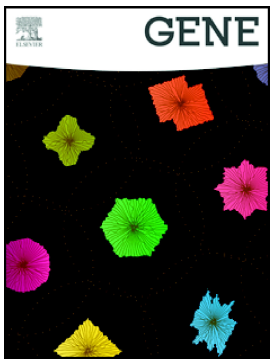

Congenital sideroblastic anemia: Advances in gene mutations and pathophysiology

Zhangbiao Long, Hongmin Li, Yali Du, Bing Han

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Congenital sideroblastic anemia: advances in gene mutations and pathophysiology

Zhangbiao Long[a*], Hongmin Li[a*], Yali Du[a], Bing Han[a†]

[a] *Department of Hematology, Peking Union Medical College Hospital, Chinese Academy of Medical Sciences and Peking Union Medical College, Beijing, 100730, China*

[†]E-mail: hanbingpumch@sina.com

[*]Zhangbiao Long and Hongmin Li contributed equally to this work

Abstract

Congenital sideroblastic anemia (CSA) is a series of rare, heterogeneous disorders, characterized by iron overload in the mitochondria of erythroblasts and ringed sideroblasts in bone marrow. In recent years, rapid development of next-generation sequencing technology brings great advance in understanding of genetic and pathophysiologic features of CSA. Based on the pathophysiology of mitochondrial iron metabolism, causative genes of CSA can be divided into three subtypes: Heme biosynthesis related; iron-sulfur cluster biosynthesis and transportation related; and mitochondrial respiratory chain synthesis related. Patients with CSA present various clinical manifestation due to relevant mutation gene and require different treatment strategies. The recognition of the causative genes and evolution of pathogenicity is critical. In this review, we summarize the recent progress in mutation genes of CSA, and its potential role in the pathogenesis, diagnosis and treatment.

Keywords: congenital sideroblastic anemia, iron metabolism, gene, pathogenesis

1. Introduction

Sideroblastic anemia is a heterogeneous group of rare disorders due to impaired heme biosynthesis or iron utilization when hemoglobin synthesis, and characterized by the presence of ringed sideroblasts in bone marrow, which reflects abnormal mitochondrial iron accumulation by the erythroblasts (Bottomley *et al.*, 2014; Furuyama *et al.*, 2002). Impaired iron utilization could disturb reduction-oxidation reaction in cellular and induce apoptosis, leading to ineffective erythropoiesis (Fleming, 2011; Harigae *et al.*, 2010), thus present