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Preface

Congenital gastrointestinal disorders. Why is it relevant to adult gastroenterologists?

This special issue of the journal aims to give a comprehensive review on the main congenital disorders of the liver, esophagus, gut, and pancreas. All these diseases are rare (prevalence <1/2000), and most of them express clinically early in life. We gathered a panel of renowned international experts in the field—all pediatric gastroenterologists or pediatric surgeons—to cover both practical issues and perspectives. There are at least five reasons why these articles are published in a general gastrointestinal (GI) journal, mostly read by gastroenterologists:

1 The genetic revolution and recent advances in prenatal diagnosis, pediatric endoscopy, and imaging as well as motility assessments result in better and earlier diagnosis and the pathophysiology understanding of many of these diseases. Practical consequences are prenatal diagnosis (*e.g.,* esophageal atresia and cystic fibrosis), family screening (*e.g.,* Wilson disease, several congenital inherited hepatic disorders, hereditary pancreatitis, and cystic fibrosis), genotype-phenotype correlation, and finally personalized medicine.
2 Dramatic improvement in care has been observed over the past 20 years (*i.e.,* neonatal care, pediatric surgery, and liver transplantation) allowing most of these children to survive through adulthood. Most congenital gastrointestinal disorders are no more pediatric diseases but adult diseases with life-long problems. Severe complications that were not observed in the past because only a few patients reached adulthood are nowadays a concern (*i.e.,* esophageal cancer in patients with esophageal atresia, cirrhosis and hepatobiliary tumor in patients with biliary atresia surviving with their native liver, and diabetes mellitus in patients with cystic fibrosis). Other complications are *de novo* diseases as consequences of treatment during childhood (*i.e.,* bone and liver disease secondary to prolonged parenteral nutrition, renal failure, lymphoma, and skin cancer after liver/intestinal transplantation). Management is multidisciplinary and best performed in specialized referral centers with expertise in relevant nutritional, medical, and surgical therapy—involving gastroenterologists—as well as psychosocial support to improve outcomes.
3 Transition at adolescence to adult medicine is, therefore, a new challenge. This is a critical period where the risk of poor compliance and loss to follow-up are high. This should be anticipated, and a good transition requires training and preparation of the adolescent and family and a multidisciplinary team involving gastroenterologists.
4 Current therapies (pharmacological, surgical, and transplantation) need to be optimized and ideally tailored to individual patients.
5 Despite recent advances in treatment (surgery, transplantation, supportive care, and drugs), there is a significant unmet medical need for many of these diseases. Very promising novel therapies are expected in the coming years. It includes cell-based therapy as an alternative to liver transplantation, targeted genetic therapy in cystic fibrosis, tissue engineering in esophageal atresia, and new pharmacological therapies in biliary atresia or Wilson disease. These approaches with a focus on individualized and disease-specific strategies have yielded promising results in preclinical studies and need to progress toward proof of concept in patients and first in human clinical trials.

We hope this special issue will facilitate a closer collaboration between adult and pediatric health care providers to improve care and outcomes of patients presenting with a congenital gastrointestinal disorder.

Last but not least, gastroenterologists should be aware that these patients have a high level of knowledge about their disease. They expect from health professionals not only competence but also listening and availability. Most patients have gone through difficult times during childhood and adolescence. Knowledge and skills are not enough; positive attitude, empathy, and compassion are also needed.

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