campaign. We have also made sure that the website can be found more easily on Google and that it can be viewed in different languages. The Family Circle has also been given more prominence on the website, from which it can easily be seen how rare MSS is. The MSS world map shows where the MSS kids all live, by clicking on a balloon on the map the profile of one of the children can be seen.

When families register we already gather a lot of information about the young patient, following which there is almost always fairly frequent contact in which a lot of information is exchanged. Parents have a real need to know more about MSS.

Besides information, sharing knowledge and experience among parents is also becoming an increasingly important part of the website. An initial version of an MSS wiki online has been created to enable parents to exchange information about MSS. In this way we bring families closer together and the website is becoming a source of useful information. Professor Raoul Hennekam and Dr. Adam Shaw provided the initial input for this. They used the comments made by the parents in an as yet unpublished general scientific article about MSS. The MSS wiki will be further developed over the coming year. We will also continue to focus on making the website as accessible as possible and thus support the goal of providing information on MSS.

TV

- **January 1** *Jeugdjournaal* (children's news) item on MSS New Year's dip 2009
- **February 28** TV L1 on the MSS Family Event
- **February 28** *The Week of... Henk-Willem Laan* (the week just before the MSS Family Event)
- **April 8** 'Hart van Nederland' national news program on Nina and the rare diseases day 2009
- **October 7** 'Hello Goodbye' on Rafaéla arriving at Amsterdam Schiphol airport from Brazil.

NEWSPAPERS

- **February 24** *De Telegraaf* on Adriana and rare diseases
- **February 25** Five questions put to Henk-Willem Laan in the AD national daily
- **March 2** An article in *Trouw* national daily about the MSS Family Event
- **March 2** Hope for children with the Marshall-Smith Syndrome (article appeared in various media)

RADIO

- **February 28** Radio L1 on the MSS Family Event
- **February 28** Villa VPRO on the MSS Family Event

The website also plays an important role in providing information to the general public.

8. Financial report

8.1 Balance

Balance sheet as per 31 December (after appropriation of result)

ASSETS	31-12-2009	31-12-2008
	€	€
Receivables		
Interest	1.844	795
	-----	-----
Cash	80.041	45.931
	-----	-----
Total assets	81.885	46.726
EQUITY AND LIABILITIES		
	€	€
Reserves		
<i>Reserves</i>		
Designated reserve	75.389	36.690
Equalization reserve	4.865	-
	80.254	36.690
	-----	-----
Provisions		
Provision for fellow-sufferer contact	1.595	-
	-----	-----
Liabilities (short-term)		
Subsidy liabilities		10.000
Other liabilities	36	36
	36	10.036
	-----	-----
Total equity and liabilities	81.885	46.726

8.2 Statement of income and expenditure

	Budget 2010	Realization 2009	Budget 2009
	€	€	€
INCOME			
Income own fundraising	80.000	37.682	24.000
Subsidies	43.500	31.900	30.250
Other income	1.000	1.110	1.000
Total income	124.500	70.692	55.250
EXPENDITURE			
Expenditure to objectives			
Research	76.250	-610	20.400
Standard of care	21.000 *	51 *	5.000
Fellow-sufferer contact	9.995 *	9.962 *	10.250
Information	7.500 *	11.717 *	10.000
Protection of interests	1.000 *	710 *	1.000
	115.745	21.830	46.650
Expenditure fundraising			
Cost for own fundraising	400	56	400
Management and administration			
Cost for control and administration	4.500 *	5.242 *	6.500
Total expenditure	120.645	27.128	53.550
Surplus	3.855	43.564	1.700

*) Subsidized with a dedicated grant fund from the Dutch Fonds PGO.

The full financial statements can be found at www.marshallsmith.org.

The financial statements are reviewed by With nonprofit Accountancy.

9. Board

9.1. Board members

In 2009 Liaan Jansen stepped down and we were pleased to welcome Hendrik de Wit as a board member. The following people currently sit on the board of the Marshall-Smith Syndrome Research Foundation:

Froukelien Schiebaan - van der Mooren, GP (1978) - Chairperson

Froukelien works as a locum GP in The Hague region. She completed her general practicespecialism at Leiden University Medical Centre (LUMC) in 2007. She studied medicine at the University of Maastricht (UM) followed by an optional internship at McGill University Montreal, Canada and an extra-curricular internship in a small rural hospital in Pakistan. For her general practice specialism she worked for two years in a hospital as a junior doctor in the pediatrics and surgery departments. In the medical field she has been active as secretary to the board of the National Organization of GPs in Training (LOVAH) and as an advisory board member of the GP and Nursing Home physicians Registration Committee (HVRC) and the Board for general practitioners, nursing home physicians and physicians for the mentally disabled (CHVG).



drs. Froukelien Schiebaan-van der Mooren

Henk-Willem Laan (1975) - secretary/treasurer

Henk-Willem works as a housing and finance manager and teaches economics at a school complex in Gouda. He is currently coordinating the construction of the two new buildings. He studied tax law and economics at the University of Maastricht from 1994 to 1998. Thereafter he worked as a tax consultant for eight years. In recent years Henk-Willem has worked for various civil and social organizations. He is currently a board member of the development organization Adopt a Goat (www.adopteereengeit.nl), member of the viewers' panel of the EO broadcasting association and member of the Management Committee of the Ichthuskerk church in The Hague. His son Joas (2006) has the Marshall-Smith Syndrome.



drs. Henk-Willem Laan

Sonja Bracke (1962)

Sonja took a higher professional degree course in nursing from 1986 - 1990 and worked for several years thereafter as a district nurse. Since 1993 she has been a manager at the Vitaal Groep. Vitaal is active in lending aids, as a home care shop and in arranging supplies for local councils under the Social Support Act (WMO 2006) in the context of the personal budget and other grants made to customers of various health insurers. Vitaal is also active intramurally in the context of the Exceptional Medical Expenses Act (AWBZ) and as a supplier of institutional facilities. At present Vitaal has four home care shops and a large showroom close to the centre of Utrecht. Sonja's daughter Nina (13 years) has the Marshall-Smith Syndrome. Since 2004 Sonja has been a member of the advisory board to the Mytyschool Ariane de Ranitz. Sonja's daughter Nina (1995) has the Marshall-Smith Syndrome.



Sonja Bracke

Wout T. Koelewijn (1976)

Wout studied International Business Administration at Maastricht. Before, during and after his degree course, he gained a lot of experience abroad. After graduating he started working for ExxonMobil in Brussels where he became expert in purchasing. With the move to a consultancy in 2004 he became acquainted with the world of healthcare and became fascinated by the major challenges facing the sector. Working closely with doctors, nurses and other parties involved, he has introduced various purchasing and change processes among many care providers. Wout now works as a senior consultant for Significant where he advises on the topic of 'Market forces in the Care Sector' for central government, healthcare insurers and care providers.

*drs. Wout T. Koelewijn***Hendrik de Wit (1979)**

Hendrik has been active in the field of marketing communications since 2000. Following his degree in Mathematics, he spent 7 years working for ABN AMRO bank in Corporate Communications and other areas, alongside which he also took a post-graduate course in Policy, Communication & Organization at Amsterdam Free University. For a period of roughly three years Hendrik worked as Online Marketing Manager at the world's biggest pharmaceutical company where he gained wideranging experience in interactive communication with patients, doctors and nursing staff. Hendrik has now returned to the financial sector where as part of the Global Marketing team of the Intertrust Group, he is responsible for the online strategy for both internal and external target groups.

*Hendrik de Wit***9.2. Tasks and activities**

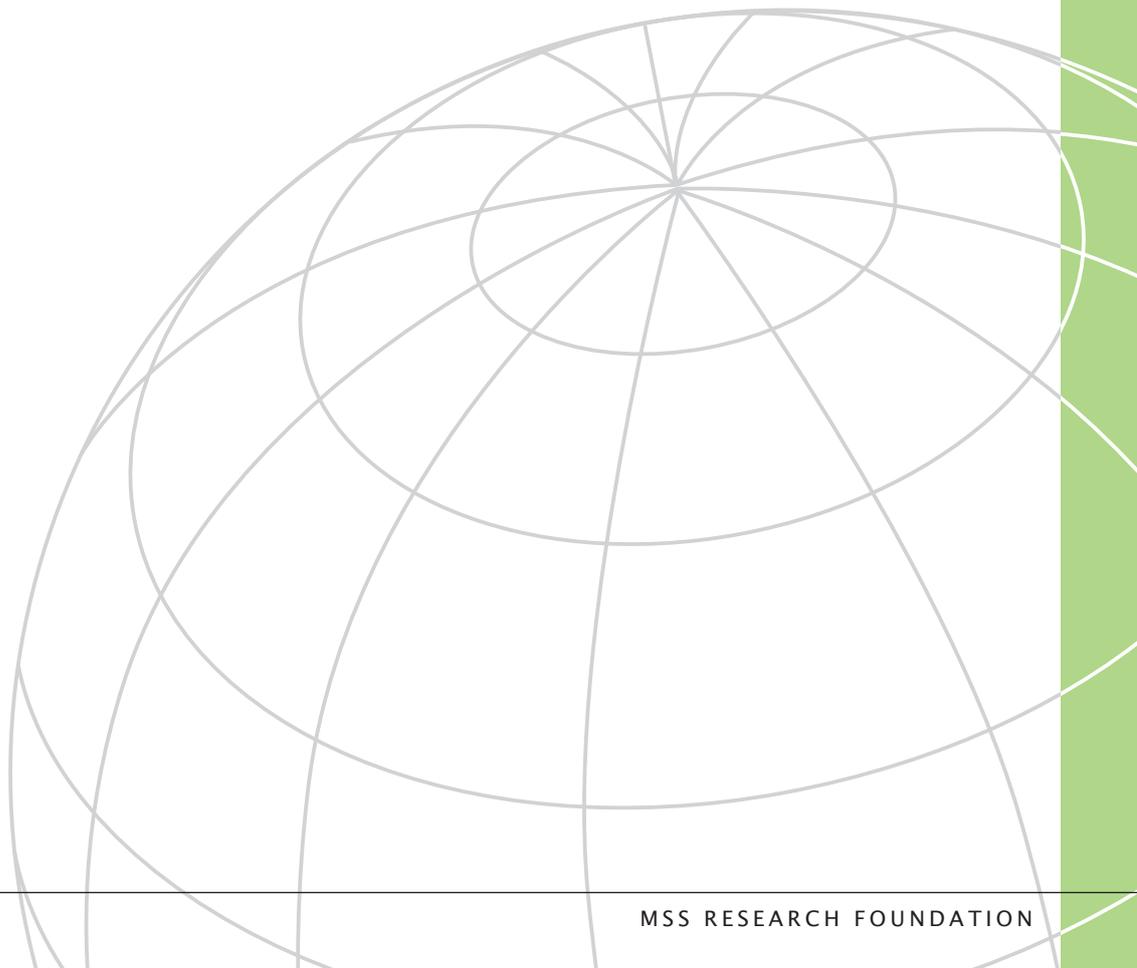
The board is responsible for managing the MSS Research Foundation. It is the board's main ambition to ensure that the goals of the Foundation are met with excellence. In 2009 the board drew up a policy plan which includes a number of longer-term objectives. The budget is set on the basis of these goals. The board oversees the proper use of the funds in line with the policy plan and the budget, and is also accountable for this.

9.3. Board meetings

In 2009 the board met four times at various locations throughout the Netherlands. One of these meetings was devoted to defining the policy for 2009 and the longer term goals. In addition, some board members also met regularly in connection with the Family Event 2009.



***Your rare
chance to have a
global fan-club***





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