Utility Of Molecular Profiling For Diagnosing Patients With Cancer Of Unknown Primary In A Population-Based Cohort.

PURPOSE: Cancer of unknown primary (CUP) patients have metastatic cancer with no identifiable primary tumor after a battery of guideline-recommended diagnostic tests. Currently, no guideline recommending organization recommends molecular profiling to identify the primary tumor. We characterized the effectiveness of molecular profiling diagnostic testing on receipt of site-specific treatments and overall survival among a population-based sample of CUP patients diagnosed between 2005 and 2015.

METHODS: Patients with cancer of unknown primary were identified in the Surveillance Epidemiology and End Results (SEER)-Medicare database (n=10,575). SEER is the cancer registry used to calculate US population-based incidence rates of cancer. Medicare database contains all billed medical procedures received before and after a cancer diagnosis. The effectiveness of the diagnostic procedures regarding overall survival was examined in patients who received the initial battery of guideline-recommended diagnostic tests and survived at least three months (to allow time to receive the molecular diagnostic testing) using Cox proportional hazards regression and mediation analysis for treatment effects.

RESULTS: Only 35.3% of CUP patients received timely guideline recommended initial diagnostic procedures. Receipt of all guideline-recommended initial diagnostic procedures was associated with a 15% reduced risk all-cause death, 36.9% of this reduction in death would remain if everyone received treatment. Receipt of molecular profiling was all associated with a nearly a 50% lower risk of death relative to comparable patients not receiving this molecular profiling, Hazard Ratio and 95%Confidence Interval, 0.67(0.56,0.80).

CONCLUSIONS: This research expands on small clinical studies demonstrating the benefit of molecular diagnostic tests to a population-based cohort of patients who received molecular testing despite a lack of clinical guideline support. This research provides evidence to guideline producing organizations of the utility of molecular provides in a large and diverse population-based cohort; namely more effective treatment and longer survival.