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Opinion

TUFTING ENTEROPATHY AT BIRTH: MECHANISMS, PATHOGENESIS, AND BIOLOGY

Evan Vas*

Department of Dermatology and Cutaneous Surgery, University of Miami School of Medicine, Miami, Florida, USA

INTRODUCTION

An infantile autosomal recessive condition known as congenital tufting enteropathy (CTE) results in severe intestinal failure, electrolyte imbalances, and impaired growth. Villous atrophy, crypt hyperplasia, and focal epithelial tufts composed of densely packed enterocytes are typical histological characteristics of CTE. This disease is thought to be caused by mutations in the EPCAM and SPINT2 genes. A deeper comprehension of the disease's pathophysiology is required due to the significant morbidity and mortality as well as the absence of direct treatments for CTE patients. Clinical aspects, disease genetics, and research model systems are all reviewed in this systematic review of the most recent CTE biology knowledge. The predicted mechanisms of CTE and its pathogenesis are given special attention because they could shed light on potential new treatments. The role of intestinal cell differentiation, enterocyte defects, a broken barrier and cell-cell junction, cell-matrix adhesion, and other aspects of intestinal homeostasis are all well-described here (see Graphical Abstract). In addition, potential mechanistic pathways that may contribute to the pathogenesis of CTE due to either loss of function or EpCAM mutation are highlighted based on the known dynamics of EpCAM signaling. These pathways give us a better understanding of this terrible disease, even though they don't fully explain it. Diarrhea continues to be the leading cause of malnutrition and the second leading cause of death among children under the age of five, as stated by the World Health Organization (WHO). Congenital diarrhea and enteropathies (CODEs), in contrast to the more treatable causes of diarrhea, are hereditary, monogenic disorders that frequently result in life-threatening intestinal failure in infants. CODES patients, especially those with epithelial cell defects, typically present within the first few months of life.

DISCUSSION

Defects in epithelial transporters, enzymes and metabolism, epithelial trafficking and polarity, enteroendocrine cell dysfunction, and/or immune cell dysregulation are all hallmarks of CODEs and share common pathophysiology. Congenital tufting enteropathy (CTE) is a severe disorder that falls under the defective enterocyte trafficking and polarity category of CODEs, along with microvillus inclusion disease. CTE, also known as intestinal epithelial dysplasia, is a rare autosomal recessive disease of infancy that causes severe, watery diarrhea, imbalances in electrolytes, and stunted growth. One of several infantile intractable diarrheal diseases, CTE affects between

1/50,000 and 100,000 live births in western Europe though it occurs more frequently in the Middle East. Patients suffer from intestinal failure, which necessitates parenteral nutrition and, in some instances, intestinal transplant, both of which carry inevitable risks of liver disease, infection, rejection, and vascular complications [1,2].

The diarrhea is both osmotic and secretory. In addition, the majority of patients never attain enteral autonomy. The small intestinal epithelium's characteristic structural changes are the basis for the diagnosis of CTE. Villous atrophy and crypt hyperplasia are typical findings, but focal epithelial tufts are the most recognizable pathologic abnormalit. CTE, also known as epithelial dysplasia, was reportedly discovered despite the fact that a disease phenotype that is historically comparable was first documented when it was referred to as familial enteropathy. Villous atrophy, crypt hyperplasia, the absence of brush-border villous enterocytes, an increase in inflammatory cells in the laminar propria, and persistent diarrhea were all reported by patients with familial The patients had a family connection, as suggested by the term "familial enteropathy. The disease was reported in two patients who were the children of consanguineous marriages confirming the hereditary nature of CTE. CTE is primarily caused by mutations in the epithelial cell adhesion molecule (EPCAM. The majority of CTE patients have mutations in EPCAM, despite the fact that SPINT2 (Serine Peptidase Inhibitor Kunitz mutations have also been reported in a syndromic form of the disease. A recent review suggests that 74% of patients reported have mutations in EPCAM, while 26% have mutations in SPINT2, despite the fact that not all patients are reported in the literature. It has been nearly 25 years since congenital tufting enteropathy was first identified, and more than a decade since the genetic etiology of the disease was discovered. Despite this, the disease's management has not changed much because the pathobiology and clinical aspects are not fully understood [3-6].

CONCLUSION

The important but little-studied pathogenesis and disease biology of CTE, as well as its possible connection to its genetic etiology, "EpCAM," were the primary topics of this review article. There is more to CTE pathogenesis than just a change in cell adhesion and barrier defects, according to the current review. Defects in epithelial cell and enterocyte function include enterocyte disorganization, impaired enzyme and metabolism, defective epithelial trafficking/polarity, and altered cell differentiation as part of the pathogenesis. Despite the fact that the disease's pathogenesis is still poorly understood, this information opens the door to a deeper comprehension as well as therapeutic options for patients.

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Correspondence Author:

Evan Vas*
Department of Dermatology and Cutaneous Surgery, University of Miami School of Medicine, Miami, Florida, USA
E-mail: patterson@dun.ac

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