ANNUAL REPORT 2007/2008





RESEARCH FOUNDATION





MSS

ASTER SKA



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1. Foreword

2008 was a special year for the MSS Foundation. A good time to look back. This is the first annual report in the history of the MSS (Marshall-Smith Syndrome) Research Foundation. Marshall-Smith Syndrome (hereafter MSS) 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.

A great deal has happened since the organization was set up on 15 December 2007. Other sufferers around the world have made contact. Scientific research has "I thought I was the only one"

Parent of a child with MSS

begun. Various Dutch media have devoted attention to children with MSS and their parents. The website played a pivotal role in all of this.

"I thought I was the only one".

We are pleased to be able to say that for the children and parents of those with MSS, this no longer has to be true. At the end of 2008 we now know of 20 children worldwide with MSS. Nevertheless, the specific problem of the rarity of the syndrome puts children and their families in a somewhat isolated position.

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 2009 towards improving the position of people with MSS, increasing awareness among the general public and promoting further scientific research.

This annual report also includes the annual accounts since the organization was set up, to 31 December 2008. The annual report and accounts have been drawn up as far as possible in accordance with the Dutch Accountants' Guideline 650 for Fundraising Organizations. The MSS Foundation thus meets almost all the criteria for the Dutch Central Bureau for Fundraising certificate for small charitable organizations. It will be decided at a future date whether or not to apply for this certificate.

On behalf of the Board of the Marshall-Smith Syndrome Research Foundation Froukelien Schiebaan-van der Mooren Chairperson

The Hague, May 2009







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 society of people with the Marshall-Smith Syndrome (hereafter referred to as MSS);
- to provide the general public with information on MSS;
- \cdot 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 are 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.

Supporting testimony

"Very limited information and having to wait too long for the diagnosis of MSS to be confirmed. Not knowing what can be done about it. A lack of knowledge amongst medical specialists but most important having to live in uncertainty. You have my sympathy. I am trying, wherever there is an opportunity, to attract attention for the issue of rare diseases. I am wishing you in the coming period of time



success and much strength." Erica Terpstra

Former freestyle swimming champion, former Minister of Health, Welfare and Sport at present chairperson of the Dutch National Olympic Committee.

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

Following the first round of fundraising, research into MSS could begin in 2008. In a joint campaign the MSS Research Foundation and the Institute of Child Health of University College London set up a wide ranging study into MSS. The aim of the research is to collect medical information from as many MSS children in the world as possible. Less than 50 cases have been described in the literature. With this information the MSS Research Foundation hopes that eventually it will be possible to answer questions such as: what is the development of the children and what problems do they have? How does a child develop MSS and what causes the problems?

Seven people were examined in the Great Ormond Street Hospital for Children in London by the research group of Professor R. Hennekam and Dr. A. Shaw of the UCL Institute of Child Health. These were children from the Netherlands and Great Britain. Information has also been collected from five other people. Six people have also been studied behaviourally. The DNA tests were analysed in more detail. The results of these initial tests were presented and explained at the Family Event early in 2009.

At the end of 2008 the research group applied for further research funding. The research protocol sets out the goals of the follow-up study. The first line of research is to examine whether chromosome defects are inherited from one of the parents of the children or whether they rarely occur (de novo). De novo chromosome changes are the most probable cause of the Marshall-Smith Syndrome. Besides the seven people examined, the research group also has DNA samples from six other Marshall-Smith Syndrome patients throughout the world. The necessary funding for this research was pledged by the MSS Research Foundation.



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

5. Patient contact

5.1. Patient contact and website

Since February 2007 the three Dutch families that each have a child with MSS have had contact with one another. The contact was made by Internet at the time. With the launch of the website *www.marshallsmith.org* the organization and the MSS Family Circle, as it is known, have become visible and accessible to everyone searching on the Internet for MSS. Apart from the three families in the Netherlands, this has led to 17 (sometimes brief) contacts with families in the following countries: Norway (1), USA (6), UK (4) Israel (1), France (2) Brazil (2) and Paraguay (1). Almost every parent who has made contact through the website is pleased with the Dutch initiative to foster contact between the families and research into MSS.

The website also has an important part to play in the research into MSS; contact with the parents of MSS children means that they can be put in touch with the doctors in London who are carrying out the research. To maintain contact between the families and provide new visitors with useful information on what the organization is doing, it is important that a properly functioning forum is set up on the website in 2009.

5.2. MSS Family Event

The first MSS Family Event was held at the end of February 2009 in the Ronald McDonald Children's Village in Valkenburg, the Netherlands. The purpose of the meeting was to foster contact between the families of patients all over the world. During the meeting the initial results of the research on MSS were announced by Professor Hennekam, Dr. A. Shaw and Dr. I. Van Balkom.

5.3. Patient organizations

Since 2008 the MSS Foundation has become a member of VSOP. VSOP is an association of 57 patient organizations for diseases with a hereditary component. In 2009 an MSS Foundation board member will attend a Eurordis meeting to represent the interests of all MSS families. Eurodis is the European organization for rare diseases.



6. Fundraising

6.1. Letter sent by parent network

The first fundraising campaign began towards the end of 2007 with a personal letter sent within the networks of the Dutch parents. The aim of this campaign was to raise $\leq 10,000-15,000$ for the initial research into MSS. This campaign was very successful. Well over the target amount was collected.

6.2. Rare Diseases Fund

Contact was made with the Rare Diseases Fund (Zeldzame Ziekten Fonds (ZZF) in early 2008. The ZZF very much wanted to contribute to the initial research on MSS. The Dutch parents received a cheque for \in 7,000 from the chairman, Charles Ruijgrok, on Rare Disease Day. The second stage of the research (requiring £8,400 or approx. \in 10,000) was also supported by the ZZF with a grant of \in 6,000.

6.3. Private fundraising activities

Several private individuals also organized various activities to raise money for the MSS Foundation:

- On Whit Monday 2008 Anouk and Sicco de Vries together with Hinke and Matthijs cycled the route of the Eleven Cities Skating Marathon (*Elfstedentocht*). Their sponsored bike ride raise the incredible sum of €1,435!
- Sonja Peters, a colleague of the Dutch mother Ana Godani arranged to be sponsored as she walked the International Four Day Marches Nijmegen (*De Nijmeegse Vierdaagse*) from Tuesday 15th to Friday 18th July 2008.
- On New Year's Day (1 January) 2009 15 brave swimmers were sponsored to dive into the cold North Sea to raise money for the MSS Foundation.

The sponsorship funds raised by Sonja Peters and the New Year's swim were presented during the MSS Family Event.







6.4. Friends of the MSS Foundation

Mid-2008 the Board decided that a regular group of donors was necessary to safeguard the future of the MSS Foundation. It is therefore possible to become a "Friend of the MSS Foundation" by donating a fixed amount per month, quarter or year. The minimum annual donation is \in 25.

6.5. PGO Fund grant

The PGO Fonds (Ministry of Health agency which provides grants to organizations for patients, the disabled and the elderly) pointed out to the MSS Foundation that it could be eligible for a patient organization institutional grant. At the end of 2008 this grant was applied for, further to which it will be awarded in 2009.

Adriana has MSS

Hi, I am Adriana and I am almost 6 years of age. I am a nice and happy girl. Really my life is not all that happy because I have the Marshall-Smith syndrome. My bones are



twice as old as I am and I have little resistance and I often am therefore not well. I often have earaches also. That hurts. Sleeping the whole night through I find already difficult but it is even harder when I have an earache. I am not able to speak yet and I am busy practicing walking on my own. I have glasses with strong lenses and which I refuse to wear. I really appreciate if you will help me and all the other children with MSS.

Regards, Adriana.

The MSS Foundation is required to spend the grant on activities related to information, contact between fellow-sufferers and promoting the interests of patients or on activities which are intended to enhance the professionalism of

the organization. The grant will be used to organize the MSS Family Event (contact between fellow-sufferers) and for professionalization, including this annual report. After the grant is awarded, the Board will develop policy in 2009 on how the remainder of the funds shall be spent.

7. Information

Providing information to the general public is one of the main goals of the organization. The major diseases receive a lot of media attention, which is generally not the case with rare diseases.

Nevertheless it is important that wide-ranging communication on the rare MSS takes place. Insurance company and care facility workers, public officials, healthcare professionals, doctors and nurses can all come into contact with MSS in the course of their work. Particularly as this group is so varied, the information provided for the wider public also has to be comprehensive.

This approach has been successful in the Netherlands, as shown by the reporting described below. Partly through cooperation with the Rare Diseases Fund, a great deal of media attention was devoted to

Items op TV

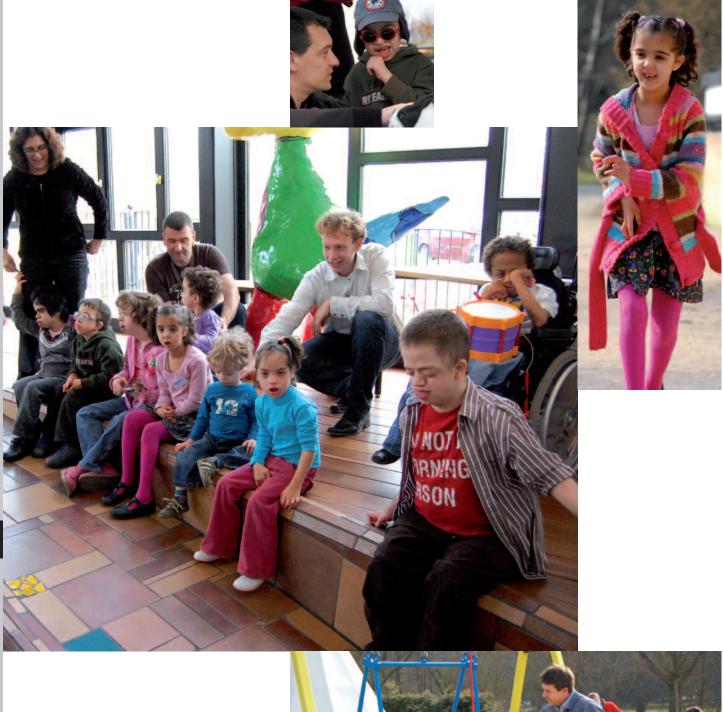
- 25 February 2008 Netwerk current affairs programme on rare diseases
- 29 February 2008 RTL News on rare diseases day
- 29 February 2008 Film made by the Rare Diseases Fund
- 21 March 2008 Netwerk current affairs programme on Joas and Nina and the trip to London for the study
- 20 November 2008 Avro broadcasting company series on children's hospitals with a programme on Joas.

Items in the press

- 27 February 2008 Nederlands Dagblad daily on Joas and rare diseases
- 28 February 2008 Algemeen Dagblad daily Leven (magazine) with an article Zorgen over Joas (Caring for Joas); on rare diseases day
- 29 February 2008 Submitted article on opinion page of Trouw daily on attention given to rare diseases
- February 2008 De Gelderlander regional paper with article on Nina
- 1 March 2008 BN/DeStem regional daily on rare diseases
- 27 March 2008 Noordhollands Dagblad regional paper with article on Adriana

Items on the radio

• 29 February 2008 Radio 1 Kassa (current affairs programme) on rare diseases day







MSS in the Netherlands in connection with the first European Rare Diseases Day on 29 February 2008. Joas and his parents, and others, quickly became the 'face' of MSS.

The Board believes in the importance of awareness of the syndrome. A rare disorder is already bad enough. Unfamiliarity with it makes it worse. This year we have managed to do something about this lack of familiarity. Raising awareness in other countries is an important goal for the coming years.



Joas has MSS

Hi, I am Joas. I was born in July 2006. Straight after my birth, I developed breathing problems for which I needed to be brought to a hospital by ambulance. I stayed in



the hospital for four weeks before being able to return home. I have had to go to hospital on many occasions for all kinds of examinations. I have also had an operation on both my eyes as I have glaucoma (high eye-pressure).

My largest problem is that I have narrowed channels of nose, throat and ears. As a result it is often difficult for me to breath, especially when I have a cold. Often I have an earache and my bones become older rapidly. Despite my problems I am always happy and have a lot of energy.

Nina has MSS

Hello, I am Nina. I am a happy girl of nearly 13 years old. have a lot of energy and am always in for an adventure. I have been through a lot

in my short life. I am not fond of doctors but they have helped me. I sleep at night with a small tube in my nose (nasopharyngeal tube) as when I sleep I cannot breath properly.



When I was a baby, for a long time I could not eat by myself. I am happy I can now. I am attending a special school and I have learnt a lot, especially better walking, speaking and playing with others. I love a party; dancing, jumping, singing, but I do have to take good care that I will not fall. I have a narrowed channel in my spinal cord and therefore falling would not be good for me.

8. Financial report

8.1. Key figures

	2008
Spending ratio relative to income:	
Spent on objective / total revenue	21.6%
Spending ratio relative to expenditure:	
Spent on objective / total expenditure	92.7%
Cost percentage for own fundraising activities:	
Cost of own fundraising activities / income from own fund raising	0.1%

8.2. 2009 Budget

Mid-2009 the Board will publish a policy plan for the coming years. The annex includes a provisional budget for 2009. This budget will be updated when the policy plan is finalized.

8.3. Financial policy

In 2009 the MSS Foundation will develop policy on the extent and purpose of reserves and funds and an investment policy will be drawn up.

8.4 Annual Accounts

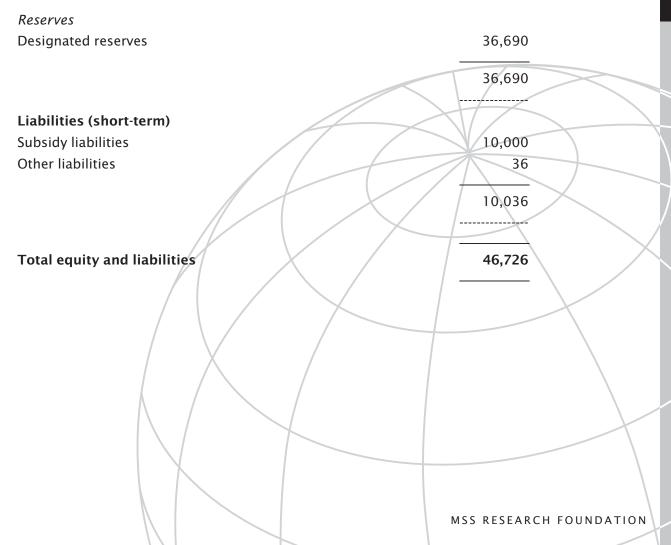
8.4.1 Balance sheet as per 31 december

(after appropriation of result)

ASSETS	31-12-2008
	€
Receivables	
Interest	795
Cash (8.4.7)	45,931
Total assets	46,726

EQUITY	31-12-2008
EQUITY	31-12-2008

Reserves (8.4.7)



8.4.2 Statement of income and expenditure

	Realisation 2007/200
INCOME	
	€
Income from own fundraising	47,065
Other income	795
Total income	47,860
EXPENDITURE	
Expendifiling to objectives	
	10,358
Research	10,358
Research Expenditure fundraising	64
Expenditure to objectives Research Expenditure fundraising Cost for own fundraising Management and administration	64
Research Expenditure fundraising Cost for own fundraising Management and administration	64
Research Expenditure fundraising Cost for own fundraising	10,358 64 748 11,170

8.4.3. Appropriation of result

Realisatior	2007/2008

Appropriation to reserves

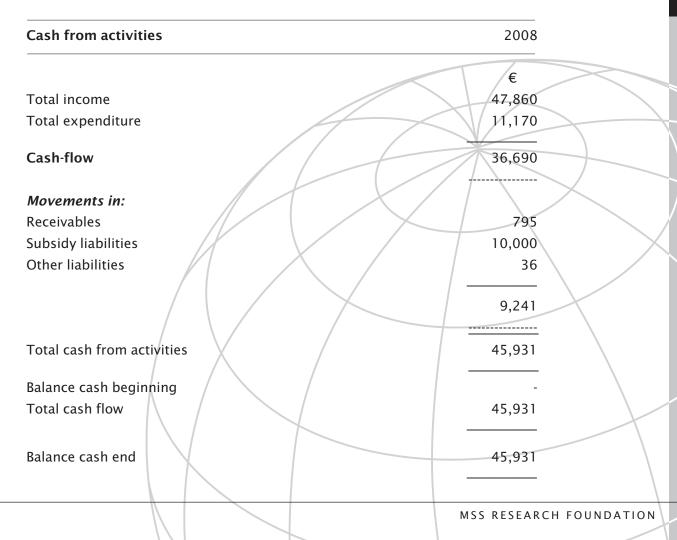
Designated reserves research

36,690

Expenditure	Research	Cost for own fund rasing	Cost manage- ment and administration	Total
	€	€	€	€
Research	10,000	-	-	10,000
Travel expenses research	358	-	-	358
Bank charges	-	-	178	178
Legal expenses	-	-	570	570
Promotional expenses	-	64	-	64
Total expenditure	10,358	64	748	11,170
Target spending ratio of income:Expenditure to objectives / total income21.6				21.6%
Target spending ratio of expenditure: Expenditure to objectieves / total expenditure				92.7%
Cost own fundraising ratio: Cost own fundraising / income from own fundraising			0.1%	

8.4.4. Overview of expenditure 2008

8.4.5 Cash flow statement



8.4.6 Principles of valuation and presentation

General principles

The annual accounts are compiled based on Guideline 650 for Fundraising organizations. Amounts are stated in Euro's.

Comparison

The first financial year started at 15 December 2007 and ended at 31 December 2008.

General principles of valuation

Assets, equity and liabilities are valued at fair value.

Receivables, cash and liabilities

Receivables, cash and liabilities are valued at fair value. Liabilities for subsidy are taken based on the decision made by the board, which has been communicated to the receiver of the subsidy.

Reserves and funds

Reserves concerns money's which are free disposable for the Foundation. The Board is able to appropriate designated reserves for a specific target.

Funds must be spent for the objective for which it has been given to the Foundation. The funds represented at the balance sheet represents the unspent part of these money's.

General principles of determination of results

The result is determined as the balance of net income and all related expenses and other expenditure attributable to the reporting year, in accordance with the valuation principles referred to above. Income is recognized in the year in which the services were provided. Losses originating prior to yearend are recognized as they can be predicted.

8.4.7 Notes to the balance sheet

Cash	2008
	€
Bank current account	12,931
Bank savings account	33,000
	45,931
Cash is free disposable.	
EQUITY	
Reserves	
Designated reserve	
Balance as at 1 January	-
Appropriation of result	36,690
Balance as at 31 December	36,690
The amount in this reserve is designated to Research exp	

In May 2009 the Foundation has been granted a subsidy for professinalising its operations. One of the actions out of this grant was to review the annual report by an auditor. The expenses of this review are taken into account in 2009 due to the fact that the grant was given in 2009.

8.4.8 Notes to the statement of income and expenditure

Realisation 2008

INCOME		
Income from own	fundraising	
Donations		36,865
Designated funds		10,200
		47,065

8.5. Accountant's review report



Stichting MSS Research Foundation Althingstraat 120 2593 SZ DEN HAAG



Introduction

We have reviewed the financial statements of Stichting MSS Research Foundation, Den Haag, for the year 2007/2008 (as set out on pages 14 to 19). These financial statements are the responsibility of the foundation's management. Our responsibility is to issue a report on these financial statements based on our review.

Scope

We conducted our review in accordance with Dutch law, including Standard 2400 'Engagements to review financial statements'. This law requires that we plan and perform the review to obtain moderate assurance about whether the financial statements are free of material misstatement. A review is limited primarily to inquiries of company personnel and analytical procedures applied to financial data and therefore provides less assurance than an audit. We have not performed an audit and, accordingly, we do not express an audit opinion.

Conclusion

Based on our review, nothing has come to our attention that causes us to believe that the accompanying financial statements do not give a true and fair view of the financial position of Stichting MSS Research Foundation as at 31 December 2008 and of its result for the year 2007/2008 in accordance with Part 9 of Book 2 of the Netherlands Civil Code.

Sliedrecht, 26 June 2009

WITH ACCOUNTANCY B.V.

J.J. van Ginkel RA

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 WITh een initiatief van Lansigt en Hak & Baak heeft vestigingen in Alphen aan den Rijn, Gouda en Sliedrecht

9. Board

9.1. Board members

The 'founding fathers' of the MSS Foundation are Gert de Jong and Gerrit Laan. The expertise that they brought to setting up the organization was invaluable. Both gentlemen stepped down as board members in October 2008. The Board of the Marshall-Smith Syndrome Research Foundation currently comprises the following people:

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

Froukelien works as a locum GP in The Hague region. She completed her general practice specialism 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 paediatrics 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).



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.



Henk-Willem Laan

Liaan Jansen (1962)

Liaan works as manager of the Ronal McDonald house in The Hague. She worked for Transavia Airlines for 10 years, initially as a stewardess and thereafter at the head office in the tax-free goods purchasing department and various departments relating to flight advice. She is currently occupied with the planning and construction of a new Ronald McDonald House on a new site in The Hague (Leyweg) further to the merger of the Juliana Kinderziekenhuis (children's hospital) with the HagaZiekenhuis hospital group. She currently sits on various advisory committees of the Ronald McDonald Children's Fund. Liaan has a passion for many sports, including running and racing cycling, as well as sailing, mountain hiking and skiing.



Liaan Jansen

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.

X

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.



Wout T. Koelewijn

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 organization are met with excellence. In June/July 2009 the Board will draw up a long-term policy plan for this purpose. On this basis a budget covering several years will be drawn up and the budget for 2009 will be finalized. The Board shall oversee the proper use of the funds in line with the policy plan and the budget, and shall be held accountable for this too.

9.3. Board meetings

Since the founding of the organization to 31 December 2008 the Board has met four times at various locations throughout the Netherlands. In addition, some members of the Board met regularly in the context of the Family Event 2009.

Your rare chance to have a global fan-club.





Contact details

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