Advances in Pediatrics 60 (2013) 217–261

ADVANCES IN PEDIATRICS

Enteropathies of Infancy and Childhood

Pierre Russo, MD

Department of Pathology and Laboratory Medicine, The Children's Hospital of Philadelphia, The University of Pennsylvania Perelman School of Medicine, 324 South 34th Street, Main Building, Room 5NW16, Philadelphia, PA 19104, USA

Keywords

• Intestinal biopsy • Esophagogastroduodenoscopy • Intestinal disorders • Children

Key points

- Esophagogastroduodenoscopy and intestinal biopsy have become routine in the investigation of pediatric gastrointestinal disorders.
- Intestinal biopsy remains the gold standard for the diagnosis of most disorders that cause chronic diarrhea and malabsorption in children.
- Many disorders that cause intractable diarrhea of infancy have been described in the past few decades.
- Understanding the biology and molecular mechanisms of some of these disorders has provided important insights into intestinal function.

INTRODUCTION AND GENERAL REMARKS

The aims of this article are to review some of the entities that cause chronic intractable diarrhea and malabsorption of infancy and childhood, illustrating their characteristic features on small intestinal biopsies and specimens. These are listed in Box 1.

Chronic diarrhea, usually defined as lasting more than 2 weeks, can present particularly difficult diagnostic and therapeutic challenges, especially when it occurs in the neonate. Intractable diarrhea of infancy is a term coined by Avery and colleagues [1] to refer to these cases that at the time remained mainly undiagnosed and were associated with a high mortality. Better recognition and understanding of many of the disorders that cause intractable diarrhea of infancy, combined with advances in parenteral nutrition, immunosuppression, and the availability of bowel transplantation has improved the survival of many of these children, thus prompting the need for timely and accurate

E-mail address: russo@email.chop.edu

0065-3101/13/$ – see front matter
http://dx.doi.org/10.1016/j.yapd.2013.04.010 © 2013 Elsevier Inc. All rights reserved.

Box 1: Enteropathies of infancy and childhood

Congenital transport and enzymatic deficiencies
- Glucose-galactose malabsorption
- Disaccharidase deficiency
- Lysinuric protein intolerance
- Abetalipoproteinemia
- Chylomicron retention disease
- Sodium chloride diarrhea
- Primary bile acid malabsorption

Inherited epithelial defects
- Microvillus inclusion disease
- Tufting enteropathy
- Enteroendocrine cell dysgenesis

Autoimmune enteropathy

Gluten-sensitive enteropathy (celiac disease)

Eosinophilic enteritis

Lymphangiectasia

Metabolic diseases and tumors

diagnosis. Furthermore, investigation of some of these disorders has led to significant advances in related fields of disease pathogenesis, transcending their relative clinical rarity. For example, the discovery that mutations in *FOXP3* underlie IPEX syndrome, resulting in the absence of naturally occurring T regulatory lymphocytes (Tregs), has opened new avenues in our understanding of the control of the immune response, autoimmunity, and immune tolerance.

INTESTINAL BIOPSY IN CHILDREN

As pediatric esophagogastroduodenoscopy (EGD) has evolved into a routine outpatient procedure, the indications for its use have changed over the last few decades. At The Children's Hospital of Philadelphia, the first-time EGD rate increased 12-fold in a 20-year interval between 1985 and 2005, with isolated abdominal pain replacing gastrointestinal (GI) bleeding as the most frequent indication [2]. Much of this increase seems to have been driven by the dramatic increase in food allergy–related disorders such as eosinophilic esophagitis, by the increased prevalence of celiac disease and its clinically atypical forms, and by the routine use of EGD in addition to colonoscopy in the evaluation of children with suspected inflammatory bowel disease. According to a recent study from a tertiary pediatric center in the United Kingdom, the most frequent indications for endoscopy in children less than 1 year old were diarrhea, failure to thrive, reflux, and rectal bleeding [3]. Histologic

Download English Version:

https://daneshyari.com/en/article/4140299

Download Persian Version:

https://daneshyari.com/article/4140299

Daneshyari.com