Researchers call for closing gap in collecting racial and ethnic data in studies of rare genetic condition

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In a review of published research papers, investigators from the Johns Hopkins Medicine Hereditary Hemorrhagic Telangiectasia (HHT)

Center have identified a substantial lack of racial and ethnic data that may be negatively impacting the treatment and diagnosis of this rare disorder in diverse patients.

The study, a systematic review of 79 U.S.-based studies, was published June 13 in the *Orphanet Journal of Rare Diseases*. The findings highlight growing concerns about research study designs that exclude or hide information about underserved populations.

HHT is a rare inherited condition caused by mutations in five genes, and the condition was first described by physician William Osler at The Johns Hopkins Hospital in the late 1800s. HHT affects one in 5,000 individuals, and is characterized by blood vessel malformations within the nose and gastrointestinal systems. Severe cases of HHT also impact the nervous and pulmonary systems, as well as the liver. Patients with such cases suffer from hemorrhagic nosebleeds, strokes and possible brain abscesses.

Individuals with HHT often suffer from intense nosebleeds and may experience broken blood vessels near the surface of the skin. All of these symptoms tend to run in families, and such familial history often leads to genetic tests that can help diagnose the condition.

Early diagnosis of HHT—and early medical interventions that cut off or prevent malformed blood vessels from growing—helps prevent severe complications such as internal bleeding. Bevacizumab, which is also used to treat some cancers and macular degeneration, is one such effective intervention. The drug has been under intense investigation in the treatment of severe HHT in recent years.

"The study started with the intention of wanting to better explore the benefit of bevacizumab," says first author Panagis Galiatsatos, M.D., M.H.S., associate director of the HHT Clinic and assistant professor of

medicine in the Johns Hopkins University School of Medicine. The research team's work refocused, says Galiatsatos, once they saw gaps in race and ethnicity data. "We started to wonder if we were looking at indications of inequitable health care both in treating and diagnosing HHT."

Researchers next selected four studies published between 2014 and 2019 investigating the value of bevacizumab as an HHT treatment and looked at the study participants' social and demographic information. In each study, they discovered that race or ethnicity data was missing, creating a potentially serious gap in the ability to understand bevacizumab's effectiveness as an HHT treatment in different ethnicities.

Such racial disparities have been established in other studies for treatments related to conditions such as high blood pressure and diabetes.

The researchers say the lack of racial and ethnic data is a "red flag" in HHT research, because such a variable may critically affect the effectiveness of treatments and the decision-making process patients engage when undergoing treatment for a chronic illness.

"I've had African American patients who will ask if this treatment will really work for them or if it is safe for someone of their race," says Galiatsatos, who says that without study data accounting for race or ethnicity, clinicians can struggle in providing answers to their patients.

Investigators say exclusions of racial and ethnic demographical information in studies could be the result of implicit bias, or unconscious thoughts and actions one may have that cause them to unknowingly confirm racial and ethnic stereotypes. The researchers worry that if people of color are underrepresented in HHT studies such as the ones analyzed, HHT cases may be underreported, too, creating moral and

ethical concerns in HHT care.

If that is the case knowledge related to the diagnosis and treatment of the condition may be skewed and exclusionary of diverse patients. Galiatsatos and the research team emphasize that these factors could contribute to a delay in diagnosis and a lack of clinical confidence and understanding in using drugs like bevacizumab to treat HHT.

The researchers suggest that future research related to HHT treatments must include a broad spectrum of patients across multiple racial and ethnic background to ensure fair and effective care, as well as the elimination of potential bias in researching, treating and diagnosing HHT.

"HHT affects people of all races and ethnicities," says Gina Robinson, M.S.N., R.N., program coordinator for the Johns Hopkins HHT Center. "The most important takeaway we found is that we have to do a better job with community engagement and providing inclusive care to all HHT patients."

More information: Panagis Galiatsatos et al, A lack of race and ethnicity data in the treatment of hereditary hemorrhagic telangiectasia: a systematic review of intravenous bevacizumab efficacy, *Orphanet Journal of Rare Diseases* (2022). DOI: 10.1186/s13023-022-02371-0

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