Utilization of care and alignment of screening with National Comprehensive Cancer Network Guidelines for patients with Lynch Syndrome

Christina M. Lowry – Kaiser San Francisco

Topic: Genetics

Objectives

The primary objective is to identify the number of patients with Lynch syndrome receiving guideline concordant care. Secondary objectives are to assess over and under-utilization of screening tests and to determine factors associated with screening practices.

Methods

Retrospective cohort study at a large integrated health care system that included patients with a single germline mutation in MLH1, MSH2/EPCAM, MSH6 or PMS2 that were tested between 01/01/2015 and 12/31/2018 with Ambry. Clinical and demographic characteristics were abstracted from the electronic medical record and chart review. Screening tests for the following cancer sites were assessed: Gynecologic (TVUS, Ca 125, EMB, hysterectomy, BSO); gastrointestinal (H. pylori, colonoscopy, endoscopy, MRCP, endoscopic US); urinary; skin; neurologic (neurologic exam, brain MRI). The National Comprehensive Cancer Network guidelines (NCCN) were matched to the year of testing as they changed over time to assess concordance and over and under utilization. Secondary outcomes include factors associated with guidelines concordant care and subsequent cancer diagnoses.

Results

We identified a total of 234 patients meeting study inclusion criteria, 46 with MLH1, 64 with MSH6, 83 with PMS2 and 41 with MSH2/EPCAM mutations. The mutation carriers were 51.6 years old on average, pre-dominantly white (59.8%) and 56% (103/234) were diagnosed with Lynch Syndrome after genetic testing for a personal cancer history. 49% (114/234) of patients received screening as recommended by NCCN guidelines. 25% of patients (58/234) received less than the recommended amount of screening. Women with MLH1 mutations had higher rates of TVUS (34%, p=0.02) and Ca 125 screening (31%, p=0.06) compared to women with the other Lynch mutations. Patients with MLH1 mutations also had high rates of GU cancer screening (UA/urology referral, 28%, p=0.04).

Conclusions

These results indicate that gaps in screening exist for patients with Lynch Syndrome. There is an opportunity for improving rates of screening tests and tracking this information. Additionally, these results highlight that patients are getting hereditary cancer diagnoses following a primary cancer diagnosis, indicating need for earlier diagnosis of these familial syndromes.

Abstract Table or Graph

SPAZFWFX-1512836-1-ANY(1).pdf