American Journal of Medical Genetics 72:247 (1997)

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.

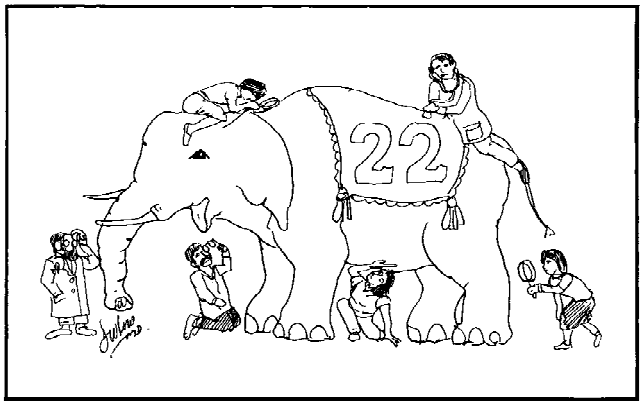

Fig. 1. The 22q.11.2 story can be likened to the old adage of a group of blind men trying to identify an elephant by each examining a separate part. Each man was accurate in describing his own area of interest, but none was able to see the big picture. Several conditions once thought to be separate are now known to be due to the 22q11.2 deletion.

REFERENCES

Burn J, Takao A, Wilson D, Cross I, Momma K, Wadey R, Scambler P, Goodship J (1993): Conotruncal anomaly face syndrome is associated with a deletion within 22q11. J Med Genet 30:822–824.

McDonald-McGinn DM, Zackai EH (1996): The history of the 22q11.2 deletion. "22q and You," a newsletter for families and professionals. Summer 1–11.

Wilson DI, Burn J, Scambler P, Goodship J (1993): DiGeorge syndrome: Part of Catch 22. J Med Genet 30:852–856.

Wulfsberg EA, Leana-Cox J, Neri G (1996): What's in a name? Chromosome 22q Abnormalities and the DiGeorge, Velocardiofacial, and Conotruncal Anomalies Face syndromes. Am J Med Genet 65:317–319.

Donna M. McDonald-McGinn*
Elaine H. Zackai
Division of Human Genetics and Molecular Biology
David Low
Division of Plastic and Reconstructive Surgery
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania
Department of Pediatrics
University of Pennsylvania School of Medicine
Philadelphia, Pennsylvania

*Correspondence to: Donna M. McDonald-McGinn, M.S., Associate Director, Clinical Genetics Center, The Children's Hospital of Philadelphia, 34th and Civic Center Boulevard, Philadelphia, PA 19104.

Received 27 November 1996; Accepted 15 January 1997