Identifikasi Metilasi Mismatch Repair Gene (MMR) pada Kanker Kolon dengan Teknik Methylation-Specific Multiplex Ligation-Dependent Probe Amplification (MS-MLPA) = Identification of Mismatch Repair Gene (MMR) Methylation in Colon Cancer with Methylation-Specific Multiplex Ligation-Dependent Probe Amplification (MS-MLPA) techniques

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Abstrak

Metilasi DNA merupakan salah satu penyebab umum inaktivasi Mismatch Repair Gene (MMR). Gen MMR memperbaiki kesalahan penyisipan/penghapusan basa nukleotida pada proses sintesis DNA. Metilasi pada promoter gen MMR memiliki asosiasi dengan pembentukan kanker kolon, sehingga metilasi tersebut perlu diidentifikasi. Identifikasi gen MMR dapat dilakukan menggunakan teknik methylation-specific multiplex ligation-dependent probe amplification amplification (MS-MLPA). Prinsip dari teknik MS-MLPA yaitu amplifikasi probe yang menempel pada sekuens termetilasi. Tujuan dari penelitian ini yaitu untuk mengoptimasi teknik MS-MLPA dan mengidentifikasi metilasi gen MMR pada kanker kolon dengan teknik MS-MLPA. Penelitian ini menggunakan 27 sampel jaringan frozen kanker kolon yang telah tersedia di Biobank Rumah Sakit Kanker Dharmais (RSKD). Sampel tersebut dianalisis menggunakan probemix Mismatch Repair Gene [ME011-C1][C1-0518] yang telah didesain khusus untuk mendeteksi pada beberapa gen MMR yakni MLH1, PMS2, MSH6, dan MSH2. Hasil penelitian menunjukkan optimasi teknik MS-MLPA telah berhasil dilakukan, sehingga identifikasi metilasi pada gen MMR telah berhasil diperoleh pada 4 sampel pasien. Gen MMR tersebut yakni MLH1 dan MSH6, dengan persentase masing-masing 75% dan 25%.

.....DNA methylation is one of the most common causes of mismatch repair gene (MMR) inactivation. The MMR gene corrects errors in the insertion/deletion of nucleotide bases in the DNA synthesis process. MMR gene promoter methylation has an association with the formation of colon cancer, so the methylation needs to be identified. Identification of the MMR gene can be done using the methylation-specific multiplex ligation-dependent probe amplification (MS-MLPA) technique. The principle of the MS-MLPA technique is the amplification of the probe attached to the methylated sequence. The purpose of this study was to optimize the MS-MLPA technique and identify MMR gene methylation in colon cancer using the MS-MLPA technique. This study used 27 samples of frozen colon cancer tissue that were available at the Dharmais Cancer Hospital Biobank (RSKD). The samples were analyzed using the Mismatch Repair Gene probemix [ME011-C1][C1-0518] which has been specially designed to detect several MMR genes, namely MLH1, PMS2, MSH6, and MSH2. The results show that the optimization of the MS-MLPA technique has been successfully carried out, so that the identification of methylation in the MMR gene has been successfully obtained in 4 patient samples. The MMR genes are MLH1 and MSH6, with a percentage of 75% and 25%, respectively. analyzed using probemix Mismatch Repair Gene [ME011-C1][C1-0518] which has been specifically designed to detect several MMR genes namely MLH1, PMS2,
MSH6, and MSH2. The results showed that the optimization of the MS-MLPA technique was successful, so that identification of the methylation in the MMR gene was successfully obtained in 4 patient samples. The

MMR genes are MLH1 and MSH6, with percentages of 75% and 25% respectively.