MSS

MARSHALL-SMITH SYNDROME
RESEARCH FOUNDATION

Marshall-Smith Syndrome

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Preface

The Marshall-Smith Research Foundation (MSSRF)[1] has an important mission based on three objectives; reinforcing the position of people suffering from the Marshall-Smith Syndrome (MSS), financing and stimulating scientific research on MSS and providing information and advice on this very rare syndrome to a broad and international public. Since 2007, the foundation has worked intensively towards realizing the above-mentioned goals together with parents, doctors and other professionals.

Right from the start, the MSSRF has built up a good relationship with many parents whose child suffers from MSS. An international network has developed consisting of parents who support each other and the MSSRF. The MSSRF is now the most important partner in scientific research and because of its valuable experience, the MSSRF is an important source of information regarding the syndrome and the need for care. The foundation considers itself as the party who, together with other relevant parties, reports on the quality of the care and the life of patients suffering from MSS, and improves the coordination of the care offered. Developing a care standard for MSS meets the objectives of the foundation. It is an obvious step in the attempt to reinforce the position of the patients and to give information and advice. The aim of the care standard is to improve the information supply and the quality of the care for MSS patients and their parents/carers.

Why a Care standard for the Marshall-Smith Syndrome?

This Care standard is based on five principles that describe the intentions and the goals of the Care standard. These principles are:

1) To report on all knowledge and information on MSS. This care standard describes the syndrome and highlights the problems arising in both acute and chronic settings.
2) To reinforce the interdependence between the patients with MSS, the symptoms and the treatment possibilities and to have a holistic view of the consequences of the syndrome and management.
3) To describe the minimum standards of good care for MSS patients, from the patient's perspective.
4) To support self-management with an active role for the parents.
5) To initiate comprehensive coordinated multidisciplinary care.

MSS is a very rare syndrome with very serious consequences for the children and their parents. Because there is not much information on the syndrome and its characteristics and not much has been published worldwide, it is not clear what is meant by good care for children and young adults with MSS.

The MSSRF has initiated a project, together with the Dutch National Alliance (VSOP) called "Care standard for MSS" as part of the project "Care standards for rare disorders: the patient in the

1 www.marshallsmith.org

central role" financed by Fund /PGO/Ministry of Public Health, Welfare and Sport (VWS) (see 1.2).

The care standard consists of evidence from published articles on MSS, information given by parents and the knowledge of doctors and other closely involved professionals currently available. In addition, information and guidelines have been used from standard management for some medical manifestations of MSS.

Which people play a central role in this care standard?

The most central role in this care standard is for the patients themselves with MSS. The difficult period just after birth; living with chronic health problems; the treatments and subsequent consequences, all offer much information and insight into MSS and the care and treatments that are required.

Children and (young) adults with the syndrome all have profound physical and intellectual disabilities. They are not able to take care of themselves and/or to be in charge of their own lives and they need their parents/carers to tell their story and to manage their care. In addition, the parents need thorough information on the syndrome, so they can make conscious choices in relation to the treatment and care for their child, but also regarding the risk to future pregnancies.

Expectations for a child suffering from MSS

The expectations for a child suffering from MSS that is born today have greatly improved since the first description in 1971, especially due to the general progress in medical care, but a great deal of improvement is still required in treatment and care. For a child born with the syndrome, it is of great importance that the disorder is recognized as early as possible, so the right investigation and treatment can be provided. With early and optimal intervention, the quality of life and the expectations for the baby improve substantially. Children with MSS can, with the right and personalized care in the long-term, reach adult age. However the outlook for patients is heavily determined by the burden of the disease and the current limitations of effective treatment.

On behalf of the MSSRF

Sonja Bracke, Managing Director

1.0 Introduction

1.1. Background care standard

A care standard describes, from the patient's perspective, the individual prevention and care for a certain chronic disorder. It not only describes the components of the care but also the (multidisciplinary) organization of the chain of care and the relevant quality criteria. (1). This care standard describes the care pathway for the Marshall-Smith syndrome and is meant for parents, carers and other parties in the healthcare sector, such as health insurers[2].

A care standard is based on current and – if in existence – evidence-based views. Where a (multidisciplinary) guideline for the chronic disorder is available, the care standard refers to the guideline and also to relevant quality indicators and quality criteria, if available.

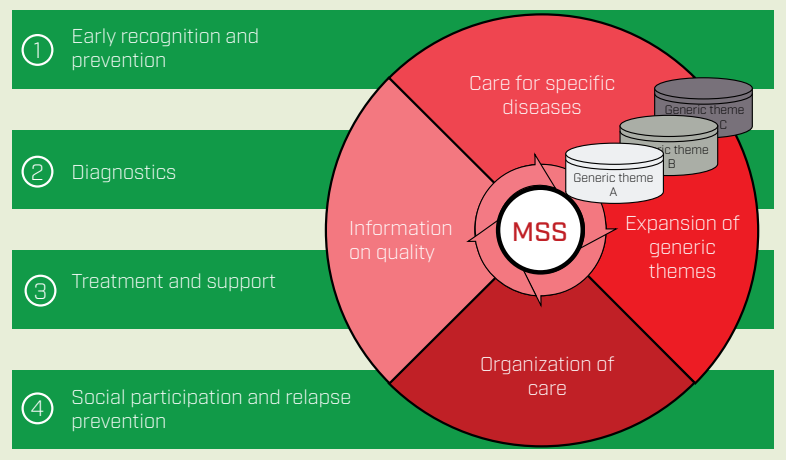
A care standard is specific for a disease and describes the disease-specific care, organization and quality standards. Besides the disease-specific care description, a care standard contains so-called generic care modules (or more concise care themes). These describe, from the patient's perspective, the care pathway of a generic component in the care that is applicable to other chronic diseases.

The Platform Coordination Care Standards (Coordinatieplatform Zorgstandaarden, CPZ[3]) introduced a format with which Dutch care standards have to comply if they wish to qualify for national recognition and registration by the Quality Institute. Based on this format, the disease-specific care continuum of a chronic disease is divided into four phases: 1) Early recognition and prevention; 2) Diagnostics; 3) Individual care plan and treatment; 4) Support, revalidation, reintegration, participation and relapse prevention. The care, organizational structure and quality information are described in each of the four phases. If necessary, each generic module of the theme is extended with disease-specific information (figure 1).

2 The care standard is a guideline for contracting the chain of care by the insurance company and for determining the claims in the insured care and the underlying funding system

3 The CPZ was founded by ZonMW at the request of VWS in 2010. The CPZ was integrated into the Quality Institute in 2013.

Figure 1. Schematic view of the structure of the care standard. The four phases of the disease specific care continuum (horizontal bars) are integrated into the chapters on the organization of the care, the quality information, the generic care themes (or modules) and their disease specific expansions.



1.2 Accountability

The Care standard for MSS has been produced within the framework of the project “Care standards for rare diseases; the patient in the central role”, as applied for and accepted by the Ministry of Public Health, Welfare and Sport/Fund PGO/CIBG (December 29, 2010). For the development of this care standard, the MSSRF joined the Dutch National Alliance (VSOP). Where possible, the care standard format of the CPZ has been followed. Where this was not possible or where it would have led to a less logical classification for MSS, it has been decided to opt for a specific classification that better meets the progression/nature of the syndrome.

The project organization consisted of several working groups: a sounding board, consisting of a project manager, first contact person from MSSRF, advisors (therapists for MSS patients) and the Think tank. Think tank members were medical specialists and therapists with experience/affinity with MSS. The duties and responsibilities of all sounding board members were described clearly: advisors gave advice based on their own specialism and experience and read, corrected, commented on the paragraphs/chapters from the different versions of the care standard relevant to their specialism. Think tank members read complete versions of the care standard, made the necessary corrections and/or comments and agreed to it. The first contact person from the MSSRF and the project manager consulted supporting members and wrote the care standard. For members of the project organization see appendix 1.

For the development of this care standard an inventory of literature and quality standards was made and supporting members were consulted (see appendix 2). Results of the consultation of the supporting members were organized systematically, compared and incorporated together in the relevant chapters of the care standard. The quality criteria (6.1) were developed, based on the systematically organized results of the consultation of the supporting members. For a short description of the consultation of the supporting members and a summary of the results please see appendix 2. The project steps have been explained on the

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website to the different workgroup members. Moreover, relevant links and information on care standards for rare disorders and care standards in general have been inserted. The first contact person from MSSRF and the project manager exchanged documents via a secure project management software program.

For the use of images of children with MSS, consent was obtained from their parents.

1.3 Legal framework

This section was made based on the advice report: 'The consequences of entering professional standards into a legal framework for the legal meaning of these standards and for the legal position of health care providers [2]. Care standards fall under the definition of quality standards, as described in the amended Patients' Rights (Care Sector) Act and other laws connected with tasks and duties in the field of quality of care (Stb.2013, 578). The amended Patients' Rights (Care Sector) Act came into force on April 1, 2014.

The amended Patients' Rights (Care Sector) Act :

- Brings no change in the current legal status of a standard. That status is that a professional person must be expected to follow an applicable standard, unless the circumstances call for a deviation (comply or explain).
- Has no consequences for the meaning of the standards in the context of the Healthcare Inspectorate or the health insurance companies

The amended Patients' Rights (Care Sector) Act is primarily focused on incorporating legal provisions regarding the Dutch National Health Care Institute (Zorginstituut Nederland, ZIN).

- Focusses on the maintenance of a public register by KI (part of ZIN).
- The quality standards that meet the Assessment Framework developed by KI are incorporated in the Register. The incorporation of a care standard in the public register of the Dutch National Health Care Institute (ZIN) has no consequences for the legal effects and the legal position of this standard. If a care standard has not yet been incorporated in the register, professionals are nevertheless expected to follow the standards. However, this implies that the care standard has been developed in conformity with "Model Care Standards "[[4].
  The care standard for MSS has not been offered for incorporation in the register because of the extremely rare incidence of this disease and, as a result of this, a lack of support for authorization of the care standard by medical scientific associations. The agreement of the Think Tank is considered as approval of the care standard.

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1.4 Control and maintenance

The care standard is owned by MSSRF. MSSRF is responsible for control and maintenance of the care standard. Important features are regular review, accessibility and readability of the care standard.

The moment this care standard was introduced a Maintenance Commission was founded (see appendix 1). The Maintenance Commission assesses the care standard periodically for:

- Current national and international developments
- Accessibility and readability

This assessment may lead to a revision. Adjustments regarding the content or the text can be made by the Maintenance Commission. The care standard will then be finalized again by the MSSRF and then agreed on by the Maintenance commission and the Think tank.

The Dutch National Alliance (VSOP) is responsible for identifying and responding to new developments within the generic care themes during the duration of the project and beyond. For a recent overview of these themes please see the website www.zorg-standaarden.net.

1.5 Readers guide

The digital copy of the care standard (in pdf and flipping book format), allows you to find specific information quickly and easily.

This form and method allow you to work with references both inside and outside the care standard. In some cases, a choice has been made for repetition (of the content) because it gives added value.

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2.0 The Marshall-Smith Syndrome

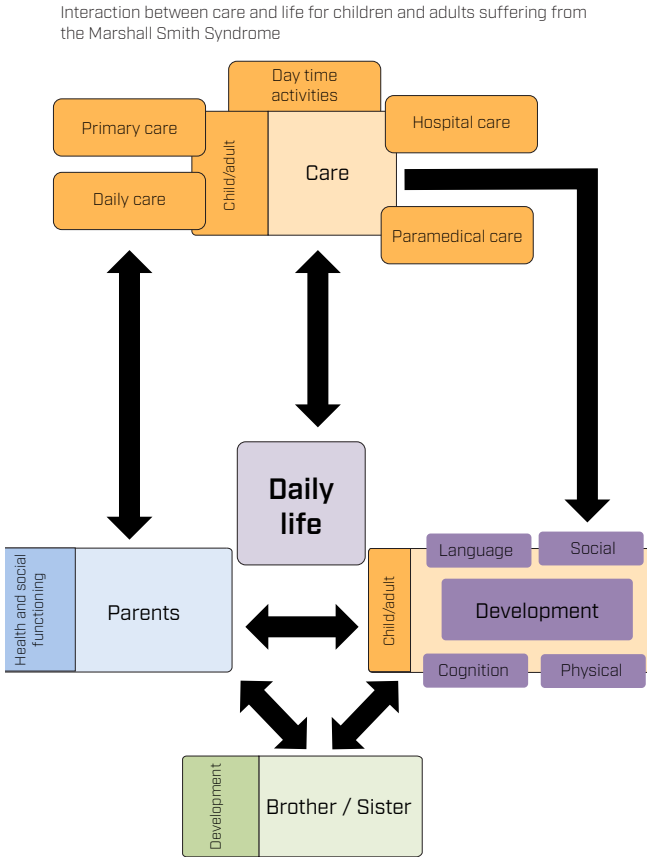
The Marshall-Smith Syndrome is an infrequently described genetic disorder with an apparently very low global prevalence [see 2.4]. The syndrome was first described in 1971 by doctors Richard E. Marshall, C.B. Grashalm and David W. Smith [5]. They described two patients who both had a similar, but unusual appearance and significant developmental delay. The appearance of the children was characterized by bulging eyes, thick eyebrows and a sunken nasal bridge. Both children had severe respiratory problems and failure to thrive and they appeared to have a very advanced bone age. One of these two children died when he was 20 months old. Between 1971 and 2010, at least 43 patients with this phenotype have been described in independent case studies in the medical literature. This entity became known as the Marshall-Smith Syndrome (MSS). The majority of the initially reported children died as toddlers or in early childhood. Later, it appeared that some children were able to live longer, particularly due to the improved treatment methods for respiratory problems. Over the past years, studies have been published that describe several patients. This is particularly a result of international collaboration between clinicians and the MSSRF, including the launch of a web-based WIKI for parents in 2010. A larger group of patients made it possible to draw more solid conclusions about the phenotype of MSS and the natural history of the syndrome. [6]

In 2010, there was a breakthrough in MSS research. A genetic study reported the presence of mutations in the transcription factor nuclear factor I/X (*NFIX*) in individuals with MSS [7] [see 2.3]. Identifying the gene associated with MSS is of great importance in making a diagnosis and gives insight and hope for possible future treatments of MSS.

2.1 Description of the disorder

Children and (young) adults with MSS have unusual facial features, are physically severely disabled and have severe intellectual disability. MSS is a chronic, progressive developmental disorder with complex problems in health and physical/intellectual development. Individuals with MSS have profound multiple disabilities (PMD) to a greater or lesser degree and this makes them vulnerable. They need personal care and supervision 24 hours a day, besides the regular treatments for the symptoms of MSS. Daily life for children and adults with MSS is characterized by intensive and complex care. The development and wellbeing of the individual with MSS is influenced by all aspects of the care network and its personnel (professional carers as well as close family and friends). Figure 2 provides a schematic description of how elements of the care and the daily life of the individual with MSS influence each other.

Figure 2. Schematic view of the interaction between caregivers, close relatives, and the daily life of the individual with MSS. The development of the child or adult with MSS is also influenced by the quality of daily life and by the interaction between caregivers and close relatives (parents, brothers/sisters). The health and social functioning of the parents, as well as the development of any brothers and sisters is also influenced by the individual with MSS.



In the chapters below, the characteristics of MSS are described (2.2) and the physical and mental development of children and adults with MSS (2.3) Possible treatment choices for the various symptoms are described in chapter 3.

2.1.1 Facial appearance

Striking features are the high and prominent forehead, shallow eye sockets, the flat middle section of the face, the prominent bones of the upper jaw and the small, retracted lower jaw. The nose is short with a low bridge, an up-turned tip and open, forward-facing nostrils. The vertical groove in the upper lip (philtrum) is long at an early age and gets shorter with increasing age (see appendix 3).

The position of the ears tends to be low-set and they have some anatomic abnormalities (see 2.1.2). The eyes appear large and the eye sockets shallow, which cause prominent or bulging eyes. Often there are long eyelashes and a blue tint to the whites of the eyes (see 2.1.3) (6). As the years pass by, the features become more prominent, especially the eyes, the nose and the thick, everted lips. The mouth often remains open at rest, revealing irregularly positioned teeth and thick gums (see chapter 3). The tongue may be large and sometimes protrudes from the mouth. Young adults of both sexes can have excessive hair growth (6). Almost all the children have curly hair even if their parents do not.

2.1.2 Ears

There are minor exterior ear malformations and the internal and external auditory meatus is very small. This affects middle ear

function and makes chronic otitis media and sensorineural or mixed hearing loss common. In some children, infection has led to a petrous apex effusion that is an accumulation of fluid [6] [carer consultation] (for treatment possibilities see 3.3.2.2)

2.1.3 Eyes

The eye sockets are shallow which make the eyes appear big and sometimes bulging; the white of the eyes in some children can be remarkably blue. The eyes are very sensitive to light and this may have an effect on the eye-hand coordination. Some of the children are short-sighted to a lesser or greater degree (myopia). A squint (strabismus) sometimes occurs. Many children have narrow or obstructed tear ducts, which means that tears cannot flow away causing persistent eye-watering and increased risk of infection. An underdevelopment of the optic nerve (septo-optic dysplasia) is occasionally seen. Increased internal eye pressure (glaucoma) which is caused by abnormalities inside the eye, has been detected in several children, in some cases in combination with septo-optic dysplasia [6] (for treatment possibilities see 3.3.3.1). Once a child is able to walk, they may have problems dealing with a transition to a different surface. The cause is unknown although parents think a possible cause may be short-sightedness or a lack of depth perception (source: carer consultation).

2.1.4 Skin and nails

The skin is often dry and eczema often occurs, particularly at the start of puberty. The nails are thick, fast growing, and are turned up or can grow into the nail bed (source: carer consultation).

2.1.5 Bones, joints, connective tissue and muscles

Bone growth; dysostosis and osteroporosis/arthritis

An important physical characteristic of MSS is dysostosis: a disturbance in normal bone development, especially in terms of calcification of the bones. There is a seemingly advanced bone maturation in part of the bones. Because of this, the bone age as evaluated by x-rays of hands and feet seems several years advanced compared to the normal skeletal age [6]. In x-rays of the hands and feet from birth until five years old, this phenomenon is visible. The unusual bone maturation is a consequence of the *NFIX* gene defect (see 2.3). The abnormalities commence when the fetus is developing in the womb. The defective gene disturbs the anatomy of the head/neck region and the limbs, affecting breathing and ability of the baby to feed normally.

Previously, MSS was regarded as an example of a syndrome with excessive growth based on the seemingly advanced bone age. ([5][8]). However, this is in fact incorrect, because in other parts of the skeleton (for example the chest and the back) the bone age is not advanced. At the age of about five years, x-rays of the hands and feet show a gradual disappearance of the advanced bone age, and it becomes more or less the same as in the remainder of the body. The seemingly advanced bone age should be seen as a phenomenon of dysostosis. After some time, there may be evidence of reduced bone density in the form of osteoporosis. This phenomenon is related to the abnormal bone formation in the hands and feet and does not occur in all children suffering from MSS. The reduced bone density causes fractures in most children,

but not in all. The reason is still unknown ({6}. In some individuals the uppermost vertebrae in the spine (the atlas and axis or C1/C2) are malformed and unstable ([5][9]).

Fractures

Bone fractures occur from early childhood until puberty and sometimes even at an older age. However, not all children get fractures, and when they do occur, the problem can vary from a single facture to frequent occurrences. They have occurred with relatively minor trauma and also on occasion spontaneously with no apparent trauma. ({6}) {source: carer consultation}. It is (still) not clear why some children get fractures and other children don't (for treatment possibilities see 3.3.2.4).

Back and neck (kypho)scoliosis and stenosis

Growth (length/height) is fairly normal in early childhood but changes from the tenth year and sometimes earlier, gradually falling far under the average [see appendix 4]. There does not appear to be a growth spurt in puberty [6].

Most children develop kyphoscoliosis of the thoracic spine in early childhood but in some cases not until around puberty. Significant curvature can develop very quickly, and is associated with a significant risk of respiratory complications and even death. If osteoporosis is present, surgical options for treatment may not be possible. Cervical spine stenosis which can cause weakness and paralysis is can sometimes be a problem [6] see 2.1.8).

Craniosynostosis

Craniosynostosis arises from a premature fusion of one or more cranial sutures. It is seen in some children with MSS [6] [9] [10] [11] [8]; for treatment possibilities see 3.3.2.4).

Connective tissue, muscles and joints

All children have reduced muscle bulk and tone. This, combined with lax ligaments contributes to delayed motor development and impaired mobility. Joints are often very flexible in the young, but contractures of the joints can also develop. Abnormal development of the joints, including the hips, is often seen, and this can encourage and/or mimic osteoarthritis in older children and young adults.[6]

The gross and fine motor development in most children is severely delayed and the standard motor milestones are not achieved. Many children are not able to learn to walk, to speak, or to feed themselves. The lack of speech development is likely to be multi-factorial, with the abnormal anatomy of the voice-box having some contribution, but with neurodevelopmental impairment likely playing a more major role [6](for treatment possibilities see 3.3.1.3 and 3.3.2.5).

Teeth

Teeth develop normally, but there is often dental overcrowding due to the small size of the jaw, and the gums often appear enlarged (gingival hyperplasia) (for treatment choices see 3.3.2.4)

Feet

Almost every child with MSS has flat feet, with low muscle tone and lax ligaments being contributory features. The fleshy pads under the toes are often very prominent, but these do not cause problems, however the nails are often very thick and difficult to cut [(see appendix 3)]. The children very often have very sensitive feet (for treatment possibilities see 3.3.2.4).

Hands

The bones of the hands appear different on an x-ray, with the bones at the tips of the fingers being very small, but the other bones in the fingers and thumbs broader than usual [(see appendix 3)]. Most children have sensitive hands and don't like being touched. [carer consultation](for treatment possibilities see 3.3.2.4).

2.1.6 Breathing

Airway obstruction

The severe breathing problems are the result of a combination of symptoms due to the upper airway obstruction. The obstruction is secondary to the combined abnormalities in the craniofacial anatomy. These include a small and receded lower jaw (retrognathia), an underdeveloped centre of the face, a low nasal bridge and closed or narrow nasal passages, and a protruding larynx distorting and limiting the airway [6] [carer consultation]. In some cases, the patient has a larynx which lacks rigidity (laryngomalacia) and an underdeveloped epiglottis [6]. This can cause coordination disorders in the pharynx. The palate is often high and the tongue is often limp and thicker than usual.

One or more of the above mentioned symptoms may cause restricted breathing of the child after birth. Only one single case of MSS without airway obstruction has been documented [12]. The airway obstruction may be less prominent, or not occur until a later time [6]. The child will display, depending on the severity of the obstruction, clearly audible and restricted breathing, snoring, arrested or interrupted breathing, retractions of the thorax and nostrils, an increased heart rate, and cyanosis. The oxygen saturation of the blood is often lower than normal, or shows frequent drops, and the level of carbon dioxide in the blood is often (highly) increased. This can cause tiredness, lethargy, drowsiness and a lack of appetite (for treatment options see 3.3.1.3).

Fatalities

Airway obstruction, obstructive sleep apnea, aspiration and its complications (e.g. chest infections/pneumonia) may cause fatalities in young children with MSS [5][13][12][8].

ALTE

During sleep, some children suffer from unexplained apnea (ALTE[4]) (carer consultation). The infant's position during sleep is

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important, the airway can be obstructed by lying supine and the tongue falling back (for treatment options see 3.3.2.5).

Infections

During their first weeks or months, the children will be staying in a hospital environment where infections are common. These children will come into contact with many viruses and bacteria, increasing the risk of infections. Open-mouthed breathing increases the risk of infections (see 2.2.4). Airway infections often cause enlarged nose and throat tonsils; these can cause and aggravate the breathing obstruction (for treatment options see 3.3.1.3 and 3.3.2.5).

2.1.7 Nourishment

Almost all infants with MSS have (severe) problems with oral feeding [carer consultation]. Hypotonia, the abnormal craniofacial anatomy and/or devices used to support their breathing all play a role in this issue [6]. Babies often have a weak or absent suck and swallowing reflex. These oral coordination issues are often prolonged and also affect speech development. Other associated problems include a risk of choking, aspiration pneumonia and gastro-esophageal reflux (for treatment options see 3.3.1.4 and 3.3.2.6).

2.1.8 Neurological characteristics

Hypotonia

Children with MSS have hypotonic torso muscles, and hypertonic peripheral muscles. This causes general body weakness and pronounced deep tendon reflexes in their limbs. This remains unchanged during their lifetime. The posture of the child after birth is characteristic; it is significantly different from the normal posture of a healthy baby. A baby with MSS tends to lie flat, rather than curled up in the fetal position, and this is likely to be due to the low muscle tone, but also allows the ribcage more freedom to expand (carer consultation). Excessive drooling is common, both due to an open mouth and reduced swallowing. Reduced swallowing before birth probably contributes to a surplus of amniotic fluid (polyhydramnios) in the final trimester of pregnancy [14][10][15]. Most children are unable to fully close their mouths. In combination with the blocked nasal passages, this leads to their mouths being continuously open [6], (for treatment options see 3.3.1.5, 3.3.2.7 and 3.4.3).

Reflexes

Reflexes such as the sucking and grasping reflexes are delayed, and in some cases the sucking reflex is absent, or disappears. Hand-eye coordination is also delayed. The deep tendon reflexes (e.g. the knee jerk reflex) is often normal or reduced in the young child but can become more brisk and pronounced with age as the muscle tone increases [6].

Stenosis

Narrowing (stenosis) of the spinal canal, due to abnormalities of the bones of the spine can occur. This does not necessarily cause any problems, but weakness in the limbs due to compression of the spinal cord has been reported [9] (for treatment options see 3.3.2.4 and 3.3.2.7).

Brain

Brain scans have identified a number of changes to the structure of the brain in some patients, although none are universal. These include an underdeveloped or absent corpus callosum, excess fluid (hydrocephalus) (carer consultation) (for treatment options see 3.3.1.5), an increase or decrease in the folds on the surface of the brain (pachygyria or polymicrogyria), and septo-optic dysplasia [6].

Seizures

In some cases the patients suffered seizures during early childhood, often without an epilepsy diagnosis. However, in some cases epilepsy does seem to be present [carer consultation][(for treatment options see 3.3.2.7).

Sleeping

Many children with MSS are restless sleepers, showing signs of problems when falling asleep and continued sleeping problems (for treatment options see 3.3.2.7). Obstructive sleep apnea may be the main cause of restless sleeping (carer consultation).

Toilet training

Most children with MSS will remain incontinent throughout their life. However, this is not universal and behavioral training can be beneficial (for treatment options see 3.3.2.7).

2.1.9 Other characteristics

Pyloric stenosis (vomiting due to thickened muscles in the stomach) has been observed in a small number of babies. Umbilical hernia and congenital heart problems can also occur[6] and an enlarged kidney (hydronephrosis) has also been reported [17]. Excessive hair growth (hypertrichosis) occurs to some extent in all patients, and one patient reportedly developed a pilonidal cyst at an early age (carer consultation). One child developed a Wilms tumor, however, no cancers have been reported in any of the other children with MSS[6].

2.2 Physical and mental development

MSS causes a complex range of congenital and developmental physical and neurological problems with lifelong consequences.

2.2.1 Sensory development

Sensory perception can be normal, but vision and hearing problems are relatively common, for example due to glaucoma or conductive hearing loss. Furthermore, sensory processing may be affected by the level of intellectual development. This can leave children feeling vulnerable, isolated, and anxious.

The patients often suffer from an over-sensitivity to touch although an apparent insensitivity and high-pain threshold is also recognized (carer consultation). The oral phase, when infants put all objects into their mouths, lasts much longer than usual. The hands and feet are particularly susceptible and some children have been diagnosed with tactile sensitivity disorder. The children may display repetitive movements, and are often very attached to one toy that seems to offer them some form of security. This

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is particularly appealing to children with diminished hearing and sight, and/or severe mental disability.

Research into the functioning of children with severe multiple disabilities (SMD) shows that there are some common characteristics: they often display severe problems in their sensory stimuli processing system, and a fluctuating level of awareness. Some stimuli do not register, or hardly register, while other stimuli cause fierce reactions. In general, the pace of information processing is low. Awareness is an important condition for learning new things, and for further development [18][19]. The functioning of the stimuli processing system is particularly important in children with MSS; these children are often more aware than other children with SMD.

2.2.2 Cognitive development

Within the group of children with MSS there are great differences in cognitive development (carer consultation) [20]. However the children all suffer from severe mental retardation, and show limitations in the processing of stimuli and information. The cognitive development is strongly linked to the motor development issues and the stimuli processing issues. Children function on a developmental age level of between 7 and 31 months on the mental scale, and between 2.5 and 38 months on the motor scale (information derived from a test with children with calendar ages between 32 and 181 months)[17]. The children are able to imitate simple activities, and their good (eidetic) memory and curiosity will sometimes allow them to achieve a performance age at the level of a 3-year-old (for treatment options see 3.4).

The diminished capacity in processing stimuli and information should be investigated. The results inform best management and therapy options to encourage the development of each individual child. The children can comprehend many things, and use their entire body, particularly their hands and fingers, to communicate (non-verbal communication). By only emphasizing the cognitive development, these children are deprived of options. They learn more through contact and experience than through their (limited) faculty of thought. Safety for the child, communication with others and research into the potential of the child form together the basis for all further advances in development of MSS individuals.

2.2.3 Motor development

Motor development in all aspects in MSS is significantly impaired. One of the main neurological causes appears to be hypotonia. All motor milestones are delayed (head control, rolling, crawling, sitting, standing and walking) and most individuals have life-long impairment in mobility. There is significant variation and some children can learn to walk with and/or without a walker, and/or can learn to ride a bike, while others cannot achieve this, leaving these children dependent on aids (carer consultation).

Children with MSS are curious, and love to move. However, they always require stimuli and guidance from people around them, both to develop gross and fine motor skills, and to discover the world around them. The limited physical and motor development limits the patient's range of motion. The child is not automatically able to start exploring the environment by themselves (for treatment options see 3.4.3).

2.2.4 Social-emotional development, interaction and behavior

Children with MSS show differences in social-emotional development, as well as different common characteristics in communication, social interaction and behavior. The children are generally friendly and tenacious, they are often cheerful and enjoy contact. The children are able to recognize their parents and carers, enjoy physical contact and are able to show affection to trusted people. In many cases, they also enjoy contact with unfamiliar people. The children are able to absorb and copy the behavior of others, and enjoy their 'normal' daily life (for treatment options see 3.4.4).

Communication

The children lack verbal communication skills and also show abnormalities in non-verbal social communication with others. In many cases of MSS this is demonstrated as a lack of reciprocal social communicative skills, limited play acting and the display of stereotypical, repetitive behaviors [20]. The severe retardation of the development of speech/language, social skills and the levels of communicative and adaptive functions can be similar to the symptoms of autism. Nevertheless, the social functioning of children with MSS is relatively good compared to those with autism [20] (for treatment options see 3.4.4).

Behavior

In general, children with MSS are compliant, and are able to form part of a family and an appropriate group. A general behavioral trait in MSS is a dislike of change of routine - with or without tantrums -, repetitive and stereotypical play, and limited fantasy play [20]. Problems within one of the development areas are directly connected to the other development areas. That is why it is important that problems in the (mental) development are signaled at an early stage, using appropriate diagnostic tools {1} and related treatments (for treatment options see 3.4).

2.3 Cause and heredity

Cause

A genetic study in 2010 reported on possible mutations in the transcription factor nuclear factor I/X (*NFIX*) for MSS [7]. The specific function of NFIX is as yet unclear, but the *NFIX* gene does play an important role in the development of the brain and skeleton. The same study discovered abnormalities (mutations) in *NFIX* in nine children with MSS. The study showed that *NFIX* mutations lead to different phenotypes, depending on the type and impact of the mutation. An earlier study of *NFIX* mutations in mice, reported that the mice had reduced growth, malformations of the spine and decreased density of the bones.

A later study reported that all the children investigated had a mutation in the *NFIX* gene [21]. The impact of the mutation differs, but it is (still) unknown exactly how this influences the phenotype. *NFIX* mutations can also cause Malan syndrome [7].

Heredity

MSS is a genetic condition caused by mutations in the *NFIX* gene that occur during the production of the gametes (sperm/ova) prior to conception. Therefore although MSS is genetic, it is not inherited, and the risk of parents having more than one child with MSS is low. Indeed worldwide it has not been reported that more than one affected child was born in the same family. Males are affected as frequently as females. No risk factors, such as advanced parental age, parental relatedness, or reduced parental fertility are known. The offspring of someone with MSS would be expected to have a 50% likelihood of also being affected, but no individuals with MSS have been known to have children. It is not known whether fertility is normal or not MSS [6].

2.4 Incidence and prevalence

The syndrome occurs almost globally [6] (carer consultation). The MSSRF registers increasingly more young children. This is likely due to the fact that the diagnosis is made earlier than before, and parents are able to contact the foundation personally through the Internet. The prevalence is approximately 1 in 4 million; this means that it is categorized as an ultra-rare disorder (1 in 2 million or more)[22].

2.5 Care phases

MSS clearly distinguishes between the period right after the delivery, characterized by an acute, often life-threatening situation, and the period after that, when the situation of the infant has stabilized and the child enters the chronic phase of the disorder (see figure 3).

The acute care phase for MSS starts at the birth of the child and lasts - depending on the health condition of the newborn – anywhere between several days to weeks/months. The final element of the acute phase is the release from hospital after the first post-natal hospital stay. The chronic phase of the care starts at the release after the first postnatal stay and continues for the rest of the patient's life. During the chronic phase the organ specific complications and developmental problems will start to surface.

Chapter 3 describes the disease specific care based on this classification (see 3.0 introduction, 3.3.1 and 3.3.2). Chapter 5 describes the aspects of organistional care for both care phases.

Figure 3. Care structure for an individual with MSS based on the care needs and the characteristics of the disorder.

Acute care phase	Chronic care phase
Post-natal period in hospital	Organ specific complications Cognitive and behavorial problems

2.6 Disease burden and psychosocial consequences

The disease burden for patients with MSS is determined by the patient's overall health status. This focuses on the disease, symptoms, limitations and the quality of life of the child/adolescent. The manifestations of all prevailing health issues and limitations of the syndrome, and the changing nature of these issues and limitations during the patient's lifetime determine the disease burden for each individual child with MSS. The disease burden is often also experienced by the parents/carers, and is often, but not necessarily proportional to the severity of the symptoms of their child with MSS. This disease standard does not classify the disease burden for MSS, but indicates whether the disease burden is relatively bigger or smaller during the acute and chronic care phases.

Observations and carer consultation have shown that children with MSS are very resilient when it comes to dealing with treatments and limitations, especially in those who do not appear to experience pain. However, intensive treatments will leave their mark on children, and contribute to the disease burden (for treatment options, see 3.4.4 and 3.4.5).

2.6.1 Disease burden during the acute phase

The acute phase requires acute specialist care. The children will often stay at the hospital for weeks or months on end, causing a high level of disease burden. During the acute phase, the physical limitations that cause the disease burden are predominantly caused by breathing difficulties, infections, and feeding difficulties. There is significant stress and anxiety on the parents and wider family. The disease burden will increase, for both children and parents, over the course of time, due to the characteristics of MSS and the invasive nature of treatments.

Breathing

The disease burden occurs after birth, caused by severe obstruction of the upper airways and presents an acute life-threatening situation. The seriousness of the obstruction determines the treatment (for treatment options see 3.3.1.3 and 3.3.2.5). The artificial airway, ventilator, feeding tube, and intravenous cannulas and infusions, limit the movements of the child, and the proximity of the parents.

Infectious diseases

During the acute phase, the child is very susceptible to infections, partly due to the obstruction of the airways and the use of ventilatory support. Infections also cause a high disease burden, because of their profound influence on the overall health and dietary condition of the patient. Infections appear to be, along with breathing and dietary issues, one of the main causes of 'failure to thrive'[5], (for treatment options see 3.3.1.3. and 3.3.2.5).

5 This means that the child has a significantly lower weight than should be expected and/or the child gains significantly less weight than is usually the norm.

Diet

Oral feeding of the child is often not possible, immediately after the birth, due to the abnormal anatomy. Not being able to take in nourishment by itself causes disease burden, not only because the baby cannot influence the feeding process, but also because it causes a disruption of an important moment of contact between parent and child (for treatment options see 2.7.3 and 3.4.5).

2.6.2 Disease burden during the chronic phase

During the chronic phase, the children are usually cared for at home with family members. The invasive treatments after the hospital stay are often continued at home, which causes a high to moderate level of disease burden. During the chronic phase, the patients are often admitted to hospital for varied amounts of time, due to surgery, infections and/or complications due to the characteristics of MSS. The disease burden in this phase is mainly determined by the problems that occur in, and the treatment the patient is given for breathing, feeding issues, infections and the increasingly more prominent physical limitations, complications, disturbed psychosocial well-being and a disturbed social-emotional development (see 2.7.3, 3.3.2 and see 3.4.4).

Breathing

The airway obstruction is usually still present in most children at this stage. In some cases, treatment will allow children to breathe unaided. These treatments contribute to the disease burden, but the results will also decrease the burden. Monitoring the breathing obstruction will remain an important element during this phase. Obstructive sleep apnea syndrome (OSAS) can occur during the chronic phase without obvious symptoms. OSAS can present as restless sleep, no appetite in the morning, behavioral problems and snoring (see 3.3.2.5).

Infectious diseases

Infections are concentrated in the ear, nose and throat area, and increase the disease burden through breathing difficulty, treatment with antibiotics and the common necessity for surgical removal of tonsils and adenoids. As the children get older, the infections will decrease (see 3.3.2.5).

Diet

The disease burden related to the dietary needs of the child in the chronic phase differs for each child. Some of them will gradually learn how to take in food orally; these children will often eat a lot and enjoy eating, without actually gaining a lot of weight. Another group of children will not be able to take in enough food and fluids orally; in order to decrease the disease burden caused by a nasal feeding tube, they will be fed through a PEG/gastrostomy tube. When this tube is placed correctly, connected to a Mic-key button, it causes little disease burden (see 3.3.2.6).

In some cases, the patient has a large and protruding tongue that hinders the eating process, and often causes excessive salivation and drooling (carer consultation).

Physical limitations

The delayed motor development is reflected in the chronic phase, and causes an increase in the disease burden. Children will not start to walk, or will start this process much later than is generally the case (4-6 years). Scoliosis, kyphosis, joint problems, fractures and an abnormal position of the feet, including flat-footedness, cause movement limitations and a related increase of the disease burden.

The progressively debilitating character of the physical issues of MSS is determining for the disease burden from the age of ten; during this phase, the motor skills will slowly decrease and the damage caused by the lipid metabolism disorder will become more noticeable. Fractures and treatments contribute to the disease burden.

2.6.3 Psychosocial consequences of MSS for child and parents

Being born with and having to live with MSS means living with the continuous presence of limitations, both physically, mentally, and psychosocially, both during the acute phase and the chronic phase. The psychosocial consequences of MSS for children are difficult to measure objectively, due to the development levels of the children. The right diagnostic tools do offer possibilities for research {1}.

Child

The normal development of a child with MSS is partially influenced by the intensive care that the child receives during the first weeks to months of its life [20]. During the acute phase, the child has no natural, constant contact with the parents, as is normally the case in a home situation. During both the acute and the chronic phase, the (prolonged) hospital stay(s) and invasive treatments cause an increased risk of post-traumatic stress disorder (PTSD) for both parents and child. If this syndrome occurs, it causes a high disease burden [20]. During the chronic phase, many children develop a fear of the hospital and treatments (carer consultation).

The moderate to serious mental retardation, with significantly limited mobility and little to no speech, causes an ever widening developmental gap between these children and their peers. The cognitive and social-emotional development is influenced by the intellectual disability (see 2.3 and 3.7). The children do not appear to experience a lot of disease burden from this element; however, the parents do suffer from this (carer consultation).

Parents

For parents, the acute phase is a very stressful time. The birth of a child that is 'different' and is often born under life-threatening conditions, and the prolonged and recurring stays in hospital often cause them to feel as if they are losing control of the situation, and this causes a feeling of helplessness. This increases the risk of PTSD for the parents. For the child's benefit, it is important that the parents do not develop PTSD, as that would also limit the risk of the child developing this disorder [23].

Having a child with MSS means a very profound change in the lives of the parents. MSS influences their entire life, the domestic environment, daily routines, psychological well-being, their

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partnership, any other children, the psychosocial environment, work, social position and participation (see 3.5 and figure 2). The parents are suddenly faced with the medical care for their child at home. In families with multiple children, the child will be a full member of the family, despite all limitations.

Parents experience a heavy disease burden during the acute and chronic phases. Handling this special situation takes a toll on their psychological resilience. Information from professionals, in order to support the parents, is therefore very important during both the acute and chronic phase (see 3.4.5 and 5.0).

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3.0 Disease-specific care

This chapter is about the proper and specific care that is needed for MSS in the acute and chronic phases (see 2.5). Patients with MSS experience long-lasting restrictions in their daily lives and the children need continual care and attention from the people around them. An essential part of the disease-specific care is the provision of information and educating the parents of a child born with MSS.

Guidelines

There are no evidence-based or consensus guidelines for multi-disciplinary care in MSS, and in practice, management is based on the supportive care of individual symptoms. Several symptoms of MSS are relatively consistent features across the patient cohort. For some features, there are well established management guidelines for management, regardless of the underlying cause. Over the last 40 years, experience has been gained in the treatment of children with MSS. In this care standard, we draw on the experience of doctors and parents for treatment, coaching and rehabilitation. In this care standard, the guidelines for the treatment and care of children with profound multiple disabilities are applied, because MSS causes profound multiple disabilities in most children and the treatment and care of people with a PMD correspond to the level of development and the symptoms.

Acute phase: introduction

For a child born with MSS, acute and proper care is needed, focussed on stabilising a possibly life-threatening situation. If necessary, the Dutch “Resuscitation of newborn infants” guideline (or similar guidelines in other countries) should be followed {5}. Following this, the objective will be to work towards a less or non-life-threatening condition, where acute problems will be under control. After the acute care phase, which can take weeks and sometimes months and which always takes place in hospital, the child will be discharged from hospital in a stable condition (see 5.2.1). The child will often go home with a feeding tube and/or breathing support. Children will breathe with the aid of a tracheostomy tube or a nasopharyngeal airway (NPA) and will sometimes also use oxygen (see appendix 7).

In 3.3.1 symptom specific treatment options are described for the physical characteristics of MSS in the acute phase.

Chronic phase: introduction

After the acute phase, the stabilized condition is very fragile during the first years of life and many health problems often arise. The main physical problems are: obstructive sleep apnoea syndrome, pneumonia and aspiration pneumonia, pulmonary hypertension, scoliosis, kyphosis and an early onset of osteoarthritis [6]. Frequent visits have to be made to paediatricians, ENT specialists and other specialists. Fewer or no visits are made to the GP or health clinic. The serious and often life-threatening health problems mean that the child often fails to thrive. For this reason, parents often have the feeling that the child is slipping through their

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fingers and those of the doctors too. Medical assistance is very often needed for the physical symptoms. Furthermore, signs of seriously delayed mental development emerge, meaning that parents need the support of first and second line professionals, such as a speech therapist, physiotherapist, occupational therapist, rehabilitation specialist and psychologist. The delay in physical, sensory, motor and cognitive-emotional development means that it is necessary for children to attend day programmes at rehabilitation centres, or at special day-care centers/schools for children with multiple disabilities, where proper assistance with the child's development is available together with an individual support plan [see 5.2.1 and 5.2.2].

In 3.3.2 symptom specific treatment options are described for the physical characteristics of MSS in the chronic phase.

Informing and educating parents

Parents act by definition in a holistic manner; they continually follow the entire development of their child from close by, see a great deal, think about things in terms of their own frame of reference and adapt their actions accordingly. Parents' expectations are built up from this perspective and it is important that professionals take the background, extent and reality of parents' expectations into consideration, as well as the strength of the expectations and the energy that parents and the child are willing to invest in realising these expectations.

The observations, experiences, feelings and expectations of parents with regard to the child should always be taken seriously. This means that the professional must listen well, ask probing questions, discuss treatments and take decisions together with the parents. A system, such as the "Pediatric Risk Evaluation and Stratification System" (PRESS), is recommended during hospital stays, but the appointment of a case manager is also important [see 5.2.1]. Parents need information about the syndrome and support in organising care at home. The information helps parents to understand what the problems are and to have more control over their lives. Support with the organisation of care means that parents can cope with the intensive care at home and keep going for a longer period of time. These are both in the interests of the child.

Peer-to-peer contact is important to many parents [carer consultation]. This contact should always be facilitated by making parents aware of the existence of the MSSRF Foundation. Parents are then in a position to make their own choices.

3.1 Early diagnosis and prevention

3.1.1 Early diagnosis

This care standard is an important guideline for early diagnosis of MSS. Early recognition and diagnosis of MSS in the acute phase is of enormous importance in initiating appropriate investigations and management.

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3.1.2 Prevention

Due to its rare and sporadic occurrence, and typical presentation after birth, prevention of MSS is not currently possible. Nonetheless, early and anticipatory recognition of symptoms and secondary complications is hoped to reduce disease burden and improve outlook.

In the case of MSS, there is care-related prevention (tertiary prevention); this is focused on the prevention of deterioration, complications, or restrictions to the child caused by MSS. The tertiary prevention is also focused on the enhancement of knowledge and the ability of the parents to cope. This is done by providing information and education during the acute and chronic phases of MSS (see 3.3 and 5.2).

3.2 Diagnosis

3.2.1 Indications for the diagnosis

A combination of presenting symptoms, clinical features and diagnostic investigations leads to the diagnosis of MSS. If MSS is suspected on clinical grounds, an X-ray of the hand can be arranged, as the appearance of the bones in the hand is very characteristic. In children with MSS, aged between 0 and 5 years, bone development is always seemingly ahead of the chronological age [5]. An x-ray of the hand showing an advanced bone age is, however, not an indication of MSS in itself. In all cases, there is evidence of a combination of symptoms, as is normal with a syndrome. If the external characteristics and the physical characteristics have been established and correspond to MSS, then genetic testing of the *NFIX* gene is indicated (see 2.3).

3.2.2 Differential diagnosis

MSS shares similarities with other syndromes, some of which are; Desbuquois chondrodystrophy, Fine-Lubinsky syndrome, Pyknodysostosis, Antley-Bixler syndrome, Ehlers-Danlos type VII, galactosyltransferase | deficiency and Lysyl hydroxylase 3 deficiency [6].

3.3 Treatment, education, rehabilitation and care-related prevention: physical characteristics

This part of the care standard describes treatment, education, rehabilitation and care-related prevention for the physical characteristics and symptoms, both in the acute and the chronic phases of MSS. The description is based on the most commonly occurring characteristics and symptoms. Care-related prevention mainly involves precautionary measures and/or investigation, in order to identify problems and prevent them from becoming worse. Rehabilitation involves development, a possible recovery or reduction of the consequences of MSS.

3.3.1 Acute care phase

Investigation and interventions during this phase are mainly focused on diagnosis and stabilization of the child in relation to the symptoms that can occur in the acute phase. These are described in paragraphs 3.3.1.1 to 3.3.1.5.

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3.3.1.1 Eyes

The eyes should be examined after birth for possible evidence of glaucoma. If glaucoma is detected, then this can be treated in accordance with the applicable guidelines from the European Glaucoma Society {10}. Late detection of glaucoma can result in severe visual loss. The treatment of this is dependent on the severity; Baerveldt implants are often used for this. The cornea of the eyes is delicate. During anesthesia, the eyes must be carefully taped shut [6], so that the corneas are not damaged

3.3.1.2 Bones, joints, connective tissue and muscles

In the acute phase, an X-ray of the hand or full skeletal survey performed to investigate the possibility of an advanced bone age and dysostosis, a disorder of the development of the bone. The joints and muscles are also examined. At present, there is no treatment available for dysostosis [see 3.3.2.4].

3.3.1.3 Respiration

Airway obstruction

During this phase, a multi-disciplinary team of specialists will carry out examinations and acute interventions in the case of a life-threatening airway obstruction.

Laryngoscopic examination, intubation and anesthesia

It is important to carry out an examination of the throat area in order to establish in how far the airways are defective and constricted and to decide upon the necessary treatment. The anesthesia needed for laryngoscopic examination and other treatments requires careful preparation by an ENT surgeon and the anesthesiologist, so that they are prepared for any possible problems that may arise during the intubation of a child with MSS. Various publications have shown that serious complications can arise in children with MSS during anesthesia ([24][25][26]). In practice, it has been demonstrated that a laryngeal mask (LMATM) is a satisfactory aid during the anesthesia of a child with MSS. Examination of the larynx and trachea can be carried out by the introduction of a flexible endoscope into the laryngeal mask which has been placed [27]. It is important to ensure the eyes are closed during anesthesia in order to reduce the risk of desiccation, damage and infection of the cornea [6].

The insertion of an endotracheal tube during intubation can normally take place in accordance with the existing guidelines for difficult airways {3}. However, the anatomy may be of such a nature that intubation through direct laryngoscopy could be difficult or traumatic. This can be caused by particular defects, such as the reduced rigidity of the larynx (laryngomalacia), a larynx situated too far forward and/or the small receding lower jaw. It is difficult to wean some children from mechanical ventilation. As far as we know, this is caused by a serious obstruction and not due to central neurological dysfunction [carer consultation]. The guideline ‘weaning from mechanical ventilation’ is applicable in the case of MSS {7}.

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Choanal atresia

Most children have a unilateral or bilateral choanal narrowing (stenosis) or atresia at the back of the nasal passage. In the case of bilateral atresia, resuscitation and immediate intervention may be necessary after birth {5}. However, atresia is usually noticed during laryngoscopic examination. The atresia can be temporarily relieved by placing a Mayo tube in the mouth. The only effective treatment is an operation to correct the defect where the atresia is opened and an open airway is secured using a nasopharyngeal tube (NPA) or a temporary stent {4}. The narrowing or atresia is caused by the defective bone development in the face that occurs with MSS.

Cervical spine instability

During intubation under anesthetic and the care of the child, it is important to consider the possibility of the atlas and the axis in the neck. Bending the head too far backwards during a throat examination or laryngoscopy for intubation can cause damage to the spinal cord [5]. It is therefore recommended to carry out endotracheal intubation with fibre-optic visualization through a laryngeal mask. A supine position is usually contraindicated for children with MSS, because the airway is not open in the supine position ([27] [carer consultation]).

Tracheotomy

A tracheotomy often takes place a few days or weeks after the birth or if, after a period of several weeks, a nasopharyngeal airway cannot be sited. In these cases, the obstruction of the airways is often too serious and cannot be solved in any other way than a tracheotomy. A tracheotomy is carried out according to standard procedure and cared for according to guideline {6}.

Nasopharyngeal tube and other treatments

Some children are treated temporarily or receive prolonged treatment with a nasopharyngeal airway (NPA), which effectively opens the upper airways. The NPA is also used sometimes during anesthesia ([28][29]{4}).

One child is being treated with a Tübingen Platte. The Tübingen Platte is a palate plate that ensures that the tongue, lying far back in the mouth due to the receding lower jaw, is held forward and does not fall back into the throat, leading to an improvement in the child's breathing. This is a tailor-made aid. The Platte provides a better resting position for the tongue, and makes it easier to swallow. This, in turn, produces a positive effect on the functioning of the Eustachian tube and the ventilation of the middle ear ([30] [carer consultation]).

Infectious diseases

During the first years of life, viral and bacterial infections cause almost permanent problems for the airways, which vary from an infection of the upper airways to severe pneumonia. Treatment involves rinsing the nose and pharynx with a saline solution and often administering antibiotics, sometimes for a considerable length of time, in order to cure bacterial infections. Admission to hospital is necessary for some children.

3.3.1.4. Nourishment

There are no guidelines available about nourishment and the intake of nourishment in children with profound multiple disabilities. It is important for a child with MSS to receive nourishment that provides sufficient energy. In the acute phase, during which the child experiences breathing problems, infections and stress due to the treatment, the child uses far more energy than a baby in a normal situation. It seems almost impossible to prevent the failure to thrive in children with MSS, which means that weight lags behind growth and the rate of growth is lower than normal. However, if attention is focused on the supply and intake of food from birth, this can help in prevention.

Offering food will give an indication of the child's abilities to ingest food. If ingestion through the mouth is not possible, the child will be fed through a nasogastric tube or – if possible – in combination with bottle feeding. Brest feeding or for bottle feeding, expressed breastmilk is preferable to formula milk [31]. The mother should be well-supported and offered advice according to the rules of the UNICEF/WHO breastfeeding certificate. The expressed breastmilk can possibly be enriched with a protein supplement, such as Neonatal BMF®.

If it is not possible to express breastmilk, nourishment will consist of formula milk, or an adapted drip-feed, such as Infatrini®. The individual situation of the child will determine what kind of nourishment the child will receive. In practice, this means that some children will only be able to begin ingesting food via the mouth after the acute phase.

In children with choanal atresia, drinking is often very difficult during the first weeks, due to obstruction in the nose (a healthy child has to breathe through the nose when it is sucking, which is impossible in the case of choanal atresia). With a step-by-step plan including the use of a Mayo tube, soother/dummy and something to drink, the child can, if possible, be carefully guided towards oral feeding.

Pre-speech therapy support is indicated for all children with MSS both in the acute and the chronic phases. Using specific knowledge and methods, the pre-speech therapist deals with problems in the oral cavity that hinder the normal development of drinking (and later eating) and can support the child and its parents, both in hospital and at home.'

3.3.1.5 Neurological characteristics

The Brain

In the acute phase, neurological investigations will take place for signs of hypotonia, structural anomalies of the brain and possible hydrocephalus. The hydrocephalus can be treated according to the guidelines for hydrocephalus 0-2 year-olds and/or hydrocephalus in craniosynostosis {8}.

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Hypotonia

Due to the hypotonia and poor head-control, the child needs to be well supported whenever it is being cared for and carried. In bed, the child can be supported by a small sandbag or a horse-shoe-shaped cushion (see appendix 7).

3.3.2 Chronic care phase

During the chronic phase, examination and interventions are focused on the symptoms that occur. It is important for a multidisciplinary team of specialists to be involved in the treatment, developmental training, rehabilitation and care-related prevention for the child with support from the parents. In many cases, the diagnosis will already have been made during this phase. In addition to the physical problems, the focus during this phase will lie on the motor, cognitive and psychosocial development of the child. The physical symptoms worsen in most children as they reach the age of 8 to 10 years old. The child needs preparation for this by a multidisciplinary team (see 5.0).

3.3.2.1 External characteristics

The external anatomical characteristics become more prominent during the chronic phase. As the cranial sutures do not always join together properly, the skull has an abnormal shape. Through the development of the permanent teeth, the prominent upper jaw and the sometimes prominent teeth become more visible. Purely cosmetic treatment is rarely offered in the Netherlands unless an improvement in function is anticipated. Other countries may have different approaches [carer consultation].

3.3.2.2 Ears

The commonly occurring ear infections can be treated according to the guidelines {13}. The placing of T-tubes in the eardrum offers symptomatic relief from infections. However, some children develop chronic ear discharge afterwards [carer consultation]. A meatoplasty may be carried out to widen the ear canal and, in some cases, it is necessary to carry out an operation on the petrosal bones (mastoidectomy) [carer consultation]. Some children require hearing aids.

3.3.2.3 Eyes

Short-sightedness can be treated with glasses or tailor-made lenses. Glasses need to be tailor-made due to the prominent eyes and small nose. Blocked tear ducts can be opened during an operation. A squint can be routinely treated. A well-fitting pair of sunglasses often offers a solution to light sensitivity [carer consultation]. It is, however, also important to be aware that photosensitivity can be a symptom of glaucoma.

3.3.2.4 Bones, joints, connective tissues and muscles

Bone growth, dystosis and osteoporosis/arthritis

After approximately the age of 5, x-rays of the hands and feet show that the bone-age is more or less equivalent to chronological age. The rest of the skeleton develops relatively normal bones, but after a while signs of osteoporosis often become apparent. This is related to the abnormal bone formation (dysostosis) in the

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hands and feet. In 2013, research was initiated into bone metabolism in MSS in Oxford (UK) using a mouse model for MSS.

In one child, hip pain caused by wear and tear has been treated with corticosteroid injections and, later on, hyaluronic acid [33]. These treatments only had a temporary effect. Several children have had a total prosthetic hip replacement [carer consultation]. Instability of the atlas and axis can be treated by fixing the vertebrae.

Fractures

Fractures are routinely treated with an external plaster cast and rarely with surgery [carer consultation]. A number of children have been treated with FOSAMAX® and/or Zometa ® for the prevention of fractures. Due to the difficulty in administering these medicines and the side effects they induce with prolonged use, this treatment has been stopped for most children [carer consultation]. Most of the children known to have fractures, but also some without fractures are having preventive treatment in the form of vitamin D and calcium. The efficacy of these treatments is not yet known [carer consultation].

Back; kyphoscoliosis, scoliosis and stenosis

Curvatures of the spine can in principle be treated in accordance with a (treatment) guideline ({14}{15}). In some cases, scoliosis has been treated with a brace. However, this treatment had little effect and was distressing. Some children with MSS have had successful back operations. A complication lies in the simultaneous prevention of the pronounced osteoporosis, meaning that the fixation of material to correct the scoliosis can be a great problem. In the case of kyphoscoliosis with stenosis of the vertebrae, an operation is quite risky, but in practice does not appear to have been impossible [34].

When considering surgery on the back and/or vertebrae, the whole back must be fully imaged, as there is often a combination of kyphoscoliosis or scoliosis with stenosis in a part or the whole of the spine. It is also important to consider the total functioning of the child and quality of life outcomes when considering any surgery.

Craniosynostosis

In the various countries where the children with MSS live, doctors follow different procedures when offering surgery for craniosynostosis [carer consultation]. In the Netherlands, the Dutch guideline provides directions for the examination and treatment of craniosynostosis in the case of MSS {11}. A number of children abroad have undergone reconstructive surgery with satisfactory results ([11][carer consultation]).

Teeth

The jaw is often too small for the permanent dentition and routine dental and orthodontic care is usually necessary [carer consultation]. It is recommended that the child visits a dentist specialized in treating children with a disability, and treatment (under anesthetic, see 3.3.1.3) usually takes place in a centre for special dental care [carer consultation].

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Feet

The 2010 study showed that all children have pes planus (flat feet) [6]. For some children, custom-made arch supports in ordinary or orthopedic shoes are sufficient. Other children may also need additional ankle or leg supports and/or fully custom-made orthopedic boots [carer consultation].

Hands

The increased mobility in the thumb and contractures in the fingers can be treated with a brace or splint [carer consultation].

3.3.2.5 Breathing

Growth, infections and other arising health problems require careful monitoring of the treatment employed. It is often assumed that natural growth in the head and neck area will allow a child to breathe without aid. In practice, it appears that this is often not the case, or has only been possible for a limited period of time (28) [carer consultation]). However, by applying new techniques in craniofacial surgery, it appears to be possible for some children with MSS to live without breathing support (see craniofacial surgery).

Airways obstruction

Many children with MSS are dependent for their entire life on a tracheostomy, positive pressure ventilation at night (CPAP), a Nasopharyngeal airway (NPA) or another aid. However, even if a child is no longer dependent on breathing support, the optimal functioning of the airways is an important point to bear in mind. Obstructive Sleep Apnea Syndrome (OSAS) is a very common symptom, particularly in the chronic phase. OSAS is investigated and treated according to the guideline {2}. In many cases, obstruction of the airways fluctuates erratically. In practice, it often occurs that an airways obstruction can develop postnatally, and an obstruction that appeared to have disappeared recurs, requiring further intensive intervention with a tracheotomy or an NPS ([source: carer consultation]{4}.

It is important to be alert to temporary obstruction of the airways in the child (while asleep). During an upper respiratory tract infection, thick mucus, or swelling of the tonsils and adenoids can quickly block the airway. A swollen tongue can also block the airway. Infections can be treated by rinsing with normal saline and, if necessary, antibiotics. Adenoids and tonsils are usually removed during childhood. In order to combat the weakening of the throat area, an operation such as an uvulopalatopharyngoplasty (UPPP), a laser-assisted uvulopalatoplasty (LAUP), or a somnoplasty as part of a total course of treatment, can be carried out. Using endoscopy, an ENT specialist can explore the smallest recesses of the nasal cavity and pharynx to determine whether one of these operations may be successful.

Infections

In the chronic phase, infections of the upper airways occur often. However, infections in the lower airways (pneumonia), also occur, either due to a viral or bacterial infection, or through the aspiration of food. Pneumonia is a frequent cause of death in children with MSS ([carer consultation][25]). In some cases, sepsis can result from bacteria entering the bloodstream. Effective treatment

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of any breathing problems reduces the risk of pneumonia with a likely improvement in general health, growth and development [source: carer consultation}. Bacterial infections are treated with antibiotics, but antibiotics are not effective against viral infections. Children will often require hospital admission for treatment and support during infections.

Immunity and vaccinations

As far as we know, the children have a normally functioning immune system. However, the children are more susceptible to infections due to airways obstructions and the failure to thrive. In accordance with the Dutch national vaccination program, vaccinations can take place and, in individual cases, can be supplemented with a pneumococcal immunization and an annual flu vaccine.

ALTE

Apparent Life Threatening Event (ALTE) needs to studied and approached on the basis of the guidelines relating to ALTE {12}. Parents' experience: by picking up the child and stimulating him/her, breathing usually starts again spontaneously. In all the children, the throat area is narrow, the tissue is lax and, in many cases, the tongue can close off the windpipe if the child is lying down. It is important to find out the most optimal position for breathing. Lying on one side is usually the best and with the help of a sand bag or a small cushion, the baby can be positioned on his/her side. At home many children are monitored by a heartbeat/saturation meter, which may reassure parents.

Craniofacial surgery

Several children in the US have undergone craniofacial surgery ([36][37]), where parts of the face/jaw have been reconstructed. In this way, the lower jaw can be fixed in a more forward position by extraction, or a more extensive operation can take place (Monobloc distraction, lower jaw distraction, LeFort iii RED distraction). With MSS, the aim of these reconstructions is to give the airways more space, so that a tracheostomy or other breathing aid is no longer necessary. This operation has already been carried out on two children, where the tracheotomy has been closed and the children no longer need aids for breathing support [carer consultation].

3.3.2.6 Nourishment

In this phase, it is important for a speech therapist to identify the options for oral nourishment. Any successful finger feeding can be continued [see 2.1.7]. Positive experiences relating to food are also of importance during this phase, and the pre-speech therapist can help devise ways for the parents to support this. Due to the breathing problems, the treatment of them and the failure to thrive, this is not easy. However, it does appear to have been possible to teach the children to ingest adequate nourishment and/or fluids orally. This is certainly the case if the swallowing function is intact and satisfactory treatment for airway problems has led to a greater appetite. Most children are interested in food and it is important to react well to the signs given by the child.

Because of the invasive treatments, the mouth and throat areas are often sensitive and there is resistance to oral feeding. With involvement from a pre-speech therapist and a dietician, an individual treatment plan can be devised which may need updating and revision as the child grows and progresses.

At the beginning of the chronic phase, most children are fed or receive supplementary nourishment via a feeding tube, sometimes through the nose. Insertion and care takes place in accordance with the guideline {9}. A PEG tube 11, possibly followed later on by a MIC-KEY button, will be placed if it appears that the child is unable to ingest sufficient nourishment and liquid via the oral route [carer consultation]. Some children in the US have undergone surgical procedures on the area around the stomach (fundoplication) due to serious reflux problems. The upper part of the stomach is then wrapped as a kind of sealing ring around the esophagus wall. The guidelines for reflux are applicable here {20}.

If, after a number of years, the ingestion of nourishment and/or liquid does not succeed via the oral route, it is possible to have special eating training [38] in a specialized rehabilitation centre. For one of the children with MSS, this training has been successful [carer consultation]. Obviously, when considering the possibility of this treatment, the child's individual situation is always taken into account.

In the end, it is not always possible for all the children to ingest enough nourishment and fluids via the oral route. Some children remain dependent on partial or full nourishment and/or fluids via a PEG tube/MIC-KEY button for the rest of their lives [carer consultation].

3.3.2.7 Neurological characteristics

Hypotonia

Children with MSS have a greater tendency to drool, due to the combination of hypotonia in the face, an abnormal jaw position and inadequate mouth closure, posture, reduced awareness of drooling and less frequent swallowing.

It is worthwhile checking to see if saliva loss can be influenced by speech therapy treatment. This should not only involve the refinement of motor activity in the mouth, but checks should also be made to see if the child can be made aware of saliva loss and learn how to deal with excess saliva and swallow it in an efficient way. In the case of moderate to severe drooling that cannot be adequately treated by therapy, saliva-reducing therapy (such as Botulinum Toxin A injections) or a surgical procedure may be suggested. For this decision to be made, a child can be registered with a multi-disciplinary 'drooling' team. In the Netherlands, this is possible in Groningen and Nijmegen.

Stenosis

Spinal stenosis occurs in a number of children, in combination with scoliosis and/or kyphosis. As far as we know, there are no typical signs of loss of neurological function with stenosis, not even in the case of severe stenosis [carer consultation]. Treat-

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ment in the form of a surgical procedure is possible, but does involve a degree of risk [34], (for treatment options see 3.3.2.4).

Seizures

At present, it is not completely clear whether epilepsy occurs in children with MSS or not. While asleep, some children do experience episodes similar to those of epilepsy which sometimes appear to be related to breathing problems and sometimes not. Most children are not treated with medication for epilepsy. Too little is known about this to offer advice for treatment [carer consultation].

Sleeping

Respiratory problems and obstructive sleep apnea are frequent causes of disturbed sleep, but even with effective treatment, a poor sleep pattern is common. The cause is unknown, but sleep problems are often recognized in individuals with learning difficulties. Melatonin is often used to treat sleeping disorders in people with an intellectual disability [39], and some children with MSS have had some benefit from Melatonin.

Toilet training

Only some children with MSS have been toilet trained or have been trained to go at certain times of day. Due to a serious lag in development, most children are completely incontinent and toilet training does not appear to be possible [carer consultation]. However, when the children reach the developmental age of three/four, it is wise to organize structured training [40] at a behavioral therapy training centre.

3.4 Treatment, training, rehabilitation and care-related prevention: physical and mental development

3.4.1 Sensory development

In children with MSS, it is important to pay attention to the processing of sensory information from the moment of birth. The sensory development of touch, movement and balance form the basis of development. Touching is the first experience between the child and the outside world and this forms a basis of trust. [41] Making a conscious effort to carry, nurse and touch the child can strengthen trust. It is always important to give these children time to process sensory information and this means that parents and carers have to adapt themselves to the child. It is also important to find out about aids and methods that can be used to enhance the processing of sensory information in the child (see 3.4.2). The guideline for treatment-oriented diagnostics in people with profound intellectual and multiple disabilities {1} is of great value in identifying the degree of development and possible disturbance. A child-rearing theory for people with PMD [42] and the “systemization of experiences” theory [18] are important supplementary aids in drawing up a personalized individual care and development plan for a child with MSS (see 3.4.4 and 5.1.3).

3.4.2 Cognitive development

It is of importance to the child that his/her own potential with regard to cognitive development, which will always be limited, and particularly his/her potential to communicate are examined. Com-

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munication is important for cognitive development (see 3.4.4). The Total Communication method can serve as an important guideline in this for diagnostics and treatment. Under daily supervision, some children learn to develop themselves with the aid of (supporting) gestures, signs and pictograms, while others are able to use a talking computer with pictograms or limited spoken language (see 3.4.4).

A method such as 'Just Experience It' can be an important aid in the daily care and development of children with MSS [19]. During the care of the child, this method can stimulate the development and cognitive development in the area of sensory-motor, senses and communication, based on specific themes in the child's surroundings [18](see 3.4.4).

3.4.3 Motor development

Interplay between sensory, motor and cognitive development

For motor and (limited) cognitive development in MSS, interplay with sensory development is an important principle. The senses enable children to observe the world around them and form the basis of sensory-motor development, where movement and experience through the processing of sensory stimuli go hand in hand. Physical exercises are important for motor development. However, due to the abnormal sensory perception, physical exercise alone will not be appropriate for children with multiple disabilities. It is advisable to examine the sensory-motor development of the child and to stimulate him/her using an individually tailored sensory-motor approach and possibly a sensory-motor information processing therapy. 14 The sensory-motor development learning pathway is appropriate for these children and can be employed at a day centre or school. It is obviously important to also connect this to the home environment.

Gross motor skills

Under the supervision of parents and a pediatric physiotherapist, many children with MSS learn the gross motor skills such as turning over, sitting, crawling, reaching and possibly standing up independently. Stimulation of balance is ultimately important for learning to walk by holding hands or walking independently. Some children with MSS do achieve this. Activities that can support the development of gross motor skills include: supporting and challenging the child in its own movements, movement on an air cushion, rocking, swimming, playing with a ball, adapted gymnastic exercises, and sensory therapy. These activities often form part of a psychomotor or sensory-motor therapy. The children can sometimes also learn to ride a tricycle or on a tandem bicycle (supervision always recommended) [carer consultation].

New issues relating to balance and walking often arise at a later stage due to the scoliosis, kyphoscoliosis and hip problems [6] [33]. X-ray examination of the hips and specific exercises taught by a pediatric physiotherapist are indicated to help retain the walking function as much as possible [carer consultation].

Fine motor skills

The support of an occupational therapist is recommended for the development of fine motor skills and for the oversensitivity experi-

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enced by many of the children. The work of the occupational therapist is based on a sensory-motor approach where, for example, hairbrush therapy is very effective in the treatment of oversensitivity to touch. Playing with sand, soft clay, shaving foam and various hard/soft materials and swimming and playing in the water also help the child in the development of fine motor skills. Some children learn fine motor skills such as cutting and pasting, which mean that simple arts and crafts activities are within reach.

Due to the severity and complexity of the disorder, the professional supervision of a multidisciplinary team is indicated. A rehabilitation specialist, a pediatric physiotherapist, a speech therapist and an occupational therapist (preferably specialized in sensory-motor development) coach and treat the child and the parents. Cooperation and an exchange of information between these professionals about the problems and progress of the child are of great importance. It is therefore preferable to combine the specific training and treatment for motor stimulation with daily activities at a specialized day centre/treatment centre. Children with profound multiple disabilities learn the most if coaching and specific treatment for motor development are integrated within the daily activities and care routines. Motor stimulation can therefore be specific and effortless at the same time. It can be integrated within the daily care routine, becoming a continual process both in the professional setting and at home.

3.4.4 Social-emotional development

MSS and profound multiple disabilities (PMD)

A large number of factors determine the social-emotional functioning of children and young people with PMD, including the physical condition and communication between those directly involved. In children with PMD, the focus often lies on the physical problems, but problems relating to the social-emotional development also play an important role in daily life and in the care of a child with PMD [43]. In children with PMD, it is important to carry out a good diagnostic evaluation ({1}[43]); also particularly focusing on social-emotional functioning and adapting the approach to and the treatment of it. What does this mean in the coaching and treatment of children with MSS? Children with MSS display moderate, and profound to very profound deficiencies in the area of social-emotional development, cognition and adaptive functioning. The children often function at a PMD level or just above. A stable state of health is obviously an important prerequisite in being able to develop. In the case of MSS, breathing and nutrition are of particular importance to a stable state of health. In addition, it is important to determine and evaluate the level and development of the social-emotional, cognitive and adaptive functioning at regular intervals, so that treatment plans for the child's individual level of development can be drawn up ({1}[43]).

Methods

Vlaskamp's method [42] offers a framework for an individual coaching plan for each child and also recommendations for a daily program. Furthermore, the specific details of work objectives give

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therapists, coaches and parents the necessary tools to support the child in his/her development. The method is based on the assumption that a child with profound multiple disabilities does have the potential to develop him/herself.

The ' systemization of experience' method [18] is a theory for people with a disability based on the development phases of a child. The phase in which a child finds him/herself determines how he/she systemizes and processes experiences. In children with an intellectual disability, these phases are similar to those experienced by children developing normally. The 'systemization of experience' theory is based on the extent to which and the way in which stimuli are experienced and demonstrates four methods of systemization which all work together. In the case of a disturbance of information from one of the systemizations, a response is needed from the person him/herself or from his/her environment. In the case of a child with MSS and PMD, the response from parents and coaches is of importance. In the treatment of the deficiencies and in all other daily activities, it is important to use the experience level of the children as a basis for coaching and treatment.

Communications

The children with MSS usually express themselves in a friendly manner and appear to be happy. There is a certain level of reciprocal interaction, which makes communication possible. The sensory disposition of the children is an important opening for the stimulation of contact and communication. In most children with MSS, there are delays in language development. They do not learn to speak with words but they all communicate non-verbally and with sounds, such as pointing and non-verbal vocalization. Examples of this are: wanting something by reaching for it or pointing at it, not wanting something by pushing it away and laughing in a funny way. Good use of the Dutch Non-Speech Test (NNST) can be made by the speech therapist to identify the level of understanding and the development of language and communication. It is recommended to make use of a method of communication. A method such as "Total Communication" is a good starting point for assessing the child's ability to communicate and the proper coordination of the child's capabilities ([18][19]). The underlying principle of the theory is that everything that the child does is a form of communication; a facial expression, listening, pointing, making a gesture and talking. With this system, therapists use gestures, signs, photos and pictograms, in addition to spoken language. A talking computer with pictograms is a good communication option for children who are able to learn how to use this.

If the child's immediate surroundings offer sufficient stimuli that are specific and inviting, most children will engage in mutual, interactive dialogue and will be able to continue in their development. The child will gradually learn to communicate actively in the present, also without speech.

Behavior

The children usually have a friendly and positive attitude and enjoy social interaction with people they know. They display very little inappropriate behavior. The children are, however, very single-minded and sometimes behave in an obsessive way towards people or objects, sometimes displaying stereotyped and repetitive behavior

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([20][carer consultation]). The lack of communication skills, the limitations in social interaction and the lack of reciprocal communication and social skills could be compatible with the definition of a disorder in the autistic spectrum [20]. Any problematic behavior that arises may be caused by a medical problem that has not yet been identified. It is therefore important to investigate this and make use of the literature available. 17

3.4.5 Psychosocial coaching of the child and parents in the acute and chronic phases

In the acute phase, it is very important for the child to be in quiet, safe surroundings with plenty of contact with the parents, in spite of the fact that circumstances surrounding a hospital stay make this difficult. In the acute phase (but also in the chronic phase) it is crucially important that parents receive enough information and that they are able to stay as close to their child as possible (see 2.6.3 and generic theme Psychosocial Care with Rare Disorders).

Nursing care according to the Newborn Individualized Developmental Care and Assessment Program (NID-CAP), which is already employed in many Dutch hospitals for the care of premature babies, is indicated for babies with MSS. Good medical and technical procedures and good psychosocial care focussed on safety, observation and coaching are of great importance to both the child and the parents. This requires an appropriate attitude by both doctors and nurses, important aspects of which include empathy, respect, trust and showing initiative.

The development of a Post Traumatic Stress Disorder (PTSD) is a great risk for both the parents and the child. Symptoms of PTSD include sleeping disorders, fear and re-experiencing, as well as anxiety for no apparent reason. EMDR therapy 18 is a possible treatment for this, although not in combination with sensory integration therapy. One child successfully underwent EMDR therapy for PTSD, which was a result of intense medical treatment during the acute phase [44].

It is important for the development of the child to be strongly attached to his/her parents and has a preventive effect on PTSD. Involving the parents in the care of the child also produces a preventive effect on PTSD, because they feel that they are actually able to do something for their child instead of being a helpless onlooker [23].

During both the acute and the chronic phases, it is very important for people to be alert to the psychological tolerance level of the parents. This is the task of the case manager, who should, in consultation with the parents, enlist the aid of professional psychosocial specialists if necessary (see chapter 5 for more details).

3.4.6 Premature death and palliative care

MSS is a chronic, progressive disease with complex complications. When the diagnosis has been established, the child and the parents are always immediately confronted with the possibility of premature death. This is due to the child's symptoms, the severity of the syndrome and publications, which up until 2010, almost always refer to death in early childhood. Nowadays, we know that

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survival rates during childhood are good, due to the satisfactory treatment of serious breathing problems. Even so, it is still the case that some children die every year due to possible complications arising from MSS.

It is important to be able to talk about the possibility of premature death. During such a discussion, parents can express their feelings and worries and the professional can provide them with the proper information, partly to give reassurance but also to be able to continue with the dialogue. At the same time, it can be established what treatments the parents would like and would not like for their child and where they would like to draw a line as far as treatments are concerned, such as non-resuscitation or non-treatment policy. Any established protocol should be revised each year.

More and more attention is being paid to palliative care in the healthcare sector. This is not the same as end-of-life care and can be started earlier in the process of a life-threatening illness. As an approach, palliative care is particularly focused on the improvement of the quality of life for the patient and his/her environment, when the symptoms of the life-threatening disease become more serious (see also the national care module Palliative Care). Palliative care for people with profound multiple disabilities requires a specific approach. The guideline 'Palliative Care for People with Intellectual Disabilities' {16} offers tools for this and the handbook 'Palliative Care for People with Intellectual Disabilities' can be a good starting point for a discussion with parents about palliative care for children and young adults with MSS (see also 4.0).

3.5 Social and Societal Participation

The individual with MSS

Children and young adults with MSS are socially engaged and enjoy having people around them and taking part in activities. They enjoy being with people (always under constant supervision) and do not appear to be restricted by their unusual appearance. They like to explore the world around them; this varies from exploring the surroundings at home or close to home when they are small to going on shopping trips, visiting a playground, zoo or theatre performance when they are older. The children often like playing rough games and enjoy travelling on a train, bus or plane [carer consultation]. However, due to physical restrictions, this is not always possible and parents cannot always manage it. It is important to allow and give the children and young adults a place in society, whether this is as a family member at home, a pupil at school or day centre or during trips into the outside world: it gives them a sense of purpose and meaning to their lives [carer consultation].

MSS has an effect on many areas of the child's/young adult's daily life; personal care, daily activities, social contacts, the ability to cope independently and mobility. With the help of those around them, the child and later the adult will adjust to life with a chronic, progressive illness and the restrictions that come with it. In doing this, the child and the parents will come up against the prejudices of society, which is not sufficiently oriented towards people with disabilities. The children are meaningful to others, precisely because they confront people and encourage them to think and reflect.

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Parents

For parents, it is a challenge to expose themselves to their own environment and the outside world with a child with multiple disabilities. Reactions from the people around them have a great effect. Parents are often more upset by rejection than the child him/herself and it is very difficult for parents to deal with this.

Many parents take on a great deal of the care themselves at the expense of paid work. [carer consultation]. In the case of paid work, possibly part-time, it is important for employers to be supportive and flexible towards the parents.

For most parents, it is very important for them to be able to share their experiences during and after the acute phase with their network, such as family, colleagues, friends and acquaintances. It is important for parents to know that they can contact people in a similar situation, the case manager (see chapter 5) and other healthcare professionals.

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4.0 Generic care

The first chapters of this care standard describe the disease specific care. These chapters refer to generic care themes for rare disorders that have been developed by the VSOP (Dutch Association of Parent and Patient Organizations). These care themes and the national generic care modules can be part of the generic chapter of a care standard. A generic module describes a generic component in the care of chronically ill patients. A generic care theme describes a generic component in the care for rare diseases. These care themes are brief descriptions of the care and information provision for and to people with rare diseases and their next of kin, focusing on a number of themes. The themes have been developed according to the format for generic modules (as much as possible) [4] and contain the indication criteria, information about treatment/counselling and quality information.

Within the MSS care continuum, the following generic care themes apply:

- Communication and information (disease specific information in 5.2.1.1, 5.2.1.2, and 5.2.2.4)
- Pharmaceutical care (under construction)
- Genetics (under construction)
- Psychosocial care (applicable to parents/caregivers, disease specific information in 5.2.1.2 and 5.2.2.3)
- Registrations/patient registers (disease specific information in 5.2.2.6)

Within the MSS care continuum, the following completed national generic care themes apply:

- Care module Palliative care in 3.4.6 – Care module Nourishment in 2.1.7, 3.3.1.4 en 3.3.2.6


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5.0 Care pathway structure

MSS is a multi-system syndrome. Care for an individual with MSS requires a proactive, multi-disciplinary approach and timely, well-coordinated communication between the caregivers and the parents/carers of the patient, and between caregivers. This is the case for both the acute and chronic care phase. This chapter describes the generic characteristics (5.1) and the disease specific characteristics of the care pathway (5.2).

The description of the disease specific care pathway is aligned to the disease specific care phase (see 2.5): it first looks at the integrated care pathway for the acute care phase. Next, the chapter will describe the care pathway of the chronic care phase. The chronic phase is characterized by an integral care network, consisting of a number of elements, including the daily care and hospital care. The description of the care pathway in this chapter will discuss the following aspects:

Figure 4. The care during the acute care phase is called 'integrated care': its elements follow each other progressively. The chronic care phase is characterized by an integral care network: coordinated multi-disciplinary care and support.

- Elements of the integrated and integral care
- Partnerships for integrated and integral care (content cooperation, coordination, communication, agreements, responsibilities)
- Selection options for the different elements of care, for parents/carers and for patients

5.1. Independent characteristics of the care standard

5.1.1 Responsibilities of the multi-disciplinary team

The care plan, both during the acute and the chronic phase, is characterized by an adequate division of the responsibilities across the multi-disciplinary team [45]. This document refers to the *'Handreiking Verantwoordelijkheidsverdeling bij samenwerking in de zorg'*, developed by the KNMG, in cooperation with nine other care professional and industry organizations, for advice on good organization of and cooperation in patient care, and for a better insight into what a patient can expect. This support document deals with both the simultaneous and the progressive cooperation between caregivers. This support document names and discusses thirteen focus points that are important in the division of responsibilities across the care-team. Appendix 2 contains a summary of these focus points.

Tasks and responsibilities of the caregivers are part of the function/task package, and the related responsibilities as they have been determined by the organizations where the caregivers work, or are connected to by way of their job position.

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5.1.2 Case manager

Contrary to the format of care standards [1], where the main caregiver is the first point of contact and coordinator for the team, but also supports the patient's self-management, this care standard appoints an additional case manager [46]. The reason for this is that in the case of MSS there is no patient self-management, and so the main caregiver must support all their needs [47]. The tasks of the case manager differ for each phase of MSS, and are described in sections 5.2.1 and 5.2.2.

5.1.3 Individual care plan

An individual care plan is [6] required for each type of care that continues more than three months[48]. The goal of the individual care plan is to determine, with the patient, the care requirements for the patient, and to monitor and adjust these needs if necessary. In case of MSS, there is an integral individual care plan that is called an 'individual care and support plan' (see 5.2.2.5).

There is no individual care and support plan for the acute phase of MSS: in this phase, the care is aimed at eliminating the life-threatening symptoms and complications, and stabilizing the health of the newborn. However, an individual care and support plan is required for the chronic phase (see 5.2.2.5).

5.2 Dependent characteristics of the care standard

5.2.1 Acute phase

The care during this phase is directed at eliminating any life-threatening symptoms and complications, and stabilizing the health of the newborn (see 2.5). In this phase, the correct diagnosis is often lacking.

5.2.1.1 Description of the integrated care

This phase is characterized by integrated care across the consecutive events from delivery to hospital discharge. Participants in the integrated care during the acute phase are all caregivers that are involved in the delivery and the subsequent hospital treatment (for example: midwife, family doctor, emergency room, regional ambulance (RAV), hospital/obstetric team). One key transfer moment takes place right after the delivery (at home or at the hospital) when the newborn is transferred to specialist neonatal care[7].

6 The Coördinatieplatform Zorgstandaarden (CPZ, coordination platform for care standards) has developed the Raamwerk Individueel Zorgplan (Framework Individual Care Plan). This document is a guideline for the development of individual care plans, aiming to improve the uniformity in format and principles (www.zorgstandaarden.nl/activiteiten-platform/lopende-projecten/raamwerk-individueel-zorgplan/).

7 During the hospital stay, several internal transfers can be required (e.g. from the ER to the IC, from the IC to the Neonatal department). The parents should be informed about each internal transfer moment and the relevant practical information (floor number, room number, visiting hours, accessibility, regulations, etc.)

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The organization and make-up of the acute obstetric care must comply with the current quality standards [49][50]: The hospital will immediately initiate the appropriate interventions (also called parallel actions) [49] by appropriately qualified obstetric professionals [51]. The obstetric professional will bring in the team, including a pediatrician, gynecologist, anesthesiologist, nurse and possibly other professionals and/or the surgical team. The pediatrician is the child's main physician. The main physician is responsible for the treatment(s) during the entire hospital stay, and has the ultimate responsibility for overall clinical management of the child [46].

During the acute care phase, it is important that the parents are informed of the required interventions and the current health condition of their baby. Ideally, the parents are not separated from their newborn, unless absolutely medically required. This is a task for the case manager (see next section, Case Manager). In case of an acute life-threatening situation, the case manager must inform the parents as soon as possible about the options for resuscitation (and any resulting complications/risks). The case manager will actively ask for the parents' wishes regarding resuscitation; these will be taken into consideration by the multi-disciplinary team.

It is desirable that at least one of the parents stays in the hospital with the newborn throughout the stay.

While adequate care is provided according to the current guidelines (see 3.3.1), the main physician will simultaneously take steps to determine the correct diagnosis. Because MSS is an ultra-rare disease, it is unlikely that the correct diagnosis will be recognized straight away. The pediatrician will - depending on his/her knowledge and experience - contact one or more expert colleagues here or abroad, and/or consult international databases about the symptoms and signs of rare diseases [33]. If the main physician's actions do not lead to (at least a temporary) diagnosis during the child's hospital stay, they will refer the patient to a colleague expert in the field of ultra-rare disorders.

After the health status of the newborn has been stabilized, and no further acute complications are expected, the pediatrician will decide to discharge the patient from hospital. Prior to discharge, there should be a final meeting at the hospital, with the parents, the main physician and the multi-disciplinary team. This meeting is intended to inform the parents about:

- the medical history to date, with the procedures required and any ongoing complications,
- the current medical condition of their infant,

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- the (preliminary) diagnosis (name and explanation of the clinical picture, the course of the disorder, possible treatment options, prognosis)

During this final meeting, the parents will receive the final report on their newborn (including the above mentioned information) and the following practical information:

- instructions and information about the care for the newborn at home during the first period after release from hospital,
- name and contact details of the caregiver(s) (district team, local nurse, pediatric nurse) that are available to support the parents from the day of discharge from the hospital,
- name and contact details of the case manager, and information about how and when this person can be contacted (office hours, after office hours, by phone, etc.),
- instructions about when the ER must be contacted,
- day and time of the follow-up appointment at the hospital or a specialist clinic,
- if referred to a specialist clinic, the parents will also receive the name, address, phone numbers of the clinic and the details of the expert specialist at that clinic,
- if referred to a specialist clinic, the transfer procedure should also be described.

The generic care theme *Communication and information* describes the generic communication and information aspects for rare conditions that may be applicable for the above mentioned points.

5.2.1.2 Case Manager

Ideally, the main physician will take on the role of case manager during the acute phase. If this is not possible, another member of the team, preferably the child's primary nurse (see paragraph one of the previous section "Description of the integrated care") will assume this role. As soon as there is no (longer) acute life-threatening danger for the newborn, the case manager will take care of the following during this phase:

- escorting the parents to the post-natal ward or other accommodation/quiet-room, and also offering support to the parents.
- Ensuring that the mother, who has just given birth, receives the appropriate post-natal care.
- informing the parents in detail about any new developments in the status or progress of the surgery/treatment of their baby.
- verbally informing parents about any further required (medical) treatment.
- informing the parents of (any) benefits and risks of any proposed subsequent treatment.
- asking the parents about their preferences and taking these into consideration during the decision-making process about (any) required subsequent treatment.

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- asking the parents about their preferences regarding ventilation, resuscitation and possible end-of-life decisions that may come up during the stay at the hospital.
- asking the parents if they need spiritual support (religious or secular counsellor), and if so, initiating such support.
- providing the parents with written information about hospital procedures for the use of medication, anesthesia and analgesia for newborns.
- providing parents with written information about incident protocols as a result of the use of medication, anesthesia and analgesia for newborns.
- providing written information about any complaints procedures (related to medical treatment, complication, personal treatment, etc.) at the hospital.
- informing the parents about the date and time of the final meeting with the main physician and the multi-disciplinary team (see above) as soon as this information is available.
- creating a list of practical information that is handed to the parents during the final meeting.

The generic care themes *Communication and information* and *Psychosocial care* will describe the generic communication, information and psychosocial care aspects for rare disorders that may apply to some of the above mentioned points.

5.2.2 Chronic phase

The chronic phase of the care starts at the discharge after the 1st postnatal inpatient stay, and continues for the rest of the patient's life. If no definitive diagnosis was made during the acute phase, this should take place as soon as possible during this phase (see 3.2).

The chronic phase of MSS includes, along with medical care (see 3.3.2), also daily support, teaching guidance and education. The chronic care phase is not just aimed at stimulating the growth and development of the baby/child and the treatment/prevention of any symptoms or complications, but encompasses all elements of life: improving the quality of life and, depending on the individual needs, improving the mental and behavioral development, social participation and developing meaningful daily activities. This phase also includes the transition from pediatric care to adult care (see 5.2.2.3). These common objectives are realized through an integral care network that is defined as: ''Care that is focused on the individual needs of the patient, with professionals from different fields or different organizations aligning their activities, creating a joint plan for the care and support of the patient.''

Participants in the integral care network in the chronic phase are all caregivers, educational employees and other professionals involved in the care, education and daily activities of a child with MSS after the first post-natal hospital stay (see 5.2.2.2).

5.2.2.1 Focus on the individual

MSS is a complex system disorder with severe mental, intellectual and physical limitations. The term Severe Multiple Disabilities (SMD) is often used for this disorder. Although there are a number of main characteristics for the disorder (see 2.1 and 2.2),

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every individual suffering from MSS has a unique combination of symptoms, physical disabilities and cognitive level, as is the case for most syndromes [53]. The disease burden differs per individual, resulting in very varied care needs (see 2.6). The severity of the different disabilities only becomes clear over the course of the first years. This means that the organization of the long-term care for MSS patients is a very complex, dynamic and individually determined process.

The nature of the disorder and the diversity of care requirements lead to a very diverse integrated care network for MSS, with a great diversity in elements and structure. In other words, there is no such thing as “a single integrated care network”. That is why this care standard has decided to describe some core elements of the integrated care network and its organization. The reformation of the long-term care system was set in motion some time before 2014 [54]. That is why we’ve decided not to place the care pathway for the long-term care for MSS patients within the most current Dutch policy setting. Instead, we’ve decided to focus on the main care needs, and the elements of the integrated care network and the structure of this network.

5.2.2.2 Elements of the integrated care network

Children with MSS require daily care. Over the years, they will have to undergo one or more hospital treatments, they will require regular care from the family doctor, and they will all need special education, focused on children with (multiple) disabilities, often in the form of day-care. Once they reach adulthood, MSS patients are unable to live or spend the day without supervision. The allied health disciplines are part of the integrated network care. Based on these core care needs, a child with MSS will encounter the following types of care and support:

- **Daily support and care**
  Children with MSS will either live at home or at a specialized residential care facility, where they are cared for and supported on a daily basis. In some cases, the child will first live at home, and when he/she reaches adulthood, the patient will transfer to a specialized care facility with residential units and facilities for daytime activities. Depending on the situation, the daily care of the child with MSS is performed by the parents at home, or by caregivers in a residential care facility. If the child lives at home and the parents are fully responsible for the daily care, structured support of other caregivers is advisable. In an ideal situation, the parents will receive help from professionals such as nurses that can offer structured help with nursing and caring for the child. The Dutch healthcare system offers parents temporary relief from the heavy burden of care. This is called ‘respite care’ or attendant care support[8].

- **Hospital care**
  Children and adults with MSS require specialist care that is provided in hospitals. The following disciplines play a role in

8 https://www.regelhulp.nl/zr/rh/webapp/kennisbank/Informatie/Logeeropvang

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the treatment of a child or adult with MSS: pediatrics (child heath), AVG (adult health), ophthalmology, neurology, ear-nose-throat (ENT), cardiology, anesthesiology, radiology, orthopedics, internal medicine, specialized dentistry, clinical genetics (see also chapter disease specific care).
Ideally, at the end of the acute care phase there will be a decision on which hospital and main physician will be responsible for the care of the child with MSS in the future. To maintain continuity and high quality care during the chronic care phase, it is important that the patient has a 'regular' hospital and main physician. The case manager is also a crucial element of care.

- **Family doctor care**
  Mild symptoms in a child or adult with MSS can be dealt with by the family doctor. When the family doctor suspects more serious issues, or the risk of complications, he or she will refer the patient to a specialist at the hospital.

- **Allied Health Professional care**
  Both children and adults with MSS might require treatment from a range of allied health professional care disciplines: speech therapy, podiatry, orthoptics, educational therapies, occupational therapy, dietetics, physiotherapy and (behavioral) psychology. If (some of) these disciplines are not available from the day care center/specialist care facility, the parents/carers can enlist the required care themselves.

- **Special education/ day care/ rehabilitation**
  Around the age of 18-24 months, a special educational specialist will chart the development of a child with MSS, to determine the most suitable support for the child.

Children with MSS who live at home can, like other children with multiple disabilities, visit one of the following types of centers from the age of two: the specialized (nursing) day care centre, the special education day centre or the child rehabilitation centre. Children who live in a specialized facility will attend the day care centre that is connected to/part of the care facility.

A day care centre for children with multiple disabilities will group children together as much as possible based on age, like the general education system. The age and individual ability will determine which group best suits the child's potential.

Some children with multiple disabilities will stay at the child day care centre until they are 18. Others will progress and transfer to the special needs school system, the so-called tyltyl schools (Dutch system). These schools focus on children with severe physical disabilities and moderate to severe mental disabilities. The tyltyl school is often linked to a mytyl school; the latter offers education to children with severe physical disabilities. The tyltyl school, mytyl school and the school for children with serious learning disabilities (ZMLK in Dutch) are all part of the 'special education cluster 3' category of special education.

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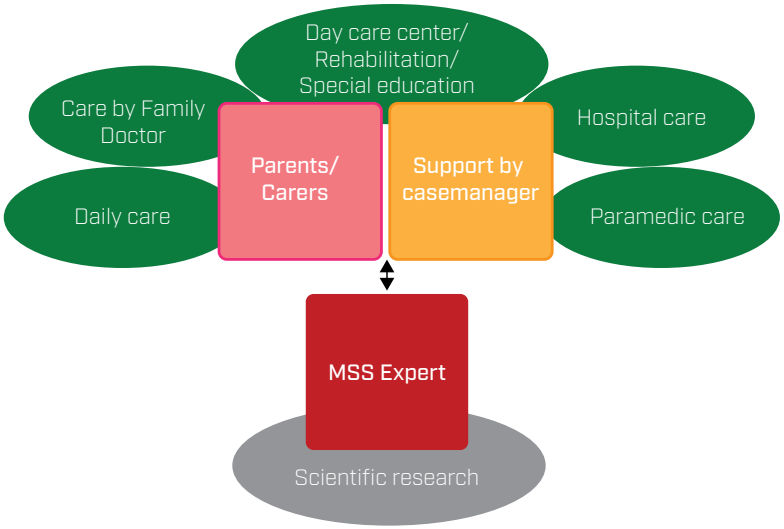
Figure 5. Diagram of the integrated care network. The support from the case managers for the parents/carers can be intense or moderate, and can occur in different forms, depending on the needs/wishes of the parents/carers. This support enables a fitting, efficient and timely delivery of integrated care. The expert is consulted for the creation/adjustment of the individual care and support plan, and he/she is invited by the parents/carers to attend the periodic meetings.



Between the ages of 18 and 20, many adolescents transfer to a day activity centre (which may or may not be part of an adult rehabilitation centre). These children will be prepared for their eventual transfer to a day care centre for adults.

5.2.2.3 Focus points for (designing) an integrated care network

The creation of an integrated care network for a child with MSS will take some time. After infancy, once the child starts visiting a specialist day care center, the need for integrated care will increase. By then, parents/carers will already be very experienced. During the creation of an integrated care network, it is important to map out the care that can be provided by the parents/carers (see also generic care theme Psychosocial care). The 'Mantelscan' is an instrument that can map out the informal care network (mantelzorg in Dutch) for someone in need of care; this can estimate the risk of overloading the informal caregiver(s). The Mantelscan document explains all the elements of the 'mantelscan test' [55]. This was created for every care professional who deals with care networks and informal caregivers. In case of informal carers for people with MSS, the scan should be performed frequently: the care requirements for people with MSS and the work load ability of informal carers can vary from time to time.

Transition from pediatric to adult care is another important element that is often considered a bottleneck by parents. The continuity of care requires a smooth transition from pediatric to adult care. Regarding the hospital care, the transition should be gradual; the knowledge and tasks of the pediatrician should gradually be transferred to the new main physician, a specialist in adult mental retardation/intellectual disabilities (in Dutch: AVG; a separate specialty in the Netherlands). The transition should take place in consultation with the parents/carers and the case manager {19}. With respect to special education / day care / rehabilitation, it is impor-

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tant that the transition is also monitored by the special education specialist, as well as by the parents/carers and the case manager.

5.2.2.4 Parents/carers, the case manager and the MSS expert

The parents/carers

Many parents want to play a strong managerial role, whilst others find it easier to delegate these roles and the tasks that come with it. The tasks in the care network are shared after consultation with the case manager, and the parents will typically:

- Remain the manager for the integrated care for their child: they take decisions on behalf of their child about the required treatment, education and (daily) activities, and are responsible for these choices, even after the child becomes an adult [56][57].
- Communicate all relevant information they get from the different care organizations to the case manager and the expert.
- Offer access to their child's (electronic) patient file and the individual care and support plan to the case manager and the expert.
- Remain responsible for the accurate, timely and complete written transfer of information to the case manager and the expert, regarding all current and relevant matters that result from their daily care for their child.

The case manager

Depending on the parents' desire to stay in control, the case manager will fulfil either a marginal or more prominent role in the integrated care network. Below is a list of the possible roles, tasks and responsibilities of the case manager; these can also be performed by the parents/carers, if they wish to take more control. The case manager:

- Is the spider in the web of complex integrated care for the child with MSS.
- Will frequently perform the informal care scan (Mantelscan) and will create the related 'eco folders' (ecomappen) according to the Mantelscan Document [36].
- Has appropriate professional qualifications and is able to advise other agencies involved in the care for people with multiple disabilities[9].
- Has access to different organizations that deliver the different elements of the integrated care network (hospital, emergency care, etc.)
- Has one focal point/contact within each organization/care industry. There are:
  - Hospital care, main physician
  - Family physician care: assistant or the family physician
  - Day activities/ rehabilitation/ special education: daily carer / rehabilitation physician/ special educationalist
  - (Virtual) expertise centre or expertise network: MSS expert
  - Allied Health Professionals: if arranged separately, contact for each service provider

9 See organisation list 2014 in appendix 6.

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- Informs and maintains good relations with all persons involved in the partnership between different organizations
- Coordinates the cooperation between the different organizations that are involved in the integrated care, and is responsible for this coordination.
- Monitors the exchange of data and the information feedback between the different care organizations.
- Is informed about the current care offerings and any changes or developments at different organizations.
- Is informed about all current (national) regulations that can apply to the child with MSS (options for benefit payments, facilities for resources, etc.) and has access to the organizations that can arrange/take care of these options.
- Ensures that the right care and arrangements are delivered and made, at the right time, and by the right organization.
- Ensures continuity of care, including the transition from pediatric to adult care.
- Monitors the quality of the care process.
- Is responsible for the correct, timely and complete transfer of written information to the parents/carers about any current and relevant matters that are the result of the care that is provided by the different care
- Has frequent personal contact with the parents/carers and informs them about any current matters regarding the integrated care network for their child

The MSS expert

- Has had appropriate medical training and specializes in ultra-rare disorders such as MSS. He/she conducts research into the disorder(s) and has published publications in several international scientific journals. He/she is known among colleagues here and abroad as an expert' in the field of MSS.
- May live and work in any country in the world. He/she works at a specialist expert facility, or as part of an expert team at an academic hospital. He/she is closely involved in the (virtual) expert centre for ultra-rare disorders (see 5.2.2.6).
- Is accessible to and available for the parents/carers. The expert and the parents/carers will agree on the method of communication, the timing and the reasons for communication (for the benefit of the care and support plan, consultation, etc.)
- Will examine the child with MSS at least once every 2-3 years.
- Is responsible for communicating new scientific developments regarding the (natural) course of the disorder and complications, and any subsequent changes in the patient's care.
- Has close ties to the MSS community (see 5.2.2.7).

5.2.2.5 Individual care and support plan

The individual care and support plan for people with MSS is part of the chronic care phase. It is an integrated plan, linked to both the medical and allied-health care, as well as the cognitive and behavioral aspects and the patient's general well-being. All caregivers that are part of this partnership will have a common, current version of this plan. The electronic medical patient file is part of the individual care and support plan. The individual care and support plan consists of four parts: (1) general information (2) tasks

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and appointments (3) electronic medical patient file (4) support plan. This final part is drafted and used at the specialized day care centre, but is part of the individual care and support plan that was made available to the caregivers who have been authorized by the parents. In an ideal situation the individual care and support plan is completely accessible electronically for all authorized persons.

(1) General information

This part of the care and support plan contains the

- personal and demographic information about the individual (name, address, social security number, hospital number, health insurance details)
- name and contact details of the legal representative and the type of representation (judicial administration, receivership, carership, other[38])
- name and contact details of the case manager, including the availability of this person
- name and contact details of the MSS expert
- address details of the care organizations, names and contact details of the key personnel at these care organizations
- address and contact details of the pharmacy.

(2) Tasks and agreements

This part of the care and support plan contains the

- tasks and responsibilities of all participants in the integrated care network
- consultations between the case managers and the key personnel at the different care organizations
- consultations between the case manager and the parents
- consultations between the case manager, the parents and the expert
- agreements about authorizations for the individual care and support plan and the electronic patient file that includes the individual care and support plan.

(3) Electronic medical patient file

The medical content of the individual care and support plan is the responsibility of the main physician (pediatrician/AVG) and the MSS expert. The information in the medical plan is essentially the content of the electronic patient file, containing the following information:

- medical history of the person with MSS (including details about the pregnancy, delivery, hospital stay(s), treatments, vaccinations, individual needs, etc.)
- current medical status and prognosis for the patient
- mobility limitations
- current use of medical aids/resources
- current medication
- allergies, nutrition and dietary requirements
- future possible (problem) issues/ points of attention
- if treatment is required/desirable:
  - concrete treatment objectives
  - options for treatment types
  - explanation about the use/necessity/follow-up of any type of treatment

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- description of treatment and follow-up care (including duration, frequency, any medication, use of resources, etc.)
- what assessment is required before and after treatment
- consequences and complications of the treatment for other elements of the care and development of the patient (e.g. consequences of back surgery for physiotherapy; use of new device for physical, cognitive development, behavior, etc.)

(4) Support plan

Specialist day care centers often use a support plan (sometimes also referred to as a development plan). This plan is part of the individual care and support plan, and is discussed, assessed and adjusted at the specialist day care center according to their usual practice. The discussion and assessment takes place in the presence of the multi-disciplinary team, the parents and the regular caregiver. The support plan, which is the responsibility of the special educationalist, is kept up to date.

5.2.2.6 (Virtual) expert centre and patient register

MSS is an ultra-rare disorder. The number of people with MSS is so small that to create a dedicated physical center of expertise - even if this is combined with other ultra-rare disorders - is not a realistic goal [52]. A virtual center of expertise offers the solution for the most significant bottlenecks in the care for patients with MSS: correct diagnosis, knowledge about the natural course of the disorder and knowledge of (possible) complications.

A virtual center of expertise for a group of ultra-rare disorders is a desirable development. The creation of such a centre can only be realized if we join forces, on an international level. The virtual center of expertise can be built around an international patient register. If such a center is realized, this would allow registered physicians digital access to the centre, and it enables the uploading of standardized and anonymous and/or indirectly identifiable data and images (with the consent of the parents/carers/patients). Other registered physicians can also consult the database. Advanced search options and matching algorithms are part of the virtual center of expertise, enabling an accelerated diagnostic process. The database also contains case studies and descriptions of the natural history of the disorders.

5.2.2.7 MSS community

Nowadays (2014) the MSS community is a well-organized, active group[10], consisting of families from all parts of the world where patients have been diagnosed with MSS. The Facebook group of family members of MSSRF registered families contains stories, experiences and questions. The group is available in multiple languages, using an on-line translation program. In cooperation with international experts, the Wiki page is also kept up to date with current scientific information about MSS, and the community also hosts biannual international family meetings.

10 More information at http://www.marshallsmith.org

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6. Quality information

The chapter about quality information shows the quality indicators that have been developed for this disorder. No disease specific quality indicators have been developed for MSS. The quality information is limited to the quality criteria for the elements and organization of the patient's care. These have been determined from the perspective of the parents/carers of children and adults with MSS (6.1.1.1 and 6.1.2.1). The quality information about the elements and organization of the care also includes the quality criteria/indicators that have been developed by different organizations/professional associations, and that are applicable to the elements/phases of the MSS care (6.1.1.2 and 6.1.2.2).

The quality criteria and/or indicators that are part of the generic themes and the generic module 'Palliative care' are not repeated in this chapter; we refer to the relevant sections in the specific theme/module (see 6.2)

6.1 Disease specific care: quality criteria for the elements and organization of care

6.1.1 Acute phase

6.1.1.1 Quality criteria from the perspective of the parents/carers

Section 5.2.1 gives a detailed description of the elements of the integrated care during the acute phase. Quality information is an integral part of this description, and is summarized below:

- A case manager is appointed (preferably the main physician) who will, as soon as the patient is in no imminent danger, carry out the activities as described in 5.2.1.2.
- The parents are informed of the required interventions and the current health condition of their baby.
- The parents are not separated from their newborn, unless absolutely medically required.
- At least one of the parents stays in the hospital with the newborn throughout the stay. The hospital will arrange for a suitable room.
- A final meeting will take place prior to the patient's release from the hospital. This meeting is attended by the parents, main physician and the multi-disciplinary team.
- During this final meeting, the parents will receive the final report on their newborn, including the practical information from section 5.2.2.1.

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6.1.1.2 Quality criteria and indicators developed by different organizations/professional associations

The following quality criteria and indicators (including a short summary) are applicable:

(1) Indicator Set Acute Obstetrics [49]:
- Agreements about the transfer within the integrated care (structural indicator)
- Periodic meetings with integrated care partners (structural indicator)
- Lead time for allocation trip assignments (process indicator)
- Agreements about parallel actions (structural indicator)
- Incident room has direct number for acute obstetrics (structural indicator)
- Lead time for ambulance service in case of report of acute obstetrics patient (process indicator)
- Presence and training qualified caregivers (structural indicator)
- Neonatal results of the delivery (result indicator)
- Presence and use of instruments for registration of complications and/or incidents (structural indicator)

(2) The first care at the hospital is guided by an obstetric professional who meets the criteria in the policy document 'Nota kwalificaties geautoriseerd obstetrische professionele acute verloskundige zorg' (qualifications authorized obstetric professional acute maternity care) [51]:
- Support during partus: assessment of cardiotocogram, amniotomy, placement skull electrode, micro blood panel, birthing support, placing and suturing episiotomy, assessment of rupture and suture of first and second degree rupture
- Diagnostics and treatment of acute obstetric situation is directed by the first line
- Protocol start of care in acute obstetric situation: shoulder dystocia, prolapsed umbilical cord, ruptured uterus, abruptio placentae, unexpected breech, postpartum hemorrhage, eclampsia and the treatment of severe hypertension
- Resuscitation of the newborn
- Resuscitation of the adult
- Knowledgeable about the organization that is providing the care

6.1.2 Chronic phase

6.1.2.1 Quality criteria from the perspective of the parents/carers

Section 5.2.2 gives a detailed description of the elements of the integral care network during the chronic phase. Quality information from the perspective of the parents/carers is an integral part of this description; below is a brief summary of this information in the form of quality criteria:
- A case manager is appointed; the roles/tasks/activities and responsibilities of this person are described in 5.2.2.4
- The Mantelscan is an annual test to determine the strengths and weaknesses of the care network [55].

- The parents/carers will be provided with the contact details for the MSS experts
- All caregivers that are part of this partnership will have a common, current version of the individual care and support plan.
- The individual care and support plan consists of four parts, as described in 5.2.2.5.

6.1.2.2 Quality criteria and indicators developed by different organizations/professional associations

If applicable to the individual MSS patient, the following symptom specific quality criteria and/or indicators apply:

Hospital care
(1) Indicator set Obstructive Sleep Apnea Syndrome (OSAS) in adults[58]
(2) Indicator set Adenoid and Tonsil Diseases [59]
(3) General quality criteria for the treatment of scoliosis [60]
(4) Indicator set Surgical treatment of lower back herniated disc and stenosis [61]

Paramedic care
(5) Quality criteria 2010-2015 speech therapy [62]
(6) Quality criteria 2010-2015 occupational therapy [63]
(7) Quality criteria 2010-2015; remedial therapy explanation [64]
(8) Quality criteria as formulated in the document "Professional competency profile of pediatric physiotherapists" [65]
(9) Quality criteria as formulated in the document "Professional competency profile of optometrists" [66]

Disability care
(9) Quality criteria for facilities for intensive child care [67]
(10) Content of care indicators in the quality framework for Mentally Disabled Persons [68]
(11) Content of care indicators in the quality framework for Physically Disabled Persons [69]

Rehabilitation care
(12) Collection of indicators 'Insight into Rehabilitation2013"[70]

Education
(13) Quality standard for special education [71]

Nursing, care and home care
(14) Quality document 2012 Nursing, Care and Home Care [72]

6.2 Generic care: quality criteria for the generic themes and modules

The quality information for each generic theme (see 4.0) is described in chapter 4 of the relevant theme. There is no chapter on quality information for the care module Palliative Care.

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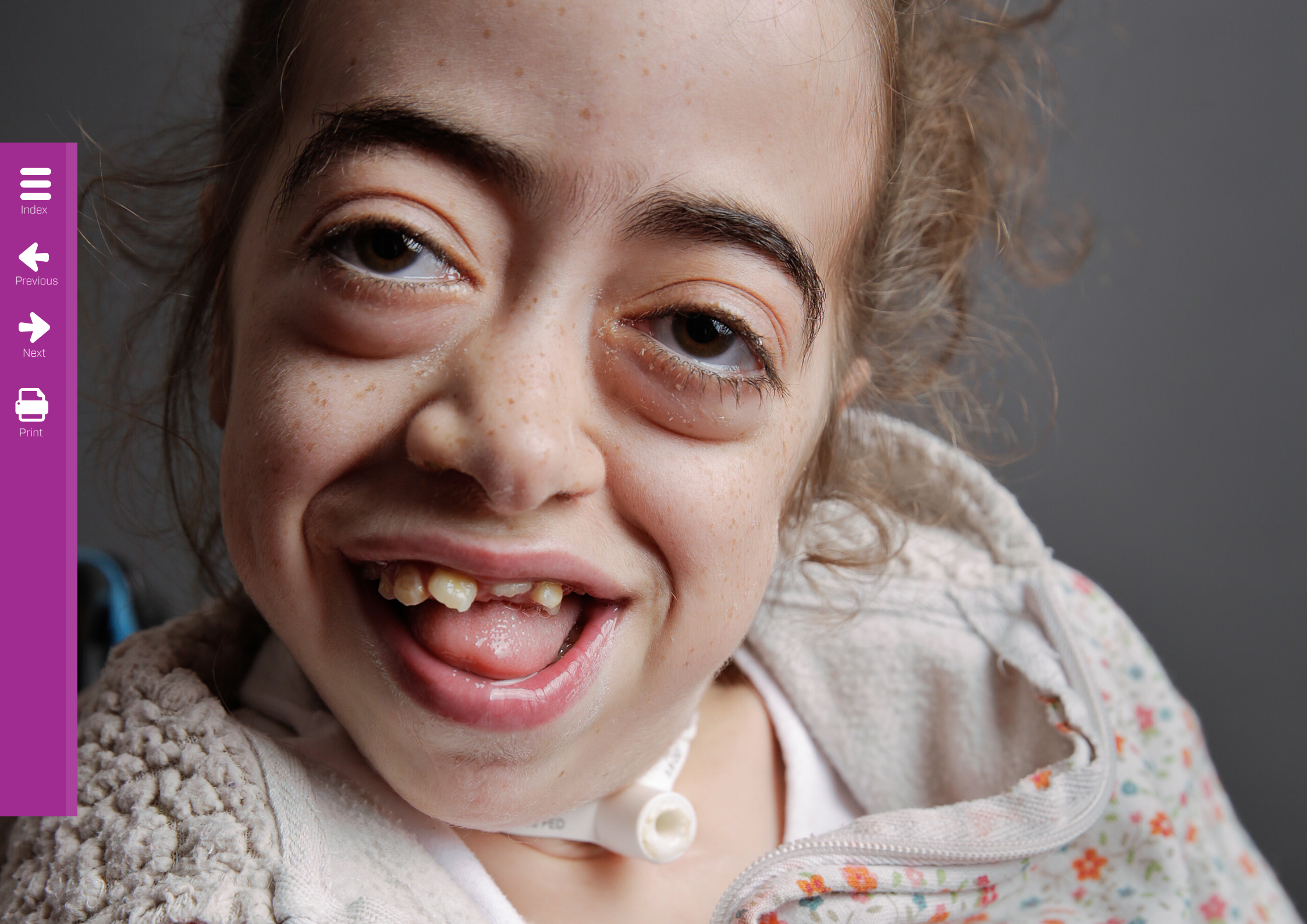

Appendix 1: Project Organisation

Development group

Sonja Bracke, managing director, MSS Research Foundation
Ildikó Vajda, policy advisor, VSOP

Think tank

Dr I (Inge) Van Balkom, child and youth psychiatrist, Jonkx Groningen
Dr. G (Gerda) Derksen-Lubsen, pediatrician, Haga Ziekenhuis/ Juliana Kinderziekenhuis Den Haag
K (Karin) Houwing, occupational therapist, Therapeuticum Artemis
Prof. dr. R (Raoul) Hennekam, professor of pediatrics and translational genetics, Academic Medical Center, University of Amsterdam
Dr. A (Adam) Shaw, clinical geneticist, Guy's Hospital, London, UK

Consultants

Dr. A (Andreas) Machotta, pediatric anaesthetist, Sophia Kinderziekenhuis Rotterdam
Dr. E (Eric) Boldingh, physical therapist, Tyltylcentrum "de Witte Vogel"
Dr. H (Henk) Blom, otorhinolaryngologist,Haga Ziekenhuis, Den Haag
Mrs. A (Annika) van Hemert, speech therapist, Nederlandse Vereniging voor Logopedie en Foniatrie
Drs. M (Marloes) Visser, AIOS intellectual disability physician, ORO
Drs. C (Coby) Wijnen, dietician, Revanet, VKS, SN

Maintenance committee

Dr. G (Gerda) Derksen-Lubsen, pediatrician, Haga Ziekenhuis/ Juliana Kinderziekenhuis Den Haag
Prof. dr. R (Raoul) Hennekam, professor of pediatrics and translational genetics, Academic Medical Center, University of Amsterdam
Dr. G (Gertjan) Kamps, otorhinolaryngologist, SJG Weert Hospital

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Appendix 2: Consultation relevant participants (Carer Consultation)

Short description and summary of results and bottlenecks

The consultation of relevant participants (known as 'carer consultation') for the care standard MSS took place through two methods: the parents of persons with MSS were either interviewed thoroughly, or took part in an online or telephone survey.
The consultation of relevant participants had multiple aims:

- Documenting the complaints and symptoms, and the applied treatment of children with MSS among members of the MSS community (see 5.2.2.7)
- Documenting the effect of different treatments and the satisfaction with the care provided
- Documenting the gaps and issues in the care for individuals with MSS.
- Documenting the broad social and psychological effects of MSS on the patient and on their next of kin.

10 families took part in an extensive personal *in-depth interview*. One family, who only spoke French, was interviewed through an interpreter. One family participated in the interview via Skype.

Based on the interviews, scientific literature, the input from the MSSRF and Dr. Van Balkom, a member of the think tank, we created *a survey*, in Dutch. This survey was available online, or upon request it was sent by mail, allowing the participants to return it in a postage paid envelope. This survey was delivered to Dutch families with children with MSS. The request to complete the survey came with an explanation about the importance of care standards. The anonymity of the participant was guaranteed through a privacy statement. The survey consisted of 404 questions, not all relevant for each respondent, they only had to answer those questions that were relevant to their situation/child.

The Dutch survey has been translated into English and was made available to the rest of the MSS community. The recipients of the English version were also asked to participate and were informed about the importance of good care standards. The anonymity of the participants was guaranteed through a privacy statement. The survey was also translated into Croatian and emailed to a Croatian family (in MS Word).

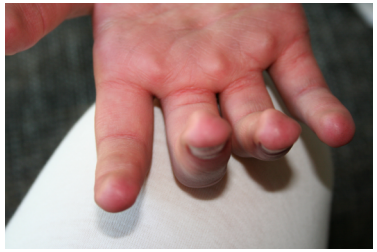
Four families indicated they preferred to complete the survey in their own language. These families (2 French, 1 German and 1 Spanish) were provided with official interpreters. In the presence of the project leader, these families were surveyed via the telephone; the interpreter was able to prepare the questions in advance. The answers were translated into Dutch simultaneously, and recorded by the project leader.

In total, 17 families answered the survey questions. These were recorded on an Excel sheet, compared and summarized in overview tables. These survey results have been incorporated in the relevant chapters (chapters 2, 3 and 6), and the source is documented as 'carer consultation'.

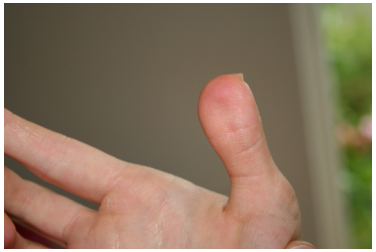
The Marshall-Smith Syndrome Research Foundation an international Family Weekend, every two years. Parents and children meet each other and share experiences. Also information about the MSS research program is shared by involved doctors.

Appendix 3: Images

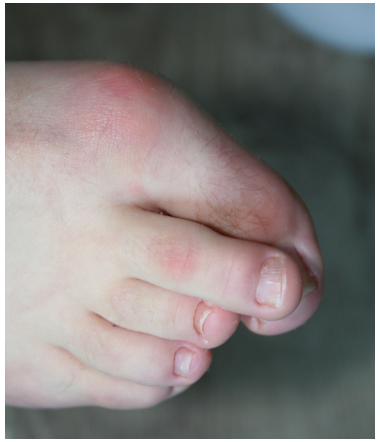
Images (pictures of children)



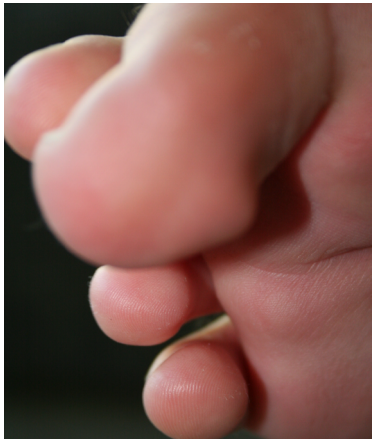
Fingers



Thumb



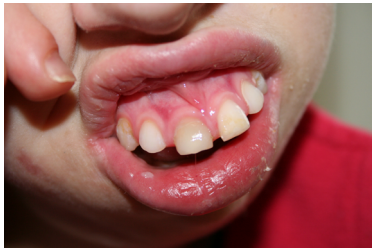
Top of the foot



Toes

Bottom of the foot

Long eyelashes, white of the eyes can be remarkably blu



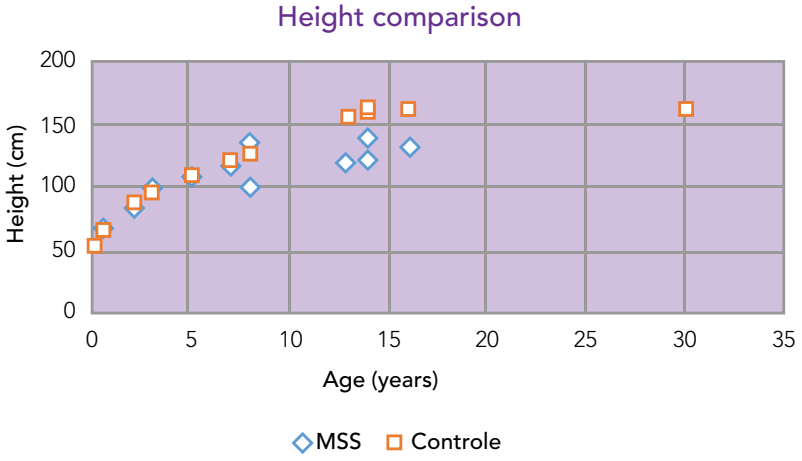
Teeth and gums

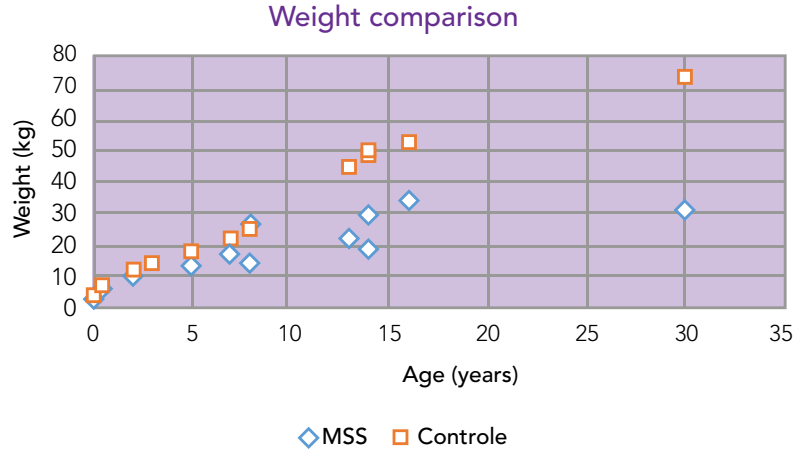
Appendix 4: Growth charts

The height and weight data of 15 of the examined 19 MSS patients has been published [54]. To enable comparison with the general population, this research used:

- WHO growth charts[11]: for Z scores for the height of girls and boys up to the age of 19, and the weight of girls and boys up to the age of 10
- The report "Anthropometric Reference Data for Children and Adults: United States, 2003–2006"; National Health Statistics Reports, 22 October 2008: for determining the height and weight of an average 30-year-old adult female.
- CDC weight charts[12]: for Z scores for the weight of boys and girls aged 11-20.

The reference values for the individual MSS patients were determined in the above-mentioned tables/documents for the corresponding gender and age[13]. These values, and the height and weight data of the MSS patients, were charted in the diagrams below.





11 http://www.who.int/childgrowth/standards/en/

12 http://www.cdc.gov/growthcharts/zscore.htm

13 Accuracy in weeks for data point 1, in months for data point 2 and subsequently in years.

Appendix 5: Guidelines and protocols

Guidelines and protocols that are referred to in the document:

{1} Expertise Group EVMB, at the Carante Groep, “Richtlijn handelingsgerichte diagnostiek bij mensen met Ernstige verstandelijke en meervoudige beperkingen.” Feb-2010.
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{12} NVK, “Guideline: Apparent Life Threatening Event (ALTE).” 2006.
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{16} Guideline Palliative care for people with mental disabilities, K. de Haan, N. van Rossum, 2009.
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{18} Guidelines for diagnosis and treatment of visual disorders in people with mental disabilities, 1997 (planned update in 2014).
{19} Transition of care in adults with mental disabilities, april 2013, NVAVG and NVKN/ VRA/ NVK.
{20} Guideline for diagnosis and treatment of gastroesophageal reflux disease in people with mental disabilities, NVAVG, 1999 and partial-guideline reflux disease in people with severe mental disabilities, A.J.E. de Veer, J.T. Bos, A.L. Francke, Nivel 2008.

Appendix 6: Supporting organizations and information materials

Supporting organizations

- **Stichting MEE** – Support for people living with disabilities (www.mee.nl/)
- **BOSK** – association for and by disabled people and their parents (www.bosk.nl)
- **Platform EMG** – knowledge network and information database for severe multiple disabilities (http://www.platformemg.nl)
- **Partnership of different associations for 'Integral help in early stages'** – help in case of developmental disorders or behavioral disorders (www.integralevroeghulp.nl)
- **Ieder(in)** – Network for people with disabilities or chronic disorders (https://iederin.nl/)

Links to information materials

- www.elkerliek.nl/Elkerliek/Patientenfolders/Prelogope-die-borstvoeding.pdf)
- www.kennispleingehandicaptensector.nl/docs/producten/Het_ziekenhuisbezoek_van.pdf
- www.medizin.uni-tuebingen.de/uktmedia/Patienten/PDF_Archiv/ZKFKG/PRS_Elterninformation.pdf
- www.ncbi.nlm.nih.gov/pmc/articles/PMC2876895/
- www.radboudumc.nl/Informatiefolders/2401-Kinderen_met_sondevoedin-i.pdf
- www.communicatiemethodenemb.nl/methoden/opvoeding-sprogrammas/
- www.nidcap.org/
- www.logerenmetzorg.nl/pages/handreiking.php
- http://www.amerpoort.nl/binaries/content/assets/brochures/kinderen/scoliose-bij-kinderen-met-emb.pdf

Appendix 7: Resources

Resources during the acute phase

In the acute phase, the children and their parents will benefit from the following resources/tools/equipment: NPA, Cpap, feeding tube, suction device. There are resources to teach a child to eat food, such as cup feeding, spoon feeding and a pre-speech cup. The most successful method during early childhood varies per child.

Resources during the chronic phase

During this phase, children will start using more resources/tools, in addition to those mentioned in the acute phase. For instance: special chairs, walking aids, standing table, special stroller, wheelchair, commode, adjustable bed with high framing, orthopedic shoes, swimming aids, chair seat on bicycle, three-wheeler bike or tandem bike.

Acknowledgements

We would like to thank in particular all the families that were willing to participate in the carer consultation. Without their input, we would not have been able to create this care standard. We also thank the consultants and the think tank, who were able to deliver the required content corrections and additions. Special thanks to Marieke van Meel for conducting the majority of the in-depth interviews, and also special thanks to Kim Wever, Adam Shaw and Raoul Hennekam for assisting in the translation of the Dutch survey into English.

Colofon

Graphic design and interactive pdf:www.bluezilla.nl
Standard of Care Marshall-Smith Syndrome 2015
English translation: 2017

Publisher
MSS Research Foundation
Oeverbiesstraat 20
Den Haag
2548 WP Nederland
Telefoon: 070- 33 56956
E-mail: info@marshallsmith.org
Website: www.marshallsmith.org

Authors: Ildiko Vajda en Sonja Bracke.

ISBN/EAN: 978-90-823046-0-2

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