Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Mini-Review

Regulation of human alpha-globin gene expression and alpha-thalassemia

D.M. Ribeiro and M.F. Sonati

Departamento de Patologia Clínica, Faculdade de Ciências Médicas, Universidade Estadual de Campinas, Campinas, SP, Brasil

Corresponding author: M.F. Sonati
E-mail: sonati@fcm.unicamp.br

Genet. Mol. Res. 7 (4): 1045-1053 (2008)
Received May 9, 2008
Accepted July 1, 2008
Published October 14, 2008

ABSTRACT. Hemoglobin and globin genes are important models for studying protein and gene structure, function and regulation. We reviewed the main aspects of regulation of human α-globin synthesis, encoded by two adjacent genes (α_2 and α_1) clustered on chromosome 16. Their expression is controlled mainly by a regulatory element located 40 kb upstream on the same chromosome, the α-major regulatory element, whose activity is restricted to a core fragment of 350 bp, within which several regulatory protein binding sites have been found. Natural deletions involving α-major regulatory element constitute a particular category of α-thalassemia determinants in which the α-globin genes are physically intact but functionally inactive.

Key words: Hemoglobin; Globin genes; HS-40; Thalassemia; Gene expression; Alpha-major regulatory element

