Representation of Racial Groups in Genomic Studies of Gastroenteropancreatic Neuroendocrine Neoplasms

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Introduction

1. Translational cancer genomics is maturing at an exponential rate, bringing invaluable findings from the bench to clinical practice. Integrated genomics will be key to discovering oncogenic pathways and therapeutic susceptibilities in gastroenteropancreatic neuroendocrine neoplasms (GEP-NENs), for which few effective therapies are available.

2. Not all populations are poised to benefit from advances in cancer genomics, as large-scale genomic analyses have consisted of mostly White/Caucasian patients.

3. The goal of this study was to evaluate ethnoracial populations represented in genomic, transcriptomic, and epigenomic studies of GEP-NENs.

Racial Representation in Cancer Genomics[1]

Methods

A systematic review of the literature was conducted with structured queries to PUBMED, EMBASE, and the Cochrane Library. Abstracts were reviewed independently by two individuals for the sequencing/genetic analysis of DNA, RNA, or DNA methylation.

Language Processing (NLP) using the python package NLTK was then used to determine the frequency of the NIH race category terms "African American," "Black" "Hispanic," "Latino," "Asian" "Native American," "Native Hawaiian," "Pacific Islander" "Caucasian," "White," as well as "Race," and "Ethnicity" in these manuscripts.

Tumor types and subject numbers by ethnoracial group were then manually evaluated. The number of subjects and the number of studies including subjects by race were evaluated with Fisher's Exact test.

Results

Figure 2. A) Number of the 313 total manuscripts meeting criterion that analyzed each GEP-NEN type. **B)** Number of specimens from each GEP-NEN type included in all 313 manuscripts that met inclusion criterion.
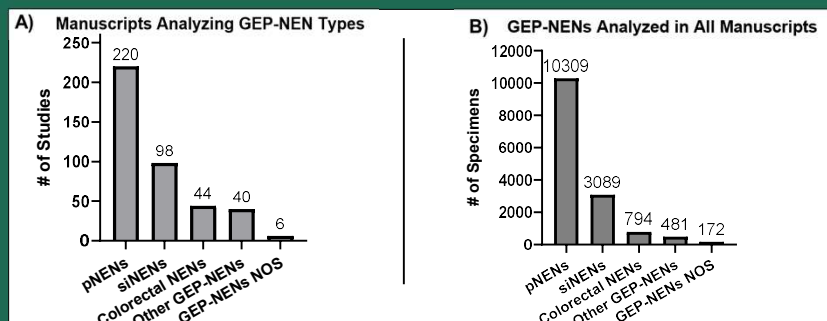

Figure 3. A) Proportion of the 313 total manuscripts that analyzed each type of biomolecule, by the inclusion of patient race information. *Of the 16 studies that included patient race information, 2 studies analyzed both DNA & RNA. **B)** Assays used to analyze biomolecules in the 16 studies that reported patient race.
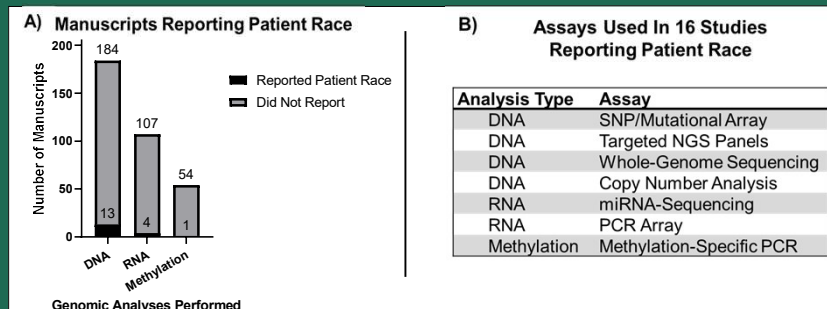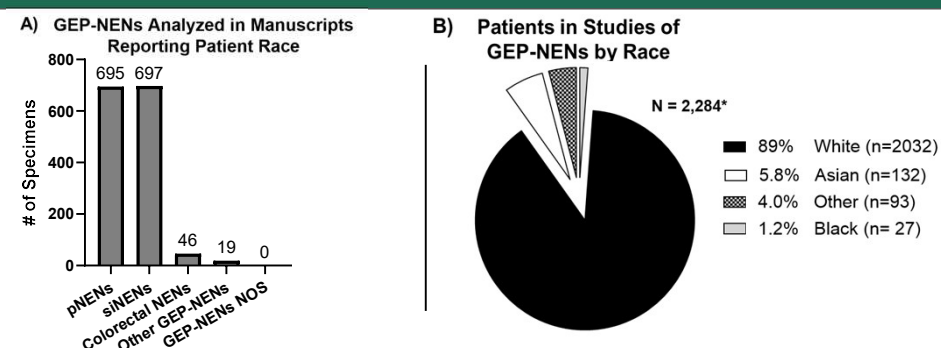

Figure 4. A) Number of each GEP-NEN type analyzed in the 16 manuscripts that reported patient race. **B)** Number of patients from each ethnoracial group that were included in (epi)genetic studies of GEP-NENs that reported patient race.

Methods

Figure 1. Studies meeting inclusion criterion for NLP analysis were screened independently by two investigators before screening by NLP and manual review to determine subject number by race.
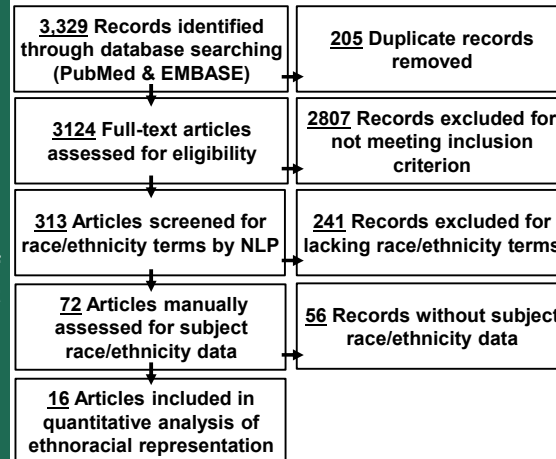
Conclusions

1. Few studies on the genomics of GEP-NENs include data on the race/ethnicity of their participants

2. There is little representation of racial minorities in genomic studies of GEP-NENs.

3. Data for these populations are integral for understanding GEP-NEN biology, generalizing findings, and improving therapy.

4. Diversity in genomic studies of GEP-NENs is imperative to uncovering the genomic or epigenomic variation in these tumors that may influence their disease, treatment, or prognosis

References:

1. Guerrero, S., López-Cortés, A., Indacochea, A. et al. Analysis of Racial/Ethnic Representation in Select Basic and Applied Cancer Research Studies. Sci Rep 8, 13978 (2018). https://doi.org/10.1038/s41598-018-32264-x

https://sites.uab.edu/neuroendocrinesurgerylab/