LETTER TO THE EDITOR

In Reply to: Alawi F. "Using rare diseases as teaching models to increase awareness"

To the Editor:

Two important publications have recently highlighted the study of rare diseases and the collaboration of studying these conditions.[1,2] A rare disease affects fewer than 200,000 people in the United States, less than 250,000 people in the European Union, and less than 50,000 patients in Japan.[1] The concept of a "rare disease," according to the World Health Organization and adopted by the Ministry of Health of Brazil, refers to a disorder that affects up to 65 people in every 100,000 individuals or 1.3 for every 2000 people (http://portalms.saude.gov.br/saude-de-a-z/doencas-raras).

As highlighted by Professor Alawi, the concept of and approach to rare diseases mandate interactions with various health care professionals.[1,2] In Brazil, within the last decade, great strides have been made to emphasize the importance of postgraduate interdisciplinary cooperation, which is a joint action by several professionals for the benefit of society (http://www.capes.gov.br/avaliacao/dados-do-snpg/cursos-recomendados-reconhecidos).

It also highlights the need for collaborations in studies in spite of difficulties in obtaining representative samples from a quantitative point of view.[2] We have conducted some investigations involving rare and unusual diseases and have experienced these aforementioned difficulties and have sought international collaborations, whenever possible.[3-5] In 2012, we investigated 25 patients from 16 families living in different countries, who manifested unexplained nephrocalcinosis and characteristic dental defects (amelogenesis imperfecta, gingival hyperplasia, impaired tooth eruption). This was the first study to identify the putative mutation in the *FAM20 A* gene associated with enamel renal syndrome,[5] illustrating the importance of multiprofessional interaction. Recently, Brooks published a review on syndromes associated with blue sclera, including malformations of the head and neck.[6] These are some of the many rare and unusual diseases that must be included in the undergraduate and postgraduate curricula.

A few years ago, I heard an important medical geneticist in Brazil saying: "Physicians more than ever need to open their patients' mouths to better understand mucosal alterations and particularly dental alterations." I believe that this task also needs to be performed from the opposite point of reference. Numerous publications in *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology* on rare and uncommon diseases have been instrumental in disseminating knowledge about these diseases and to enable collaborations that promote better understanding of those diseases.

In Brazil, 164 postgraduate courses (MSc and PhD degrees) are offered; 19 of these offer oral pathology and oral medicine as areas of concentration, and only 2 are specific to oral medicine and oral pathology (School of Dentistry, Paulista State University of São José dos Campos, UNESP and School of Dentistry, State University of Campinas, FOP, UNICAMP) (http://www.capes.gov.br/avaliacao/dados-do-snpg/cursos-recomendados-reconhecidos).

Although the fields of oral medicine and oral pathology have been successful in international collaborations in the fields of technical and scientific production,[7,8] only a limited number of groups are dedicated to the study of rare diseases with orofacial involvement. Articles, such as the ones mentioned at the beginning of this letter,[1,2] help not only to identify rare diseases but also to provide enhanced insights into their diagnosis and etiopathologic correlations, with the ultimate goal of achieving improved clinical outcomes.

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