Perbandingan sensitivitas dan spesifisitas a-globin strip assay dengan polymerase chain reaction (PCR) sebagai baku emas dalam mendeteksi mutasi thalassemia = Comparison of sensitivity and specifity between globin strip assay and (PCR) as the gold standard in diagnosing thalassemia / Dian Puspita Sari

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Abstrak

[ABSTRAK
Latar belakang: Metode PCR rutin untuk mendeteksi mutasi pada thalassemia α seperti PCR multi kompleks dan restriction fragment length polymorphism (RFLP) membutuhkan proses yang lama dan reagen yang banyak serta biaya yang besar. Saat ini telah dikembangkan metode baru yaitu tes strip (α-globin strip assay), yang dapat mendeteksi 21 macam mutasi gen globin -α secara simultan dalam satu paket reaksi dan hanya membutuhkan DNA dalam jumlah sedikit.

Tujuan : Mengetahui nilai sensitivitas dan spesifisitas metode α-globin strip assay dalam mendeteksi mutasi thalassemia-α.

Metode penelitian: Penelitian merupakan uji diagnostik yang dilakukan dengan metode belah lintang yang membandingkan pemeriksaan α -globin strip assay dan PCR rutin dalam mendeteksi mutasi gen pada thalassemia α. Pada tahap I disertakan 17 pasien yang berobat ke pusat thalassemia di RSCM dan Lembaga Biomolekular Eijkman Jakarta pada bulan Oktober 2014 sampai Maret 2015, kemudian tahap II disertakan 18 anggota keluarga inti subjek pada tahap I. Pada semua subjek dilakukan pemeriksaan hematologi termasuk indeks eritrosit, morfologi darah tepi, analisis Hb, PCR rutin dan α -globin strip assay.

Hasil penelitian dan pembahasan: Ditemukan tujuh jenis mutasi yang terdiri dari: 1) delesi 1 gen 3,7kb; 2) non delesi Cd59; 3) non delesi HbCS; 4) delesi 2 gen SEA; 5) mutasi campuran 3,7kb/Cd59 ; 6) mutasi campuran Cd59/HbCS; 7) mutasi campuran SEA/HbCS. Metode α-globin strip assay memiliki nilai sensitivitas dan spesifisitas sebesar 100%.

Kesimpulan : Metode α-globin strip assay akurat mendeteksi mutasi thalassemia-α dengan tingkat sensitivitas dan spesifisitas sebesar 100%.
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ABSTRACT
Background : Routine PCR methods in detecting mutations that occur in α thalassemia such as multi-complex single tube PCR and PCR restriction fragment length polymorphism (RFLP) require a lengthy process and utilize large amount of reagents and are costly. α-globin strip assay is a new method in detecting α thalassemia related mutations that is able to detect 21 types of globin-α mutations simultaneously in a single reaction and requires only small amount of DNA.

Objective: To determine the sensitivity and specificity of α-globin strip assay compared to routine PCR in detecting α thalassemia associated mutations.

Methods: A cross sectional diagnostic study was performed comparing α-globin strip assay and routine PCR in detecting mutations related to α thalassemia. Phase I of the study includes 17 patients treated for α thalassemia at RSCM and Biomolecular Eijkman Institute between October 2014 and March 2015, phase II includes 18 close relatives of patients recruited in phase I. All subjects underwent hematological examination including erythrocyte indices, peripheral blood morphology, Hb analysis, routine PCR and α ?globin strip assay.

Results: Seven kind of mutations were identified including 1) deletion of one gene 3,7 kb; 2) non-deletion of CD59; 3) non deletion of HbCS; 4) deletion of two genes SEA; 5) mixed mutation of 3,7kb/CD59; 6) mixed mutation of CD59/HbCS; 7) mixed mutation of SEA/HbCS. α-globin strip assay has sensitivity and specificity of 100%.

Conclusion: α ?globin strip assay accurately detect mutations in α thalassemia with 100% sensitivity and specificity., Background : Routine PCR methods in detecting mutations that occur in α thalassemia such as multi-complex single tube PCR and PCR restriction fragment length polymorphism (RFLP) require a lengthy process and utilize large amount of reagents and are costly. α-globin strip assay is a new method in detecting α thalassemia related mutations that is able to detect 21 types of globin-α mutations simultaneously in a single reaction and requires only small amount of DNA.

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