Editorial Comment: Editor’s Foreword to a Special Issue “Elements of Morphology: Standard Terminology”

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This Special Issue of American Journal of Medical Genetics comprises an Introduction and six articles on “The Elements of Morphology”. I would suggest to our readership that these pieces represent a timely and substantial contribution to the field of medical genetics. These six articles provide recommendations for the description and definition of human phenotypic variations in the same way that the International Standing Committee on Human Cytogenetic Nomenclature accomplished this for human cytogenetics [ISCN, 2005] and the Nomenclature Working Group proposed the description of human sequence variations [den Dunnen and Antonarakis, 2001].

Taking up somewhat where an international working group on malformations left off in the 1980s [Spranger et al., 1982], the working group that authored the articles in this issue set out 4 years ago to develop consensus definitions and nomenclature for variations of phenotype. These articles are the direct result of that work. The group consisted of over 30 clinical geneticists from the United States, Canada, Europe, and Australia. The process of developing these consensus definitions was time consuming and stringent: recruitment of committee chairs, formation of six working committees, drafting of assigned definitions, attendance at two meetings, hundreds of emails, perusal of over 400 figures, 22 months of revisions, and five web-based conference calls.

As Editor of the Journal, I would like to point out that the articles published herein are the final product of a unique peer-review process: All of the participants provided input for the wording of the definitions for each section. The committee chairs reviewed and critiqued the proposals of the other reports. The almost-final versions represented the consensus recommendations of this ongoing process of reviewing and editing. As a last step, four of the six committee chairs edited and reviewed the final manuscripts of each region (Head/Face, Periorbital, Ear, Nose/Philtrum, Lips/Mouth, and Hands/Feet) for internal consistency and style.

The American Journal of Medical Genetics is pleased to publish these articles, which will be indefinitely available on the Journal’s web page at http://www.wileyinterscience.com/journal/ajmg.

REFERENCES


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