Myths and Misdiagnoses of Proteus Syndrome

Leslie G Biesecker
National Human Genome Research Institute, Bethesda, MD, USA

In a recent correspondence in the journal titled “Proteus syndrome: unveiling the anesthetic myths,” De et al. described anesthetic considerations for a patient who they claimed was affected with Proteus syndrome. Unfortunately, one of the myths of Proteus syndrome is that all patients claimed to have this disorder indeed have Proteus syndrome. The patient they describe is very unlikely to have Proteus syndrome. The patient they describe is very unlikely to have Proteus syndrome based on the character of his overgrowth and the nature of his vascular anomalies. As we have demonstrated in a large review, only a minority of patients claimed to have this disorder actually have it, according to clinical criteria. Since that review was published, the molecular basis of Proteus syndrome has been delineated, so there should no longer be ambiguity regarding this diagnosis. Molecular testing should be mandatory for any patient claimed to have a genetic disorder of known molecular etiology if they are to be published in the scientific literature. While molecular testing can be costly and is not universally available, editors and peer-reviewers should insist on it as the literature will otherwise be populated by reports of patients with incorrect diagnoses, leading to incorrect conclusions and management by the readers of these articles, as is the case here.

This patient has an uncharacterized overgrowth syndrome and there are no data to support the conclusion that it is Proteus syndrome. Thus, the conclusions of the publication are fundamentally incorrect.

Acknowledgements

The author is supported by the intramural research program of the U.S. National Human Genome Research Institute, but the opinions expressed here are his own.

References


Received: 15 November 2017; Accepted: 13 February 2018.
Corresponding Author: Leslie G Biesecker, MD, National Human Genome Research Institute, 50 South Dr., Room 5140, Bethesda, MD 20894, USA. Tel: +1-301-402-2041, E-mail: lesb@mail.nih.gov