ture, it is not possible to approach a statistical estimate with certainty. It is interesting to note from their table that they ultimately found 147 patients with Asperger syndrome in the existing reports, with 13 (9%) positive tests. It is notable that this is still in the reported range of positive studies from individual reports.

One point that we would raise as different from their interpretation is in the dismissal of several positive tests as "unlikely" to be etiologically causative or "comorbid." We suggest that the identification of the six chromosome anomalies should be considered as possibly/probably related. In particular, the association of 22q11 deletions and autisms is well-enough documented that in our opinion, it should not be dismissed.

Another point of note is that all existing studies share some sort of selection bias, by nature of the clinical source of patients ascertained. Such bias has often been cited as leading to an overestimate of the diagnostic yield. Still, recent studies that have not found Fragile X in their patients have suggested that preselection (either intentional or not) may remove patients with Fragile X and lead to an underestimate.[1,2]

Finally, the foundation for what are made as recommendations based on an existing (albeit incomplete) body of literature comes down to the proverbial "lumper" versus "splitter" bias of the genetics provider making the recommendations. In the latter's mind, one should not make a recommendation for genetic testing in Asperger syndrome until there are specific studies that have addressed that particular issue. Alternatively, a synthesis of the literature coupled with an understanding of what a spectrum means could lead one to recommend studies for all those who fall into the spectrum until there is evidence to the contrary. With the goal of providing a unifying diagnosis for as many patients as possible, we fall into this category.

G. Bradley Schaefer, MD
University of Arkansas for Medical Sciences
Arkansas Children's Hospital
Little Rock, Arkansas

Nancy J. Mendelsohn, MD
Division of Medical Genetics
Children's Hospitals & Clinics of Minnesota
Minneapolis, Minnesota

Response to letter by Chodirker and Chudley

To the Editor:

We have read the letter to the editor by Drs. Chodirker & Chudley entitled "Routine Genetic Testing for Asperger Syndrome" with great interest. We thank them for their thoughtful comments and recommendations given toward a complex issue. We also appreciate the opportunity to respond to their letter.

Our initial response is, in general, agreement with the basic premise put forth. That is, there is a paucity of published studies that have specifically looked at a diagnostic yield when Asperger syndrome is selected out from the rest of the Autism Spectrum Disorders.

Given the absence of such reports, Drs. Chodirker and Chudley reviewed the literature in search of documentation of genetic testing abnormalities and persons with Asperger syndrome. What they found was a handful of cases of patients with Asperger syndrome and abnormal genetic tests. Given the small number of cases that could be extracted from the litera-

References

1. Abdul-Rahman OA, Hudgins L. The diagnostic utility of a genetics evaluation in children with pervasive developmental disorders. *Genet Med* 2006;8:50–54.
2. Herman G. Genetic evaluation of the child with isolated autism. Proceedings of the Annual Meeting of the American College of Medical Genetics 2008:Abstract #313.