2 May 2021

To the Editor, Washington Post:

As an international group of clinicians and scientists devoted to the study of 22q11.2 deletion syndrome, we would like to correct several factual errors and provide important clarifications to the story published 1 May 2021 on-line in the Washington Post, under the headline, “Doctors said the boy was suffering from teenage psychosis. What he really had was a rare genetic condition.”

The genetic condition, 22q11.2 deletion syndrome (previously known by several names, including DiGeorge syndrome and velocardiofacial syndrome), is associated with elevated risk for developing several treatable psychiatric illnesses, including schizophrenia. Indeed, about one in every four individuals born with a deletion (piece missing) on the long arm of chromosome 22 develops this well-recognized psychiatric illness.

Making the correct diagnosis is important in order to receive the most effective treatment. This is the same whether the associated condition is congenital heart disease - still called a congenital heart defect even if associated with 22q11.2 deletion syndrome - or is a psychiatric illness like schizophrenia. The signs and symptoms and course of illness have been shown over and over again to be exactly the same as in anyone else with schizophrenia. To say otherwise is simply wrong - and may be part of the longstanding fears, stigma, and discrimination associated with psychiatric illness.

There is much scientific evidence and clinical experience to support the fact that individuals with a 22q11.2 deletion who develop a psychiatric illness, in general respond well to standard medications and management, as for anyone in the general population.

For schizophrenia, the mainstay of successful management are the antipsychotic medications that can in fact be life-saving for individuals with this serious illness. As for all treatments, there are side effects to be aware of, and specialist expertise is often optimal for assessing the balance of any adverse effects with the effectiveness of these standard medications. This is particularly the case in a complex, multi-system condition like 22q11.2 deletion syndrome.

The experimental compound, metyrosine, is not a recommended treatment for schizophrenia or related psychiatric illnesses, whether associated with a 22q11.2 deletion or not. A clinical trial of metyrosine registered just one patient and was terminated.

To state that the psychiatric illnesses associated with 22q11.2 deletion syndrome are not the same as for others in the general population, implying that we do not know what they are or how best to manage them, and to further suggest that standard treatments are ineffective, could be actively harmful to many people - including those who do not have this genetic condition.

The journalistic standard of this article was disappointing to see in a venerable media source like the Washington Post. We would respectfully request an Editor's Note to address these issues.

Anne S. Bassett, MD, FRCPC
Professor, University of Toronto, and Director, Dalglish Family 22q Clinic, University Health Network, Toronto, Canada

Donna M. McDonald-McGinn, MS, LCGC
Clinical Professor of Pediatrics, Perelman School of Medicine, of the University of Pennsylvania, and Director, 22q and You Center, Children's Hospital of Philadelphia, Philadelphia, USA

Bernice Morrow, PhD
Professor, Albert Einstein College of Medicine, New York, USA

Peter Scambler, PhD
Professor, University College London and Great Ormond Street Institute of Child Health, London, UK

Ann Swillen, PhD
Professor, Department of Human Genetics, KU Leuven, Leuven, Belgium

Trustees, on behalf of the 22q11.2 Society