Portuguese Prevalence of Pediatric Chronic Intestinal Failure

To the Editor: Intestinal failure (IF) is defined as the inability of the gut to absorb sufficient water, macronutrients (carbohydrate, protein, and fat), micronutrients, and electrolytes, and thus a requirement of intravenous supplementation or replacement in order to sustain life (1). Short bowel syndrome is the most common cause of IF (2). A pediatric review dated of 2012, from Germany, reported that the prevalence of chronic IF (CIF) in children is 13.7 per million (3).

The condition is incompatible with life in the absence of the safe application of specialized and individualized medical therapy (4). Long-term home parenteral nutrition (PN) has dramatically improved the life expectancy and quality of life in children with CIF. The considerable costs, however, of PN bags and burden of necessary-associated healthcare, such as central venous catheters, with consequent risks of infection and thrombosis, should not be dismissed. New drugs or treatment strategies are essential for these patients.

In order to be registered in a database to benefit from clinical trials and new treatment strategies, the Portuguese Pediatric Gastroenterology, Hepatology and Nutrition Society (SPGP) proposed this Pediatric CIF multicenter study. In December 2018, all units responsible for the follow-up of all Portuguese children with CIF met in the first national CIF meeting of SPGP. The aim of the meeting was to enable the pediatric national multicenter group to increase knowledge of the field and share their experiences as well as to merge their data, including to determine the prevalence of CIF in Portuguese children, which had been unknown until recently. The newly developed database identified 51 children/adolescents (under 18 years of age) requiring PN in order to live because of CIF with a male to female ratio of 1.1 : 1. Using data from the National Statistics Institute in 2018, Portugal had 1 960 310 inhabitants less than 18 years old. Therefore, the data suggested a prevalence of CIF in Portugal of 27 patients per million inhabitants younger than 18 years.

The majority of the 51 patients had short bowel syndrome (n = 38). The most frequent cause for short bowel syndrome was intestinal volvulus (16) followed by congenital intestinal atresia (11), gastroschisis (6), and necrotizing enterocolitis (5). Ages ranged from 3 months to 17 years old. At the time of submission of this letter, all but 2 of the children remained on PN with partial enteral nutrition. Six paediatric patients were still in the hospital (1 for social reasons). Seven patients started teduglutide during 2019 resulting in 2 discontinuing PN and the remaining reducing their requirement for PN (1 reducing the frequency to 4 days/week and 2–3 days/week as well as 2 reducing the daily PN volume).

The remainder of the 51 patients had chronic intestinal pseudo-obstruction (6), Hirschsprung disease (2), Mitchell Riley syndrome (2); very early onset Inflammatory Bowel Disease (VEOIBD) with Interleukin 10 receptor and Syntaxin-Binding Protein 3 deficiency (2) and Ewing sarcoma (1). Both children with VEOIBD have subsequently had haematopoietic stem cell transplantation, of which one recently came off PN but the other died. The child with the Ewing Sarcoma suffered an intestinal obstruction with peritonitis and sepsis requiring intestinal resection and although he had more than 60 cm of residual small intestine continued to need PN.

Our data provides the first ever data about the prevalence of CIF and associated PN use in Portugal. It allows the SPGP to compare these data with other European countries and provides the opportunity to improve the medical care and quality of life for Portuguese patients and their families. Finally, it provides the Portuguese government a tool to develop a future strategy to deal with this complex medical condition.

Acknowledgments: The SPGP multicenter study on chronic intestinal failure won the SPGP 2018 research grant sponsored by Danone Specialized Nutrition Portugal and the Jaime Salazar de Sousa SPGP 2019 research grant sponsored by Nestlé, Portugal. We acknowledge Carla Maia, MD, Hospital Pediátrico de Coimbra, for attending the national CIF meeting in December 14, 2018 in Braga, Portugal.

[*†]Henedina Antunes, [‡]Sara Nóbrega, [‡]Miguel Correia, [‡]António Pedro Campos, [‡]Raul Silva, [§]Paula Guerra, [||]Paula Mourato, [||]Ana Isabel Lopes, [¶]Helena Mansilha, and [¶]Mónica Tavares

[*]*Gastrenterology, Hepatology and Nutrition Pediatric Unit and Clinic Academic Center, Hospital de Braga, Braga*

[†]*Life and Health Sciences Research Institute (ICVS); ICVS/3B's - Associate Laboratory and School of Medicine, University of Minho, Braga, Portugal*

[‡]*Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisboa*

[§]*Centro Académico Hospitalar S. João, Porto*

[||]*Departamento de Pediatria do Centro Hospitalar Universitário de Lisboa Norte, EPE (CHLN), Lisboa*

[¶]*Unidade de Nutrição, Centro Materno Infantil do Norte (CMIN), Porto, Portugal*

REFERENCES

1. Allan P, Lal S. Intestinal failure: a review. *F1000Res* 2018;7:85.
2. Batra A, Key SC, Johnson MJ, et al. Epidemiology, management and outcome of ultrashort bowel syndrome in infancy. *Arch Dis Child Fetal Neonatal Ed* 2017;102:F551–6.
3. Krawinkel M, Scholz D, Busch A, et al. Chronic intestinal failure in children. *Dtsch Arztebl Int* 2012;109:409–15.
4. Merritt RJ, Cohran V, Raphael BP, et al., Nutrition Committee of the North American Society for Pediatric Gastroenterology, Hepatology and NutritionIntestinal rehabilitation programs in the management of pediatric intestinal failure and short bowel syndrome. *J Pediatric Gastroenterol Nutr* 2017;65:588–96.

Two Novel *PNLIP* Mutations Causing Congenital Lipase Deficiency in Identical Twin Boys

To the Editor: Congenital pancreatic enzyme deficiencies are rare causes of exocrine pancreatic insufficiency in children. There is only 1 described mutation in the pancreatic lipase gene, *PNLIP*, reported in the literature. In 2014, Behar et al (1) described a homozygous missense mutation at amino acid 221 in *PNLIP* causing congenital lipase deficiency in 2 brothers of consanguineous parents. In 2015, the mutation was studied in cellular and