

Hereditary Nonpolyposis Colorectal Cancer: an Ominous but Enlightening Scenario

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Case

A 41 year old Middle Eastern female presented to the ED with complaints of abdominal pain and menorrhagia. A CT scan revealed an edematous uterus and a pelvic mass. Colonoscopy and D and C were positive for adenocarcinoma. Further evaluation by genetic and immunohistochemical (IHC) testing confirmed the diagnosis of Hereditary Nonpolyposis Colorectal Cancer (HNPCC).

Strategies

A retrospective case analysis investigating the modern diagnosis, natural history, and follow up care associated with HNPCC with a focus on laboratory features and input directly from Henry T Lynch, MD.

Reason for Presentation

There exists a need for physicians to become more familiar with the most common inherited form of colorectal cancer. HNPCC accounts for up to 5% of the annual colorectal cancer incidence. It was first described in 1913 and was revisited in 1971 by Dr Henry T Lynch. Since that time we have developed new diagnostic criteria and follow-up regimens that have a positive impact on patient outcome.

Conclusions and Recommendations

The early detection and intervention allows for a more favorable outcome not only for the patient being treated for HNPCC but also for their family. Increased provider awareness of diagnostic criteria will facilitate the recognition of what may be an underdiagnosed entity with significant morbidity and mortality.

References

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