### **DISPARITIES IN GENETIC TESTING FOR NEUROLOGICAL DISORDERS**

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### **Background and Purpose**

The role of genetic testing in the diagnosis and management of neurologic conditions has increased dramatically in recent years. Genetic testing is now indicated for appropriately selected patients with many neurologic conditions including ALS, epilepsy, myopathy, neuropathy, dementias, and ataxias, among others.<sup>1</sup>

Healthcare disparities are unfortunately widespread in the US healthcare system, but disparities in the utilization of genetic testing for neurologic conditions have not been

### Likelihood of attending a neurogenetics visits (pathway step 1), restricted to top seven referring diagnoses

| Demographic             | OR (95% CI)        |   | P <sub>adj</sub> |
|-------------------------|--------------------|---|------------------|
| Race                    |                    |   |                  |
| White                   | ref.               | • | ref.             |
| Black                   | 0.67 (0.57 - 0.79) | • | 5 x 10⁻          |
| Asian                   | 1.2 (0.87 - 1.62)  |   | 0.64             |
| Some Other Race         | 1.21 (0.94 - 1.54) |   | 0.50             |
| Ethnicity               |                    |   |                  |
| Not Hispanic or Latino  | ref.               | • | ref.             |
| Hispanic or Latino      | 0.91 (0.65 - 1.25) | • | 0.98             |
| Socioeconomic status    |                    |   |                  |
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In neurology, non-White patients have less access to neurologic care than White patients and worse health outcomes across many neurologic conditions and subspecialties.5,6

### <u>Methods</u>

- The Penn Neurogenetics Group analyzed retrospective data from patients who underwent genetic evaluation and testing through our institution's neurogenetics program.
- We tested for differences between demographic groups at three steps of the genetic evaluation pathway: (1) attending a neurogenetic evaluation, (2) completing genetic testing, (3) receiving a diagnostic result.
- Our group compared patients on this genetic evaluation pathway to the population of all neurology outpatients at our institution to examine the hypothesis that access to



## Likelihood of completing genetic testing (pathway step 2), restricted to top seven referring diagnoses

| Demographic            | OR (95% CI)        |                                    | $\mathbf{p}_{adj}$ |
|------------------------|--------------------|------------------------------------|--------------------|
| Race                   |                    |                                    |                    |
| White                  | ref.               | $\bullet$                          | ref.               |
| Black                  | 1 (0.79 - 1.27)    |                                    | 1.00               |
| Asian                  | 0.93 (0.59 - 1.47) |                                    | 0.98               |
| Some Other Race        | 0.94 (0.65 - 1.36) |                                    | 0.98               |
| Ethnicity              |                    |                                    |                    |
| Not Hispanic or Latino | ref.               | $\bullet$                          | ref.               |
| Hispanic or Latino     | 0.99 (0.61 - 1.6)  | e                                  | 1.00               |
| Socioeconomic status   |                    |                                    |                    |
| Quartile 4 (highest)   | ref.               | $\bullet$                          | ref.               |
| Quartile 3             | 1.02 (0.84 - 1.26) |                                    | 0.98               |
| Quartile 2             | 1.03 (0.85 - 1.25) |                                    | 0.98               |
| Quartile 1 (lowest)    | 1 (0.8 - 1.25)     |                                    | 1.00               |
| Insurance              |                    |                                    |                    |
| Private                | ref.               | $\bullet$                          | ref.               |
| Medicaid               | 0.95 (0.67 - 1.34) |                                    | 0.98               |
| Medicare               | 0.96 (0.81 - 1.15) |                                    | 0.98               |
| Sex                    |                    |                                    |                    |
| Male                   | ref.               | $\bullet$                          | ref.               |
| Female                 | 0.97 (0.84 - 1.12) |                                    | 0.98               |
|                        | ſ                  | 0.50 1.0                           | 20                 |
|                        |                    | Under-represented Over-represented |                    |

and results of genetic testing vary according to race, ethnicity, sex, socioeconomic status, and insurance status for adults with neurologic conditions.



### **Results**

Between 2015-2022, a total of 128,440 patients were seen in our outpatient neurology clinics and 2,540 patients underwent genetic evaluation.

Black patients were less than half as likely as White patients to be evaluated and this disparity was similar after controlling for other demographic factors.

Patients from the least wealthy quartile of zip codes were less likely to be evaluated.

# Likelihood of a diagnostic result (pathway step 3), restricted to top seven referring diagnoses

| Demographic            | OR (95% CI)        |           | $\mathbf{p}_{adj}$ |
|------------------------|--------------------|-----------|--------------------|
| Race                   |                    |           |                    |
| White                  | ref.               | $\bullet$ | ref.               |
| Black                  | 0.87 (0.57 - 1.31) |           | 0.98               |
| Asian                  | 1.18 (0.54 - 2.39) |           | → 0.98             |
| Some Other Race        | 1.56 (0.9 - 2.62)  |           | → 0.37             |
| Ethnicity              |                    |           |                    |
| Not Hispanic or Latino | ref.               | $\bullet$ | ref.               |
| Hispanic or Latino     | 1.41 (0.66 - 2.82) | •         | → 0.81             |
| Socioeconomic status   |                    |           |                    |

Among patients who underwent evaluation, there were no disparities in the likelihood of completing genetic testing, nor in the likelihood of a diagnostic result after adjusting for age.

#### **Conclusion**

- We observed unequal utilization of our clinical neurogenetics program for patients from marginalized and minoritized demographic groups, especially Black patients.
- Among patients who do undergo evaluation, all groups benefit similarly from genetic testing when it is indicated. Understanding and removing barriers to accessing genetic testing will be essential to healthcare equity and optimal care for all patients with neurologic disorders.

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| Quartile 4 (highest) | ref.               |      | •                     |                | ref. |
|----------------------|--------------------|------|-----------------------|----------------|------|
| Quartile 3           | 0.95 (0.68 - 1.35) |      | •                     |                | 0.98 |
| Quartile 2           | 1.02 (0.74 - 1.42) |      |                       |                | 1.00 |
| Quartile 1 (lowest)  | 0.89 (0.61 - 1.31) |      | •                     |                | 0.98 |
| Insurance            |                    |      |                       |                |      |
| Private              | ref.               |      | •                     |                | ref. |
| Medicaid             | 0.66 (0.33 - 1.23) | <    | •                     |                | 0.64 |
| Medicare             | 0.68 (0.49 - 0.93) | <    | •                     |                | 0.07 |
| Sex                  |                    |      |                       |                |      |
| Male                 | ref.               |      | •                     |                | ref. |
| Female               | 0.97 (0.77 - 1.23) |      | • • •                 |                | 0.98 |
|                      |                    |      |                       |                |      |
|                      |                    | 0.50 | · 1.0                 | 2.0            |      |
|                      |                    |      | Under-represented Ove | er-represented |      |

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