

Poster 23: Inclusion of Medically Underserved Populations in a Cohort with Developmental and Epileptic Encephalopathies



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Background

- Genetic Determinants of Neurological and Developmental Disorders (GDNDD) identifies genetic variants in developmental and epileptic encephalopathies (DEE)
- Individuals with epilepsy are more likely to represent underserved populations
 - Nationally, children with seizures are:
 - More likely to live in poverty
 - 36% public insurance (Medicaid, Medicare)
 - 16% African American and non-Hispanic
 - 23% Hispanic
- Due to limited diversity in genetic research, rare variants can have unclear significance in people from diverse ancestral backgrounds

Objective

- Enroll ≥ 60% of participants from medically underserved populations (historically underrepresented in research) for genomic sequencing in a DEE cohort
- UNC Child Neurology clinic treats >1,700 children/year
 - Primary referral for NC (75/100 counties Tier 1 or 2)
 - ~75% public insurance
 - ~20% African American
 - 15% Hispanic
 - 12% Non-English preferred language

Methods

- Single center prospective study
- Recruited individuals with DEE and negative genetic testing and their biological relatives (trios)
- Medical providers reported barriers to enrollment
- Used new and existing recruitment and enrollment techniques (Table 2)

Table 1. Definition of underserved

Diversity Category	Represented in Research	Underrepresented in Biomedical Research (definition)
Race and Ethnicity	Identify as White and non-Hispanic	Identify as other than White and non-Hispanic (i.e. Asian; Black, African, African American; Hispanic, Spanish, Latino; Native Hawaiian, Pacific Islander; Middle Eastern, North African)
Geography	Reside in urban metropolitan areas	Reside in rural and non-metropolitan areas
Household Income	>200% Federal Poverty Level	Annual Household Income ≤200% of the Federal Poverty Level
Access to care	Can obtain/pay for medical care	Cannot easily obtain or pay for medical care as needed

Adapted from The All of Us Guide for Diversity and Inclusion

Results

FIGURE 1. LANGUAGE

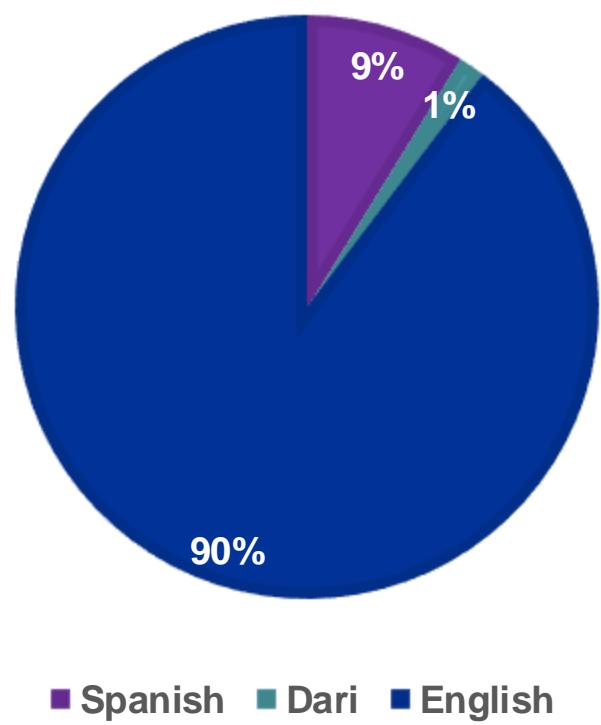


FIGURE 2. RURALITY*

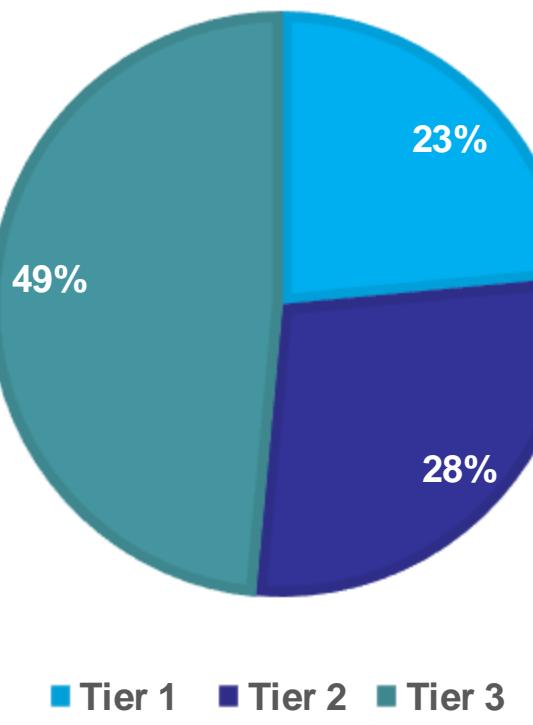


FIGURE 3. INSURANCE

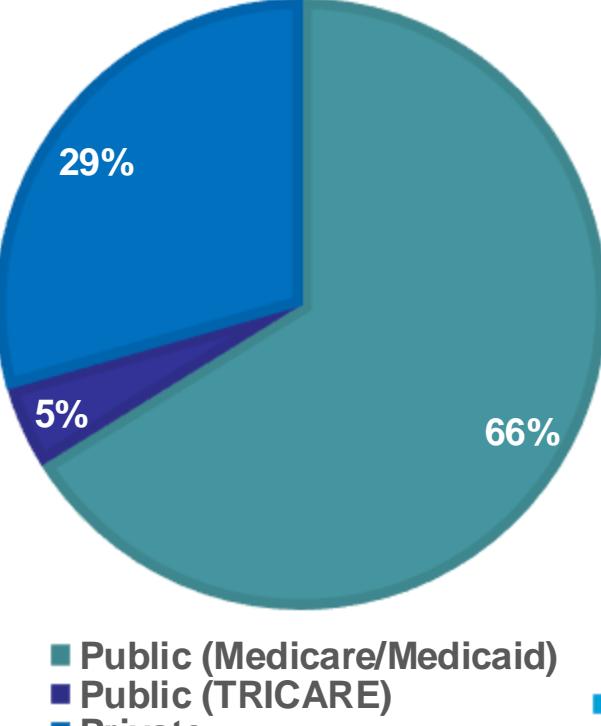


FIGURE 4. ETHNICITY

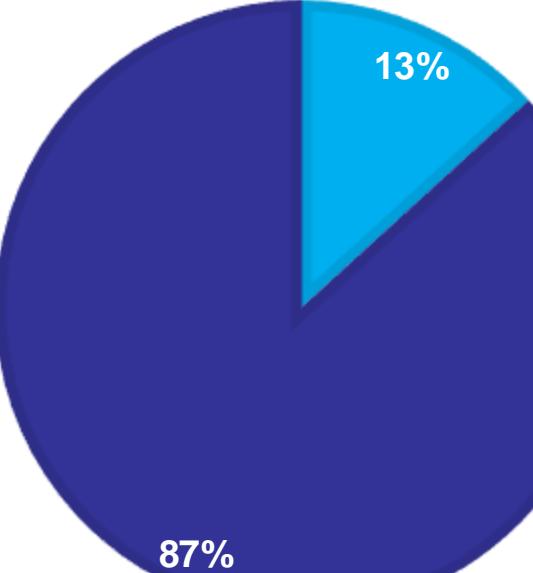
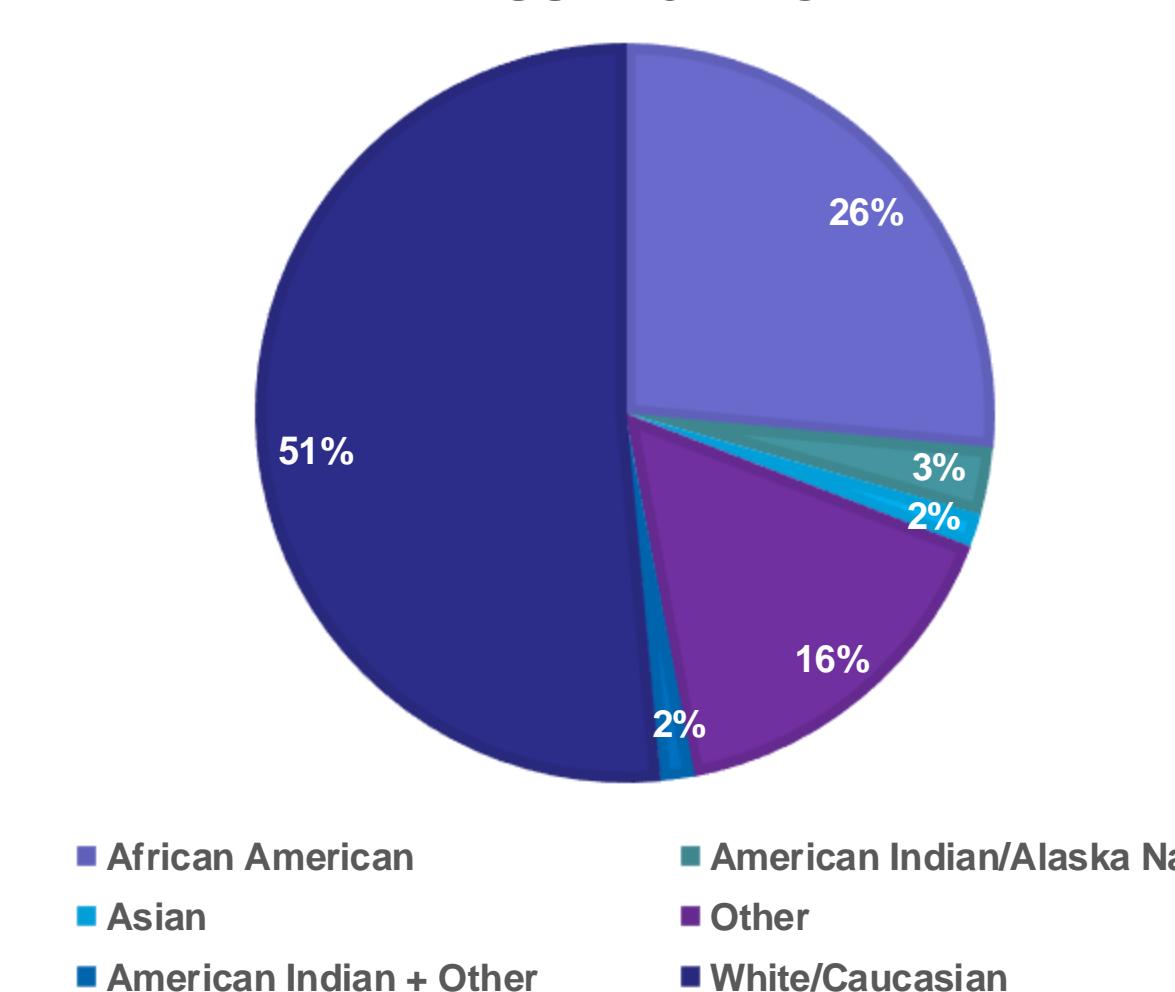


FIGURE 5. RACE



Results continued

- 68 probands with DEE enrolled (n = 158 total)
- 84% (n = 57) from underserved populations
 - 10% (n = 7) with non-English preferred language
 - 51% (n = 35) from tier 1 or 2 designated counties
 - 66% (n = 45) with public insurance
 - 50% (n = 34) identified as racial/ethnic minorities**
- Pathogenic variants detected in 2 participants enrolled using mitigation strategies
 - Likely pathogenic GRIN2A variant
 - Barrier: non-English language (Spanish), rurality
- Pathogenic somatic MTOR variant
 - Barrier: non-English language (Dari), rurality

Conclusions

- Individuals with epilepsy are:
 - More likely to represent underserved populations
 - Less likely to be included in genetic research
- Population of medically underserved individuals enrolled (>60%) is comparable to that of our clinic
- GDNDD demonstrates the feasibility of prioritizing ancestrally diversity and inclusion of medically underserved individuals in genetic research, per NHGRI Strategic Vision

References

- Mapes BM, Foster CS, Kusnoor SV, et al. Diversity and inclusion for the All of Us research program: A scoping review. *PLoS One*. 2020;15(7):e0234962
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