2017 CGA Annual Meeting - Abstract Submission

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Abstract Title	Concordance of Multi-Gene Panel Testing with Prior Microsatellite Instability and Immunohistochemistry Analyses
Type of Submission	Poster or Podium
Syndrome Type	Lynch
Category Type	Clinical
Has this abstract been presented at another meeting as poster or podium presentation?	Yes
If yes, please indicate which conference or meeting.	InSiGHT 2017
Enter relevant disclosures below; enter "none" if you have no relationships to disclose.	Full time employee of Ambry Genetics

If I provide recommendations involving clinical medicine, they will be based on evidence that is accepted within the profession of medicine as adequate justification for their indications and contraindications in the care of patients. All scientific research referred to, reported or used in support of justification of a patient care recommendation will conform to the generally accepted standards of experimental design, data collection and analysis.	Agree
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Enter your abstract text in the field below.	Background: Microsatellite instability (MSI) and immunohistochemistry (IHC) analyses are acceptable screening methods, but their sensitivity and specificity for diagnosing Lynch syndrome are each less than 90%. We aimed to compare test results in a multi-gene panel testing (MGPT) cohort of individuals with prior MSI and/or IHC (tumor testing). Methods: Cases with MGPT performed at Ambry Genetics and tumor testing performed elsewhere were reviewed and classified as concordant if tumor testing matched MGPT results, discordant if they did not match MGPT results, and atypical if they had both concordant and discordant features. Results: Tumor testing and MGPT results (N=3850) were concordant in 73.1%, discordant in 24.6%, and atypical in 2.3% of cases (Figure 1). Of discordant cases (N=947), 85.7% had abnormal IHC with no constitutional MMR gene mutation, while 10.8% had high MSI, no or normal IHC, and no MMR mutation, 2.5% had normal tumor testing with an MMR mutation, and 1.0% had loss of protein(s) discordant from the MMR mutation. Results stratified by protein(s) missing, including the percent unexplained, are outlined in Table 1. Conclusions: Nearly 25% of tumor testing results were discordant from MGPT results. Possible explanations include MLH1 promoter methylation not ruled out, two somatic MMR mutations, unclassified variants, mutations not detected with current technology, and inaccurate tumor testing. IHC results did not always predict the MMR gene mutation and MMR mutations were identified even when both MSI and IHC analyses were normal. These results support the utility of MGPT and somatic MMR gene testing in these patients.
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Abstract Table and/or Chart	MSI-IHC Concordance CGA abstract Figure 1.pdf
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