

SINGLE NUCLEOTIDE POLYMORPHISMS OF EXON 1 AND EXON 2 AND HAPLOTYPES OF MBL GENE (MBL₂) IN CHILDREN WITH REPEATED RESPIRATORY TRACT INFECTIONS IN CHINESE

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Background and aim: Mannose-binding lectin (MBL) is an important component of innate immunity. Earlier studies have reported that low plasma MBL levels determined by the polymorphism of mbl₂, were more common in children with recurrent infections. However, seldom study covered the haplotypes and exon2. In this study, we investigated the single nucleotide polymorphisms and haplotypes of promoter, exon1 and exon2 in children with repeated respiratory tract infections.

Methods: MBL plasma concentrations were measured with ELISA method, and the polymorphism of mbl₂ were determined with sequence analysis method.

Results: More individuals with low plasma MBL levels were found in RRTI group when compared with the healthy group. The variant allele frequencies of codon 54 in healthy group was 0.167 and in RRTI group was 0.283, the difference was significant. No mutation was found at codon 52 and 57. The variant frequencies at -550 and -221 sites in healthy children were 0.348 and 0.095 respectively, while in RRTI children, the frequencies were 0.542 and 0.117 instead. Less haplotypes of HYP A in RRTI group (0.463) was found when compared with the healthy group (0.619), while the frequency of LYP A was higher in RRTI group (0.256) than that in healthy group (0.114). Two heterozygous for T to A mutations at 84 codon and one heterozygous for G to A mutation at 101 codon in exon 2 were found in RRTI children.

Conclusion: Genotypes of mbl₂ resulting in low circulating MBL levels were associated with increased susceptibility to RRTI in Chinese children.