Research Summary

Interrogating ICD Coded Electronic Health Records to Characterize the Epidemiology of Prosopagnosia (Face Blindness)

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Study reveals new insight about who is diagnosed with prosopagnosia.

We are delighted to share results from a recently published study on prosopagnosia. If you are a member of the prosopagnosia support community, you belong to a large community of individuals around the world who help researchers answer important health questions and discover new medical treatments. Without your research partnership, clinical research would not be possible.

Here, we share the summary of results of a study that was recently published in the *Journal of Clinical Translational Science*. We hope it helps you understand and feel proud of your key role in medical research and discovery.

What is this study about?

- People with prosopagnosia (face blindness) have great difficulty perceiving, recognizing, or memorizing faces, even those of very familiar people. Living with prosopagnosia can negatively impact the affected person's quality of life.

- Before conducting this study, little was known about the epidemiology of prosopagnosia. We didn't necessarily have good ways of tracking the characteristics of the population such as gender, age, race, and/or which other diagnoses or conditions people with prosopagnosia also have.

- We used information from a collection of more than 7 million de-identified electronic health records to study the characteristics of people with prosopagnosia, and find out how common the diagnosis is.

What people were included in this study?

- We studied people from the greater New York City area whose de-identified health records were included in the Clinical Data Research Network (Insight) dataset. The CDRN is a large electronic health records dataset which contains de-identified electronic heath record information from 8 large healthcare organizations in the New York City area. It includes health information and diagnoses for more than 7 million people.

- We developed a method to search the database using a combination of terms and diagnoses that allowed us to detect individuals who had been given the diagnosis of prosopagnosia, and extract information about their characteristics from the database. This is called the "case-cohort".

- We used the same search formula in a matched "case-control" group of people who were of similar age, race, gender as the people with prosopagnosia, but did not have prosopagnosia. Comparing how often other medical conditions occur in people with prosopagnosia compared to the control group allowed us to study which other illness and conditions people with prosopagnosia are more likely to have.

- For each case we detected, we searched the database for an additional 100 controls, individuals who would not carry the diagnosis code for prosopagnosia but would match each case in age, gender and hospital-site, where they had been seen.

This study included:
- **902 individuals who met the search definition of prosopagnosia (51% women/49% men and average age of 48 years old) (case-cohort group)**
- **100,973 individuals without prosopagnosia (case-control group)**
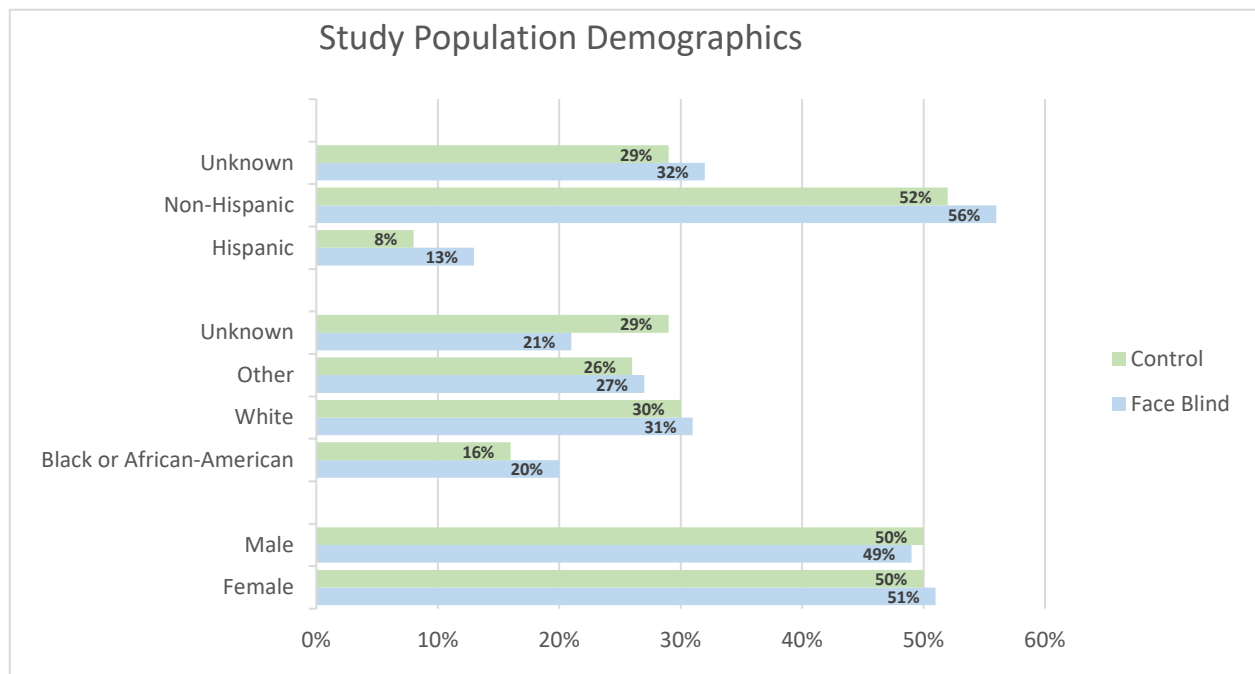

Figure 1. This figure shows the characteristics of the people with the diagnosis of prosopagnosia, and those in the control-cohort. The two groups are closely matched in age, race, ethnicity.

- Data extracted included information on age and gender, as well as race, ethnicity and the presence of diagnoses of a select number of comorbid conditions of interest, such as depression, anxiety, epilepsy, and stroke. Using this strategy we were able to reveal the condition's diagnostic rate in the New York area and learn more about what comorbid conditions people with prosopagnosia have.

What were the results of the study?

- We discovered that prosopagnosia does not get diagnosed as often as we thought. While, it has been reported that prosopagnosia may affect up to 1 in 33 individuals, we discovered that only 12 in 100,000 individuals had been diagnosed with the condition.

- We further discovered that individuals who carried the diagnosis code for prosopagnosia were more likely to also be diagnosed with conditions, such as depression, anxiety, personality disorder, and/or epilepsy.

- We also discovered that individuals of Hispanic ethnicity were more likely to be diagnosed with prosopagnosia compared to the other ethnic groups reported.

- Moreover, we found that the likelihood of carrying the diagnosis code for prosopagnosia increased with the number of comorbid condition diagnoses.

Study Population Selected Diagnoses

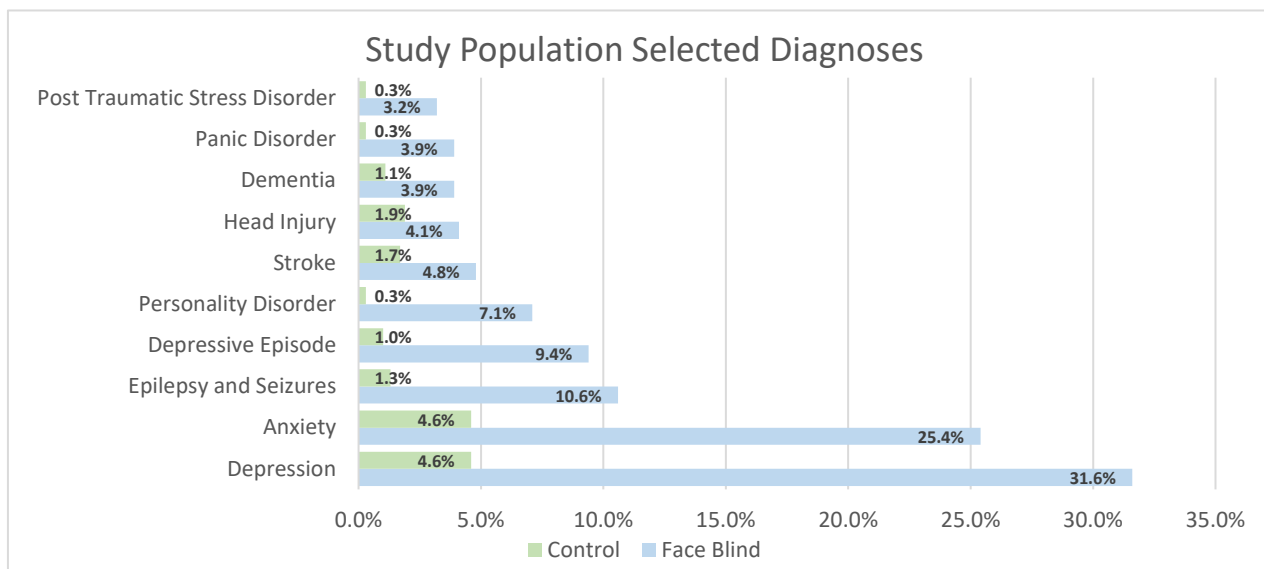| Condition	Control	Face Blind
Post Traumatic Stress Disorder	0.3%	3.2%
Panic Disorder	0.3%	3.9%
Dementia	1.1%	3.9%
Head Injury	1.9%	4.1%
Stroke	1.7%	4.8%
Personality Disorder	0.3%	7.1%
Depressive Episode	1.0%	9.4%
Epilepsy and Seizures	1.3%	10.6%
Anxiety	4.6%	25.4%
Depression	4.6%	31.6%

Figure 2. This figure shows a series of medical conditions (vertical axis) and how often these diagnoses were made (horizontal axis) for the people diagnosed with prosopagnosia (blue bar) and those in the control-cohort (green bar). Many of the medical conditions were much more common in the people with prosopagnosia than they were in the control-cohort.

How has this study helped patients and researchers?

- These results are important for prosopagnosics, their caregivers, and family members, as well as health care providers.

- Findings from this study are important as they will raise awareness on prosopagnosia and inform the medical community about the other conditions that are often associated with it.

- Results from this study will help guide further investigations of the relationships between prosopagnosia and other conditions, potentially leading to better care and diagnosis.

- The results of such a large study—while representative of the population overall—may not be true for every individual.

What were the limitations of this study?

For this study, we used electronic health records that were coded using the ICD-9 clinical diagnosis coding system. This medical coding system did not contain a specific diagnosis for prosopagnosia. This means that individuals with a related but distinct diagnosis called "simultanagnosia" may have been included in this study. This is a limitation of the medical records system.

Are there plans for further studies? Or what happens next?

A newer medical coding system, which provides a specific diagnosis code for prosopagnosia, is available and will improve the results of similar studies conducted in the future.

Who paid for this study?

This study was supported in part by grant # UL1 TR001866 from the National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH), and the Patient Centered Outcomes Research Institute (PCORI) PCORnet grant to the NYC-CDRN grant # CDRN-106-03961.

Where can I find more information about this study?

More information may also be available by looking up the official study title "**Interrogating an ICD Coded Electronic Health Records Database to Characterize the Epidemiology of Prosopagnosia**", or by going to *Journal of Clinical Translational Science.*

For more information about prosopagnosia visit:
- https://seek.rockefeller.edu/beyond-recognition/
- https://faceweb.org

For information about research studies at Rockefeller, go to www.RUCARES.org

For information about research studies on prosopagnosia at Rockefeller, go to
http://clinicalstudies.rucares.org/protocol.php?id=409&cat=53

This research was important. Thank you for helping us understand more about prosopagnosia. If you have questions, please contact us.

Thank you for being interested in this research. Without research participants there can be no clinical research.