# Race- and ethnicity-based insurance coverage gaps for genetic testing for cancer in the greater Pittsburgh region

by

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## Abstract

Cancer has a large public health impact because it directly or indirectly affects all individuals, regardless of race and ethnicity. Hereditary breast and ovarian cancer (HBOC) and hereditary colorectal cancer (hCRC) often cluster in families and are implicated in 5-10% of breast cancers and 2-5% of colorectal cancers. Research has determined the variants in select genes that are likely to cause HBOC and hCRC, and individuals can receive genetic testing to determine if they are at an increased risk of developing these cancers. However, there are gaps in knowledge of and testing for these variants by race and ethnicity. There are also disparities in insurance coverage for individuals of different races and ethnicities. Identifying individuals with HBOC and hCRC is relevant to public health because cancer treatment is a burden on the healthcare system, and understanding risks of hereditary cancers can guide surveillance and management options. Laboratory billing claims data of individuals presenting at a UPMC facility between 2014-2019 was analyzed to determine whether there were gaps in genetic testing or inequities in insurance coverage for the genetic testing by race and ethnicity for HBOC and hCRC. The results showed that genetic testing for HBOC was more likely to be covered by an insurance company than hCRC, as 25.61% of HBOC tests were covered compared to 9.67% of hCRC tests. Furthermore, minoritized racial and ethnic groups were underrepresented in the study in comparison to census data in the region, composing only 6.31% of the study population, and overrepresented in Medicaid, with 27.5% of Black individuals and 6.67% of Asian individuals receiving coverage

through Medicaid compared to 4.3% of white individuals. The results confirm that race- and ethnicity-based insurance coverage gaps exist for genetic testing for cancer, showing the importance of developing interventions to increase knowledge of and access to genetic testing for minoritized racial and ethnic groups.

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## Preface

First, I am deeply grateful to my essay advisor Jodie Vento for her assistance at every stage of this project. I would also like to extend my sincere thanks to the rest of my essay committee – Dr. Coleman Drake, Dr. Elizabeth Felter, Dr. Lindsay Sabik, and Kathleen Vitullo – for their insightful comments and suggestions on how to improve my essay. I would like to express my sincere gratitude to Maureen May for providing CPT codes for data collection and ICD codes to further analyze the data. I would also like to thank Samantha Pettersen for providing information about the Pennsylvania Medicaid landscape as well as Coralys Carcana-Barbosa and Imani Beard for reviewing and editing my background chapter. Furthermore, I would like to thank the Pitt Health Record Research Request for providing me with my data for this project. Finally, I am extremely grateful for my family and MPH cohort for all the support I received throughout this process.

## 1.0 Introduction

Having adequate, consistent health insurance is a major determinant of long-term health outcomes (Bernstein, 2010), and it is imperative to develop a system of widespread coverage to address present-day inequities. The current health insurance system in the United States (US) is a patchwork system, with health insurance coming from a wide variety of sources, including employment, the federal government, or the marketplace. This leaves many people uninsured through gaps in the system and disproportionately impacts minoritized racial and ethnic groups. Although the introduction of the Affordable Care Act (ACA) in 2010 helped to fill in some of these gaps, there are nevertheless millions of people who still lack adequate health insurance (Sommers, 2020).

Studies have shown that being uninsured, underinsured, or lacking continuous coverage leads to worse health outcomes (Short, Graefe, Swartz, & Uberoi, 2012). Many of these gaps come from having a low socioeconomic status, including not having enough money to cover out-of-pocket expenses or having a job that does not include health insurance benefits (Short et al., 2012). People lacking insurance or continuous coverage often lack access to and do not utilize preventative cancer screenings, like pap smears and colonoscopies (Bernstein, 2010). Not being able to take advantage of these services means they are more likely to go to the doctor with more severe and often untreatable forms of disease.

Present gaps in the health insurance system include disparities based on gender (Witter, Govender, Ravindran, & Yates, 2017), race, and ethnicity. These gaps often come from social and structural hurdles, including employment type and language barriers. Lacking quality coverage decreases the likelihood that the person will receive the preventative care they need to identify

early signs of cancer, like mammograms and colorectal cancer screening (Bonafede et al., 2019). Studies have shown that having stable health insurance increases the likelihood that cancers will be caught early and at a treatable stage (Kapoor, 2014). Furthermore, lacking health insurance or having dual enrollment in Medicare and Medicaid decreased the likelihood that an older person would participate in colorectal cancer screening programs (Guessous et al., 2010). Foreign-born residents and non-citizens of the US are less likely to participate in screening programs to identify early stages of cancer, as many of them cannot access health insurance and government-subsidized insurance programs (Reyes, 2015).

Currently, inequities in the ordering patterns and uptake of genetic testing for hereditary cancers exist along racial and ethnic lines. Doubeni et. al. (2010) noted that even though there was an overall increase in screening and genetic testing for colorectal cancer from 2000-2010, lower proportions of Black and Hispanic individuals underwent screening and testing than whites. Race/ethnicity and English proficiency level were two major factors in whether someone got tested for colorectal cancer (Doubeni, Selby, & Gupta, 2021). Racial inequities can also intersect with socioeconomic status. Black individuals are disproportionately likely to be below the poverty line and therefore less likely to have access to care and be tested for hCRC (Johnston, Yeo, Clark, & Stewart, 2021). Provider bias and lack of knowledge about genetic testing also leads to doctors not recommending that minorities receive colorectal cancer screening and genetic testing (Johnston et al., 2021). The provider's implicit biases about the patient's race, educational level, socioeconomic status, and unemployment status, among others, often drives nonrecommendation (Johnston et al., 2021).

In addition to being more likely to lack health insurance, minorities disproportionately have a higher incidence of cancer diagnosis and mortality but less utilization of genetic testing. Muller

et al. (2018) noted that minority populations are less likely to be referred for genetic evaluation for colorectal cancer, even though there were similar rates of tumor analysis between races. Even in equal access settings, Black individuals are less likely to undergo colorectal cancer screening by any method, including colonoscopies, than non-Hispanic whites (Jackson, Oman, Patel, & Vega, 2016). For hereditary breast and ovarian cancers, Jones et al. (2021) found that Black women diagnosed with breast cancer at the age of 50 or younger had the highest rate of pathogenic or likely pathogenic variants, including a hereditary *BRCA* variant (Jones, 2021). However, genetic testing is under-utilized in clinical settings for the US population due to a lack of education and knowledge about genetic services (Allen, 2019), differences in insurance status, healthcare providers' implicit biases, and social inequities.

### 1.1 Specific Aims

To analyze the impact of race and ethnicity on genetic testing for hCRC and HBOC, the utilization of genetic testing across these demographic areas will be quantified through performing descriptive statistics on the data set, which is stratified by CPT code for different testing panels for genes found in hCRC and HBOC. The proportions of people from each race and ethnicity group will be compared to census data to determine if there are any racial or ethnic groups that are over- or underrepresented in genetic testing for these conditions. Furthermore, the impact of insurance type within this cohort will be examined. Past studies have found that marginalized racial and ethnic groups are less likely to use genetic testing and have quality health insurance that allows them to access genetic testing for cancer.

Aim 1: Assess utilization of genetic testing for a personal and family history of hCRC and HBOC by race and ethnicity to determine whether any racial or ethnic groups are disproportionately using these services.

Aim 2: Assess whether the insurance status of individuals undergoing genetic testing is reflective of the overall insurance mix within the region to determine whether any racial or ethnic groups are disproportionately uninsured or underinsured and highlight any current inequities in insurance coverage.

The results and conclusions of this project will assess any disparities in accessing genetic testing for cancer and show the importance of policy on the local, state, and federal levels in reducing these disparities. By exploring and identifying gaps in utilization for cancer genetic testing in the greater Pittsburgh region, the existing literature will be strengthened by focusing on a specific region of the country that has not been extensively studied. Increasing access to genetic testing for cancer will strengthen the role of public health genetics in the greater Pittsburgh region because more Pittsburgh residents will not only be able to know their risk and treatment options but share their results with their families to help them get the preventative care they need in the future.

## 2.0 Background

Genetic testing for hCRC and HBOC helps an individual determine which treatment and management options would be best for them. Individuals are eligible to receive genetic testing if they have a personal history of that cancer, a family history of that cancer, or a combination of the two. However, barriers that limit access to genetic testing exist at all levels of society, including inadequate insurance coverage and inequities in utilization of genetic testing services.

### 2.1 Genetic basis of cancer

Cancer is caused by a pathogenic variant in a gene, usually a protooncogene, a tumor suppressor gene, or a mismatch repair gene, that leads to uncontrolled growth and proliferation. Through genetic testing, scientists have determined which pathogenic variants in which genes are most likely to cause certain types of cancers. In addition to being given treatment for their cancer symptoms, individuals with cancer are often offered genetic testing to determine the specific variant causing the cancer.

Cancer falls into two large categories in terms of the nature of the genetic change. The largest and most common type of variant is a sporadic variant, composing 75-85% of all cancers ("Review of Cancer Genetics," 2016). A sporadic variant generally happens within the cells of an individual's body and happens during the individual's lifetime. The pathogenic variant in these genes were not inherited from one of their parents, nor is it passed to their children. The risk for these variants, and, therefore, the development of cancer, generally increase with age and are

mediated by factors in an individual's environment, medical history, and lifestyle ("Review of Cancer Genetics," 2016). These variants originate from the breakdown of the cellular machinery responsible for proofreading and checking the DNA for mistakes following replication. Because these variants are often a result of wear and tear within their cells, individuals who have sporadic cancer variants usually develop cancer later in life.

Hereditary cancer composes 5-10% of all cancers ("The Genetics of Cancer," 2017; "Review of Cancer Genetics," 2016). In these cancers, the variant that increases the individual's cancer susceptibility is often passed down to them from a parent and came from a variant in the germline, or egg/sperm cells. Individuals who carry a pathogenic variant in a hereditary cancer gene often develop cancer at an earlier age and have family members who have been affected by the same type of cancer ("Review of Cancer Genetics," 2016). Genetic testing for hereditary cancers often start with the individual affected with cancer, and their first- and second-degree relatives are usually tested afterwards to see if they have the same cancer-causing variant ("Review of Cancer Genetics," 2016). Because hereditary cancers tend to cluster in families, it is important that individuals have access to and take advantage of early screening and testing programs to help them monitor the development of cancers.

Hereditary colorectal cancer (hCRC) and hereditary breast and ovarian cancer (HBOC) are two of the most common types of hereditary cancers in the US, with 2-5% of all colon cancer cases and 5-10% of all breast cancer cases being attributed to hereditary and familial causes (Carroll, Cremin, et al., 2008; Jasperson, Tuohy, Neklason, & Burt, 2010). Understanding if there is a genetic etiology for each cancer can shape treatment, surveillance, and risk for both the individual with cancer and subsequent generations.

Because certain cancers tend to cluster in families, individuals often rely on family histories to determine their cancer risk. A family history of a specific cancer can indicate that the cancer syndrome is hereditary; for example, women in families with a history of pre-menopausal breast cancer would be considered at increased risk for a hereditary breast cancer. Individuals with a family history of colorectal cancer and HBOC may have variants in the same genes that their family members have and, therefore, are at an increased risk of developing cancer. Knowing the risk of developing cancer is important to make decisions about future care. These individuals may choose to undergo elective surgery to reduce their risk of developing certain cancers; for example, women at high risk for HBOC could choose to have a prophylactic mastectomy. In individuals with a family history of colorectal cancer and HBOC, preventative care, like colonoscopies and mammograms, begin at an earlier age than they would for individuals in the general population. This is important because early detection of these cancers increases the likelihood of successful treatment, including traditional methods like chemotherapy or other options like Tamoxifen, following cancer diagnosis (Soni, Simon, Cawley, & Sabik, 2018). Additionally, an individual knowing their risk of certain hereditary cancers can affect their decisions when family planning.

### 2.1.1 Hereditary colorectal cancer

hCRC is a cancer syndrome that occurs in 2-5% of all colon cancers (Ma et al., 2018). It is caused by variants in the germline that are passed to subsequent generations and can often be traced back to specific variants in cancer susceptibility genes (Ma et al., 2018). Individuals with variants in these genes have increased susceptibility for colorectal cancer and often develop the condition at an earlier age than the general population.

Lynch syndrome is a type of hereditary cancer that leads to an increased risk of developing colorectal cancer. It is the most common type of hCRC and is implicated in 2-4% of all colorectal cancers (Ma et al., 2018). These patients have a 50% lifetime risk of developing colorectal cancer and are diagnosed, on average, at age 45 (Ma et al., 2018). Lynch Syndrome is caused by variants in DNA mismatch repair (MMR) genes, which generate proteins that are responsible for proofreading and fixing mismatched base pairs following DNA replication (Ma et al., 2018). Lacking these proteins leads to issues in repairing DNA and increased microsatellite instability (Ma et al., 2018). Variants and changes in function of the MMR proteins can be found through both immunohistochemistry staining and genetic testing (Ma et al., 2018).

The most common variants in MMR genes that cause Lynch Syndrome occur in *MLH1*, *MSH2*, *MSH6*, *PMS2*, and *EPCAM* (Ma et al., 2018). These genes all change the structure and function of MMR proteins, which impairs the DNA-repair capabilities and leads to increased instability within the DNA.

In addition to Lynch syndrome, there are other types of hCRC, including familial adenomatous polyposis (FAP), that are not explored in this study.

### 2.1.2 Hereditary breast and ovarian cancer

HBOC is a cancer syndrome that is found in 5-10% of all breast and ovarian cancers (Kobayashi, Ohno, Sasaki, & Matsuura, 2013). It is primarily caused by pathogenic variants in the *BRCA1* and *BRCA2* genes, which increases an individual's susceptibility to developing breast, ovarian, and, to a lesser extent, other cancers (Petrucelli, Daly, & Pal, 2016). Unlike most breast cancers, both men and women with a variant in *BRCA1/2* are at an increased risk for developing

breast and other cancers (Petrucelli et al., 2016). Furthermore, a family history with male breast cancer is a risk factor for a germline variant in *BRCA1/2*.

*BRCA1/2* are tumor suppressor genes and are essential to repairing double-stranded breaks in the DNA (Kobayashi et al., 2013). Variants in these genes are characterized as "loss-of-function", indicating that they can no longer perform their function if there is a change (Kobayashi et al., 2013). Germline cancer-causing variants within *BRCA1/2* are often caused by pathogenic point mutations within the coding region of the gene (Kobayashi et al., 2013). While most variants in HBOC are point mutations, 8-15% of deleterious HBOC variants in families are caused by large genomic rearrangements and are often undetectable with current genetic testing capabilities (Kobayashi et al., 2013).

A variant in these *BRCA1/2* increases an individual's lifetime risk of developing breast cancer by 60-80% and ovarian cancer by 20-40% (Kobayashi et al., 2013). Determining which variants are found within each individual can be done through genetic testing, often panel testing that includes *BRCA1/2* and other breast cancer susceptibility genes (Lumish et al., 2017). Individuals with a family history of breast and ovarian cancer often see the disease clustered in families with an early age of onset, usually before menopause in women (Kobayashi et al., 2013).

### 2.2 US insurance system

The US healthcare system is fragmented with many components. Healthcare services are provided through a patchwork of private and public insurance plans and federal, state, and local governments. Healthcare institutions and providers are often disconnected from one another, which leads to difficulties when an individual changes their insurance plan. Approximately 51%

of Americans get their health insurance through their employer and have a commercial, or private, plan (Morrisey, 2020; "Visualizing Health Policy: Recent Trends in Employer-Sponsored Insurance," 2014). Other plans are public and come from the federal, state, or local government. Older individuals receiving Social Security and individuals with disabilities who do not or cannot work – and thus, cannot receive insurance through their employer – receive health insurance through Medicare, a public insurance plan that was enacted in 1965 that is offered through the federal government and regulated by the Center for Medicare and Medicaid Services (CMS). Individuals receiving Medicare often have benefit packages similar to those on private insurance plans, though the federal government and an individual's lifetime payments into Social Security are what helps fund health insurance coverage ("Basic Introduction to Medicare," 2021).

Low-income individuals and some individuals with disabilities are eligible for Medicaid, another government-subsidized health insurance program. Medicaid was introduced at the same time as Medicare and is also overseen by CMS (*Program History*, 2021). It seeks to increase access to health insurance for low-income individuals; however, it is operated by each state and leads to differences in implementation across the country (*Program History*, 2021).

The introduction of the Patient Protection and Affordable Care Act (ACA) in 2010 worked to close those gaps and included provisions for Medicaid expansion. In most states, Medicaid now covers adults with an income up to 133-138% of the federal poverty level, which fills in long-standing gaps in coverage (*Program History*, 2021). Furthermore, the ACA standardized eligibility and benefits for Medicaid among individuals in the population (*Program History*, 2021). The ACA's enactment decreased the uninsured rate from 16.8% in 2013 to 10.9% in 2019 (Garfield, 2020) and increased access to insurance coverage for the population by removing barriers surrounding pre-existing conditions.

As part of the ACA, each state had the option to expand Medicaid and develop their own provisions for coverage. Pennsylvania expanded Medicaid in 2015, which closed the coverage gap throughout the state and resulted in a 43% reduction in the uninsured rate between 2013 and 2017 (Norris, 2020). Before Medicaid expansion, Medicaid was only available to specific groups, including low-income children and adults with disabilities ("Medicaid Eligibility," 2021). However, as part of their Medicaid expansion, Pennsylvania residents could qualify for Medicaid with income as the sole determinant of eligibility ("Medicaid Eligibility," 2021). Additionally, Pennsylvania passed a statute, PH-95, to increase insurance coverage for children with disabilities regardless of family income (Advocates, 2020).

Individuals who are not eligible for Medicaid following its expansion and do not receive insurance through their employer or Medicare are eligible to participate in the insurance exchange or marketplace, in which individuals can purchase plans through the state or federal government. Federal and state governments offer subsidies for individuals so they can find affordable health insurance that meets their current healthcare needs. About 19 million non-elderly individuals purchase health insurance directly from an insurer on the marketplace (Claxon, 2014). Of those 19 million, however, about 8 million have supplemental insurance from a current or former employer, Medicaid, or another government program (Claxon, 2014). Many of the individuals receiving insurance from the marketplace receive tax credits and waivers from the federal or local government (Claxon, 2014). As of 2017, Pennsylvania residents who received insurance through the marketplace had the opportunity to select from 6 different insurance companies that participated in the exchange (Cox, 2016).

Insurance coverage for genetic testing is determined by the policy within the insurance plan that a person has or by medical director review in circumstances where policies do not exist.

Coverage for genetic testing is increasing overall nationwide, as it is becoming more of a mainstream medical procedure (PMC, 2020). Furthermore, genetic testing for hereditary cancer is among the most covered procedures across insurance companies (*Current Landscape of Genetic Testing*, 2018).

The most recent local coverage determination (LCD) from Medicare for the coverage of genetic testing for Lynch Syndrome, a form of hCRC, covered any genetic testing on genes that cause Lynch Syndrome performed on or after October 1, 2015 (CMS, 2015). In their National Coverage Analysis (NCA) decision memo, they outline which of their beneficiaries are eligible to be covered for Cologuard tests and how often they can take the test (CMS, 2014). The coverage analysis group for Medicare did not approve all methods for DNA testing for colorectal cancer, however; they stated that any other stool DNA tests that are not explicitly mentioned in their NCA are not covered (CMS, 2014).

Medicare covers genetic testing for *BRCA1/2* and published their most recent LCD on April 11, 2016 (CMS, 2016). Their coverage guidelines specifically mention the prevalence rate in the general population and the prevalence in high-risk populations, including Ashkenazi Jews (CMS, 2016). The coverage policy includes individuals with a personal history of breast cancer (CMS, 2016). However, population genetic testing, genetic testing without a personal or family history of HBOC, and screening for individuals under 18 years of age are not included in this LCD (CMS, 2016).

Medicaid coverage for genetic testing for hCRC and HBOC varies by state (ACS, 2021). Currently, all but three states' Medicaid programs cover genetic testing for *BRCA1/2* (Moddell, 2021). However, plans that cover genetic testing for HBOC and hCRC often include them in the same document. The UnitedHealthcare Plan's Community Plan that is offered in Pennsylvania has

regulations and information for testing of *BRCA1/2* in one section and outlines coverage for other hereditary cancers, including Lynch Syndrome, in more general section (UnitedHealthcare, 2021a). The policy contains regulations for when a test is considered medically necessary and incorporates both a personal and family history of these cancers into their criteria (UnitedHealthcare, 2021a). This is consistent with the text of the ACA: genetic counseling and testing for *BRCA1/2* for individuals with a family history of HBOC is explicitly mentioned in the coverage policy, while it does not specifically address hCRC (Moddell, 2021).

Many commercial insurance plans cover genetic testing for hereditary cancers and include both hCRC and HBOC in their coverage policies. The passage of the ACA included coverage for screening for hCRC (Doubeni et al., 2021); however, genetic testing for hCRC is not clearly outlined. In Pennsylvania, many insurance companies cover genetic testing for cancer genes or are in the process of developing their oncology panel policies (PMC, 2020). Currently, many panel tests exist for HBOC susceptibility genes, including *BRCA1/2*, among other genes (*Current Landscape of Genetic Testing*, 2018). The commercial coverage policy for United Healthcare clearly outlines their coverage policy in a similar manner as their community plans (UnitedHealthcare, 2021b). Just like the community plan, the commercial plan explicitly states when genetic testing for these cancers is considered medically necessary and incorporates both personal and family history into their criteria ((UnitedHealthcare, 2021b). The criteria for "medically necessary" can vary by insurance company, but these are starting to become standardized across insurance companies based on scientific evidence.

The ACA filled in some of the gaps of the health insurance system through Medicaid expansion, especially for genetic testing for cancer. For the first time, many individuals with lower income and a lower educational level, who historically had the highest cost barriers for healthcare

services, could access screening and genetic testing to help them find and diagnose their cancers (Sabik & Adunlin, 2017). Even with the introduction of the ACA and Medicaid expansion, however, there are nevertheless still gaps in the system that disproportionately affect individuals of marginalized racial and ethnic groups (Moddell, 2021). These gaps leave people uninsured or underinsured and unable to access genetic testing for cancer.

### 2.3 Disparities in cancer genetic testing and mortality

The results from testing for variants in cancer-susceptibility genes can lead to modifications in how an individual manages their care. Although testing for hCRC and HBOC is fairly straightforward and accurate, utilization of genetic testing for these conditions varies along racial and ethnic lines. A major obstacle to performing genetic testing for marginalized racial and ethnic groups is lacking adequate insurance coverage, which makes it difficult to access affordable genetic testing for these cancers (Moddell, 2021).

Currently, inequities exist between population groups in cancer genetic testing and mortality. Black populations have a higher cancer-specific mortality rate than white populations, especially among those with Medicaid insurance (Pan et al., 2017). Part of the higher cancer-specific mortality rate of Black populations come from their decreased likelihood to participate in screening programs to catch their cancers early and, therefore, are more likely to present with more advanced-stage cancers upon their diagnosis (Spinks et al., 2012). Studies have shown that Latino and Black individuals are two and three times, respectively, less likely to receive genetic testing for *BRCA1/2* when they presented with breast and ovarian cancer and *MLH1*/*MSH2* when they presented with colorectal cancer (Moddell, 2021). An individual is less likely to undergo screening

and genetic testing for colorectal cancer and HBOC for a variety of factors, including being aged 50-64, having an income of 139% of the federal poverty line or less, lacking a usual source of care, being uninsured, being non-Hispanic Asian, and not having consulted a doctor in a year (Hall et al., 2018). Additionally, women are less likely to be screened and receive genetic testing for colorectal cancer if they have less than a high school education, and non-Hispanic white women are less likely to participate in screening and receive genetic testing for HBOC than other populations (Hall et al., 2018).

### 2.3.1 Colorectal cancer rates

Colorectal cancer is the third most common cancer in the United States, with a disproportionate amount of the Black population being affected by this condition (Dharwadkar et al., 2020). Its incidence is higher for both Black men and women than it is for white men and women in the United States (Doubeni et al., 2021; O'Keefe, Meltzer, & Bethea, 2015). Because of the differing social and societal factors affecting these populations, like interpersonal and systemic racism, survival rates for colorectal cancer care are more similar by race than any other factor, including socioeconomic status (O'Keefe et al., 2015). Mortality rates for colorectal cancer among Hispanic populations, however, are lower than that of both Black and white populations (O'Keefe et al., 2015).

Although colorectal cancer incidence is higher among Black populations, they are less likely to receive adequate care and attention from practitioners regarding options for genetic testing. Black patients with a history of hCRC are less likely to be asked for a multi-generational family history in a hospital than white patients at the same hospital (Garland, Cioffi, Kirelik, Pascual, & Borum, 2021). Black patients who are at risk of colorectal cancer are not consistently

identified as being at risk for hCRC and referred for genetic testing (Garland et al., 2021). Furthermore, both Hispanic and Black patients were less likely to be referred for genetic testing for colorectal cancer than non-Hispanic whites among the same hospital cohort (Muller, 2018). Even in study cohorts with comparable variants in heritable colorectal cancer genes, Black participants were less likely to be referred for genetic testing and counseling (Dharwadkar et al., 2020). Differences in germline testing rates between Black, white, and Hispanic populations came from under-utilization of germline genetic testing services, which could impact the inequities in early-onset colorectal cancer for minoritized individuals compared to white individuals (Dharwadkar et al., 2020).

Certain variants affect the Black population at a higher proportion than other variants for hereditary colorectal cancer syndromes. Guindalini et al. (2015) found that the majority of Lynch Syndrome cases were caused by variants in the MLH1 gene, with MSH1 being the second most common variant in the Black population. The proportions of mutation rates are inconsistent with previous studies that have only been conducted on majority European populations, and this knowledge could help increase the diagnosis of Lynch Syndrome in Black populations (Guindalini et al., 2015).

### 2.3.2 Hereditary breast and ovarian cancer rates

Breast cancer is one of the most common cancers in the United States, with the most common hereditary etiologies involving *BRCA 1* and *BRCA 2*. Breast cancer is the most common cancer among those who are uninsured or covered by Medicaid (Abdelsattar, Hendren, & Wong, 2017). Though breast cancer affects all racial and ethnic groups, mortality is higher in Black women than in white women (O'Keefe et al., 2015; Pallock, 2019). Treatment advances have led

to increases in lifespan and a decrease in the mortality rate; however, the decline in mortality is slower for Black women than white women (O'Keefe et al., 2015).

In addition to having a higher rate of mortality, there are disparities that Black women and women of color are more likely to experience involving testing and screening for breast cancer. Compared to the general population, hormone-negative, also called triple-negative, breast cancers are found at higher rates in Black women (Daly & Olopade, 2015; O'Keefe et al., 2015). Even though researchers and clinicians have established that Black women have higher rates of triple-negative breast cancer, the genetic abnormalities causing these cancers are under-researched (Daly & Olopade, 2015). Black women generally have lower rates of *BRCA 1/2* pathogenic variants than non-Hispanic, non-Jewish white women because of the number of variants of uncertain significance and polymorphisms that are under-researched within their population (Daly & Olopade, 2015). Disparities exist both during and after screening and testing services, even when minoritized groups use them, that contribute to underuse of services. Black women often experience delays in follow-up after abnormal mammogram results, which results in delays, misuse, and underuse of treatment compared to white women (Daly & Olopade, 2015). Furthermore, white women are often notified of breast cancer diagnoses quicker than minoritized women, especially Black women, even when they all have the same insurance coverage (Daly & Olopade, 2015).

### 2.4 Impact of health insurance

Having adequate insurance is a strong indicator of whether an individual can access genetic testing for cancer, and due to the patchwork insurance system in the US, many individuals fall into

what is known as the coverage gap. In recent years, the ACA has helped to fill in some of the gaps (Sommers, 2020). The uninsured rate declined significantly after the passage of the ACA for people aged 18-64, and Medicaid expansion allowed more people to access genetic testing services for cancer (Zhao, 2018). In spite of the dramatic increase in those with insurance coverage, however, a significant number of Americans nevertheless remain uninsured (Zhao, 2018).

Underinsurance is also a barrier to accessing genetic testing for cancer. An individual is underinsured if they have some form of health insurance but still face a large financial burden when trying to access care (Lavarreda, Brown, & Bolduc, 2011). Underinsured individuals could potentially have difficulty in paying for hereditary cancer testing out-of-pocket, leading to increased barriers to accessing this testing.

Gaps in the system are often caused by social and structural barriers that limit an individual's access to quality health insurance. Most notably, the largest gaps in insurance coverage are caused by socioeconomic status and race/ethnicity.

### 2.4.1 Gaps by socioeconomic status

Socioeconomic status is defined by an individual's education level, income, assets and wealth, and occupation, among other factors. Individuals who have lower income and a lower educational level are disproportionately likely to be uninsured compared to those with a higher income and a higher educational level (Spinks et al., 2012).

An individual's job often determines the health insurance benefits they receive, as the majority of Americans receive their health insurance through their employers ("Visualizing Health Policy: Recent Trends in Employer-Sponsored Insurance," 2014). However, not all Americans have a job that includes health insurance (Short et al., 2012). Many of the individuals whose job

does not include health insurance as a benefit work in lower-paid, service-sector jobs. Furthermore, these individuals cannot always make health a priority because they do not have the same workplace benefits, such as sick days, as individuals with jobs that include employer-sponsored insurance (Alcaraz et al., 2020). Additionally, these individuals generally have increased out-of-pocket costs for health procedures, including genetic testing and cancer screening, which makes them less likely to present to the doctor for these procedures (Smith, Nicolla, & Zafar, 2014).

### 2.4.2 Gaps by race and ethnicity

Studies have documented that individuals of minoritized racial and ethnic groups are disproportionately likely to be uninsured compared to white individuals (Spinks et al., 2012). Social, societal, and structural barriers within the US have caused these race- and ethnicity-based gaps in insurance coverage. Residential segregation, fueled by decades of redlining and legislation barring minoritized individuals from living in the suburbs, caused the clustering of Black individuals into more economically deprived areas (O'Keefe et al., 2015). These areas are often food deserts and have fewer recreational facilities, as well as lacking as much nature and green space as majority-white areas (O'Keefe et al., 2015). Furthermore, the environments in which these individuals live are often less maintained and have hazards, like higher levels of toxic chemicals, that increase their risk of developing cancer (Alcaraz et al., 2020). In addition to living in more dilapidated areas, individuals of minoritized racial and ethnic groups often have a deep mistrust of the healthcare system due to years of unethical human subjects research on their communities (Doubeni et al., 2021).

### 2.4.3 Effects on cancer genetic testing and mortality

Lacking health insurance leads to worse outcomes for cancer prognosis. Studies have shown that cancer survival improves when an individual has health insurance (Abdelsattar et al., 2017). Being able to access insurance, therefore, is a major determinant of the quality of an individual's care once they are found to have cancer.

The ACA expanded access to insurance for all populations, and in states that expanded Medicaid, screening rates for breast and colorectal cancers increased, and mortality from these cancers decreased (Choi et al., 2015). However, white individuals benefitted more from increased access to healthcare than Black individuals, as their cancer-specific mortality rates decreased at a more significant rate than that of Black individuals (Pan et al., 2017). Furthermore, programs developed through the ACA that were intended to increase genetic testing and early detection of cancers were under-utilized by minoritized racial and ethnic groups (O'Keefe et al., 2015). For example, though the National Breast and Cervical Cancer Early Detection Program targeted vulnerable populations to encourage them to get screening, they only had a 30% and 18% utilization rate, respectively (O'Keefe et al., 2015).

## 2.5 Inequities in genetic testing and utilization of services

Overall utilization of genetic testing for cancer has increased nationwide in recent years (PMC, 2020). Many factors indicate whether an individual will undergo genetic testing to determine the genetic basis of their cancer. Insurance type is one of the most significant indicators of whether an individual will have access to genetic testing for cancer. Genetic testing for cancer

is limited by the type of insurance a person has, with the market generally following trends set by Medicare (Morrisey, 2020). Women aged 65 years or older on Medicare or employer-based insurance had the highest rates of genetic testing for breast cancer (Zhao, 2018). Men on public insurance, which includes Medicare and Medicaid, had the highest testing rates for colorectal cancer (Zhao, 2018).

Expansion of Medicaid following the introduction of the ACA led to an increase in cancer diagnoses, especially in the early stages (Soni et al., 2018). However, testing is limited in states that did not expand Medicaid and only expanded testing in Pennsylvania following the state's Medicaid expansion in 2015 (Choi et al., 2015). Furthermore, even though Pennsylvania has a high proportion of insurance companies covering genetic testing for cancer, their utilization rate for these services are not equal, indicating that other barriers likely impact access and utilization of these services (PMC, 2020).

Another barrier for cancer genetic testing is lack of access to preventative care services, like mammograms and colorectal cancer screening, that are often needed to detect early signs of cancer, and lacking health insurance further decreases the likelihood of this occurring (Bonafede et al., 2019; Kapoor, 2014). The inequitable distribution of cancer care resources between hospitals serving majority-white populations and majority-minoritized racial/ethnic populations means that there are differences in how individuals at each hospital are treated following an appointment (Alcaraz et al., 2020; Pallock, 2019).

Providers are also inconsistent in their treatment of individuals of different racial and ethnic groups. Many healthcare providers do not recommend that individuals of minoritized racial and ethnic groups receive genetic testing for cancer due to their implicit biases about the patient's race, educational level, socioeconomic status, and employment status (Johnston et al., 2021). Even

though rates of tumor analysis for colorectal cancer between racial groups are similar, individuals in minoritized racial or ethnic groups are less likely to be referred for genetic testing (Muller, 2018). Black women who have a personal history of breast cancer are often diagnosed later than white women and are not always referred to genetic counselors (Kurian et al., 2017). When providers order genetic tests for Black women, they often order the tests without a referral to a genetic counselor and pre-test counseling (Kurian et al., 2017). Additionally, Black women with a family history of HBOC are less likely to undergo genetic counseling and testing for variants in BRCA1/2 than white women with the same family history (Daly & Olopade, 2015).

Additionally, genetic testing is under-utilized overall in clinical settings across the US, often due to a lack of education about genetic services among healthcare providers and the general population (Allen, 2019). Kurian et al. (2017) discussed that many patients with breast cancer receive genetic testing without ever seeing a genetic counselor. Furthermore, half of average-risk patients with a variant of uncertain significance (VUS) undergo bilateral mastectomy, suggesting that surgeons have a limited understanding of what a VUS is and underscoring the importance of educating providers about genetics and genetic counseling (Kurian et al., 2017). A study conducted by Carroll et al. (2008) showed that 91% of family physicians and obstetricians/gynecologists knew that genetic testing exists for HBOC; however, significantly fewer numbers of these physicians knew that genetic testing existed for hCRC. These physicians were more likely to refer patients for genetic testing for these conditions if they were aware of these tests (Carroll, Cappelli, et al., 2008). However, physicians from many other specialties lack knowledge and awareness of genetic testing, and their gap in knowledge leads to their non-recommendation of genetic testing for their patients (Allen, 2019). In fact, a physician's referral of a patient to a genetic counselor or

for genetic testing is most strongly based on a patient's inquiry about these services (Daly & Olopade, 2015).

With patient inquiry being the largest motivating factor in a physician's referral, gaps in education about genetic services in different communities lead to inequities in utilization of these services. Black communities often lack knowledge about the role and importance of genetic counseling and testing in healthcare and in cancer genomics as a whole (Daly & Olopade, 2015). This gap in knowledge results in Black individuals utilizing genetic testing at a lower rate than white individuals.

## 3.0 Materials and Methods

To determine the extent of the race- and ethnicity-based insurance coverage gaps for cancer genetic testing, laboratory billing claims data were analyzed. The data was collected through the Health Records Research Request (R3) that operates through the University of Pittsburgh.

### 3.1 Data Collected

An honest broker through R3 provided data about individuals who had an encounter at any University of Pittsburgh Medical Center (UPMC) facility and had a laboratory order for genetic testing ordered between January 1, 2014, and December 31, 2019. The honest broker used current procedural terminology (CPT) codes that coded for different genetic tests for HBOC and Lynch Syndrome, a type of hCRC. These codes are summarized in Table 1.

**Table 1.** CPT codes with genes and description

| CPT code | Gene | Description of Procedure |
|---|---|---|
| 81292 | MLH1 | *MLH1* gene full sequencing |
| 81294 | MLH1 | *MLH1* gene duplication/deletion variant |
| 81317 | PMS2 | *PMS2* gene full sequencing analysis |
| 91319 | PMS2 | *PMS2* gene duplication/deletion variants |
| 81403 | EPCAM | Mopath procedure level 4 |
| 81295 | MSH2 | *MSH2* gene full sequencing |
| 81297 | MSH2 | *MSH2* gene duplication/deletion variant |

| | | |
|---|---|---|
| 81298 | MSH6 | *MSH6* gene full sequencing |
| 81300 | MSH6 | *MSH6* gene duplication/deletion variant |
| 81162 | BRCA1, BRCA2 | *BRCA1* and *BRCA2* gene full sequencing<br>*BRCA1* and *BRCA2* full duplication/deletion<br>New in 2019 |
| 81163 | BRCA1, BRCA2 | *BRCA1* and *BRCA2* gene full sequencing analysis<br>New in 2019 |
| 81164 | BRCA1, BRCA2 | *BRCA1* and *BRCA2* gene full duplication/deletion analysis<br>New in 2019 |
| 81211 | BRCA1, BRCA2 | *BRCA1* and *BRCA2* full sequence analysis<br>*BRCA1* and *BRCA2* common duplication/deletion variants<br>Expired in 2019 |
| 81213 | BRCA1, BRCA2 | *BRCA1* and *BRCA2* gene analysis<br>*BRCA1* and *BRCA2* uncommon duplication/deletion variants<br>Expired in 2019 |

*MLH1*, *PMS2*, *EPCAM*, *MSH2*, and *MSH6* are all genes that are associated with Lynch Syndrome, while *BRCA1* and *BRCA2* are genes that are associated with HBOC. Furthermore, there are multiple codes for *BRCA1* and *BRCA2* that code for the same or similar genetic tests because the CPT codes changed in 2019.

The data was analyzed by CPT code for this project. Within the data for each code, there is an individual patient ID and demographic information about each individual. The demographic information included the patient's race, ethnicity, sex, and zip code. The data also included the patient's financial class, which is the type of the insurance they had during their encounter, and an ICD9 and/or ICD10 code that differentiated between whether the individual in the encounter got a genetic test following a personal history of cancer or a family history of cancer.

## 3.2 Study Population

The study population included anyone who presented at any UPMC facility in the Pittsburgh region and surrounding areas for hereditary cancer testing. There were 2,604 unique individuals included in the study. Each individual included in the data set was at least 18 years of age or older at the time of the encounter. Sex, race, ethnicity, zip code, and insurance category for each individual and encounter were included in the data set. Furthermore, each individual's diagnosis code(s), ICD9 and/or ICD10 codes, were also included.

## 3.3 Data Analysis

The data were analyzed via Stata and interpreted graphically using Microsoft Excel functions. The analysis consisted of descriptive statistics. This included demographic breakdowns by sex, race, and ethnicity as well as determining the numbers of individuals who had each insurance category (commercial, Medicare, Medicaid, or self-pay), which was then further analyzed by cross-referencing race, ethnicity, and personal and family history of cancer data. See Appendix B for a description of the data set and Stata codes used for analysis.

## 3.4 Ethical Considerations

This study was reviewed by the University of Pittsburgh Internal Review Board (IRB), who determined that this study was not human subjects research.

## 4.0 Results

Data analysis sought to determine the demographic breakdown of individuals in the sample, as well as to determine insurance type that each individual had per encounter. ICD9/10 codes were analyzed to see if an individual had a personal history of HBOC and/or hCRC, a family history of HBOC and/or hCRC, or both. The data were divided by CPT code and analyzed to see if there were any statistically significant correlations between the variables.

### 4.1 Demographic breakdown

The study consisted of 2,604 unique individuals ranging in age from 21 years old to 97 years old, with an average age of 58.5 years old. Age was calculated by either subtracting the birth date from the death date or, if there was no death date, subtracting birth date from the date when the data set was received, which was November 16, 2021. Most individuals were between the ages of 51 and 70. Of the 2,604 participants, 279 were deceased at the time of data analysis.

The sample was composed of mostly females, with 2,172 individuals (83.41%) identifying as female and 432 individuals (16.59%) identifying as male. This likely occurred because the study looks at genetic testing for breast cancer, which affects women more than men. Additionally, *BRCA1/2* are more common than the hCRC genes that are included in the study.

In terms of racial composition, the sample was disproportionately composed of individuals who were white, with 2,427 individuals (93.20%) identifying as white. The next largest race group was individuals who were Black, with 120 individuals (4.61%) identifying as Black.

In terms of the ethnic composition, the sample was disproportionately composed of individuals who did not identify as Hispanic/Latinx, with 2.464 individuals (94.62%) identifying as not Hispanic/Latinx and 14 individuals (0.54%) identifying as Hispanic/Latinx.

The demographics of the individuals in the data set are summarized in Table 2.

**Table 2.** Demographics of study participants (n=2,604)

| | **Frequency** | **Percent (%)** |
|---|---|---|
| ***Age*** | | |
| 21-30 | 92 | 3.53 |
| 31-40 | 257 | 9.87 |
| 41-50 | 400 | 15.36 |
| 51-60 | 632 | 24.27 |
| 61-70 | 654 | 25.12 |
| 71-80 | 412 | 15.82 |
| 81-90 | 131 | 5.03 |
| 91-100 | 29 | 1.11 |
| ***Sex*** | | |
| Female | 2,172 | 83.41 |
| Male | 432 | 16.59 |
| ***Race*** | | |
| Asian | 15 | 0.58 |
| Black | 120 | 4.61 |
| Native American | 2 | 0.08 |
| White | 2,427 | 93.20 |
| Unknown | 40 | 1.53 |
| ***Ethnicity*** | | |
| Hispanic/Latinx | 14 | 0.54 |
| Not Hispanic/Latinx | 2,464 | 94.62 |
| Unknown | 126 | 4.91 |

Compared to the demographic composition of Pittsburgh, a disproportionate number of white individuals were included in this study. According to the most recent census, white individuals represent 80.53% of the population ("Pennsylvania Population 2021," 2021). However, white individuals compose 93.69% of the sample. Black and Latinx individuals, furthermore, are underrepresented in the sample. Comparisons between the population racial demographics and the study racial demographics are summarized in Table 3.

**Table 3.** Study vs Population Racial Composition

| | Study Composition (%) | Population Composition (%) |
|---|---|---|
| White | 93.20 | 80.53 |
| Black | 4.61 | 11.18 |
| Asian | 0.58 | 3.41 |
| Native American | 0.08 | 0.22 |

The percentages do not add to 100% with the study composition because those who declined to answer the question and those who did not specify their race were not included in the table. Furthermore, the population composition given in the census includes those of two or more races and other race that was not specified. Native American in the population composition includes those who identify as Native Hawaiian and Pacific Islander.

Hispanic/Latinx individuals were also underrepresented in the sample. The Kaiser Family Foundation estimated the percentage of Hispanic/Latinx individuals in the Pennsylvania population to be 7% ("The Pennsylvania Health Care Landscape," 2016). However, Hispanic/Latinx individuals only composed 0.54% of the study sample.

### 4.2 Genetic testing and insurance

There was a total of 2,604 unique individuals with a total of 2,668 genetic tests ordered. 97.427% of individuals only had one CPT code associated (which likely represents one genetic test), 2.496% had two CPT codes associated, and 0.077% had three CPT codes associated. The most common CPT code that was ordered for genetic testing for HBOC was the test code 81211, followed by 81162. Both tests are for full sequencing of *BRCA1/2* and analysis of deletions/duplications within the gene. The most common test ordered for hCRC was the test code

81403, which codes for Mopath procedure level 4 for the *EPCAM* gene. The frequency of CPT code usage is summarized in Table 4. This does not include duplicate samples or samples that were re-registered for insurance purposes.

**Table 4.** CPT code breakdown (n=2668)

| CPT Code | Frequency |
|---|---|
| 81162 | 754 |
| 81164 | 16 |
| 81211 | 1,091 |
| 81213 | 20 |
| 81292 | 17 |
| 81294 | 14 |
| 81295 | 107 |
| 81298 | 1 |
| 81317 | 6 |
| 81403 | 642 |

Most individuals used a self-pay method to cover their genetic test, followed by commercial insurance. Insurance breakdown for the study is summarized in Table 5 and includes the insurance used for all genetic tests.

**Table 5.** Insurance type breakdown (n=2668)

| Insurance type | Frequency |
|---|---|
| Commercial | 402 |
| Medicaid | 76 |
| Medicare | 93 |
| Military | 1 |
| Self-pay | 1,566 |
| Unknown/Other | 530 |

Individuals of different races and ethnicities had different proportions of insurance types for the genetic tests included in the study. This is summarized in Table 6. The 40 individuals who declined to disclose or did not specify their race were not included. Only the insurance type for the first CPT code was included.

**Table 6.** Insurance types by race and ethnicity (n=2564)

| | **Commercial** | **Medicaid** | **Medicare** | **Military** | **Self-Pay** | **Unknown/Other** |
|---|---|---|---|---|---|---|
| Asian | 4 | 1 | 0 | 0 | 8 | 2 |
| Black | 13 | 15 | 1 | 0 | 68 | 23 |
| Latinx | 3 | 0 | 0 | 0 | 11 | 0 |
| Native American | 0 | 0 | 0 | 0 | 2 | 0 |
| White | 368 | 60 | 88 | 1 | 1,411 | 485 |

CPT codes that coded for a genetic test for HBOC were combined to determine the frequency of each insurance type for this type of cancer, and the same was done for CPT codes that coded for hCRC. The insurance type for each type of cancer was determined by CPT code, not diagnosis code. Even though the majority of individuals for both tests used a self-pay method for both types of cancer, a greater proportion of individuals who had genetic testing for HBOC used insurance to cover the cost of their test. This is summarized in Table 7.

**Table 7.** Genetic testing for HBOC and hCRC – insurance type (frequency and percent) (n=2668)

| **Insurance** | **Frequency – HBOC** | **Frequency – hCRC** |
|---|---|---|
| Commercial | 336 (17.9%) | 66 (8.4%) |
| Medicaid | 66 (3.5%) | 10 (1.3%) |
| Medicare | 78 (4.1%) | 15 (1.9%) |
| Military | 1 (0.05%) | 0 (0%) |
| Self-pay | 1007 (53.5%) | 559 (71.1%) |
| Unknown/Other | 394 (20.9%) | 136 (17.3%) |

### 4.3 Multiple Tests

Three hundred fifteen study participants had multiple CPT codes associated with their unique study ID, leading to a greater number of genetic tests ordered than participants. One hundred forty-six of these individuals, or 46.3% of those with multiple CPT codes associated with their unique study ID, had the same test ordered multiple times on the same date, indicating that the providers could have used stacked codes or billed for the same code twice. Additionally, 102 individuals, or 32.4% of those with multiple CPT codes associated with their unique study ID, had the same test ordered on multiple dates. This indicates that these samples were likely re-registered for insurance reasons. For this study, any individual that had the same CPT code used more than once was presumed to just have one genetic test. Given these assumptions, it is likely that 68 individuals likely had more than one test ordered, as they had multiple CPT codes associated with their unique study IDs. Of those with multiple tests ordered, 40 individuals (59.7%) had different tests ordered on a different date. This indicates that these individuals likely had more than one test done. Seventeen individuals (25.4%) had two unique CPT codes and one individual (1.5%) had three unique CPT codes associated with their unique study IDs all entered on the same day, indicating that they likely had multiple tests ordered on the same day. The remaining sixteen individuals had a combination of multiple CPT codes with some repeating, indicating re-registration of some samples.

### 4.4 Personal and Family History of Cancer

Eligibility criteria for genetic testing for HBOC and hCRC often includes a personal or family history of these cancers. To determine who had a personal or family history of these cancers, the data was filtered with definitions corresponding ICD9/10 diagnosis codes. Individuals with an ICD code description including a family history of breast or ovarian cancer were counted as those with a family history of HBOC, and those with an ICD code description including a family history of cancer within the digestive system or the gastrointestinal system were counted as those who had a family history of hCRC. Participants who had either a personal history of breast/ovarian cancer or a neoplasm within the breast were considered to have a personal history of breast cancer; those with a personal history of colon cancer or a personal history of colon cancer or a neoplasm within the colon, rectum, or anus were considered to have a personal history of colon cancer. Additionally, those with any ICD10 code of C00-D49 were also included. These code for malignant and benign neoplasms of the organs associated with hCRC and HBOC, which indicate a personal history of breast and/or colorectal cancer. The ICD9/10 codes and definitions that were used to determine which participants had a personal and family history of HBOC and hCRC are listed in Appendix C.2.

Seven hundred thirty-three individuals had no personal or family history of either cancer that was captured in the administrative data used for this study. Seven hundred sixteen individuals had a personal history of one or more cancers. Six hundred thirty individuals had a family history of one or more cancers, and three hundred six individuals had a combination of personal and family history for both sets of cancers. Table 8 displays the proportions of personal and family history for each cancer type, which includes individuals with different combinations of personal and family

histories for each cancer type. A table with the counts for each combination of personal and family history for each cancer type is in Appendix C.3.

**Table 8.** Frequency and percent of personal and/or family history diagnoses

| Genetic Testing Type | # Individuals with: | | |
|---|---|---|---|
| | Personal Hx | Family Hx | Personal and Family Hx |
| HBOC | 690 (26.5%) | 627 (24.1%) | 506 (19.4%) |
| hCRC | 268 (10.3%) | 301 (11.6%) | 143 (5.5%) |

Participants with family histories of these cancers got tested at a younger age. 52.2% of 21-30-year-olds and 54.5% of 31-40-year-olds have a family history of HBOC, while only 2.2% and 12.8% had personal histories of HBOC, respectively. This indicates that those with a family history of cancer will likely take advantage of opportunities to undergo genetic testing at a younger age.

There were overall lower rates of personal and family histories for hCRC compared to HBOC. Fewer individuals had a family history for hCRC; however, the rate of individuals with a personal history of hCRC is fairly consistent across ages, ranging between 3.4% for those aged 91+ and 13.4% for those aged 51-60. This shows that those with a family history of hCRC are receiving consistent genetic testing across all ages.

Figure 1 shows the proportions of individuals within each age range who have a personal and family history of each cancer type.

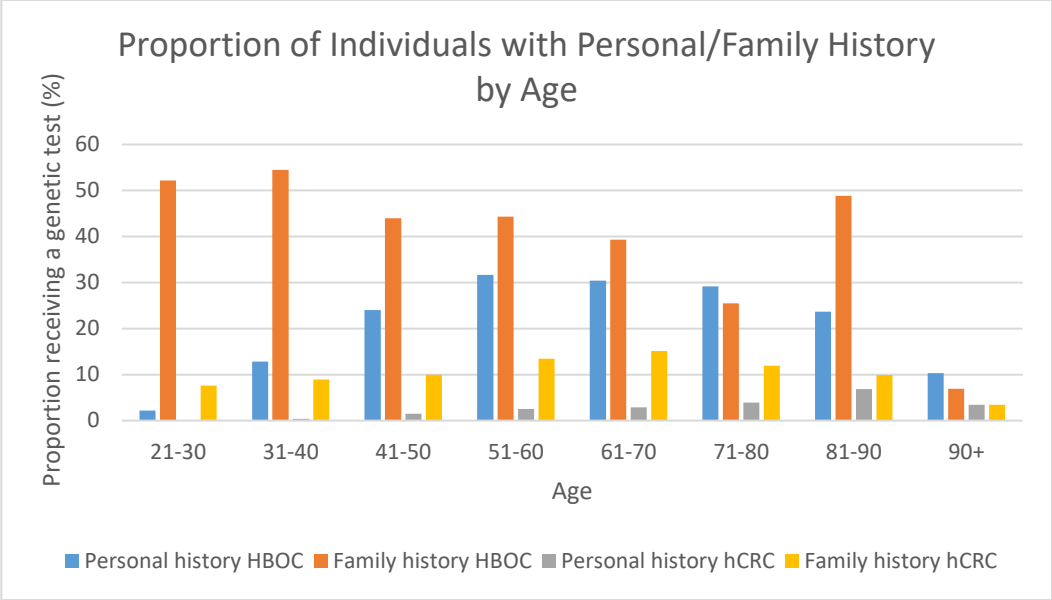


**Figure 1 Proportions of individuals with personal or family history by age**

## 4.5 Missing Data

No data points were found for CPT codes 91319, 81297, 81300, and 81163. 91319, 81297, and 81300 all code for duplication/deletion tests for genes associated with hCRC, and 81163 codes for a *BRCA1/2* full gene sequencing test. Race and ethnicity data were missing for individuals, and some either declined to disclose their race or ethnicity or did not specify their race or ethnicity. Individuals also had missing data on their insurance type.

# 5.0 Discussion

The results of this study illuminated the gaps in insurance coverage for cancer genetic testing based on race and ethnicity, demonstrating that there are inequities in knowledge of and access to care. By understanding the extent of these inequities, interventions can be developed to fill in these gaps and increase access to genetic testing for minoritized racial and ethnic groups.

## 5.1 Genetic testing and insurance

Most individuals in the study received testing for HBOC, with 68.7% of all the genetic tests in the study being for *BRCA1/2* genes and 31.3% of the genetic tests being for genes involved in hCRC. These findings are consistent with the prevalence of breast cancer and colon cancer in the US, which, in 2018, were 22 per 1000 women and 0.4 per 1000 people ("Cancer Stat Facts: Colorectal Cancer," 2021; "Cancer Stat Facts: Female Breast Cancer," 2021). The greater prevalence of HBOC compared to hCRC in the US indicates that there will be a greater number of genetic tests for HBOC.

For all genetic tests that were included in this study, most individuals listed self-pay as the method of payment for their genetic test, with almost 3 out of 4 individuals using this method to cover their test. However, it is unlikely that these individuals paid for these tests directly. This could include testing kits and direct billing of the laboratory, which falls outside the scope of this project.

Commercial insurance was the second most common type of insurance listed, followed by Medicare and Medicaid, respectively. Though it appears that most individuals lack insurance, this is likely not the case. There is a possibility that the individual's insurance plan does not cover genetic testing, the lab billed the insurance company directly, or the individual actually paid for the test themselves. However, this falls outside of the scope of the data set and this project.

A greater proportion of individuals receiving genetic testing for HBOC had it covered by insurance compared to those with hCRC, with 25.61% of genetic tests being covered by an insurance company for HBOC as compared to 9.67% of genetic tests for hCRC. This is consistent with many insurance coverage policies for hereditary cancers, as genetic testing for HBOC is explicitly covered in policies discussing genetic testing for cancer, while genes that are associated with hCRC are often mentioned with a host of other genes in the policy. Furthermore, the ACA expanded access to genetic testing for HBOC, which would increase the number of individuals on government-subsidized health insurance being able to access genetic testing (CMS, 2016).

It appears that individuals disproportionately used a "self-pay" method to cover their genetic tests, but that does not mean that these individuals lack health insurance. This may have occurred because these individuals' samples were sent to a lab in a testing kit, and the genetic testing lab billed the patient or their insurance directly. This action occurs outside the scope of the data set and is not reflected in the results and does not necessarily mean that everyone who used a self-pay method for their genetic test is uninsured.

### 5.2 Race/ethnicity and insurance coverage

Some racial and ethnic groups were disproportionately represented among certain insurance types. Previous studies have indicated that Black individuals are disproportionately likely to be uninsured or have government-subsidized insurance like Medicaid (Spinks et al., 2012). In this study, Black individuals disproportionately used Medicaid to cover their genetic test, with 27.5% of individuals in the study using this form of insurance to cover their genetic test compared to 4.3% of white individuals. Black individuals only composed 4.63% of study participants, while white individuals composed 93.69% of individuals in the study. This difference in concentration of Black study participants using Medicaid to cover their test is likely partially due to differences in sample size within the study; however, this result is consistent with previous findings.

### 5.3 Race/ethnicity and genetic testing access

Due to the disproportionate number of white individuals included in the study, it is possible that there are gaps in knowledge and access to cancer genetic testing for minoritized racial and ethnic groups. White individuals compose 93.69% of the study population, which is 15.73% greater than their composition in the Pittsburgh population. This means that all other racial and ethnic groups comprise less than 7% of the study sample.

Asian individuals are the most underrepresented in the study, with their representation in the study being 83% less than their composition in the general Pittsburgh population. Both Native American and Black individuals were underrepresented by approximately 60%. Native American

individuals' representation in the study was 63.6% less than the population composition and Black individuals' representation was 58.8% less than the population composition. This indicates that there are gaps in access to or awareness of genetic testing for cancer.

Past research has shown that Black individuals often lack knowledge of genetic testing for cancer, especially because they often have variants that have not been extensively studied (Daly & Olopade, 2015). However, it is difficult to accurately determine the scope of knowledge of genetic testing among individuals. Assessing gaps in knowledge about genetic testing falls outside the scope of this study. A way to address the gaps in knowledge of genetic testing is to invest in the health education of minoritized individuals and present information about genetic testing for cancer in culturally competent ways. Furthermore, healthcare providers should be required to have a supplemental genetics course as either part of their medical school curriculum or following their graduation, as provider knowledge of genetic testing increases the likelihood of their recommending genetic testing (Carroll, Cappelli, et al., 2008).

The results of this study further indicate that there are likely gaps in access to genetic testing for individuals of minoritized racial and ethnic groups. Minoritized racial and ethnic groups are underrepresented in the study, indicating that there could be barriers to accessing genetic testing for these individuals. Previous studies have shown that Black individuals and individuals of other minoritized racial and ethnic groups often lack access to genetic testing, whether it be due to financial barriers, implicit bias among providers, or a lack of investment in their health. Though it is hard to know the true extent of the gaps in access, this study confirms that there are nevertheless gaps in access to genetic testing along racial and ethnic lines.

Upon completion of the follow-up study, healthcare providers, insurance providers, and policymakers should develop interventions to increase access to genetic testing services for

medically underserved populations. Interventions could include policymakers investing in more health centers in areas close to medically underserved populations and encouraging and incentivizing healthcare providers to work in these areas. These interventions would help to close some of the gaps in access for these populations.

### 5.4 Family history, genetic testing, and age

The ACA's regulations deemed that insurance coverage for genetic testing for cancer would include those testing based on a family history of HBOC (UnitedHealthcare, 2021b). These regulations are consistent with the results of this study. The results show that the ACA regulations have allowed individuals of younger ages with a family history of HBOC to access genetic testing, as over half of the individuals aged 21 to 40 years had a family history of HBOC and received testing.

The large number of individuals with a personal and family history of HBOC receiving genetic testing shows that the expansion of the ACA has allowed more individuals to access testing at a younger age. Being able to access this testing allows younger individuals to make more informed decisions regarding their care, including starting screening at earlier ages and any other preventative care that they need. Additionally, offering genetic testing to younger individuals, especially those aged 21-40, can affect their decisions when family planning because they know their risk of developing cancer and passing the same variant on to their child.

## 5.5 Limitations

Limitations within the study include missing data on specific CPT codes and in the data set overall. During the data collection process, no data was found for CPT codes 91319, 81297, 81300, and 81163. This could mean that these tests were not done throughout this time or other tests were ordered. 91319, 81297, and 81300 code for *PMS2* duplication/deletion variants, *MSH2* duplication/deletion variants, and *MSH6* duplication/deletion variants. All of these genes also had a full gene sequencing test ordered, so the variants that would have been found in these tests could have been included in the scope of the other tests that were performed. 81163 codes for *BRCA1/2* full sequencing analysis and was a new code in 2019, so there likely would not have been as many data points for this due to the dates that are included in this data set. Furthermore, there was another full sequencing test for *BRCA1/2* that included duplication/deletion analysis that was new in 2019, so it is likely that the other test was ordered instead. Additionally, how genetic testing is coded is quite variable. For multi-gene panels, there are many iterations of CPT codes or code combinations that could be utilized. Therefore, we can make assumptions that certain codes fall broadly under HBOC and hCRC; however, it is difficult to make those determinations with certainty. Furthermore, multiple CPT codes per individual could be due to: stacked billing codes for one genetic test, multiple genetic tests, billing errors, and/or new registration dates obtained after insurance authorization.

There were missing data for the race and ethnicity of individuals as well as individuals either declining to answer their race and ethnicity or not specifying their race or ethnicity. There was also missing data on financial class. This means that any correlation between insurance type, genetic testing utilization, and race and ethnicity would be affected and not as accurate as it would be if all of the data were available for analysis. Furthermore, there is not as comprehensive of a

view on who has which type of insurance and what was used for genetic testing, which affects the generalizability of the data.

Another limitation is the different testing recommendations for individuals with a family history of hCRC and HBOC and a personal history of hCRC and HBOC. For individuals with a family history of HBOC and hCRC, screening for these conditions begins at an earlier age than the general population so the condition can be caught early. There are also fewer restrictions on genetic testing for individuals with a family history of these conditions. Furthermore, ACA coverage for genetic testing for *BRCA1/2* for individuals with a family history of HBOC has increased access to testing in this population. Additionally, many direct-to-consumer testing companies offer testing for *BRCA1/2* variants, so any individuals who use this test would not be included in the scope of this data.

In addition to these limitations, there are limitations that could affect this data that fall outside of the scope of the data set. Individuals also could have a primary and a secondary type of insurance, which is not reflected in this data set. Some labs use alternative billing practices that cannot be captured within the scope of this data set. Additionally, some samples could have been billed wrong, so that would not be included within the data. Samples could also have been re-registered for insurance reasons, which would make the genetic testing date different from the clinic date. An individual's insurance status could have changed before or after their appointment, and this change might not be included in the data. Any change in insurance status for these individuals would affect the proportions of individuals with each type of insurance.

### 5.5.1 Limits to the generalizability of the work

This research was conducted in a specific region of the country, so the conclusions drawn about inequities in access are only generalizable to the greater Pittsburgh region. These results cannot be generalized to the entire country. Furthermore, the overrepresentation of white individuals in the study limits the generalizability to the entire Pittsburgh region, as the study sample and the population do not have the same proportions of racial and ethnic groups.

## 5.6 Future Directions

This study uses quantitative methods to evaluate gaps in access to genetic testing for HBOC and hCRC and insurance coverage for this genetic testing for individuals of different racial and ethnic groups. A follow-up qualitative study could answer questions about why the gaps exist between racial and ethnic groups and identify any systemic barriers currently in place. These results can be used to develop interventions to address these inequities and close the gap between white individuals and individuals of minoritized racial and ethnic groups.

The Pittsburgh region has multiple hospital systems; this study explored the genetic testing laboratory data from only UPMC facilities. A future study could expand on this work and use data from all hospital systems in the Pittsburgh region to see if these issues are specific to UPMC facilities or are truly systemic.

One future direction for this study is to evaluate the levels of knowledge of genetic testing by race and ethnicity to determine the extent of these gaps and develop culturally appropriate interventions to increase knowledge of genetic testing for cancer. This can be done through both

qualitative and quantitative methods, whether it be through a survey given to individuals at primary care or specialty doctors' appointments that is then analyzed or focus groups being conducted among different racial and ethnic groups. Researchers could then compare the results of each racial and ethnic group's knowledge of genetic testing to a reference group – likely white individuals – to determine the extent of the knowledge gap between different racial and ethnic groups.

In future studies, data could be further stratified to evaluate an individual's personal or family history by race and ethnicity. This would determine if there are any racial or ethnic groups that are disproportionately likely to have a personal or family history of HBOC and hCRC. Furthermore, Black individuals are less likely to have a variant in BRCA1/2, so future studies could expand the genes that were tested to see if there is an increase in the number of nonwhite individuals with a personal or family history of HBOC (O'Keefe et al., 2015). Including variants that are more likely to be found in nonwhite individuals would provide a more robust prediction of the true number of individuals with personal and family histories of HBOC and hCRC within each racial and ethnic group.

Researchers could also explore whether there are any inequities in referrals for genetic counseling and cancer genetic testing through analyzing referral patterns of individuals of different racial and ethnic backgrounds from the UPMC hospital system. Any inequities in these referral patterns would determine the necessity for an intervention and a standardization across hospitals and healthcare facilities for the referral of patients to genetic testing.

### 5.6.1 Implications for practice in the field

To address inequities in access to cancer genetic testing, individuals and companies within multiple industries must make changes to their current policies and procedures. One current

inequity is the lack of knowledge on cancer variants that affect nonwhite populations. Genetics researchers should prioritize researching variants that affect nonwhite populations and develop genetic tests that have equal effectiveness and efficacy to those that already exist. Having highly accurate, sensitive, and specific tests that are tailored towards nonwhite populations will lead to increased trust between underserved racial and ethnic groups and genetics researchers. This will not only lead to more informative results but allow them to feel more empowered and increase the likelihood that medically underserved populations will want to receive genetic testing, leading to a higher utilization of genetic testing for these communities.

Genetic counselors also play an important role in expanding access to genetic testing for cancer. Right now, the genetic counseling field is predominantly composed of white women (Chien, 2020). A first step to addressing these inequities would be to increase the diversity within the field, as most people feel more comfortable when they see providers who look like them. Additionally, genetic counselors are experts in genetic conditions and have expertise in effective communication with individuals. Genetic counselors should use their knowledge and platform to spread awareness about genetic testing and educate medically underserved populations about their options for genetic testing for cancer.

A large gap currently exists within the insurance landscape. To ensure that all individuals have access to genetic testing, insurance companies and government-sponsored insurance programs must first work to expand access to coverage for individuals of all racial and ethnic groups, especially those who are medically underserved. Insurance companies across the United States should perform outreach campaigns and work to provide coverage for medically underserved populations. Furthermore, many genetic tests for cancers are not explicitly covered by an insurance company. Private insurance companies and government-sponsored insurance

programs should work to develop a comprehensive cancer genetic testing coverage policy so their beneficiaries know what is covered.

In addition to expanding coverage policies, insurance companies and government-sponsored insurance programs should contract with genetic counselors to work to expand access to genetic testing. Cancer genetic counselors perform pre- and post-test counseling and deliver the results of genetic testing to their patients. Contracting with genetic counselors will allow patients to receive adequate education before and after their test with a clear, accurate explanation of their results. Previous studies have shown gaps in referrals to genetic counseling, so selective contracting with genetic counselors will help increase access to genetic testing for all racial and ethnic groups.

It is also imperative to present information to medically underserved communities in culturally appropriate ways. Genetics professionals must work with community leaders when preparing information campaigns to make sure that their message is understood and well-received by these communities. Community leaders have already earned the trust and respect of their communities; working with them will build trust between the medical institution and these communities. This will begin to repair the relationship between medical institutions and the populations they have mistreated for centuries and educate them about the health benefits that genetic testing can have. Improving this relationship by working with community leaders will likely increase the likelihood that minoritized racial and ethnic groups will want to receive genetic testing.

### 5.6.2 Suggested next steps

Inequities in access to genetic testing is a widespread problem and is not limited to the greater Pittsburgh region. This study should be replicated in other regions of the United States to see if the patterns that were found in this study are similar nationwide. These studies should occur in major cities and geographic regions of the country. When conducting these future analyses, the health insurance and healthcare infrastructure as well as the racial and ethnic composition of the area should be considered.

Once these analyses are completed, action should be taken on the state level. The state should first work to develop interventions to increase access to insurance coverage and genetic testing. One of the ways this can be done is to expand the reach of the state's insurance marketplace through partnering with community organizations to target medically underserved communities. State officials should collaborate with community leaders in all underserved communities to develop information campaigns that will disseminate facts about the state insurance program, including how it is structured, benefit packages, and how to enroll in it. This could be done through distributing written materials, both in print and online, and through workshops, all of which should be conducted at locations that are frequented by community members, like community centers. States should also incentivize private insurance companies to perform outreach campaigns within underserved communities and increase enrollment.

Should the results of the studies be similar between states, an intervention to address these gaps should occur at the federal level. The federal government has made progress in developing healthcare interventions through the passage and implementation of the ACA in 2010, but there are nevertheless still ways to improve the healthcare landscape to address these inequities. Like a state-level intervention that spreads information about the state-based marketplace, there should

be a similar campaign to share information about the federal exchange. The federal government should invest more money into enrollment campaigns that target medically underserved populations. Furthermore, Medicare should adopt a comprehensive cancer genetic testing coverage policy to increase the likelihood that an individual can access cancer genetic testing; this would impact the insurance market and increase the likelihood that private insurance companies adopt this policy as well.

As genetic testing and a push for personalized medicine continue to proliferate the healthcare landscape, it is imperative that action is taken at both the state and federal level to develop policies for equitable access to genetic testing and ensure current gaps in access to genetic testing by race and ethnicity are eradicated.

## 6.0 Funding

Funding was provided by the MPH in Public Health Genetics program.

# Appendix A IRB Approval

University of Pittsburgh | Institutional Review Board
Office of Research Protections

## NOT HUMAN RESEARCH DETERMINATION

| | |
|---|---|
| Date: | October 20, 2021 |
| Review Type: | Initial Study |
| IRB: | STUDY21070152 |
| PI: | Haley Director |
| Title: | Race- and Ethnicity-Based Insurance Coverage Gaps for Genetic Testing for Cancer in the Greater Pittsburgh Region |
| Funding: | None |
| Documents Reviewed: | • Director.Exemption_Secondary Data.Specimens.docx, Category: IRB Protocol;<br>• Honest Broker Form.pdf, Category: Honest Broker Signed Agreement; |

The Institutional Review Board determined that the proposed activity is not research involving human subjects as defined by DHHS and FDA regulations.

This determination applies only to the activities described in the IRB submission and does not apply should any changes be made. If changes are made and there are questions about whether these activities are research involving human in which the organization is engaged, please submit a new request to the IRB for a determination. You can create a modification by clicking **Create Modification / CR** within the study.

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, Amy Fuhrman.

*Please take a moment to complete our Satisfaction Survey as we appreciate your feedback.*

Human Research Protection Office 3500 Fifth Avenue, Suite 106 Pittsburgh, PA 15213 www.hrpo.pitt.edu

**Appendix Figure 1 IRB Approval**

# Appendix B Coding Information

**Appendix Table 1 Stata Data names**

| Data name | Definition |
|---|---|
| study_id | Each individual's unique study ID |
| birth_date | Each individual's birth date |
| death_date | For individuals who died, date of death<br>For individuals still living, date of receipt of data set (11-16-2021) |
| gender | Each individual's sex |
| race | Each individual's race |
| ethnicity | Each individual's ethnicity |
| age | Each individual's age. Calculated in Excel. |
| proc_code1 | First procedure code |
| ins1 | First insurance/financial class |
| proc_code2 | Second procedure code |
| ins2 | Second insurance/financial class |
| proc_code3 | Third procedure code |
| ins3 | Third insurance/financial class |
| proc_code4 | Fourth procedure code |
| ins4 | Fourth insurance/financial class |
| proc_code5 | Fifth procedure code |
| ins5 | Fifth insurance/financial class |
| phist_bc | Personal history of HBOC |
| famhist_bc | Family history of HBOC |
| phist_crc | Personal history of hCRC |
| famhist_crc | Family history of hCRC |
| zip_code | Each individual's zip code from place of residence |
| city | City of residence based on individual zip code |

**Appendix Table 2 Stata codes**

| Stata code | Definition |
|---|---|
| count age if <31 | Individuals with ages under 31 (21-30) counted |
| count if age <41 | Individuals with ages under 41 (21-40) counted. Number subtracted from previous result to get ages 31-40. |
| count if age <51 | Individuals with ages under 51 (21-50) counted. Number subtracted from previous result to get ages 41-50. |
| count if age <61 | Individuals with ages under 61 (21-60) counted. Number subtracted from previous result to get ages 51-60. |
| count if age <71 | Individuals with ages under 71 (21-70) were counted. Number subtracted from previous result to get ages 61-70. |
| count if age <81 | Individuals with ages under 81 (21-80) were counted. Number subtracted from previous result to get ages 71-80. |
| count if age <91 | Individuals with ages under 01 (21-90) were counted. Number subtracted from previous result to get ages 81-90. |
| count if age >90 | Individuals with ages over 90 (91-97) counted |
| summarize age | Summary table with maximum, minimum, and average age generated |
| hist age, freq | Histogram with age distribution generated |
| tab gender | Table with frequencies of individuals in each gender generated |
| tab race | Table with frequencies of individuals of each race generated |
| tab ethnicity | Table with frequencies of individuals of each ethnicity generated |
| tab death_date | Table with the total number of individuals who had died |
| tab proc_code1 | Table with frequencies of first procedure code |
| tab proc_code2 | Table with frequencies of second procedure code |
| tab proc_code3 | Table with frequencies of third procedure code |
| tab proc_code4 | Table with frequencies of fourth procedure code |
| tab proc_code 5 | Table with frequencies of fifth procedure code |
| tab ins1 | Table with frequencies of first insurance type |

| | |
|---|---|
| tab ins2 | Table with frequencies of second insurance type |
| tab ins3 | Table with frequencies of third insurance type |
| tab ins4 | Table with frequencies of fourth insurance type |
| tab ins5 | Table with frequencies of fifth insurance type |
| bysort proc_code1: tab ins1 | Table with first insurance type sorted by first procedure code |
| bysort proc_code2: tab ins2 | Table with second insurance type sorted by second procedure code |
| bysort proc_code3: tab ins3 | Table with third insurance type sorted by third procedure code |
| bysort proc_code4: tab ins4 | Table with fourth insurance type sorted by fourth procedure code |
| bysort race: tab ins1 | Table with first insurance type sorted by race |
| by race: tab ins2 | Table with second insurance type sorted by race |
| by race: tab ins3 | Table with third insurance type sorted by race |
| by race: tab ins4 | Table with fourth insurance type sorted by race |
| bysort ethnicity: tab ins1 | Table with first insurance type sorted by ethnicity |
| tab zip_code | Table with frequencies of each zip code |
| tab city | Table with frequencies of each city |
| tab phist_bc | Table with frequencies of a personal history of HBOC |
| tab famhist_bc | Table with frequencies of a family history of HBOC |
| tab phist_crc | Table with frequencies of a personal history of hCRC |
| tab famhist_crc | Table with frequencies of a family history of hCRC |
| tab phist_bc famhist_bc | Table with frequencies of individuals with a personal and family history of HBOC |
| tab phist_crc famhist_crc | Table with frequencies of individuals with a personal and family history of hCRC |
| tab phist_bc phist_crc | Table with frequencies of individuals with a personal history of HBOC and hCRC |
| tab famhist_bc famhist_crc | Table with frequencies of individuals with a family history of HBOC and hCRC |
| bysort race: tab phist_bc | Table with frequencies of personal history of HBOC sorted by race |
| By race: tab famhist_bc | Table with frequencies of family history of HBOC sorted by race |

| | |
|---|---|
| by race: tab phist_crc | Table with frequencies of personal history of hCRC sorted by race |
| by race: tab famhist_crc | Table with frequencies of family history of hCRC sorted by race |
| bysort ethnicity: tab phist_bc | Table with frequencies of personal history of HBOC sorted by ethnicity |
| by ethnicity: tab famhist_bc | Table with frequencies of family history of HBOC sorted by ethnicity |
| by ethnicity: tab phist_crc | Table with frequencies of personal history of hCRC sorted by ethnicity |
| by ethnicity: tab famhist_crc | Table with frequencies of family history of hCRC sorted by ethnicity |
| bysort phist_bc: tab age | Table with frequencies of each age sorted by yes/no personal history of HBOC |
| bysort famhist_bc: tab age | Table with frequencies of each age sorted by yes/no family history of HBOC |
| bysort phist_crc: tab age | Table with frequencies of each age sorted by yes/no personal history of hCRC |
| bysort famhist_crc: tabe age | Table with frequencies of each age sorted by yes/no family history of hCRC |

# Appendix C Further Data Analysis

## Appendix C.1 Insurance type by CPT code

**Appendix Table 3 Insurance type, CPT code 81162**

| Insurance | Frequency |
|---|---|
| Auto | 1 |
| Commercial | 111 |
| Medicaid | 16 |
| Medicare | 38 |
| Self-pay | 215 |

**Appendix Table 4 Insurance type, CPT code 81164**

| Insurance | Frequency |
|---|---|
| Commercial | 5 |
| Medicaid | 2 |

**Appendix Table 5 Insurance type, CPT code 81211**

| Insurance | Frequency |
|---|---|
| Commercial | 213 |
| Medicaid | 48 |
| Medicare | 40 |
| Military | 1 |
| Self-pay | 781 |

**Appendix Table 6 Insurance type, CPT code 81213**

| Insurance | Frequency |
|---|---|
| Commercial | 7 |
| Self-pay | 11 |

**Appendix Table 7 Insurance type, CPT code 81292**

| Insurance | Frequency |
|---|---|
| Commercial | 4 |
| Medicaid | 1 |
| Self-pay | 9 |

**Appendix Table 8 Insurance type, CPT code 81294**

| Insurance | Frequency |
|---|---|
| Commercial | 1 |
| Medicaid | 1 |
| Self-pay | 12 |

**Appendix Table 9 Insurance type, CPT code 81295**

| Insurance | Frequency |
|---|---|
| Commercial | 37 |
| Medicaid | 5 |
| Medicare | 5 |
| Self-pay | 50 |

**Appendix Table 10 Insurance type, CPT code 81298**

| Insurance | Frequency |
|---|---|
| Self-pay | 1 |

**Appendix Table 11 Insurance type, CPT code 81317**

| Insurance | Frequency |
|---|---|
| Commercial | 4 |
| Self-pay | 2 |

**Appendix Table 12 Insurance type, CPT code 81403**

| Insurance | Frequency |
|---|---|
| Commercial | 20 |
| Medicaid | 3 |
| Medicare | 10 |
| Self-pay | 485 |

## Appendix C.2 Personal/Family History by ICD9/10 code

**Appendix Table 13 ICD9/10 Codes for Personal/Family History**

| Code/Code Family | Definition |
|---|---|
| ***ICD9*** | |
| 150-159 | Malignant neoplasm of digestive organs |
| 174-175 | Malignant neoplasm of female/male breast |
| 179-189 | Malignant neoplasm of genitourinary organs |

| | |
|---|---|
| V10-V19 | Persons with potential hazards related to personal and family history |
| ***ICD10*** | |
| C15-C26 | Malignant neoplasms of digestive organs |
| C50 | Malignant neoplasm of breast |
| C51-58 | Malignant neoplasms of female genital organs |
| D10-D49 | Benign neoplasms, except benign neuroendocrine tumors |
| Z77-Z99 | Persons with potential health hazards related to family and personal history and certain conditions related to health status |

## Appendix C.3 Personal/family history information

**Appendix Table 14 Personal/Family History**

| | **Frequency** |
|---|---|
| No history | 733 |
| Personal history HBOC | 523 |
| Family history HBOC | 496 |
| Personal history hCRC | 98 |
| Family history hCRC | 70 |
| Personal/family history HBOC | 380 |
| Personal/family history hCRC | 95 |
| Personal history HBOC and hCRC | 54 |
| Family history HBOC and hCRC | 64 |
| Personal history HBOC, family history hCRC | 104 |
| Personal history hCRC, family history HBOC | 42 |
| Personal history HBOC, family history HBOC/hCRC | 63 |
| Personal history hCRC, family history HBOC/hCRC | 25 |
| Personal history HBOC/hCRC, family history HBOC | 49 |
| Personal history HBOC/hCRC, family history hCRC | 9 |
| Personal/family history HBOC/hCRC | 14 |

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