10. Ulusal Pediatrik Hematoloji Kongresi
3 – 6 Haziran 2015, Ankara

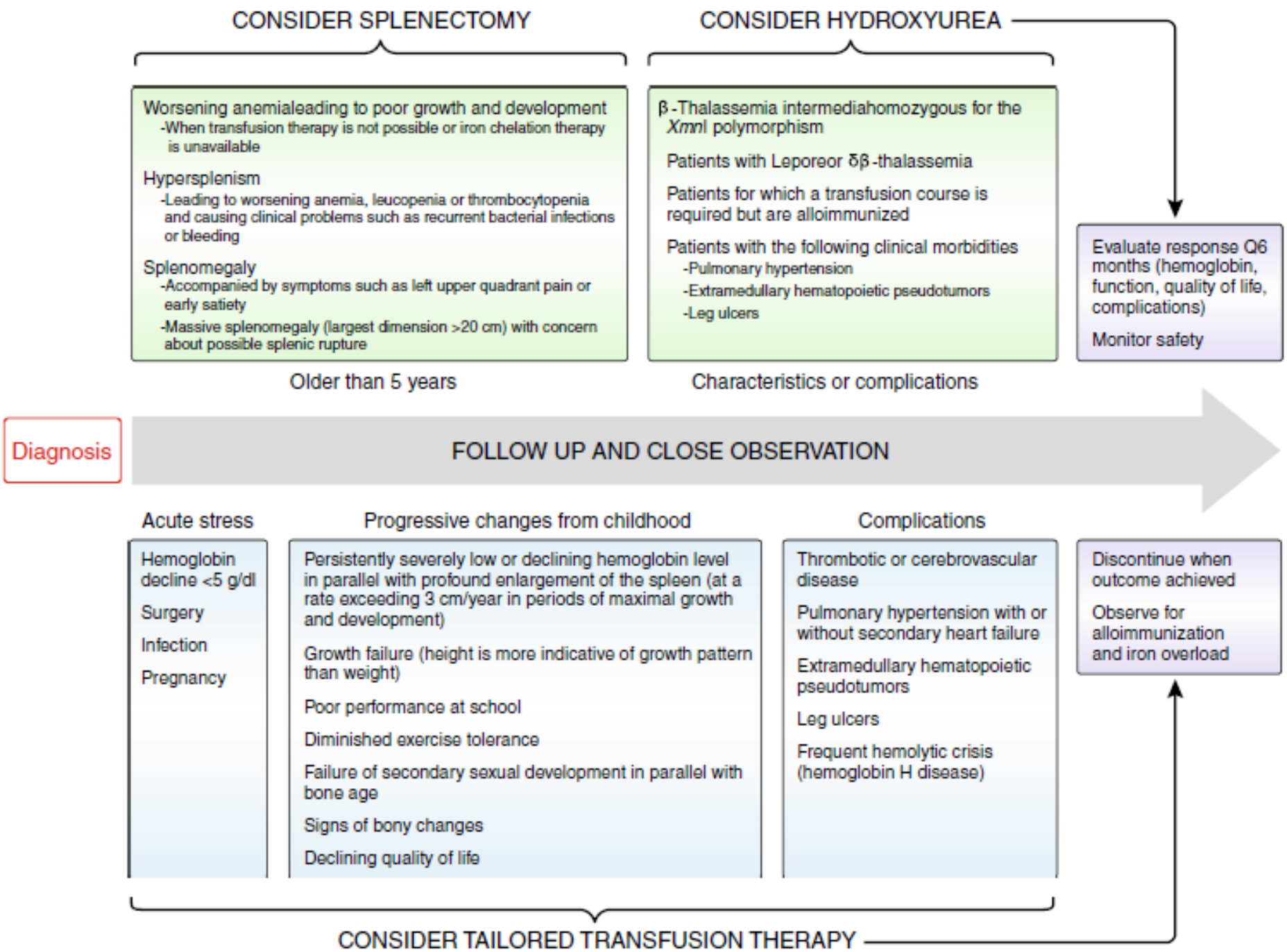
[HALE ÖREN]

BEYANI

Araştırma Destekleri/ Baş Araştırıcı	Sunumum ile ilgili çıkar çatışmam yoktur.
Çalıştığı Firma (lar)	Sunumum ile ilgili çıkar çatışmam yoktur.
Danışman Olduğu Firma (lar)	Sunumum ile ilgili çıkar çatışmam yoktur.
Hisse Senedi Ortaklığı	Sunumum ile ilgili çıkar çatışmam yoktur.
Konuşmacı Bürosu	Sunumum ile ilgili çıkar çatışmam yoktur.
Onursal Ödenti (ler)	Sunumum ile ilgili çıkar çatışmam yoktur.
Bilimsel Danışma Kurulu	Sunumum ile ilgili çıkar çatışmam yoktur.

Sunumumda aşağıda yer alan ruhsat dışı ilaç ve tıbbi cihazlar ile ilgili bilgi yer almaktadır: “Yoktur”

Ali T.Taher, Maria Domenica Cappellini Drugs (2014) 74:1719–1729.



REVIEW ARTICLE

Leaders in Hematology
review series

β-thalassemias: paradigmatic diseases for scientific discoveries and development of innovative therapies

Stefano Rivella

Department of Pediatrics Hematology-Oncology Department of Cell and Developmental Biology Weill Cornell Medical College New York, NY, USA

ABSTRACT

β-thalassemias are monogenic disorders characterized by defective synthesis of the β-globin chain, one of the major components of adult hemoglobin. A large number of mutations in the β-globin gene or its regulatory elements have been associated with β-thalassemias. Due to the complexity of the regulation of the β-globin gene and the role of red cells in many physiological processes, patients can manifest a large spectrum of phenotypes, and clinical requirements vary from patient to patient. It is important to consider the major differences in the light of potential novel therapeutics. This review summarizes the main discoveries and mechanisms associated with the synthesis of β-globin and abnormal erythropoiesis, as well as current and novel therapies.

 doi:10.3324/haematol.2014.114827
Manuscript received on January 16, 2015. Manuscript accepted on February 20, 2015.
Correspondence: str2010@med.cornell.edu

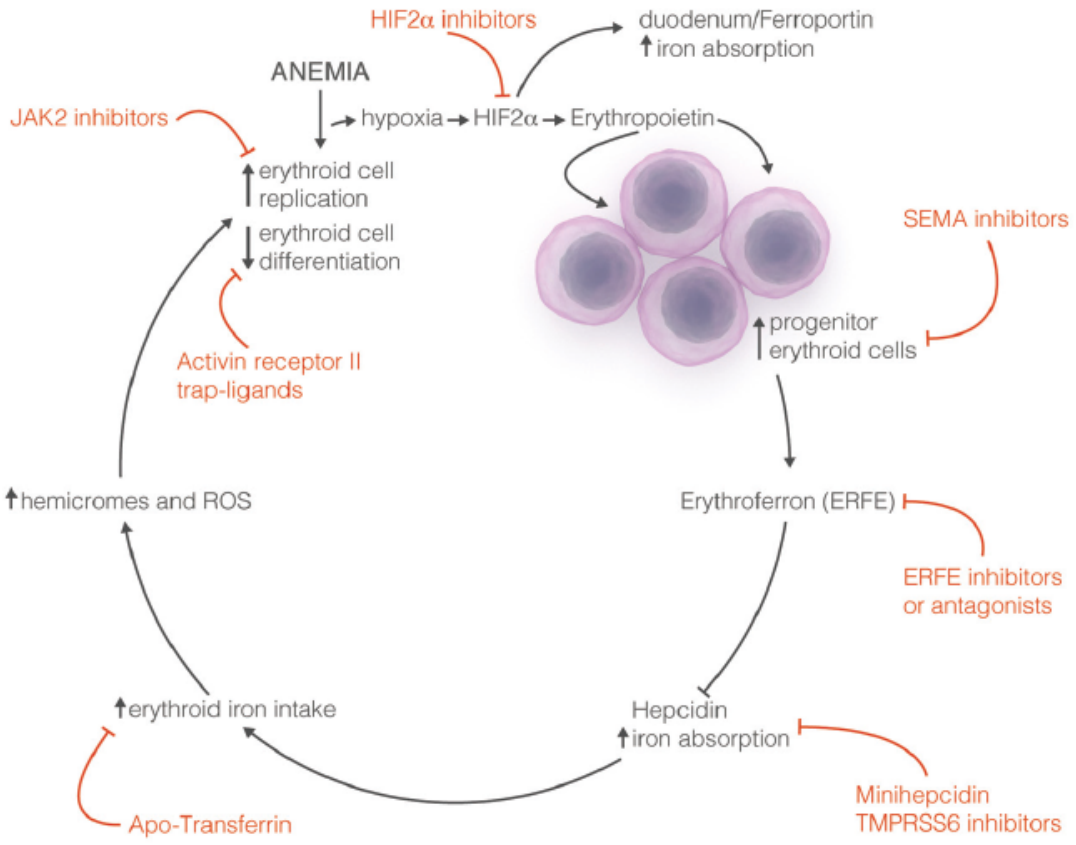


Figure 4. Illustration of the relationship between anemia, hypoxia, Epo, erythropoiesis and iron metabolism in β-thalassemia. Hypoxia, through HIF2a, contributes to augmented iron absorption by increasing expression of Fpn, Dmt1 and DcytB in the duodenum. Epo, ROS and Growth differentiation factor 11 (Gdf11) alter erythropoiesis, increasing cell proliferation and decreasing cell maturation, contributing to the extramedullary hematopoiesis. As the number of erythroid progenitors increases, more ERFE and less hepcidin are produced, leading to increased iron absorption and increased Tf-sat. Altogether, these modifications contribute to the pathophysiology of ß-thalassemia, and exacerbate the ineffective erythropoiesis and iron overload over time. The diagram also shows potential targets and therapeutics that might benefit β-thalassemia, as discussed in the text.