Letter to the Editor

What’s in a Name? The 22q11.2 Deletion

To the Editor:

We read with great interest the publication by Wulfsberg et al., “What’s in a Name? Chromosome 22q Abnormalities and the DiGeorge, Velocardiofacial, and Conotruncal Anomalies Face syndromes.” We appreciated their analogous comparison of the evolution of the 22q11.2 deletion to the anonymous Indian fable depicting a group of blind men trying to describe an elephant by each examining a separate part. “Originally described as three distinct phenotypes... it is now clear that there is only a single broad and variable phenotype associated with deletion of the DGCR...”

This concept was discussed in 1993 by Wilson et al., “We think that these conditions are all part of one clinical spectrum and that the diagnostic label depends upon the age of presentation and the predominant clinical manifestation.” Burn et al., in the same journal, wrote, “It seems increasingly likely that these eponymous syndromes will be phenotypic variations on a theme.”

Having thought along the same lines, we would like to submit “our elephant” (Fig. 1), which was published in our 1996 summer newsletter, as an accompaniment to Dr. Wulfberg’s article. We hope that this helps to illustrate the point.

REFERENCES


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Received 27 November 1996; Accepted 15 January 1997

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