ABSTRAK

Latar Belakang : Konstipasi fungsional merupakan masalah kesehatan yang sering ditemukan pada anak di seluruh dunia yang dapat menurunkan kualitas hidup dan menyebabkan biaya tinggi terhadap pelayanan kesehatan. Polimorfisme beberapa gen tertentu diduga memiliki peranan dalam terjadinya konstipasi fungsional pada anak.

Tujuan Penelitian : Mengetahui pengaruh polimorfisme gen *serotonin reuptake transporter (SERT)* dan gen *β2-adrenergic receptor (β2AR)* terhadap kejadian konstipasi fungsional pada anak.

Metode Penelitian : Penelitian ini adalah penelitian kasus-kontrol terhadap 116 subyek dengan pendekatan laboratorium untuk melihat hubungan polimorfisme gen *SERT* dan gen *β2AR* dengan konstipasi fungsional pada anak. Polimorfisme gen *SERT* ditentukan dengan metode PCR, sedangkan polimorfisme *β2AR* ditentukan dengan TaqMan probe. Uji *chi square* digunakan untuk menilai hubungan dua variabel.

Hasil Penelitian : Frekuensi distribusi polimorfisme gen *SERT*: SS 46,4%, LS 3,6%, LL 1,8%, XLXL 3,6%, dan SXLL 44,6%. Pola genotipe LS dan XLXL menurunkan risiko, sedangkan genotipe SXLL meningkatkan risiko terjadinya konstipasi fungsional pada anak. Terdapat hubungan yang bermakna secara statistik antara polimorfisme *SERT* dengan konstipasi fungsional pada anak ($p < 0,001$). Frekuensi distribusi polimorfisme gen *β2AR*: AA 25,0%, GA 19,6%, dan GG 55,4%. Tidak terdapat hubungan yang bermakna secara statistik antara polimorfisme *β2AR* dengan konstipasi fungsional pada anak ($p = 0,143$). Pasangan polimorfisme SS GA dan XLXL GG menurunkan risiko terjadinya konstipasi fungsional. Terdapat hubungan bermakna antara kombinasi kedua gen dengan konstipasi fungsional pada anak ($p = 0,001$).

Kesimpulan : Terdapat hubungan bermakna secara statistik antara genotipe *SERT* dengan kejadian konstipasi fungsional pada anak ($p < 0,001$). Tidak didapatkan hubungan bermakna secara statistik antara genotipe *β2AR* dengan konstipasi fungsional pada anak ($p = 0,143$). Terdapat hubungan bermakna secara statistik pada analisis kombinasi genotipe *SERT* dan *β2AR*.

Kata kunci : Polimorfisme gen *SERT*, Polimorfisme gen *β2AR*, konstipasi fungsional, anak

ABSTRACT

Background: Functional constipation is a common health problem that is often found in children around the world that can reduce quality of life and cause high costs for health services. Polymorphisms of certain genes are thought to have a role in the occurrence of functional constipation in children.

Objective : This study was performed to investigate the relationship between polymorphisms of serotonin reuptake transporter (SERT) gene and β2-adrenergic receptor (β2AR) gene in children with functional constipation.

Methods : This study was a case-control study of 116 subjects with a laboratory approach to see the relationship between the SERT gene polymorphism and the β2AR gene with functional constipation in children. SERT gene polymorphism was determined by PCR method, whereas β2AR polymorphism was determined by TaqMan probe. Chi square test is used to assess the relationship of two variables.

Result : Distribution frequency of SERT genotype polymorphism: SS 46.4%, LS 3.6%, LL 1.8%, XLXL 3.6%, and SXLL 44.6%. The genotype pattern of LS and XLXL decreases risk, while the SXLL genotype increases the risk of functional constipation in children. There was a significantly correlated between SERT polymorphism and functional constipation in children ($p < 0,001$). The distribution frequency of β2AR genotype polymorphisms: AA 25.0%, GA 19.6%, and GG 55.4%. There was no significantly correlated between β2AR polymorphisms and functional constipation in children ($p = 0,143$). The combination genotype pattern of SS GA and XLXL GA decreases risk of functional constipation in children. There was a significant relationship between the combination of the two genes with functional constipation in children ($p = 0.001$).

Conclusion: There was a significantly correlated between SERT polymorphism and functional constipation in children. There was no significantly correlated between β2AR polymorphism and functional constipation in children. There was a significantly correlated between combination of the two genes with functional constipation in children.

Keyworld : SERT gen polymorphism, β2AR gen polymorphism, functional constipation, children.