The aim of this analysis was to investigate the prevalence of breast cancer in patients with mutations in the individual mismatch repair (MMR) genes that cause LS. We also investigated the family histories of patients with mutations in the individual MMR genes.

Current controversial question in the literature: Is breast cancer (BC) associated with Lynch syndrome (LS)?

The current study included 2372 subjects with a MMR mutation included if they only had single-site testing for MMR genes and were also identified and the proportion of patients with breast cancer. In our cohort, the average age at diagnosis of breast cancer was slightly higher in patients with MSH6 mutation carriers than in other MMR mutation carriers. Perhaps MSH6 mutation carriers are able to age without getting a Lynch-related cancer and thus have more opportunity to develop breast cancer. In our cohort, the average age at diagnosis of breast cancer was slightly higher in patients with MSH6 mutations.

Another approach to accepting or rejecting that breast cancer is associated with Lynch syndrome as a whole, it may be more appropriate to define breast cancer risks by specific MMR gene or a broader panel of genes.

The higher prevalence of breast cancer in MSH6 may be partly explained by its lower penetrance for Lynch-related cancers than MLH1 and MSH2. Perhaps MSH6 mutation carriers are able to age without getting a Lynch-related cancer and thus have more opportunity to develop breast cancer. In our cohort, the average age at diagnosis of breast cancer was slightly higher in patients with MSH6 mutations.

DISCUSSION

The mean, median, minimum, and maximum ages of diagnosis were calculated for patients with MMR mutations who had breast cancer.

The mean age of diagnosis for patients was 51.5 in MSH2, 51.9 in MLH1, 52.2 in MSH6, 57.6 in MSH6 and 73 in EPCAM. MSH6 mutation carriers had a relatively increased age of breast cancer diagnosis.

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