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CASE REPORT

Case study: nutrition and hydration support in a child with Cornelia de Lange and short bowel syndrome on home parenteral nutrition

Cecile van Niekerk, Christa Ellis* and Claire Martin

Department of Human Nutrition, University of Pretoria, Pretoria, South Africa
**Correspondence: christa.ellis@up.ac.za*

Cornelia de Lange syndrome (CdLS) (NIPBL variant) is a rare genetic disorder, characterised by intellectual and congenital abnormalities, ultimately resulting in growth and developmental delays. This case report describes a 7-year-old boy presenting with CdLS. The patient had a percutaneous endoscopic gastrostomy (PEG) placed at the age of 2 due to malnutrition, feeding difficulties, and gastro-oesophageal reflux disease. At 5 years he presented with a midgut volvulus, necessitating surgical removal of necrotic bowel resulting in short bowel syndrome (intact colon, no ileo-caecal valve and 90 cm of small bowel remaining). Over the 3-month hospitalisation period the patient was weaned from total parenteral nutrition (PN) to a home oral diet and PEG feeds in combination with supplemental PN. All meals were fed orally and finished via the PEG. Combined feeding (oral, enteral, and parenteral) management resulted in a 1.5 kg (9.9–11.4 kg) weight gain over the 3-month hospitalised period. Mid upper arm circumference improved from −5.7 Z-score to −2.7 Z-score. Despite increases in food intake and PEG feeds, a PN dependency index of 68% indicated a continued reliance on supplemental PN. This unique case illustrates the simultaneous feeding via three administration routes while transitioning from hospital to home-based care.

Keywords: Cornelia de Lange syndrome, parenteral nutrition, short bowel syndrome

Introduction

Cornelia de Lange syndrome (CdLS) is a rare genetic disorder with a prevalence varying between 1 in 10 000 and 1 in 30 000 live births.[1] It is characterised by multisystem involvement, with physical, cognitive, and behavioural effects.[1] Although CdLS exists on a spectrum, classical presentation includes distinct craniofacial features and limb malformations.[2]

CdLS increases the risk of gastrointestinal (GI) malformations, such as duodenal atresia and congenital diaphragmatic hernia.[2] In patients with short bowel syndrome (SBS), nutritional management involves parenteral nutrition (PN), with a variety of factors influencing duration of PN dependency.[3]

Case study

Patient C was diagnosed with CdLS (NIPBL variant) at birth. Cleft palates are prevalent in 20% of CdLS,[2] and Patient C had a cleft palate repair between 1 and 2 years of age. At the age of 2, he was diagnosed with severe gastro-oesophageal reflux disease (GERD) following an upper gastro-intestinal endoscopy. This diagnosis was anticipated, given that 71% of individuals with CdLS are affected by GERD.[4]

GERD is known for persistent feeding difficulties that worsen over time.[2] As a result of the GERD, Patient C was only able to tolerate pureed foods. In Patient C, the presence of GERD, compounded by multiple surgeries (Figure 1) resulted in a diagnosis of severe acute malnutrition (SAM) at age two.

At the age of two, a percutaneous feeding gastrostomy (PEG) was placed post failed Nissen fundoplication, to assist with his feeding and growth. He exhibited delayed growth, lagging motor development milestones, hearing loss, and weakened immunity. It is important to note that children affected by CdLS commonly present with impaired growth. Their birthweight is usually below the 5th percentile and their height, weight, and head circumference will usually remain below the ranges for the general paediatric population. Patient C's growth is also influenced by his NIPBL variant, which usually affects growth more than the SMC1A variant. Growth hormone secretion is usually normal in CdLS.[2]

Nutritional management after intestinal resection: early acute and intermediate postoperative phase

In June 2022, at the age of 5½ years, he presented with a medical emergency, due to intestinal malrotation and midgut volvulus. Intestinal necrosis necessitated removal of the terminal ileum including the ileocaecal valve. He was left with an intact colon and 90 cm of small bowel, classifying the short bowel syndrome (SBS) as jejuno-colic type II.[3] During the early acute phase of PN support (0–48 hours post-surgery) the aim was to manage his fluid status and re-establish his normal physiological state.[3]

When he presented with the midgut volvulus he was growing on the 50th percentile weight and height-for age on CdLS-specific growth charts.[5] During the two months of hospitalisation, his weight decreased from 11.4 kg to 9.9 kg. His mid upper arm circumference (MUAC) plotted on the −5.7 Z-score (Table 1). At less than -3 Z-score, this classified him as having severe acute malnutrition (SAM) according to the World Health Organization.[6] High ileostomy output (> 1l/day with magnesium and sodium depletion), severe failure to thrive, liver dysfunction, and infection delayed the progression onto full enteral nutrition (EN) feeds. During this time, he also required multiple resuscitations, ventilation, and a tracheostomy, which was ultimately reversed as his clinical condition improved and he was able to breathe independently.

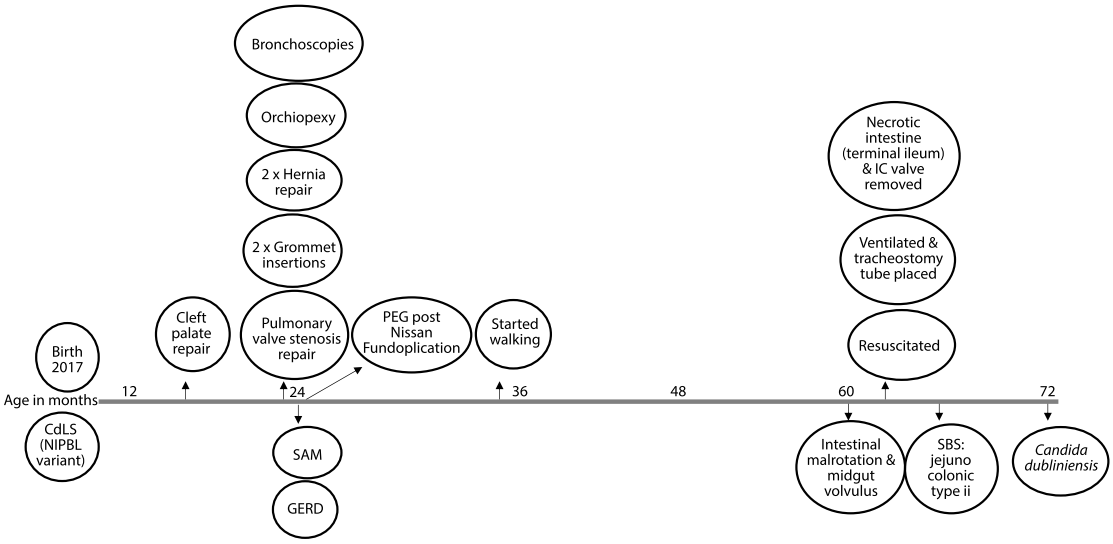

Figure 1: Patient C timeline of key medical events.

Nutritional management after intestinal resection: late postoperative phase

At the end of August 2022 (two months later), he was transferred to a specialised Paediatric Intestinal Failure unit for advanced nutritional support, to address increasing nutrient deficits, promote tissue repair and catch up growth.[3]

At this point in September 2022, he was receiving PN at 90 ml/kg/day which supplied 137 kcal/kg/day, consisting of 2 g/kg/day amino acids, 2 g/kg/day SMOF® lipid, and 9.5 g/kg/day carbohydrates. When bowel sounds returned, and stool output was 4–5 semi-loose stools daily, enteral glutamine and extensively hydrolysed EN was started at 0.5 g/kg/day and 30 ml/kg/day respectively. Over the next 3 months, he progressed to a polymeric oral nutrition supplement and soft pureed foods via his PEG. By December 2022, his nutrient requirements were met with 60% PN and 40% EN. The EN was mainly from ±100 ml soft pureed meals three times per day, ± 65 ml soft pureed snacks twice per day, 190 ml polymeric nutritional supplement and 200 ml/day oral rehydration solution (ORS).

One of the primary goals in SBS is to achieve euhydration. The normal mechanisms that regulate homeostatic fluid balance are altered in patients with SBS.[7] Hypertonic and hypotonic fluids are both problematic for patients with SBS as both exacerbate stool losses. ORS is recommended to improve hydration in patients with SBS. Most of Patient's C intake was per mouth, and the PEG was used as required, usually for breakfast and those foods and drinks that he did not want to take per mouth.

Due to his dependency on PN (cycled to minimise hepatic damage)[8,9] to meet ±60% of his nutritional requirements (1.5 g/kg/day protein, 55 kcal/kg/day total energy, 70 ml/kg/day fluid from TPN), he was assessed for home PN using suitability criteria from the European Society for Pediatric Gastroenterology Hepatology and Nutrition (ESPGHAN guidelines).[3]

Intestinal rehabilitation with home PN and EN

In December 2022, 108 days from his initial SBS diagnosis, he was discharged on home PN, which was administered with an 18-hour cycle. Although food variety was limited, and fruit and dairy were excluded, he tolerated 3 × 150–180 ml pureed meals and 3 pureed snacks at ± 100 ml. He also required ORS at 200–300 ml/day to maintain euhydration. Slowly, small changes were made to test his tolerance of specific fruit- and lactose-free dairy products. As food intake increased, his PN cycle was reduced to 16 hours by March 2023.

By April 2023, food intake increased to 180–200 ml and 125 ml pureed meals and snacks respectively, with the inclusion of lactose-free milk products and small amounts of fruit. His nutrient intake from both EN and PN was 100–120 ml/kg/day which provided 2.5–3.0 g/kg/day protein, and 100–120 kcal/kg/day total energy. His stool output remained steady (3–4 soft, formed stools) and his growth improved as indicated by an improved MUAC of – 3 Z-score from – 4 Z-score (see Table 1). With this improved intake and absorption, PN was decreased to provide ±50% of his requirements.

Unfortunately, at the end of April 2023, PN was stopped for 5 days due to *Candida dubliniensis* infection of the central line. Although food intake remained constant, without the PN and a higher stool output his weight decreased. When PN was restarted, he was discharged on the plan of 50% nutrients from PN and 50% from oral intake. His stool output, however, remained high and losses were more than intake could sustain. Management included increasing his PN to meet 60% of his requirements, decreasing food portions to 150–180 ml servings, increasing ORS to 750 ml/day and adding loperamide. His stool output decreased and weight improved thereafter.

By October 2023, 15 months after his midgut volvulus, he was tolerating PN at 50% of his requirements and a shorter cycle of PN and a new ORS was attempted. However, he did not tolerate either and became dehydrated. The Parenteral Nutrition Dependency index (PNDI) score was calculated at 68%, which indicated that he was still dependent on PN.[7] His nutritional management remained unchanged for the following four months.

In March 2024, at 7 years of age and almost 2 years post SBS diagnosis, his oral food and drink intake was ± 150% of nutrient requirements, which is not unusual and due to adaptive hyperphagia.[7] It did, however, worsen his diarrhoea and keeping a

Table 1: Postoperative anthropometric measurements

Date	**30.6.22**	**31.08.22**	**26.09.22**	**28.10.22**	**21.11.22**	**27.04.23**	**1.05.23**	**10.05.23**	**15.09.23**	**16.10.23**	**13.03.24**
Phase	**Early postoperative phase**	**Intermediate postoperative phase**	**Late postoperative phase**	**Intestinal rehabilitation phase**							
Age (months)	64	65	66	67	68	74	75	76	78	79	83
Weight (kg)	11.4	9.9	10.8	11.1	11.4	11.2	10.9	10.8	13.6	12.7	14.4
Height (cm)	95	98	99	100	100	101	101	102	103	103	104
MUAC (cm)	10.9	11	12	12.3	13.2	12.7	11.5	12.1	13.5	13.3	14.2
MUAC (Z-score)	−5.8	−5.7	−4.4	−4.0	−3.08	−3.9	−5.7	−4.9	−3.2	−2.9	−2.7
Classification according to MUAC	SAM	SAM	SAM	SAM	SAM	SAM	SAM	SAM	SAM	MAM	MAM
BMI (kg/m^2)	12.6	10.3	11.0	11.1	11.4	10.9	10.6	10.4	12.8	11.9	13.3
WFA Percentiles on CdLS growth chart	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile	> 5th centile
HFA Percentiles on CdLS growth chart	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile	50th centile

*SAM: severe acute malnutrition (MUAC -3 Z-score and below).
*MAM: moderate acute malnutrition (MUAC -2 Z-score and below).

food diary helped to focus dietary education. Protein provided ±15% of total energy for catch-up growth and development. Carbohydrate and fat contributed 50% and 35% of his EN intake respectively. He consumed foods from all food groups except concentrated sugar sources and lactose-containing dairy products as these continued to increase stool output. He was tolerating pureed meals and snacks both orally and via his PEG, providing 50% of his requirements. A 12-hour cycle of PN provided the remainder of his nutrient requirements. He also received 750 ml of ORS daily, taken in small doses between meals to improve absorption.[7,9] Every six months, his diet is re-evaluated according to his specific growth requirements and currently he is thriving and developing appropriately for a child with CdLS.

Discussion

Feeding difficulties and poor growth are commonplace in CdLS.[2] Nutrition therapy for SBS is based on meticulous fluid administration and individualised PN and EN. There is limited information on macro- or micronutrient requirements for developmentally delayed children with SBS. The Schofield equation is often used to determine the resting energy expenditure.[8] In Patient C, because he was growing on the 50th percentile (CdLS growth chart) prior to his diagnosis of SBS, weight for age at the 50th percentile rather than actual weight was used to provide additional calories required for catch-up growth.

PN is the first line of treatment in SBS, aiming to promote somatic growth until intestinal autonomy is achieved.[8] For physiological adaptation of the remaining intestine, feeding into the GI tract is key.[3,7]

EN promotes site-specific compensatory changes that maintain the absorptive function of the remaining small bowel. As the timing and composition of EN influence EN autonomy, EN in Patient C was initiated early after intestinal resection. Nutritional strategies such as glutamine, complex proteins, enteral fat, and soluble fibre were used to aid with intestinal adaptation. Sodium from the ORS influenced the diarrhoea symptoms and pancreatic–biliary function and was intended to favour the intestinal adaptation process.[10] Feed volume was gradually increased based on tolerance as assessed by stool number and volume, vomiting, and abdominal distention.[7–9] Unfortunately he has not yet managed to achieve intestinal autonomy after 2 years of PN in combination with oral intake as indicated by the PNDI score.

Conclusion

Patient C is currently achieving his desired growth velocity, but potential for complications in CdLS with SBS exists. D-lactic acidosis is a rare but serious intermediate complication of paediatric SBS,[11] while Barret's oesophagus (due to uncontrolled GERD), can develop later. Due to the high risk of growth faltering in patients with CdLS, growth monitoring forms a vital part of management. Lifelong medical and multidisciplinary care will improve his health care and increase his quality of life.

Disclosure statement – No potential conflict of interest was reported by the authors.

ORCID

Christa Ellis http://orcid.org/0000-0001-9653-539X
Claire Martin http://orcid.org/0000-0001-9517-8968

References

1. Kline AD, Grados M, Sponseller P, et al. Natural history of aging in Cornelia de Lange syndrome. *Am J Med Genet Part C Semin Med Genet*. 2007;145C:248–60. https://doi.org/10.1002/ajmg.c.30137
2. Kline AD, Moss JF, Selicorni A, et al. Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. *Nat Rev Genet*. 2018;19:649–66. https://doi.org/10.1038/s41576-018-0031-0
3. Norsa L, Goulet O, Albertini D, et al. Nutrition and intestinal rehabilitation of children with SBS: a position statement of the ESPGHAN committee on nutrition. Part 1: from intestinal resection to home discharge. *JPGN*. 2023;77:281–97.
4. Huisman SA, Mulder PA, Redeker E, et al. Phenotypes and genotypes in 51 individuals with SMC1A variants. *Am J Med Genet*. 2017;173:2108–25. https://doi.org/10.1002/ajmg.a.38279
5. Kline AD, Barr M, Jackson LG. Growth manifestations in the Brachmann-de Lange syndrome. *Am J Med Genet*. 1993;47:1042–9. (CdLS growth charts). https://doi.org/10.1002/ajmg.1320470722
6. WHO. *Guideline: updates on the management of severe acute malnutrition in infants and children*. Geneva: World Health Organization; 2013.
7. Roberts K, Shah N, Parrish CR, et al. Navigating nutrition and hydration care in adult patient with short bowel syndrome. *Nutr Clin Pract*. 2023;38:S59–S75. https://doi.org/10.1002/ncp.10951
8. Norsa L, Goulet O, Albertini D, et al. Nutrition and intestinal rehabilitation of children with SBS: a position statement of the ESPGHAN committee on nutrition. part 2: long term follow upon home parenteral nutrition. *JPGN*. 2023;77:298–314.
9. Saayman BD, Millar AJW, van Niekerk E. Nutritional interventions and outcomes of children with SBS in a tertiary hospital setting in SA. *SAJCN*. 2023;36(2):76–83.
10. Caporilli C, Gianni G, Esposito GF, et al. An overview of short-bowel syndrome in pediatric patients: focus on clinical management and prevention of complications. *Nutrients*. 2023;15:2341–64. https://doi.org/10.3390/nu15102341
11. Nes E, Knell J, Keefe G, et al. Factors associated with D-lactic acidosis in pediatric intestinal failure: A case-control study. *J Pediatr Gastroenterol Nutr*. 2024;78(2):217–22. https://doi.org/10.1002/jpn3.12075

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