Follow-up study of three cases of congenital microgastria

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Abstract

Current knowledge on congenital microgastria is limited due to its extreme rarity, and the paucity of nutritional and quality of life follow-ups. Patients affected by congenital microgastria cases followed at out center were screened, and general and nutritional status were evaluated at follow-up visits through validated questionnaires. Three cases were included: one patient died because of a complex syndromic picture where microgastria was imperatively approached conservatively. The remaining cases underwent Hunt-Lawrence at 2 and 17 months. After 2 years and 27 years postoperatively, both patients are on full oral intake. The 28-year-old patient did not reach a BMI higher than 18. She rated her quality of life as unimpacted, with a Gastrointestinal Quality of Life Index of 111. In the other case, parents reported about their 2-year-old child an Infant Gastrointestinal Symptom Questionnaire of 13, corresponding to "no distress". Our findings confirm the literature trend supporting the role of early surgery in microgastria to improve outcomes. We presented the nutritional status and quality of life in two cases of congenital microgastria operated according to Hunt-Lawrence at a 2-year and 27-year distance, which is the longest follow-up reported to date.

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Introduction

Congenital microgastria is an extremely rare condition[1-4] and actual knowledge of its management is currently still very limited. In the majority of cases, it is found as associated with other anomalies, with only 6 cases of isolated congenital microgastria reported.[5] Affected patients present with a small, underdeveloped, and tubularized stomach, with minimal or absent reservoir capacity leading to nutritional deficiencies with mortality in severe cases.[1-7] The final therapeutic goal is to achieve a satisfactory caloric intake according to the patient's nutritional requirements. Hitherto, a lack of consensus regarding the best treatment options exists due to the paucity of follow-up reports available.[1,6,7] Conservative strategies exploit tailored regimens of total enteral nutrition, either by nasogastric tube (NGT), nasojejunal tube (NJT), or gastrostomy/jejunostomy. According to Ruckzynsky *et al.*,[1] conservative treatment is usually reserved for severe cases with a high comorbidity burden and poor preoperative surgical fitness. Therefore, high mortality rates are associated with conservative therapy (47% *versus* 15%), which has been postulated as a selection bias.[1] On the other hand, surgery aims to create a neo-stomach compliance with proper nutritional intake. The Hunt-Lawrence (HL) pouch and Total Gastric Dissociation (TGD) are the most frequently described surgical techniques.[1-15] HL technique accomplishes gastric augmentation by the creation of a jejunal pouch which is anastomosed to the stomach as a Roux-en-Y.[3,5] A proximal portion of

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the jejunum is transected and bent into a "U shape". A reservoir is created by performing a later-lateral anastomosis of the two adjacent jejunum loops within the "U". Following transposition of the newly created pouch through a mesocolic window, a terminal-lateral anastomosis is then performed between the base of the new reservoir and the native stomach. An end-to-side jejuno-jejunostomy is built distally to restore bowel continuity as per Roux-en-Y. An alternative technique for developing the reservoir, which involves a longer portion of the jejunum, is the so-called "Omega loop". This time, the jejunum is brought directly to the stomach through the mesocolic window without jejunal transection, and an end-to-side gastro-jejunostomy is achieved at about half the length of the loop. The distal segment of the "omega" is manipulated to fashion a side-to-side jejuno-ileostomy. On the other hand, the TGD technique entails the retrocolic-retrogastric transposition of an isoperistaltic Roux-en-Y segment of jejunum which is pulled through the transverse mesocolon. An end-to-end gastro-jejunostomy is achieved. Bowel continuity is then restored by end-to-side jejuno-jejunostomy.[7-9] A pyloroplasty can be also performed in case of concerns for vagal nerve integrity or stomach emptying. In a recent meta-analysis reporting about 71 previously published cases of microgastria, the type of treatment was described for only 46 patients.[1] Among them, 27 were treated surgically with the HL pouch being the most commonly performed surgery with a mortality rate of 6%.[1] Among the reported follow-ups, HL pouch seems to be associated with nutritional acceptable outcomes, though it has been advocated that it is more effective the earlier the surgery is.[5] As mentioned in Ruckzynsky's metanalysis [1], which is the baseline of knowledge for congenital microgastria to date, one of the limitations discussed was the scarcity of available nutritional data and outcome of growth.[1] In fact, only one long-term follow-up has been presented for a patient with suboptimal somatic growth despite eating normal meals, having undergone an HL pouch 18 years prior for microgastria.[5] We present the follow-up of three congenital microgastria cases treated at one center and aimed to determine the related quality of life and perspective of the living patients by the employment of validated gastrointestinal system-specific questionnaires.

Materials and Methods

In ethical compliance with the Helsinki Declaration and after the Institutional Review Board's (Research Ethics Committee, 25/2022, 08/02/2022) approval at A. Meyer Children's Hospital, a retrospective analysis was conducted on patients treated for congenital microgastria in 1990-2022. Inclusion criteria were as follows: i) patients with "congenital microgastria" as definitive discharge diagnosis; ii) patients amenable to be reached either by phone or by post; iii) patients and their families informed and agreed to sign the research consent form. Exclusion criteria were as follows: i) uncertainty of diagnosis (*i.e.* "no other specified gastric developmental anomaly"); ii) impossibility in completion of the quality-of-life questionnaire. For each included patient, we recorded prenatal and postnatal history, type of treatment (conservative/surgical), type of surgery performed, postoperative surgical outcomes, re-admissions, postoperative attended clinic follow-ups, and deaths. To update the follow-up, a remote face-to-face appointment was scheduled. The follow-up data involved the patient's general clinical and nutritional status and growth curves. Quality of life, gastrointestinal health, and food aversion have been investigated by two validated questionnaires: the Infant Gastrointestinal Symptom Questionnaire (IGSQ) has been used below 2 years of age, via parent interviewing; The Gastrointestinal Quality of Life Index (GIQLI), instead, have been administered to patients/parents for patients above 3 years of age.[16-22] The IGSQ is a validated and reliable method for the assessment of infant gastrointestinal-related behaviors and has been advocated as helpful to advance parent education as well.[16] It is based on a 13-item index score aiming to assess infants' GI-related signs and symptoms noted by parents over the previous week. The investigated domains are stooling, spitting up/vomiting, flatulence, crying, and fussiness.[16] The items are rated 1 to 5, with higher values indicating worse outcomes. The final score is given by the sum which ranges between 13 (no GI distress) and 65 (extreme GI distress).[16] According to the authors,[17,18] the score of 30-35 can be set as a reasonable cut-off for meaningful digestive distress. The GIQLI questionnaire is a system-specific quality-of-life instrument that is commonly used in gastrointestinal disorders.[19-22] GIQLI is meant to assess patient-reported outcomes on GI-specific health-related quality of life. It has been broadly used in general surgery as a useful tool to prove a higher post-procedural index of quality of life compared to the pre-procedure status. It is based on 36 items in five domains: GI symptoms, emotion, physical function, social function, and medical treatment. Each item scores 0-4, where 0 equals to extremely low outcome.[19] The final score (given by the sum) ranges between 0 and 144 where the higher the total score, the better the quality of life.[19,20] A healthy control group scored 86 GIQLI in gut health in a previously published series.[21] For both IGSQ and GIQLI, patients' answers were recorded point by point at the time of the remote consultation. The follow-up time was calculated from the date of surgery to the time of death or remote consultation follow-up (done on 24/10/2022).

Results

Three patients have been treated at our center for congenital microgastria in the period 1995-2020. The full clinical dataset is shown in Table 1. Two out of three patients were females. All patients were born at term and with no prenatal suspicion of gastric developmental anomaly. Birth weight was within normal limits in two cases, whereas it was 2385 g in one case (Patient 2, Table 1). This patient was affected by Di George Syndrome and congenital heart disease. In one case, congenital microgastria was found as isolated (Patient 3, Table 1). In one case diagnosis was made at laparotomy (Patient 1, Table 1), and, in two cases, following an upper Gastro-Intestinal (GI) contrast study for severe Gastro-Oesophageal Reflux Disease (GORD) symptoms (recurrent regurgitation, post-prandial vomit and respiratory distress). Conservative treatment was attempted in all our cases and, in one case, it was the only reasonable option when considering their comorbidity burden and fitness to major surgical reconstruction procedures (Patient 2, Table 1). This patient was initially treated by continuous Enteral Nutrition (EN), then escalated to total Parenteral Nutrition (PN), and eventually died in the hospital at 8 months of age for acute respiratory failure after being in hospital since birth.

Patient 1 in Table 1 was born at term, of 3850 g weight and 54 cm length (above the 75th percentile). At birth, she was hospitalized for respiratory distress which was attributed to aspiration pneumonia, and stayed in hospital for two weeks. After discharge, the parents started noting frequent regurgitations and progressive deterioration of the respiratory function leading to a new hospitalization at two months of age. A diagnosis of left diaphragmatic eventration and left lung hypoplasia was established, and, at 2.5

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months of age, she underwent a left subcostal laparotomy and diaphragmatic plication. At laparotomy, the microgastria and asplenia were highlighted for the first time and a gastrostomy tube was put in place for later conservative treatment. The postoperative course was marked by respiratory failure due to the secondary laringotracheomalacia and hypoplastic left lung requiring prolonged intubation and tracheostomy positioning (which was then removed at 3 years of age). The patient was then discharged at 10 months of age with EN per gastrostomy. She was showing a scarce interest in eating (suggestive of a mild form of food aversion) and experiencing diarrhea or frequent bowel movements when increasing the EN amount. At 12 months of age her weight was 5 kg and length 60 cm (below the 10th percentile). As a consequence, the decision was made to proceed with the surgical management. Surgery was delayed for two months due to intercurrent C. Difficile diarrhea. At 17 months of age, in 1995, she underwent a gastric reconstruction according to HL (further intraoperative details were not retrieved), gastrostomy closure, and NGT positioning. Postoperatively, she started feeding by mouth on Post-Operative Day (POD) 1 and showed a good tolerance to increasing feeds. The postoperative course was marked by multiple respiratory infections that delayed the discharge significantly. She couldn't be sent home earlier than 6 months postoperatively, despite fully tolerating the oral intake. Upon discharge, the patient was reattached to the care of the referring physician. She was followed by different specialists at our center through annual day hospitals to assess nutritional status and general growth. Throughout early infancy, a combination of night PN and EN per SNG was later required to support the growth. PN was discontinued at the age of three, while the NGT was left in place for night-time EN until the age of 6. The patient's infancy was marked by recurrent respiratory infections. She underwent an aortopexy at 15 years old and multiple bronchoscopies with three left main bronchus stent positions. At the age of 12 due to failure to thrive and severe anemia, a new NGT for nocturnal EN was administered until the age of 16. The patient is now 28 years old and currently weights 48 kg for 161 cm height (BMI 18.5). She reports having a varied diet and enjoying food very much. She lives her life independently. Seldomly, especially after rich meals and eating fast, she experiences abdominal distension and pain, followed by diarrhea (dumping syndrome). Recently, because of tachycardia and palpitations, she was found with severe anemia which is now controlled with iron injections and vitamin B12 and folic acid supplements. She has never felt her treated microgastria has impacted her quality of life as people around her (family and friends) have always been supportive. The GIQLI questionnaire is currently rating 111 for her at a 27-year postoperative follow-up.

Patient 3 in Table 1 was also initially approached conservatively by an in-hospital combined regimen of PN and EN per NGT. However, due to persistent severe feeding intolerance and postprandial vomit, he underwent an upper GI contrast attempted at 46 days of life 15 ml volume of enteral contrast. The procedure was stopped due to the establishment of severe reflux and vomiting requiring the conclusion of the procedure. The radiologist however

Table 1. Clinical dataset of three patients treated for microgastria at one center.

	Patient 1	Patient 2	Patient 3
Gender	Female	Female	Male
Ethnicity	Caucasian	Caucasian	African
Prenatal diagnosis	N/A	None	None
Birth	At term, aspiration pneumoniae	At term	Respiratory distress, feeding intolerance
Birth weight	3850 g	2385 g	3500 g
Comorbidities	Left diaphragm eventration, with hypoplastic left lung and laringotracheomalacia. Asplenia. Growth hormone deficiency	Di George syndrome, tronchus arteriosus type1, Del 22q11.2	Pulmonary hypertension (of unknown origin) requiring medical treatment
Presentation	GORD, respiratory failure	Severe GORD	Respiratory distress, severe GORD symptoms
Diagnosis	At laparotomy	Upper GI contrast, showing a small stomach	Upper GI contrast revealing gastric volume of 15ml at 46 days of life
Treatment type	Initial conservative management. After failure, surgery was performed at 17 months of age in 1995	Conservative	Initial conservative management. After failure, surgery was performed at 2 months of age (in 2020)
Further treatment details	Enteral nutrition by gastrostomy. Then, Hunt-Lawrence pouch	Continuous enteral nutrition by NGT. Escalated to total PN at 4 months of age	PN and enteral nutrition by NGT. Then, Hunt-Lawrence pouch
Full enteral autonomy	Full oral intake tolerated in the early postoperative	Oral intake not tolerated	Full oral intake at 14 months
Postoperative complications	Respiratory infections	N/A	None
Discharge	After 6 months	N/A	POD 92
Follow-up (distance)	27 years	N/A	25 months
Follow-up (growth)	BMI 18	N/A	12 kg (50th percentile) and height is 87 cm (75th percentile)
Quality of life questionnaire	GIQLI 111	N/A	IGSQ 13
Death	N/A	At 8 months of age in 2006, in-hospital	N/A

N/A, not available; GORD, gastro-oesophageal reflux disease; GI, gastro-intestinal; NGT, nasogastric tube; PN, parenteral nutrition; POD, post-operative day; BMI, body-mass index; GIQLI, gastrointestinal quality of life index; IGSQ, infant gastrointestinal symptom questionnaire.

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reported a high suspicion of gastric underdevelopment which was confirmed at the laparotomy done at two months of age in 2020. An HL procedure was carried out by division of the jejunum at 10 cm from the Treitz ligament and its transposition through a meso-colic window to reach the anterior aspect of the rudimental stomach. A latero-lateral double-layered hand-sewn gastro-jejunal anastomosis was performed and a trans-anastomotic NJT was positioned. An omega pouch was created by termino-lateral jejuno-jejunal anastomosis, with an anti-peristaltic segment and an isoperistaltic one of about 5 cm each. Intestinal continuity was eventually restored through a Roux-and-Y procedure. Registered operative time was of 105 minutes and no intraoperative or post-operative complications were recorded. On POD 10, EN per NGT was re-started on a step-up schedule. The patient was discharged on POD 92 with an oral intake of 90 ml three hourly and overnight PN which was then stopped at 14 months. Upon discharge, the follow-up was performed at 1, 6, and 12 months with day hospitals managed by several specialists, namely a gastroenterologist, a neonatologist, a dietician, and a pediatric surgeon.

The patient is now 2 years old and 25 months postoperatively. Parents report the child in good clinical conditions with no regurgitations, diarrheoa or food aversion. His weight is currently 12 kg (50th percentile) and his height is 87 cm (75th percentile). The IGSQ questionnaire is currently rating 13 him, revealing a satisfactory quality of life and no underlying distress.

Discussion

Congenital microgastria figures in a rudimental stomach, with a tubular or saccular shape.[1-3,10-15] The reservoir capacity is null or minimal, as well as the secretory function. The underlying etiopathogenesis is related to an embryological intestinal developmental defect. Most commonly it involves other organs, according to the time of the injury.[14] In fact, isolated microgastria is reported as an exceptionally rare event, for which we report one case.[5,12-15] As opposed, syndromes that present microgastria along with other defects are the microstrip-limb reduction defect, the VACTERL complex, and the Pierre-Robin sequence.[12-15] Prenatal diagnosis is usually accomplished by lack of stomach visualization; Though, although none of our patients were known with microgastria before birth. Presenting symptoms vary amongst the severe GORD-like spectrum such as feeding intolerance, post-prandial vomiting, regurgitation, recurrent aspirations and respiratory symptoms, failure to thrive, and, in most severe cases, death. An upper GI swallowing contrast or a CT scan may provide the definitive diagnosis. Eventually, a secondary megaoesophagus may be visualized in delayed diagnoses. Consistently, in two of our cases severe GORD symptoms led to the direct visualization of a smaller stomach by an upper GI swallowing contrast, unfortunately, we were not able to display imaging pictures of academic interest because of the impossibility of retrieving images from 2006 in our system for patient 2 and occurrence, immediately after contrast injection, of severe reflux and vomiting requiring sudden conclusion of the exam and neonatologist intervention in Patient 3. The therapeutic goal aims to achieve full enteral autonomy either by a wait-and-see approach when mild microgastria, or by creating a neo-stomach with a major surgical reconstruction procedure. Considering the rarity of the condition with less than 60 cases described, gold standards and best treatment options tailored to the patient's specific anatomy are lacking. The conservative approach is established thanks to a combination of PN and EN (per NGT, NJT, gastrostomy, or jejunostomy). The purpose is to provide nutritional support while awaiting further gastric growth, stimulating dilatation and compliance, thus inducing tolerance to the enteral load. Sometimes, when there is a high comorbidity burden, this option is only available as the patient might not survive major procedures. In fact, according to Ruckzynsky *et al.*, mortality is higher in patients who were treated conservatively, which reflects the biased population. This was also the case of our Patient 2 in Table 1. In this case, the cause of death was recorded as acute respiratory failure. The already mentioned surgical techniques (TGD and HL pouch) are associated with an acceptable degree of symptom resolution and a good growth pattern at follow-ups,[1,5,6,9] though reports detailing nutritional and long-term outcomes are very few.[1] In our case series, Patient 3 underwent an early HL surgery and fully recovered his nutritional status which anyhow has shown a deflection at two months of age preoperatively with his weight having stepped down to the 25th percentile from the 50th percentile at birth. On the other hand, Patient 1 underwent the same technique at the age of 17 months and after the establishment of a severe growth deficit. Then, throughout infancy and adolescence, from the nutritional point of view, she has been struggling to keep up with her nutritional requirements having necessitated some sort of nutritional support for the majority of her life until the age of 16 years.

The findings are consistent with data supporting the definitive recovery of the oral intake tolerance post-surgery when compared to the conservative approach, but this is not fully accompanied in adulthood by a full reversal of the physical retardation set in infancy.[5] In the specific, our Patient 1 in Table 1 has never been able to reach a BMI higher than 18 and is affected by iron and vitamin B12 malabsorption. Nonetheless, despite multiple hospitalizations and long-term EN per NGT, she rated her quality of life as unimpacted and showed a GIQLI index of 111 which is fairly above the average of 86 stated by healthy volunteers.[21] Moreover, IGSQ for Patient 3 scored 13, which is the value reported by the author as "no GI distress". Health-related quality of life and patient-reported outcomes are now vital components of medical assessment as a comprehensive evaluation of the physical, emotional, and social functioning status of the human body. Our finding is comparable to a recent case report presented by the Australian group[22] of an 18-year follow-up of a patient affected by microgastria. Similarly, the patient remained under the 3rd percentile of growth but reports good quality of life. However, in our case, a decision was made not to expose the patient to further radiation, and no upper GI contrast studies were performed.

We presented the patient/parent's perspective of two cases operated for congenital microgastria at one institution in 1995 and in 2020 by employing two different validated quality-of-life tools that help estimate the disease impact on patients and families, detecting changes in their functional status and general well-being and comparing different treatments.

Interestingly, it has been reported that poorer preoperative gastrointestinal conditions result in maximum GIQLI gains.[23] An intrinsic limitation of studying extremely rare conditions such as congenital microgastria is the reproducibility of results would in a larger population.

Conclusions

Thanks to the information derived from the literature, which is confirmed by our cases, we support the concept that the outcome in congenital microgastria, except in syndromic cases, may significantly depend on an earlier surgery. However, the occurrence of dumping syndrome and iron and vitamin B12 malabsorption may be addressed either by the procedure itself or by a higher severity

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of stomach underdevelopment. Clarifying the origin of the residual intestinal symptoms may be the rationale for further studies. From our retrospective analysis and follow-up, we presented the nutritional status and quality of life in two cases of congenital microgastria operated according to Hunt-Lawrence at a 2-year and 27-year distance, which is the longest follow-up reported to date. The follow-up showed that the quality of life of these patients is (with the limitations of the extremely low number population) unimpacted. Patient's/parents' perspective, which is an important endpoint in healthcare and valuable for both researchers and clinicians, offers a new insight into this extremely rare condition by showing the impact of healthcare interventions on patients' lives rather than just on their bodies.

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