Analisis urutan basa gen PIK3CA Ekson 20 pada sampel jaringan kanker payudara yang berasal dari Sumatera Barat = PIK3CA base sequence analysis of exon 20 in the breast cancer tissue from West Sumatera

Ida Yus Sriyani, author

Abstrak

ABSTRAK
Pemeriksaan mutasi gen PIK3CA penting dilakukan karena mutasi pada gen tersebut menyebabkan penderita kanker payudara dengan subtipe HER 2 mengalami resistensi terhadap terapi trastuzumab. Kit komersial pendeteksi mutasi gen PIK3CA yang beredar di pasaran dibuat berdasarkan genotipe kaukasia/Amerika. Penelitian telah dilakukan untuk mengetahui profil mutasi gen PIK3CA ekson 20 dari penderita kanker payudara di Sumatera Barat. Hasil sekuensing menunjukkan mutasi gen PIK3CA ditemukan 5 dari 68 sampel uji (7,35%) dan paling sering terjadi pada subtipe luminal. Mutasi tersebut berupa silent mutation T1025T (4,41%) dan missense mutation H1047R (2,94%). Hasil penelitian menunjukkan bahwa mutasi gen PIK3CA Ekson 20 tidak berasosiasi dengan keberadaan reseptor ER, PR, dan HER 2. Mutasi yang ditemukan dalam penelitian ini dapat digunakan sebagai referensi pembuatan kit deteksi mutasi gen PIK3CA meskipun tidak berasosiasi dengan parameter patologiklinik.
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ABSTRACT
Examination of PIK3CA mutations in breast cancer patients is important because contributed to trastuzumab resistance problem in breast cancer subtype HER 2. Since commercial kit to detect PIK3CA mutation which now widely used were made based on caucasia/America genotype, not based on genotype Indonesia. The research has been conducted to determine the profile of the PIK3CA exon 20 mutations of the breast cancer patients in West Sumatra. Sequencing result showed that 5 out of 68 samples PIK3CA (7.35 %) had PIK3CA mutations and mostly occur in the luminal subtype. Mutations found were silent mutation T1025T (4.41 %) and missense mutation H1047R (2.94%). Meanwhile, this research results that PIK3CA mutation in exon 20 does not associated with the presence of ER, PR, and HER 2 reseptors respectively. However, mutations found in this research was expected to use as a reference in devloping detection kit of PIK3CA mutation regardless the negative association with clinical pathology parameters;