

INTISARI

Latar belakang. Penyakit Hirschsprung (HSCR) merupakan penyakit genetik dengan karakteristik tidak adanya sel ganglion pada pleksus Meissner dan Auerbach di traktus intestinal. Insidensi HSCR di Yogyakarta adalah 1 per 3250 dengan pasien terbanyak pada periode neonatus. Standar baku emas diagnosis HSCR adalah pemeriksaan histopatologi spesimen biopsi rektal ketebalan penuh yang menunjukkan aganglionosis pada pleksus submukosa dan mienterikus serta hipertrofi serabut saraf. Temuan hipertrofi serabut saraf berkorelasi dengan enterokolitis paska operasi pada pasien HSCR dan digunakan dalam mendiagnosis zona transisi *pull-through* dalam operasi HSCR. Tidak ditemukannya hipertrofi serabut saraf pada kondisi aganglionosis dapat meningkatkan kecurigaan bahwa pasien mengalami HSCR segmen panjang.

Tujuan. Untuk mengetahui akurasi, sensitivitas, spesifisitas, nilai ramal positif, dan nilai ramal negatif pengecatan HE dibandingkan S100 dalam mendeteksi hipertrofi serabut saraf pada penyakit Hirschsprung di RSUP Dr. Sardjito.

Metode. Penelitian observasional dengan rancangan studi potong lintang dilakukan pada spesimen biopsi rektal pasien diduga mengalami HSCR di RSUP Dr. Sardjito tahun 2017-2022. Tes Mc Nemar digunakan untuk menganalisis perbedaan performa diagnostik antara pengecatan HE dan S100 dan nilai indeks Cohen's Kappa digunakan untuk menganalisis kesepakatan antar kedua metode pengecatan tersebut.

Hasil. Terdapat 31 pasien HSCR, kelompok usia neonatus dan 29 hari - ≤ 1 tahun. Hasil akurasi, sensitivitas, spesifisitas, nilai ramal positif, dan nilai ramal negatif pengecatan HE adalah 90.32% (95% CI: 0.81 – 0.99), 96.30% (95% CI: 0.81 – 0.99), 50.00% (95% CI: 0.68 – 0.93), 92.86% (95% CI: 0.83 – 0.97), dan 66.67% (95% CI: 0.19 – 0.95) secara berurutan. Uji diagnostik tidak menunjukkan perbedaan yang signifikan secara statistik ($p=1.00$) dengan indeks Cohen's Kappa adalah 0.518 (95% CI: 0.044 – 0.992).

Kesimpulan. Akurasi pengecatan HE dibandingkan IHC S100 dalam mendeteksi hipertrofi serabut saraf pada penyakit HSCR adalah 90.32%. Tidak terdapat perbedaan performa diagnostik yang signifikan secara statistik antara kedua pengecatan serta kedua pengecatan memiliki tingkat persetujuan moderat (sedang).

Kata Kunci. Hirschsprung, hematoksilin eosin, imunohistokimia S100, hipertrofi, akurasi

ABSTRACT

Background. Hirschsprung's disease (HSCR) is a genetic disease characterized by the absence of ganglion cells in the Meissner and Auerbach plexuses in the intestinal tract. The incidence of HSCR in Yogyakarta is 1/3250 with the highest number of patients in the neonatal period. The gold standard for the diagnosis of HSCR is histopathological examination of a full-thickness rectal biopsy specimen showing aganglionosis of the submucosal and myenteric plexuses and hypertrophy of nerve fibers. The finding of nerve fiber hypertrophy correlates with postoperative enterocolitis in HSCR patients and is used in diagnosing the transitional zone pull-through in HSCR surgery. The absence of hypertrophied nerve fibres in conditions of aganglionosis can raise the suspicion that the patient has long-segment HSCR.

Objective. To determine the accuracy, sensitivity, specificity, positive predictive value, and negative predictive value of HE staining compared to S100 for hypertrophic nerve fibers detection in Hirschsprung's disease at RSUP Dr. Sardjito.

Methods. Observational study with a cross-sectional study design was carried out on rectal biopsy specimens of patients suspected of having HSCR at Dr. Sardjito General Hospital Yogyakarta in 2017-2022. Mc Nemar's test was used to analyze the difference in diagnostic performance between HE and S100 staining and the Cohen's Kappa index value was used to analyze the agreement between the two staining methods.

Results. There were 31 HSCR patients divided into neonatal age group and 29 days - ≤ 1 year. The accuracy, sensitivity, specificity, positive predictive value, and negative predictive value of HE staining were 90.32% (95% CI: 0.81 – 0.99), 96.30% (95% CI: 0.81 – 0.99), 50.00% (95% CI: 0.68) – 0.93), 92.86% (95% CI: 0.83 – 0.97), and 66.67% (95% CI: 0.19 – 0.95) respectively. The diagnostic test showed no statistically significant difference ($p=1.00$) with the Cohen's Kappa index being 0.518 (95% CI: 0.044 – 0.992).

Conclusion. The accuracy of HE staining compared to IHK S100 for hypertrophic nerve fibers detection in HSCR is 90.32%. There was no statistically significant difference in diagnostic performance between the two stains and both stains had a moderate level of approval.

Keywords. Hirschsprung, hematoxylin eosin, S100 immunohistochemistry, hypertrophy, accuracy