

ISSN 0972-3757

International Journal of
HUMAN GENETICS

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Int J Hum Genet, 17(3): 97-102 (2017)
DOI:10.31901/24566330.2017/17.03.01

Novel Tag SNPs of Beta-Globin Gene Cluster in Chinese Han Population: Biological Marker for Genetic Backgrounds and Clinical Studies

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KEYWORD Beta-thalassemia. Bioinformatics Tool. Genetics. Linkage Analysis. Polymorphism

ABSTRACT The beta-globin gene cluster is located on chromosome 11. SNPs database was published by Hap Map Project. Here, novel tag SNPs on beta -globin gene cluster in Chinese Han population was reported on using Haploview 4.2. The haplotype block and LD analysis showed three blocks that were constructed with 35 common SNPs. All common SNPs were generated with the Tag SNPs using Haploview's Tagger with LD threshold $r^2 \geq 0.8$, run with aggressive tagging; 2- and 3 marker haplotypes and default LOD cutoff 3.0. The results showed that seven SNPs capture all 35 common SNPs. Seven tag SNPs including rs4910736, rs5010979, rs4910548, rs11036634, rs7480526, rs10837628, rs11036635 could be used to predict the genetic background of beta-globin gene defect or clinical correlation with beta-thalassemia in a Chinese or Asian population. Further study should test these tag SNPs for genetic background and a clinical relation between the biological marker and beta-thalassemia disease.