



How the lack of ethnic diversity in GWAS contributes to inaccurate risk estimates of genetic risk for minority groups

Sadie Ibbotson-Brown and Candida Goodnough

Palo Alto High School



INTRODUCTION

- There is an issue surrounding a lack of diversity in genetic studies.
- 78% of genome-wide association studies come from populations that come of European descent when in reality, these groups only make up 16% of the global population (Sirugo, 2019).
- Studies taken from mainly European populations can result in a disparity in the level of health care provided to these minority groups who were not accurately researched in the study.

RESEARCH METHODOLOGIES

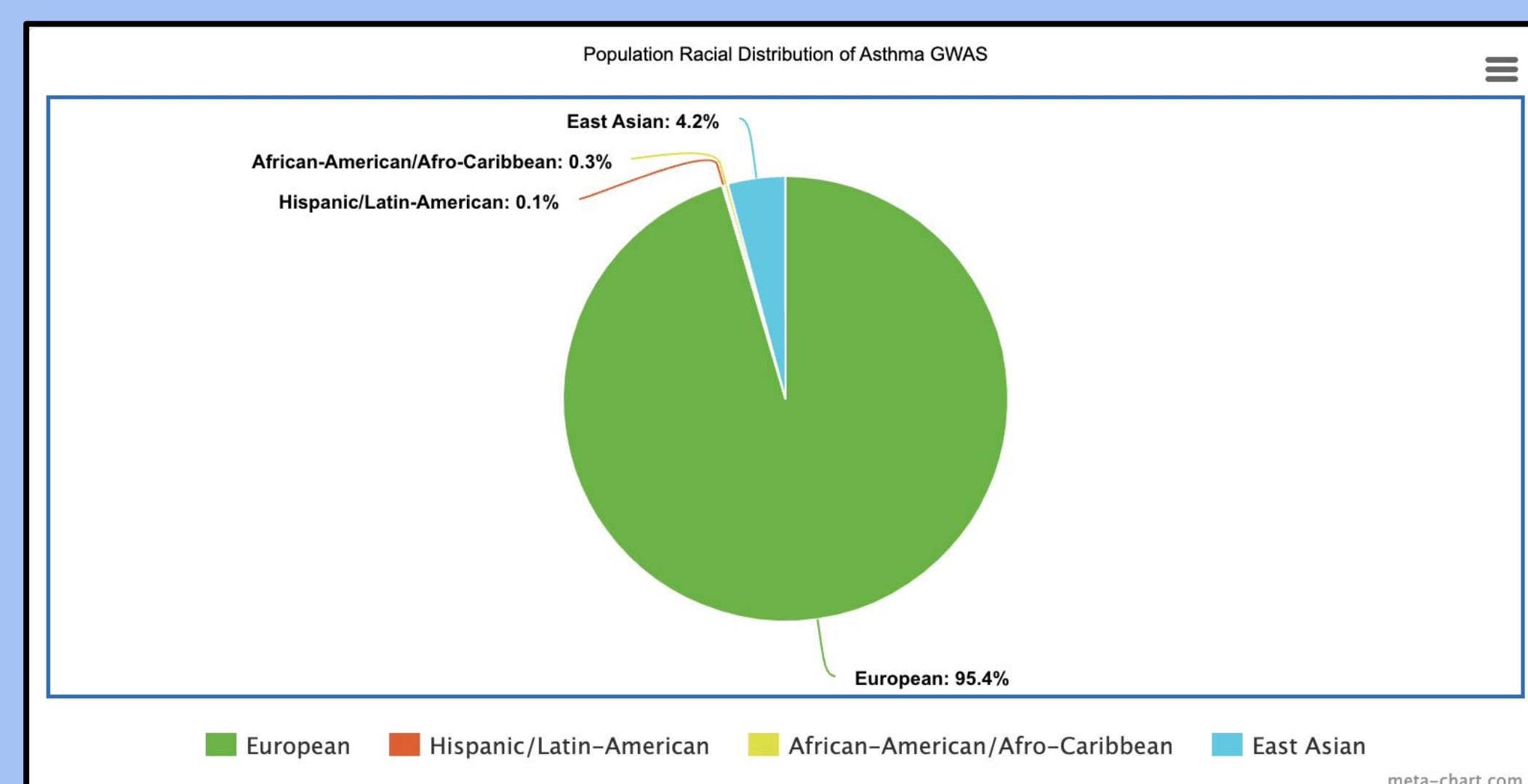
- This study utilized Correlational Research and Descriptive Research for its inquiry approaches.
- The first data set was collected from the [GWAS Catalog](#) and focused on the ethnic/racial makeups of various populations of GWAS studies.
- Secondly, the CDC's health statistics were examined to see the distribution of death rates from each condition based on race and the racial distribution of people with the condition (incidence of that condition).
- Through this research, it can be determined if there is a correlation between studies from mainly European populations and higher death rates from that particular condition for minority groups.

CONCLUSIONS AND ANALYSIS

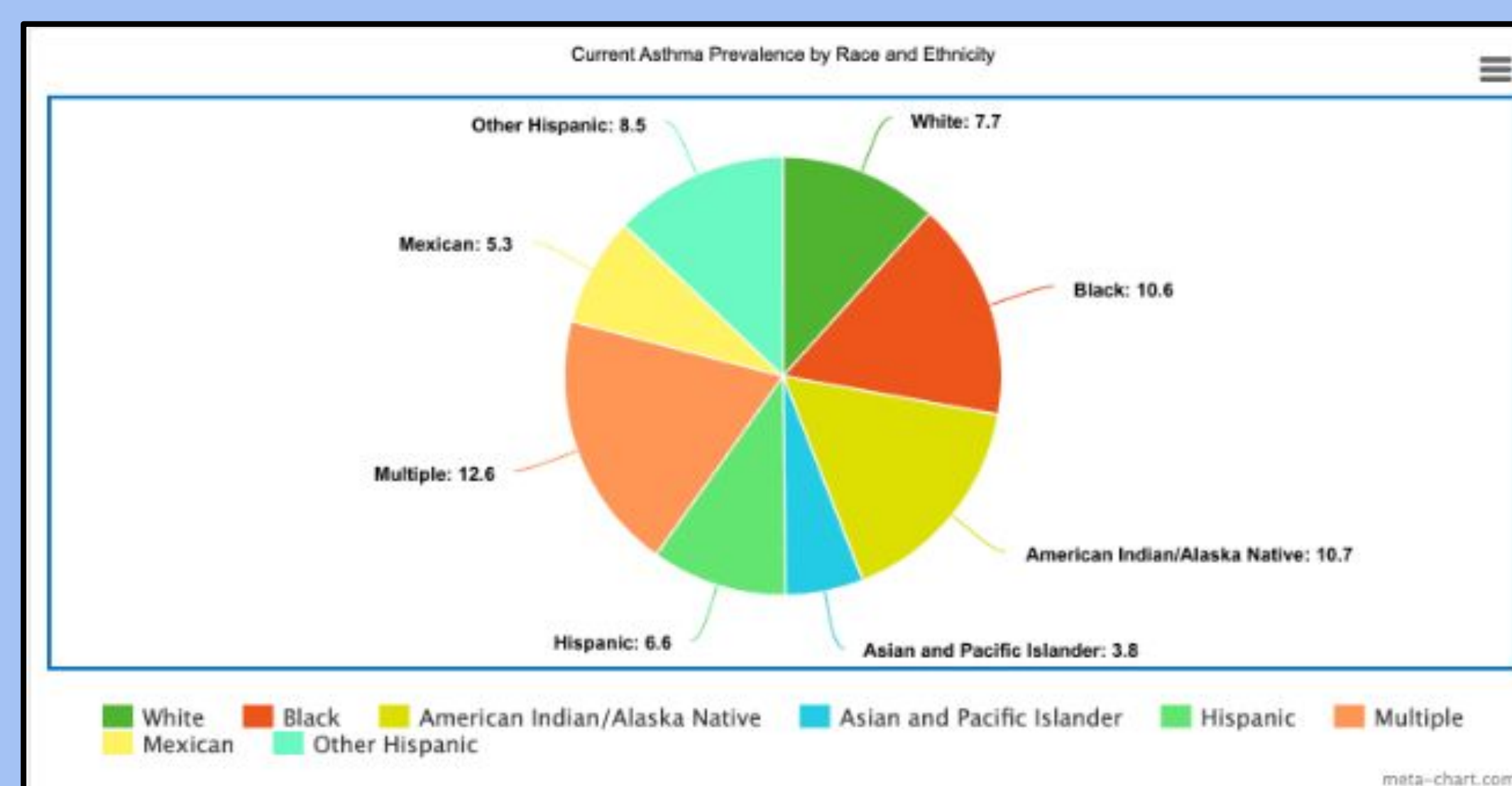
- These graphs show that there is a clear racial disparity in the populations studied in genome-wide association studies for breast carcinoma and asthma
- In the case of asthma, people of European ancestry make up over 95% of the GWAS populations, and there are less White people with asthma than there are Black people, American Indian/Alaska Natives, or Hispanic people.
 - There is a higher percentage of White people dying from asthma than from minority groups, but Black people are the next highest group, at 28.4%. A potential cause of this could be the fact that Black people only make up 0.3% of GWAS populations for asthma.
- People of European ancestry make up over 90% of the GWAS populations for breast carcinoma, and there are more White women with breast cancer than women from minority groups. However, when this is contrasted with the death rates for breast cancer, it can be seen that Black women are more likely to die from breast cancer than White women. This could be caused by Black women being underrepresented in GWAS populations.

DATA AND FINDINGS

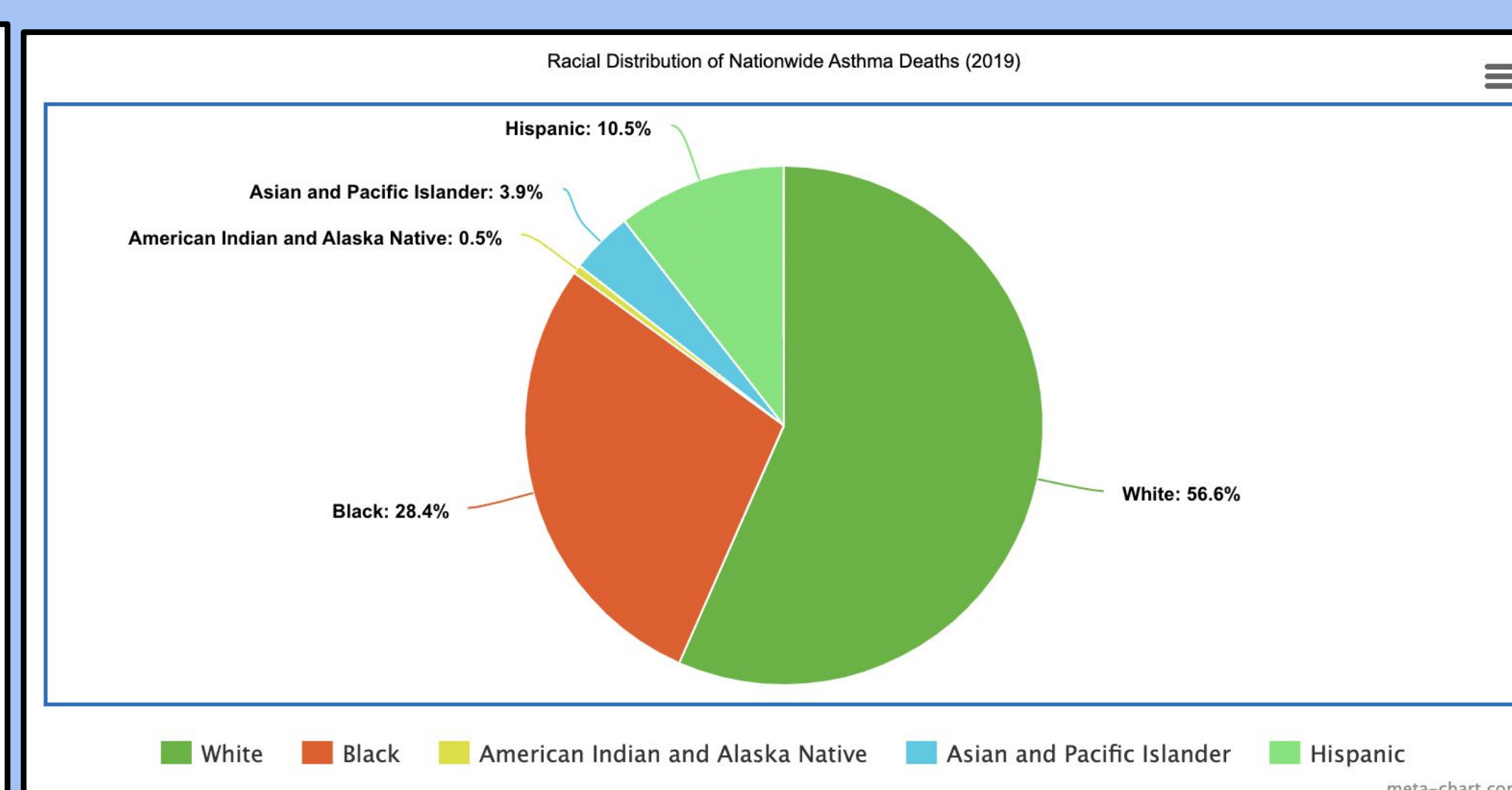
Asthma Data



This pie chart shows the racial distribution of all 85 genome-wide association study populations for asthma, a condition that is known to disproportionately affect minority populations. Hispanic/Latin American subjects made up 0.1% of this overall data. People of European ancestry make up over 95% of the GWAS populations.



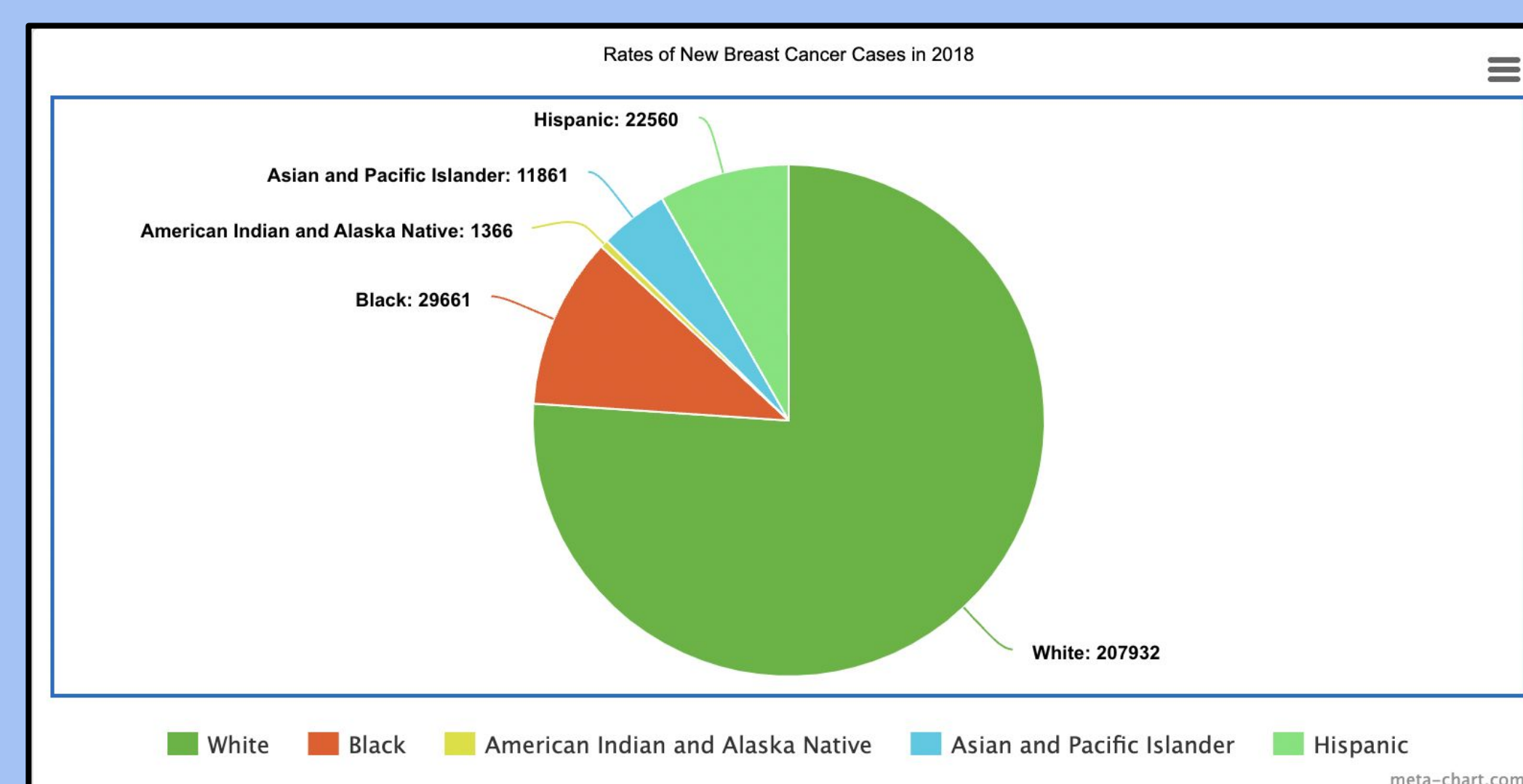
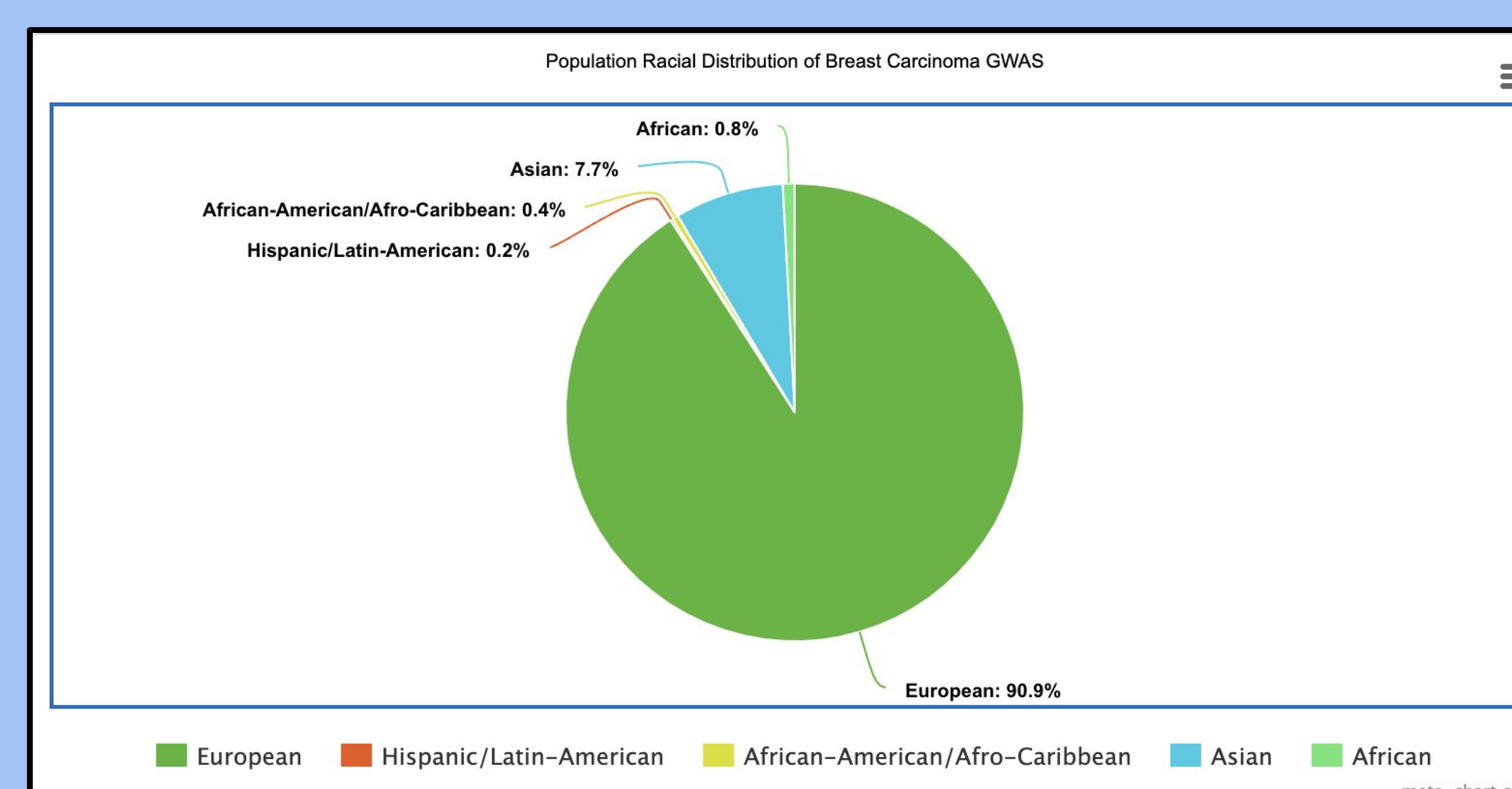
This pie chart displays the racial distribution of nationwide asthma data. As shown by the graph, there are less White people with asthma than there are Black people, American Indian/Alaska Natives, or Hispanic people.



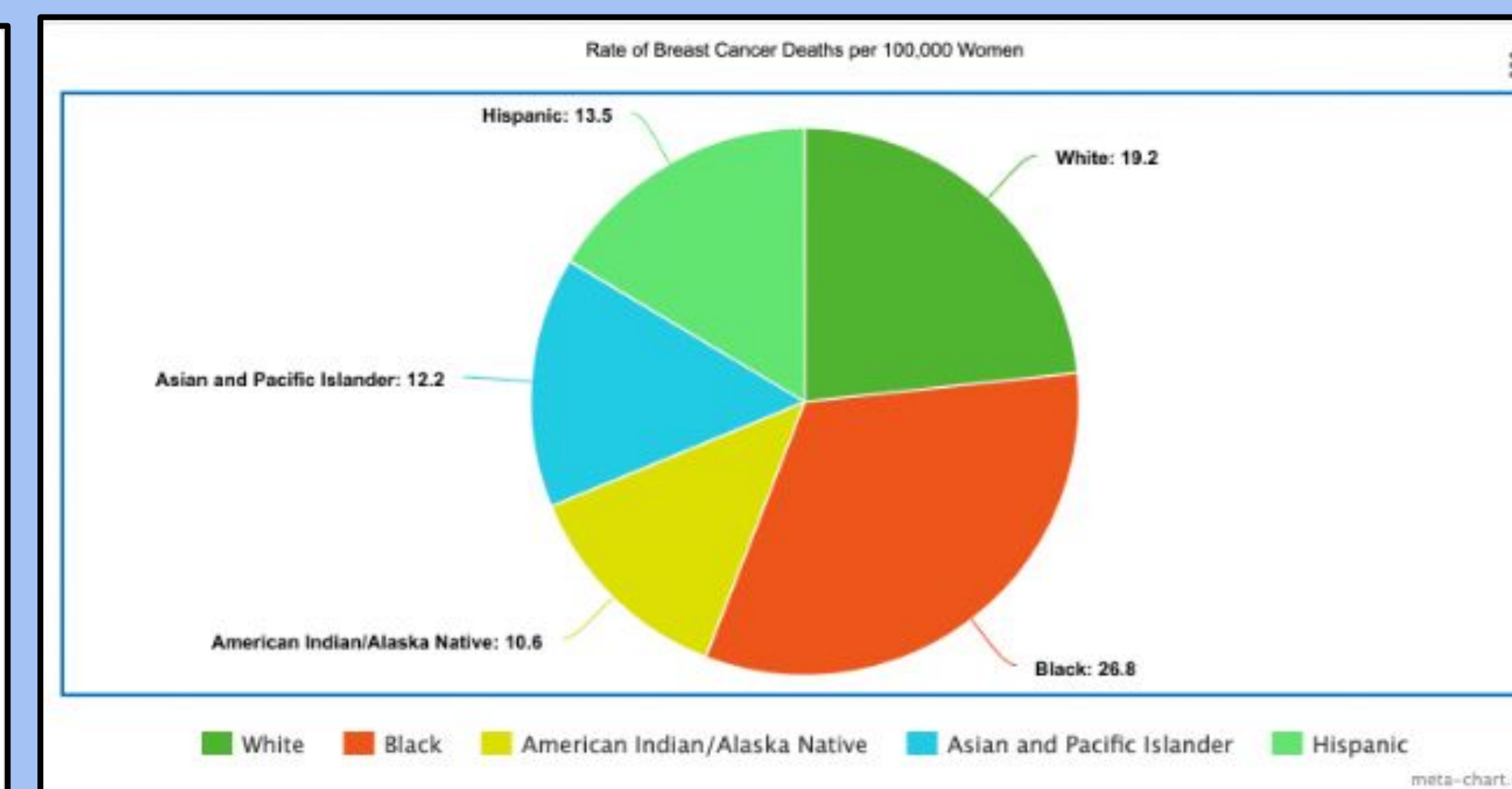
Conversely, this pie chart displays the racial distribution of the rate of asthma-related deaths in 2019.

Breast Carcinoma Data

This pie chart shows the racial distribution of all 117 genome-wide association study populations for breast carcinoma, a condition that is known to disproportionately affect Black women. People of European ancestry make up over 90% of the GWAS populations.



Conversely, this pie chart displays the racial distribution of nationwide breast cancer data. As shown by the graph, there are more White people with asthma than there are Black people, American Indian/Alaska Natives, or Hispanic people.



Conversely, this pie chart displays the racial distribution of the rate of breast cancer deaths for every 100,000 women from nationwide data. As shown by the graph, Black women are much more likely to die from breast carcinoma than White women, a disparity that is not reflected by the population distributions.

IMPLICATIONS AND NEXT STEPS

- These findings suggest that this disparity exists for more than just breast carcinoma and asthma.
- In most GWAS studies, people of European ancestry make up most of the study populations, and it's clear that this is causing inequalities in healthcare that is based off of these studies.
- Without a push for diversity in GWAS, these inequalities will continue, and people of minority groups will continue to be more likely to be at risk from these conditions due to not being accurately studied in research on these conditions.
- In the future, genetic scientists and researchers should prioritize diversity when conducting genome-wide association studies.
- Without diverse study populations, it's clear that minority groups do not receive as equal and effective treatments as White people do, resulting in healthcare disparities further down the line.
- Healthcare inequalities starts at the GWAS level, and this research shows that geneticists must conduct GWAS with people from all races and minority groups in order to create equal healthcare for all based on their studies.

ACKNOWLEDGEMENTS / REFERENCES

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Works Cited:

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