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Statewide Genomic Screening in South Carolina: Coverage and Positivity Across Rurality and Social Vulnerability

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1. Background

The U.S. Centers for Disease Control and Prevention (CDC) identifies hereditary breast and ovarian cancer syndrome (HBOC), Lynch syndrome (LS), and familial hypercholesterolemia (FH) as Tier One applications for genomic screening, given their substantial potential to improve public health outcomes. To advance population-level genomic screening, the statewide initiative In Our Deoxyribonucleic Acid (DNA) South Carolina (SC) was launched in SC on November 8, 2021, with the goal of enrolling at least 100,000 individuals. Ensuring representation across all counties, spanning the urban-rural continuum and communities with varying levels of social vulnerability, was a critical priority to build a cohort reflective of the state's diverse population.

2. Goal

The goal was to monitor and communicate enrollment in In Our DNA SC (overall and stratified by rurality and social vulnerability) to ensure representation across the state's diverse communities.

3. Solutions and Methods

We developed a user-friendly data visualization dashboard to display patient enrollment across all 46 SC counties. Deidentified patient-level data were aggregated to the county level and geo-linked with urban-rural classifications and social vulnerability measures. Rural-Urban Continuum (RUC) codes were derived from the U.S. Department of Agriculture's Economic Research Service, while county social vulnerability scores were obtained from the CDC's Social Vulnerability Index (SVI). Enrollment rates (per 100,000 population) were calculated to enable comparisons in coverage. The data pipeline was managed in R and Tableau was used to build dynamic dashboards with interactive features.

4. Outcomes

A total of 85,973 individuals has been recruited so far across all 46 counties in SC (Figure). County-level maps illustrated enrollment (counts and rates per 100,000) for HBOC, LS, and FH testing. In SC, the majority of counties (18 of 46) are classified as RUC code 2 (metro areas with populations of 250,000 to 1 million), and there are no counties assigned RUC codes 5 or 7. Coverage rates exceeded 1,500 per 100,000 across all RUCs, with the highest coverage observed in RUC 3 for HBOC (3,128.1), LS (2,284.2), and FH (2,284.2). Test positivity rates (based on valid test results) were highest in RUC 8 for HBOC (747.7), RUC 6 for LS (698.5), and RUC 3 for FH (637.3). By social vulnerability score quartiles, the distribution of counties was SVI 1 [0.00–0.25]: one county; SVI 2 [0.26–0.50]: 11 counties; SVI 3 [0.51–0.75]: 11 counties; and SVI 4 [0.76–1.00]: 12 counties. Coverage rates exceeded 2,000 per 100,000 across all SVI quartiles, with the highest coverage in SVI 1 for HBOC (3,110.2), LS (2,261.1), and FH (2,261.1). Test positivity rates were highest in SVI 3 counties for HBOC (759.9), in SVI 4 for LS (483.3), and in SVI 3 for FH (676.1).

5. Lessons Learned and Future Directions

This statewide initiative achieved broad enrollment across diverse counties, demonstrating strong coverage across rural-urban classifications and social vulnerability quartiles. The findings highlight meaningful variation in test positivity rates, underscoring the importance of continued monitoring to guide equitable genomic screening efforts.

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Figure: Enrolled for Genetic Testing