

EKSPRESI mRNA GEN *LBX1* PADA PASIEN ADOLESCENT IDIOPATHIC SCOLIOSIS DI RSUP DR. SARDJITO YOGYAKARTA

INTISARI

Latar Belakang

Adolescent Idiopathic Scoliosis atau AIS merupakan suatu kelainan yang ditandai dengan deformitas struktural spinal, yang memengaruhi sekitar 1-4% anak-anak dan remaja. Prevalensi AIS yang cukup tinggi pada anak-anak dan remaja serta masih kurangnya pemahaman mengenai etiologi dan patogenesis pasti dari AIS menjadi salah satu alasan dari beberapa penelitian yang telah dilakukan di luar Indonesia terhadap AIS. Penelitian sebelumnya menyebutkan bahwa terdapat lokus pada kromosom 10q24.31 dekat gen *LBX1* yang memiliki asosiasi kuat dengan kondisi AIS. Gen *LBX1* memiliki peranan dalam diferensiasi *spinal cord* dan mengode protein *LBX1* yang berperan dalam migrasi sel prekursor otot, sehingga gen *LBX1* menjadi gen kandidat biologis yang kuat terhadap AIS. Asosiasi gen *LBX1* dengan AIS telah diteliti dan dibuktikan sebagai gen suseptibilitas mayor terhadap AIS pada populasi etnis Jepang, Han, dan Kaukasia.

Tujuan

Penelitian ini bertujuan untuk mengetahui tingkat ekspresi mRNA dari gen *LBX1* pada pasien AIS di RSUP Dr. Sardjito dan mengetahui perbedaan tingkat ekspresi mRNA gen *LBX1* antara pasien AIS dengan subyek kontrol.

Metode

Penelitian dilakukan dengan rancangan studi observasional analitik potong lintang. Sampel didapatkan dengan menggunakan teknik biopsi jaringan otot paravertebral melalui tindakan bedah. Kuantifikasi ekspresi mRNA gen *LBX1* dilakukan dengan menggunakan *Real-time* qPCR.

Hasil

Hasil penelitian menunjukkan terdapat perbedaan peningkatan ekspresi mRNA gen *LBX1* antara pasien AIS di RSUP Dr. Sardjito dengan subyek kontrol ($p=0,012$), dengan rerata ekspresi mRNA gen *LBX1* pada kelompok pasien AIS lebih rendah dibandingkan dengan subyek kontrol tanpa skoliosis.

Kesimpulan

Rerata ekspresi mRNA gen *LBX1* pada kelompok pasien AIS cenderung lebih rendah dibandingkan dengan subyek kontrol tanpa skoliosis. Temuan ini berbeda dengan hasil dari penelitian pendahulu dan menunjukkan kemungkinan pengaruh yang kompleks antara faktor genetik serta epigenetik terhadap AIS.

Kata kunci: *Adolescent idiopathic scoliosis*, ekspresi mRNA, *LBX1*, otot skelet, Indonesia, otot paravertebral

THE mRNA EXPRESSION OF *LBX1* GENE ON ADOLESCENT IDIOPATHIC SCOLIOSIS PATIENTS IN RSUP DR. SARDJITO YOGYAKARTA

ABSTRACT

Background

Adolescent Idiopathic Scoliosis or AIS is a disorder characterized by spinal structural deformity, affecting about 1-4% of children and adolescents. The high prevalence of AIS in children and adolescents as well as the lack of understanding of etiology and the definite pathogenesis of AIS is one of the reasons some studies about AIS have been conducted outside Indonesia. Previous studies have suggested that there is a locus on the 10q24.31 chromosome near the *LBX1* gene that has a strong association with AIS conditions. The *LBX1* gene plays a role in spinal cord differentiation and encodes *LBX1* proteins that play a role in the migration of muscle precursor cells, thus making the *LBX1* gene a strong biological candidate genes of AIS. The *LBX1* gene association with AIS has been studied and proven as a major susceptibility gene against AIS in the ethnic populations of Japan, Han Chinese, and Caucasian.

Objectives

This study aims to investigate the level of mRNA expression of the *LBX1* gene in AIS patients in RSUP Dr. Sardjito and investigate the difference in *LBX1* gene mRNA expression levels between AIS patients and control subjects.

Methods

The study was conducted with cross-sectional analytic observational study design. Samples were obtained by using biopsy technique of paravertebral muscle tissue through surgical action. Quantification of *LBX1* gene mRNA expression was performed using Real-time qPCR.

Results

The results showed that there was a difference in the increase of *LBX1* gene mRNA expression between AIS patients in RSUP Dr. Sardjito and control subjects ($p = 0.012$), with mean mRNA expression of *LBX1* gene in the AIS group of patients was lower than control subjects without scoliosis.

Conclusions

The mean mRNA expression of *LBX1* gene in the AIS group of patients tended to be lower than the controls without scoliosis. This finding differs from the results of the predecessor's research and suggests the possibility of a complex influence between genetic as well as epigenetic factors on AIS.

Key words: Adolescent idiopathic scoliosis, mRNA expression, *LBX1*, skeletal muscle, Indonesia, paravertebral muscle