Polimorfisme Gen ACTN3 R557X dan Q523R terhadap Variasi Pola Skeletal pada Sub-Populasi Indonesia (Kajian Laboratoris & Foto Sefalometri Lateral) = Correlation of ACTN3 R557X and Q523R Gene Polymorphism on Skeletal Patterns in Indonesian Sub-Populations (Laboratory & Lateral Cephalometry Study)

Ninis Yekti Wulandari, author

Abstrak

Pendahuluan: Alpha-actinin-3 (ACTN3) merupakan protein pengikat aktin yang mempengaruhi kinerja otot serta proporsi jenis serat otot. Secara spesifik, ACTN3 bertindak sebagai isoform spesifik dari protein fast twitch yang hanya diekspresikan pada serat otot tipe II dan merupakan bagian dari alat kontraktil serat glikolitik pada otot rangka manusia, termasuk otot mastikasi. Penelitian terdahulu telah menghubungkan antara jenis serat otot ke dalam perkembangan kraniofasial yang dapat menyebabkan terjadinya maloklusi. Tujuan: Mengetahui korelasi antara polimorfisme gen ACTN3 R557X dan Q523R terhadap pola skeletal pada sub-populasi Indonesia. Metode: Subyek merupakan pasien dengan maloklusi skeletal kelas I, II dan III yang menjalani perawatan ortodontik di RS Fakultas Kedokteran Gigi Universitas Indonesia. Amplifikasi sekuens DNA dilakukan pada folikel rambut pasien dengan menggunakan Polymerase Chain Reaction (PCR), sedangkan analisis polimorfisme genetik gen ACTN3 dilakukan dengan Restriction Fragment Length Polymorphism (RFLP). Pola skeletal ditentukan berdasarkan analisis radiografi sefalometri awal menggunakan sudut ANB (Anteroposterior), Facial axis dan Sudut Gonion (Vertikal). Kesimpulan: Tidak terdapat korelasi antara polimorfisme gen ACTN3 R557X dan Q523R dengan maloklusi skeletal kelas I, II, dan III, pertumbuhan vertikal wajah serta arah pertumbuhan 1/3 muka bawah. Namun ditemukan bahwa ACTN3 R557X dan Q523R pada sub-populasi Indonesia mengalami linkage disequilibrium.
.....Background: The protein alpha-actinin-3 (ACTN3) is an actin-binding protein that influences muscle performance and the proportion of muscle fiber types. Moreover, it also acts as a specific isoform of fast twitch protein that is only expressed in type II muscle fibers and forms part of the contractile apparatus of fast glycolytic fibers in human skeletal muscle, including masticatory muscle. Previous study has incorporated muscle fiber type to craniofacial development that may lead to malocclusion. Aim: To determine the correlation between polymorphisms of ACTN3 R557X and Q523R gene on skeletal patterns in Indonesian Sub-Populations. Methods: Subjects were patients with class I, II and III skeletal malocclusion undergoing orthodontic treatment at the Faculty of Dentistry Hospital, University of Indonesia. DNA sequence amplification was carried out on the patient's hair follicles using Polymerase Chain Reaction (PCR), while genetic polymorphism analysis of the ACTN3 gene was carried out using Restriction Fragment length Polymorphism (RFLP). The skeletal pattern was determined by initial cephalometric radiographic analysis using the ANB angle (Anteroposterior), Facial axis and Gonion angle (Vertical). Conclusion: There is no correlation between the ACTN3 R557X and Q523R gene polymorphisms with skeletal malocclusion class I, II, and III, vertical facial growth and growth direction of the lower third of the face. However, it is found that ACTN3 R557X and Q523R in the Indonesian sub-population experience linkage disequilibrium.