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

Sonawane, Kalyani, PhD, Judge, Daniel P, MD, Moore, Ella, BS, Norman, Samantha MPH, Allen, Caitlin G, PhD, MPH

MUSC Hollings Cancer Center - Charleston, SC

MUSC Medical University of South Carolina | Hollings Cancer Center, An NCI-Designated Cancer Center

BACKGROUND

- CDC identifies **Hereditary Breast Ovarian Cancer Syndrome (HBOC), Lynch syndrome (LS), and Familial Hypercholesterolemia (FH)** as Tier 1 genomic conditions where early detection can save lives.
- South Carolina's rural and socially vulnerable populations face barriers that may limit **population-wide genomic screening (PWGS)** benefits.
- Since 2019, SC Medicaid has expanded coverage for BRCA testing and now includes whole-exome sequencing, whole-genome sequencing, and hereditary cancer panels.
- In 2021, MUSC launched ***In Our DNA SC*** to provide free Tier 1 genetic screening, aiming to enroll 100,000+ people with emphasis on underserved groups.
- **OBJECTIVE**: *Show how implementation science and informatics enable PWGS and assess coverage and positivity by rurality and social vulnerability*

METHODS

In Our DNA SC is a free statewide genetic screening program run by MUSC with Helix.

Recruitment & Screening:

- Participants enrolled via MyChart with e-consent.
- Samples collected in clinics, community events, or at-home kits.
- Community events broadened statewide reach.
- Helix sequenced samples for CDC Tier 1 genes (HBOC, LS, FH).

Informatics Infrastructure:

- Program data integrated into *SC-SPOT* for geographic and equity monitoring.
- Data linked to county rurality (RUC) and social vulnerability (SVI).
- Screening and positivity rates calculated & visualized in Tableau.

ACKNOWLEDGMENTS

The research reported in this study was supported by the US National Library of Medicine (G08LM014413), and Hollings Cancer Center Cancer Control Seed Funds and the National Cancer Institute (P30CA138313).

Questions? Citations request? *Email: sonawane@musc.edu*

RESULTS

Table 1. *In Our DNA SC* program: total consented, screening rate, and test positivity rate by urbanicity/rurality.

RUC code*	No. of counties	Total Consented	HBOC		LS		FH	
			Screened†	*Positive*‡	*Screened*†	*Positive*‡	*Screened*†	*Positive*‡
1	3	3,974	1,037.2	347.0	**699.1**	268.5	**699.1**	492.2
2	18	59,545	1,887.3	1,109.7	1,357.7	354.6	1,357.7	522.4
3	5	9,442	**1,906.9**	932.5	**1,400.7**	349.5	**1,400.7**	576.7
4	4	4,130	1,424.4	623.2	1,036.2	331.0	1,036.2	413.7
5	None	-	-	-	-	-	-	-
6	9	3,360	1,252.7	477.5	791.4	627.3	791.4	418.2
7	None	-	-	-	-	-	-	-
8	6	1,242	1,104.8	735.3	861.4	NR	861.4	420.2
9	1	185	1,340.9	NR	839.6	NR	839.6	NR

Abbreviation: RUC, Rural-Urban Continuum; NR, Not Reportable due to extremely small sample size.
*Rural-Urban Continuum codes classify counties into nine categories, from 1 (most urban; yellow) to 9 (most rural; dark green) based on metro population size, degree of urbanization, and proximity to metropolitan areas. They distinguish metropolitan counties by the size of their metropolitan area and nonmetropolitan counties by their level of urbanization and proximity to a metropolitan area.
†Screening rates (per 100,000 population); ‡Positive test results are calculated as the number of positives per 100,000 tests.

Table 2. *In Our DNA SC* program: total consented, screening rate, and test positivity rate by overall social vulnerability and social vulnerability themes.

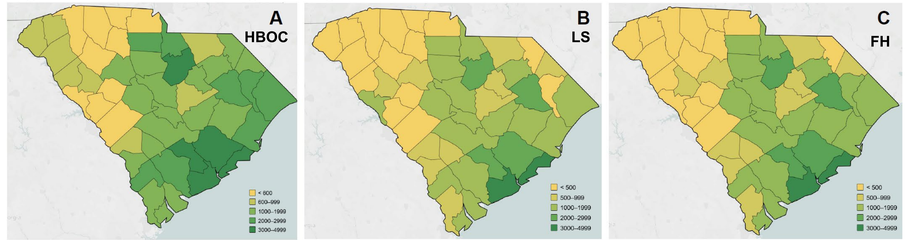
Social Vulnerability*	No. of counties	Total Consented	HBOC		LS		FH	
			Screened†	*Positive*‡	*Screened*†	*Positive*‡	*Screened*†	*Positive*‡
Overall								
Quartile 1	12	47,474	1,954.8	1,049.8	1,422.1	320.0	1,422.1	535.5
Quartile 2	11	19,115	1,673.2	957.3	1,188.4	447.3	1,188.4	455.9
Quartile 3	11	10,080	1,257.6	997.7	888.4	351.7	888.4	602.9
Quartile 4	12	5,209	1,521.7	618.8	988.1	400.7	988.1	400.7
Housing Characteristics								
Quartile 1	12	46,846	2,002.5	1,051.2	1,469.8	317.9	1,469.8	536.7
Quartile 2	11	18,145	1,424.4	988.0	995.3	391.4	995.3	527.9
Quartile 3	11	11,980	1,558.7	884.6	1,096.6	486.1	1,096.6	458.3
Quartile 4	12	4,907	1,501.9	756.4	977.2	308.6	977.2	424.4
Housing and Transportation								
Quartile 1	12	34,538	2,038.0	1,213.4	1,517.1	324.7	1,517.1	519.5
Quartile 2	11	25,296	1,635.5	801.0	1,142.0	360.8	1,142.0	544.5
Quartile 3	11	18,984	1,701.3	970.3	1,195.9	411.5	1,195.9	516.5
Quartile 4	12	3,060	907.1	418.1	589.0	387.6	589.0	276.9
Minority Status								
Quartile 1	12	24,752	1,220.6	909.4	860.5	318.5	860.5	458.7
Quartile 2	11	38,369	2,590.8	1,106.4	1,895.9	386.6	1,895.9	546.2
Quartile 3	11	7,611	1,812.8	837.7	1,318.9	256.5	1,318.9	676.1
Quartile 4	12	11,146	1,485.5	923.1	1,018.7	416.3	1,018.7	448.4
Socioeconomic Status								
Quartile 1	11	38,160	1,912.9	948.1	1,356.7	357.1	1,356.7	510.2
Quartile 2	11	24,901	1,759.0	1,121.7	1,312.4	342.4	1,312.4	513.6
Quartile 3	12	13,775	1,424.4	1,039.6	1,005.3	371.3	1,005.3	532.2
Quartile 4	12	5,042	1,473.4	592.0	960.4	413.8	960.4	564.3

*Social vulnerability quartiles indicate lowest to highest vulnerability: *Quartile 1 (0.00-0.25), Quartile 2 (0.51-0.75), Quartile 3 (0.26-0.50), and Quartile 4 (0.76-1.00).*
†Screening rates (per 100,000 population); ‡Positive test results are calculated as the number of positive tests per 100,000 results.

82,420 participants enrolled as of July 23, 2025, from all 46 South Carolina counties.
50,897 participants completed screening.

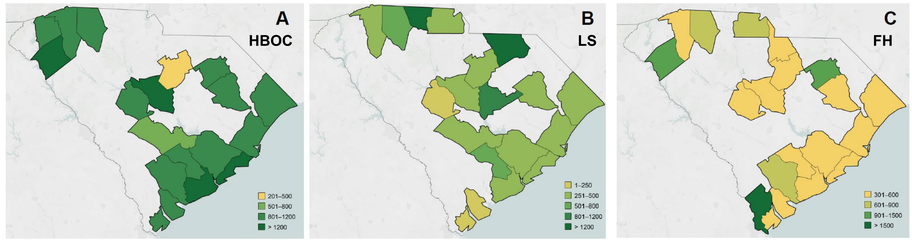
Adults aged 18–64 (71.6%)
Female (72.6%)
White (73.1%)
Non-Hispanic individuals (81.2%)

Figure 1. Genetic screening coverage rates, *In Our DNA SC* program.



The screening coverage rate per 100,000 was highest for Charleston County for HBOC (4932.5; **Panel A**), LS (3751.4; **Panel B**), and FH (3751.4; **Panel C**). Screening coverage rates for HBOC were lowest in McCormick County (115.7) and in Cherokee County for LS (142.9), and FH (142.9).

Figure 2. Positive genetic screening rates, *In Our DNA SC* program.



Positivity rates per 100,000 were highest for HBOC in Anderson County (2835.5) (**Panel A**), for LS were highest in Cherokee (4838.7) **(Panel B)**, and FH were highest in Jasper (2304.1) **(Panel C).**

CONCLUSION

- A **statewide PWGS program**, supported by an implementation framework and informatics platform, reached people across many geographic and social groups of South Carolina.
- Differences in positivity rates may reflect variations in **underlying risk or differences in who can access testing.**
- Combining implementation science with strong informatics systems is likely **essential for expanding equitable PWGS in rural and socially vulnerable communities.**