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A Grain of Normality

Weaning a Child With Marshall-Smith Syndrome From the Feeding Tube: A Case Report

Thomas Trabi, MD, Hannes Beckenbach, MSc, Stefan Ring, MD, Marguerite Dunitz-Scheer, MD, and Peter J. Scheer, MD

Abstract: *This case reports highlights of the feeding problems in Marshall-Smith syndrome and the weaning from a tube in a 5-year-old patient. The patient, Maya, showed the typical signs of this rare disease. The diagnosis of Marshall-Smith syndrome was made at the age of 2½ years. Because of respiratory problems and a high risk of aspiration, Maya got a percutaneous endoscopic gastrostomy (PEG) tube at the age of 5 months. After stabilization of her respiratory situation, Maya was able to eat from a strictly medical point of view. Maya was admitted to the authors' ward dependent on a PEG tube, but her parents wanted her to be weaned from the tube. In contrast to other patients with Marshall-Smith syndrome, Maya's typical respiratory problems stabilized, and on the basis of the little experience published in the literature, Maya seems to have a relatively good life expectancy. After careful analysis of her medical state, Maya was integrated into the specialized tube-weaning program. Tube weaning was successful, and after 3 weeks of treatment, Maya was able to gain weight by exclusive and self-regulated oral intake. In conclusion, this case report shows that weaning a child with Marshall-Smith syndrome from a feeding tube is possible and gives these children the chance to gain weight by oral intake, as any healthy child does. The reachable eating skills depend on the grade of disability. In the authors' clinical experience, weaning from a tube shows a positive effect on psychomotor development and reduces the mortality rate and the rate of complications associated with tube feeding itself.*

Keywords: Marshall-Smith syndrome; tube feeding; tube weaning; development; biphosphonates

Marshall-Smith syndrome (MSS) is a rare genetic disease, described by Marshall et al[1] in 1971. The syndrome comprises accelerated skeletal maturation, relative failure to thrive, unusual facies, motor delay, and mental retardation. The most specific findings are accelerated osseus development, including long and thin tubular bones and specific alterations of phalanges and scoliosis (see Figure 1). Up to now, 38 cases of MSS have been described in the literature. In the latest published study, the findings were summarized as an osteochondrodysplasia.[2] Most of the patients suffered from severe pulmonary problems, and in most cases, the children died from pneumonia.[3]

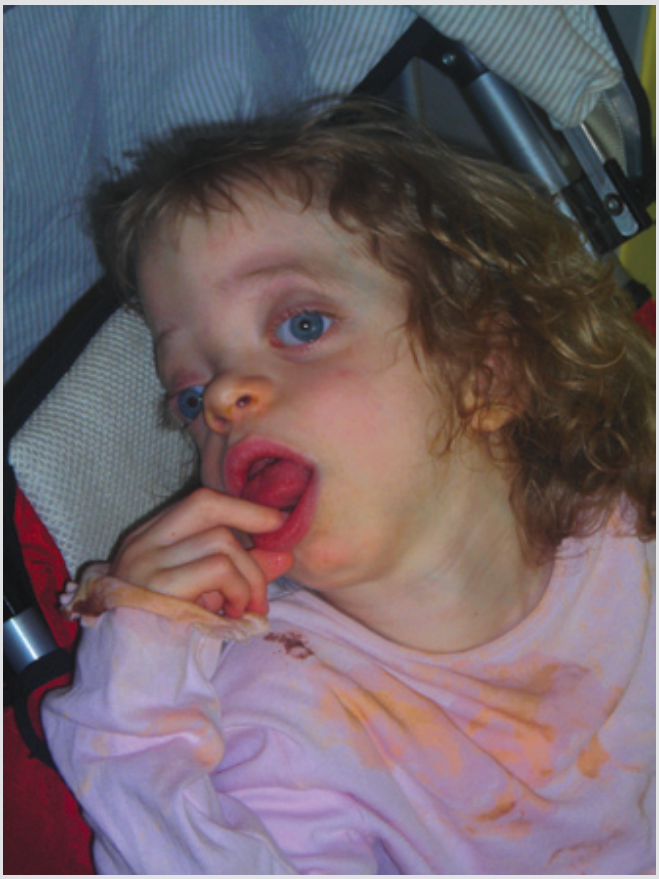
Feeding problems are not especially addressed in the available case reports, and information about feeding children with MSS is rare. In cases in which respiratory problems were subordinated, children had severe feeding problems and were fed by tube, according to information about feeding that is available in the present literature. The main problems in MSS are palatal and laryngeal malformations, causing swallowing problems and dysphagia. The swallowing dysfunctions are associated with a high risk of aspiration. Patients without respiratory problems are suggested to have a longer life expectancy.[4] At the moment, there is 1 patient reported in the literature who achieved an age of 19 years.[2]

In our unit, we specialize in feeding problems in infants and children. Within this specialization, we wean a lot of tube-dependent children from feeding tubes. Feeding tubes are commonly used in children for them to survive a period of decreased oral intake, and tube dependency is a growing problem worldwide. Most of these tubes are important to feed children and to sustain severe injuries or diseases. Some of

DOI: 10.1177/1941406410364241. From the Medical University Graz, University Hospital for Children, Psychosomatic Unit, Graz, Austria. Address correspondence to Thomas Trabi, MD, Medical University Graz, University Hospital for Children, Psychosomatic Unit, Auenbruggerplatz 30, 8036 Graz, Austria; e-mail: thomas.trabi@gmx.at.

Figure 1.

Typical aspect of a patient with Marshall-Smith syndrome. Patient has actually returned from play picnic.



these children stay dependent on tubes,[5] although there is no real medical reason for tube feeding. It has been shown that tube feeding itself increases the mortality of children.[6] In addition, quality of life is decreased, and in our experience, the children often show developmental delay caused by tube feeding. We believe that this delay can be explained as a secondary result of limitations in the normal social life of children, caused by special feeding techniques and social isolation. According to our specialized treatment, about 300 children have been weaned from feeding tubes.[7] The patient presented here was the first one with MSS. The successful treatment is specified by respecting the children's autonomy and is based on nondirective play therapy.

Patient Presentation

Maya was born as the first child (first pregnancy) of 2 healthy, nonconsanguineous parents. End of pregnancy was complicated by polyhydramnios, and Maya was delivered by vacuum extraction in the 37th gestational week. Her birth weight was 2170 g (4th percentile), length was 50 cm (60th percentile), and head circumference was 33.5 cm (40th percentile). Apgar score was 2/4/5/7, and pH of allantoic vein was 7.28. After delivery, she was respirated artificially, and muscular tone was decreased. After a few minutes, Maya had been intubated, and a nasogastric tube (NGT) was given to her for feeding. The first radiograph of the thorax showed total atelectasis of the left lung and partial atelectasis of the right lung. Maya was transferred to the neonatal intensive care unit. She was suffering from choanal stenosis, macroglossia, hypotonia, stenosis of the upper airways, prominent forehead, protuberant eyes, and a high peaked palate. Initially, Maya was suspected of suffering from Dandy-Walker syndrome. At age 5 months, Maya had been weaned from a ventilation tube, and because of laryngomalacia, she received a denture including a skid, which made it possible for her to breathe autonomously.

A percutaneous endoscopic gastrostomy (PEG) was implanted in Maya at age 5½ months for her to gain weight. After implantation of the PEG, Maya was suffering from gastroesophageal reflux (GER), and a fundoplication was done at the age of 7½ months. Her body weight was 4900 g (<3rd percentile), and length was 65 cm (40th percentile). Sonographic scans of the abdomen and head at this age were normal. An x-ray of the abdomen showed a malrotation of the duodenum and small bowel. Sphrintzen-Goldberg syndrome could be excluded genetically.

At the age of 25 months, Maya was checked again at a university hospital. Within this check, MSS was suspected for the first time. At that time, Maya was suffering from recurrent pulmonary infections and pneumonia, and she was treated with antibiotics. Within this period, Maya was repeatedly placed on oxygen. A magnetic resonance tomography (MRT) scan of the brain showed a hypoplasia of the vermis cerebelli and a hypoplastic viscerocranium. Dandy-Walker syndrome could be excluded within this MRT scan.

Two months later, a genetic examination was done, and according to her typical dysmorphism, she was suspected of suffering from MSS. By genetic examination of the *FBN1* gene and *TGFBR1* and *TGFBR2* genes, Marfan syndrome was excluded. Within this check, biphosphonate therapy suggested a positive effect on bone density.

At the age of 33 months, kyphosis and scoliosis got worse, and intensive physiotherapy was started. Supported by parents and physiotherapy, Maya started to crawl and to pull herself up. Because of hypotonic musculature and

hypermobility of articulars, an orthopedic device was given, which made it possible for her to stand alone.

Since birth, Maya has suffered from severe respiratory problems and has shown good weight gain when fed by tube. Tube weaning was no topic for the parents during this period.

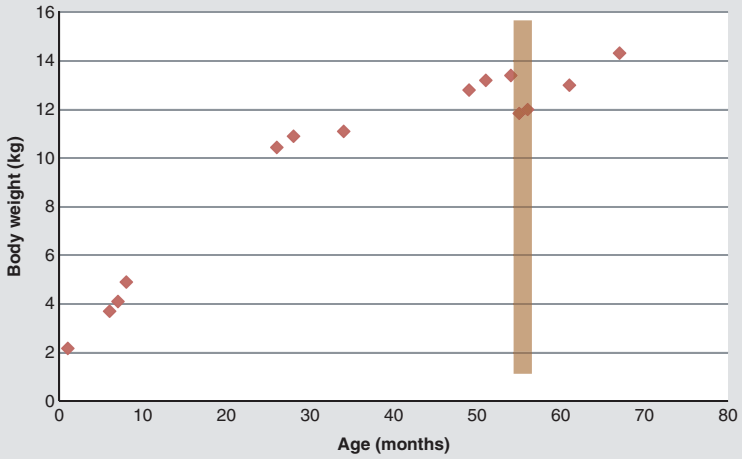
Being fed by tube, Maya continued to gain weight. At the age of 4 years, her weight was 12.8 kg (3rd-10th percentile), and length was 100 cm (25th-50th percentile). At that time, an x-ray showed a very thin tibial epiphyseal plate and a bone age of 7.8 years (using the charts of Greulich and Pyle[8]). In addition, a hypoplasia of maxilla was found. An orthopedic examination showed hyperelasticity of all joints, contracture of the left knee, and flatfeet.

At the age of 50 months, body weight was 13.2 kg, and she was admitted to adjust the upper denture. At that time, Maya was developing well and did not show any breathing problems. Stabilization of her pulmonary situation was explained by maturation of anatomical structures. Her chondral larynx was more stable, and maturation of muscles and coordination could provide a better swallowing function and a decreased rate of aspiration. For her parents, tube feeding proved to be the major problem now because Maya suffered from problems with the PEG itself. The tube was repeatedly blocked, and it was necessary to change it twice. In addition, the area of implantation was inflamed very often. Also, Maya's parents recognized her limitations in interaction caused by tube feeding. Maya was not able to eat with the other children or with her brother. Maya began to taste some food but could not increase her intake supported by her parents alone. Both parents wanted their child to eat normally, and so they contacted our unit.

Maya was admitted for tube weaning at the age of 53 months with a body weight of 13 400 g. At admission, she was fed by PGT and received 1200 mL pureed food and 1200 mL water (about 1500 kcal/d). She only took about 100 mL mashed fruits per day orally. On admission, all lab values were in normal range, and she showed no signs of undernutrition. Other parameters of undernutrition (eg, triceps skinfold or midarm circumference) were not measured. Her weight chart can be seen in Figure 2.

Figure 2.

Weight chart of the patient: the dark area marks the time of tube weaning.



She was integrated into our multidisciplinary tube-weaning program and received speech language pathology (SLP), occupational therapy, and physical therapy. She also took part in the daily play picnic, a specialized eating therapy based on psychoanalytical nondirective play therapy with various kinds of food. Within this therapy, various kinds of food are presented on multicolored dishes on the floor. All children have the ability to explore and taste the food in the way they want to. Parents are present but are requested not to force their children to eat, so the children can explore the food at their own speed and in their own manner.[5]

The combination of various therapies is clarified in Figure 3, which shows a typical timetable of the program.

Within our specialized, multidisciplinary therapy, Maya was able to increase her oral intake, and tube feeding was dropped very quickly and could be stopped totally on the 10th day of treatment. In contrast to our clinical experience and our expectation, Maya never showed anxiety toward food, and her parents never forced her to eat. Her body weight decreased to a minimum of 11 840 g on day 10 and then remained stable by oral intake. Her maximum weight loss was 1560 g and was within the expected range. We are accepting a weight loss of up to 10% of body weight, which is comparable to weight loss after birth. Learning to eat was complicated by swallowing problems. Supported by high frequent, daily SLP therapy,[9] she was able to swallow without aspiration. SLP therapy focused on safe swallowing and on coordination of swallowing. Various textures were tried, and Maya had the chance of improving her swallowing function gradually, so that she was able to minimize aspiration during the therapy. Maya was discharged after 19 days of treatment, exclusively fed orally with a body weight of 12 000 g.

"If a child with MSS or any other severe disability is fed by tube, this situation should be examined very carefully, and the possibility for tube weaning should be clarified"

Figure 3.

Typical schedule of the weaning program. PhT, physical therapy; OT, occupational therapy; SLP, speech language pathology; Psy, psychological counseling; PTDev, developmental psychotherapy; Nutr, nutrition counseling; PTP, parent psychotherapy.

Time	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday	Sunday
8–9	Ward round (daily)						
9–10	PhT	OT	SLP	PhT	SLP	Free for family activities	
10–11		PhT		Staff-Conference			
11–12	SLP		OT		PhT		
12–1	Play picnic (daily)						
1–2	Psy	Baby swimming	Baby swimming	PTDev	Nutr		
2–3	Ward round (daily)						
3–4	Nutr	PTP			OT		
evening	Family time						

Maya's parents and brother were present the whole inpatient stay, a fact that influences the outcome very positively. In our experience, siblings often influence the outcome by exemplifying eating and enjoying food. We also could see excellent parent-child interaction in this family. It has been shown in former works that the interaction has a strong impact on eating behavior.[10] Within the stay, we tested Maya to get an impression of her developmental age using the Child Development Inventory.[11] Maya showed a general developmental age of 15 months. She achieved the highest score of 18 months in social skills and the lowest score with 9 months in gross motor skills. The results have to be handled with care because we do not have any comparable data of patients with MSS, but it is obvious that Maya was developmentally delayed.

Six months after treatment, Maya could increase her oral intake up to 1000 mL (about 1300 kcal) per day. She continued to gain weight and had a body weight of 13 kg (see Figure 2). Her parents reported that she also improved her motor skills, and the removal of the PGT is planned within the next month. At the last examination 12 months after weaning, Maya had a weight of 1434 kg. Her parents reported good development.

Discussion

Feeding tubes are able to help children sustain periods of decreased oral intake; in most cases they are associated with severe illness or injuries. We are sure that tube feeding saves a lot of lives worldwide, and in most cases, the removal of the tube is done very easily. In some children, a tube is needed for feeding for their whole lives. We are working with the children between these two extremes who cannot be weaned from the tube easily and do not need tube feeding from a medical point of view. We call these children tube dependent.

In such an uncommon disease as MSS, the literature is scarce. There are only a few case reports and reviews available.[2] The present literature focuses on respiratory problems and the mineralization defect. Feeding problems in MSS have never been addressed explicitly. Because swallowing functions seem to be disturbed in many of these children, we believe that feeding must be a common problem in MSS. In some case reports, it is mentioned that the children are fed by tube, but it is not reported if some of these ever have been weaned.

As in other children with tube dependency, mortality is caused more by tube feeding itself.[6] In our opinion, this should be the main reason for tube weaning. If a child with MSS or any other severe disability is fed by tube, this situation should be examined very carefully, and the possibility for tube weaning should be clarified.

Following the current guidelines, weaning is usually begun in the United States when a child is over the 25th weight-for-age percentile. In our experience, children sometimes cannot achieve this body mass index (BMI), especially the diseases such as MSS, in which a low BMI is commonly present. These percentiles are made for healthy children, and as far as we know, special percentile charts for disabled children are available only for some diseases (eg, cerebral palsy), so the commonly used charts cannot be used for children with severe diseases, as in this patient. In our experience, disabled children often show a better weight gain by oral intake in comparison with tube feeding, so we believe that weaning in special cases should be tried, especially if the patient has undernutrition according to commonly used charts or guidelines. In such cases, it is important to think about alternatives if weaning and weight gain do not work. In this case, we left the PGT for the following months to have an immediately available way to feed her if weight gain could not be achieved by oral intake. Williams et al[3] stated that aggressive management of failure to thrive may improve the prognosis of MSS. In our opinion, the most aggressive way to manage nonorganic failure to thrive (NOFT) is to wean the patient from the feeding tube. A sufficient oral intake without aspiration may be the most successful way to support patients with MSS and to improve their prognosis.

To us, Maya seemed to be an ideal candidate because she did not show any signs of undernutrition—although her weight was below the 5th weight-for-age percentile. Her parents wished to wean her, and examination did not find any contraindications. In a retrospective study, we could show that the severity of disability does not influence the

outcome of the weaning program negatively.[7]

In our experience, tube weaning also shows positive effects on psychomotor development. There is no literature available at the moment, so we started a study focusing on the changes of psychomotor development before and after tube weaning. We hope that we will be able to present first results soon.

Apart from the negative side effects of tube feeding and the decreased quality of life, tube feeding is an enormous socioeconomic factor that is suggested to cost more than US$30 000 per year.[12] Our program—which costs about US$15 000 per patient—is often criticized as too expensive. In comparison with the cost of tube feeding, this discussion seems to be dispensable.

In conclusion, after careful review of the available literature, patients with MSS may have 3 major problems: pulmonary complications and recurrent infections, accelerated bone maturation and its associated complications, and feeding problems.

Pulmonary problems seem to be the life-limiting factor. In cases of stable pulmonary situation—as in the presented patient—feeding problems are the main goal for treatment. And for long-time survival, a successful treatment of bone maturation is needed. In our case, we were able to treat the feeding problem—tube dependency—successfully and weaned the child from the tube. To prevent osseus complications (pathological fractures), we suggested treating her with biphosphonates (Zometa; Novartis, East Hanover, New Jersey) because it has been shown in children with osteogenesis imperfecta that biphosphonates are able to reduce the rate of fractures and do not show any negative side effects, especially no influence on growth.[13]

Parents and physicians often are concerned about weight loss during tube weaning. In our program, we accept a weight loss of about 10% of preweaning body weight. Most children are able to gain this 10% in 1 or 2 weeks.[5,7]

This case report had the aim of focusing on feeding problems in MSS. Apart from pulmonary problems, feeding seems to be the second problem of this rare disease. Treatment of tube dependency may help to improve the life expectancy and life quality of these children. As we could show in this case, tube weaning is possible in such a severe disability, and weight gain can be achieved by oral intake. ■

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