



REVISTA DO HOSPITAL DE CLÍNICAS DE PORTO ALEGRE E
FACULDADE DE MEDICINA DA UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL

REVISTA HCPA 2007;27 (Supl 1) :1-292

27^a Semana Científica do Hospital de Clínicas de Porto Alegre

14º Congresso de Pesquisa e Desenvolvimento em Saúde do Mercosul
10 a 14 de setembro de 2007

Anais

MOLECULAR ANALYSIS OF COLORECTAL AND ENDOMETRIAL TUMORS FROM PATIENTS AT-RISK FOR LYNCH SYNDROME

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Intoduction: About 20% of colorectal tumors and 5% of endometrial tumors are hereditary. The recognition of at risk individuals for syndromes of hereditary predisposition to these tumors allows to implement strategies of specific prevention and accompaniment for these patients. Lynch Syndrome (also called HNPCC) is the most frequent form of hereditary colorectal cancer. In typical HNPCC families multiple generations are affected for CCR at early onset (~45 years). Endometrial cancer is the second malignance in affected families and is also the most frequent tumor in women who have the Syndrome. Microsatellite Instability (MSI), Imunohistochemistry (IHQ) and Methylation Analysis are usefull to complete the clinical diagnosis of these patients. **Objectives:** To characterize colon and endometrial tumors from at risk patients for Lynch Syndrome throught MSI, IHC and Methylation analysis of MLH1, MSH2 and MSH6 genes and to correlate its presence with clinical markers. **Materials and Methods:** Patients with Bethesda criteria and patients with endometrial cancer diagnosed before the age of 50 years, are included. DNA was obtained from paraffin-embedded normal and tumor tissue using MagneSil Genomic Fixed Tissue System kit (PROMEGA). To MSI analysis we use MSI Analysis System Version 1,1 kit (PROMEGA), that contains the pannel of the five recommended markers. IHC analysis is realized with specific antibodies against protein products of MLH1, MSH2 and MSH6 genes. Methylation analysis is carried out using MS-MLPA technique with ME-MLH1 kit (MRC-Holland, Amsterdam). **Results and Conclusions:** At the moment, 51 patients were included. From these, 11 of them have MSI analysis done, 40 have IHC and 5 have Methylation Analysis complete. To the date of the event, we expect to analyse the rest of the patients and concluce that this techniques are important tools to complete the clinical diagnostic of these patients.