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PATIENT-DRIVEN EFFORT LEADS TO FIRST-EVER PUBLISHED CONSENSUS STATEMENT TO DIAGNOSIS AND TREAT RARE DISEASE DADA2

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The DADA2 Foundation led group of three patients & 35 expert researchers, clinicians from 18 countries to answer the question, "What is the optimal approach to diagnose and treat DADA2?"

Efforts give hope to today's 600+ diagnosed patients and to those yet to be diagnosed

Nashville, Tenn. (May 31, 2023) – The DADA2 Foundation announced today the publication of a Consensus Statement to help diagnosis and treat the rare disease, Deficiency of Adenosine Deaminase 2, also called DADA2. Entitled, "Evaluation and Management of Deficiency of Adenosine Deaminase 2 – An International Consensus Statement", the article can be found for free in the *Journal of American Medical Association (JAMA)* Network Open Access.

The Consensus Statement was developed as a result of the DADA2 Foundation's efforts to foster collaboration among 35 expert clinicians and researchers from around the world. The DADA2 Consensus Committee developed research questions based on data collected from prior International Conferences on DADA2, organized by the DADA2 Foundation in 2016, 2018, and 2020.

The resulting Consensus Statement is the first-ever in medical literature to describe the approach to the diagnosis and management of DADA2. The DADA2 Foundation made sure it was open access to help clinicians learn about, better diagnose, and treat DADA2. The paper is also a springboard for new lines of future scientific discovery. For patients, the guidance is critical to securing diagnosis and starting treatment before such devastating complications, such as a stroke, occur.

"The Consensus Statement is a game-changer for clinicians and families who often times scratch their heads for years to identify DADA2," says Chip Chambers, MD, founder and president of the DADA2 Foundation and father to two children with DADA2. "It comes just in time for doctors around the world to pause and consider if their patient who has suffered with mysterious

symptoms could possibly carry this genetic disease and in some patients be treated swiftly with a repurposed drug already on the market until a cure is developed."

The published Consensus Statement covers three main areas:

- **Screening:** DADA2 is among the most common autoinflammatory diseases in the world. This adds to a 2021 prevalence study which showed that 1 in 236 individuals can carry a mutation in the ADA2 gene, which would result in potentially 35,000 undiagnosed patients throughout the world (published by the DADA2 Foundation, the Broad Institute at MIT, and Boston Children's Hospital).
- **Diagnostic Testing:** The diagnosis can be established by genetic studies and/or an ADA2 blood test.
- **Disease Management:** The inflammatory features of DADA2 are treatable using available medications that are widely used to treat arthritis and inflammatory bowel disease. More broadly, the statement provides consensus from international experts to help patients secure medication approval. Other patients with bone marrow failure may require a bone marrow transplant or patients with immunodeficienty may require gamma globulin replacement.

About DADA2

DADA2 is an inherited rare genetic disease that leads to very low ADA2 enzyme activity in the body. It is often a difficult disease to diagnose because symptoms come in a wide array, including widespread inflammation and non-descript recurring fever, rash, pain, and fatigue with or without immunodeficiency. When not treated and in more severe cases, patients can have strokes or, in certain instances, may need a bone marrow transplant. Eight percent of DADA2 patients pass away due to their disease or complications. Genetic testing and/or ADA2 enzyme testing, which is only available at two sites in the U.S., are used to diagnosis DADA2.

DADA2 was discovered in 2014 by Dan Kastner, M.D., Ph.D., when he served as Scientific Director of the National Human Genomic Institute (NHGRI) at the National Institutes of Health (NIH), as well as a group of clinicians and researchers in Jerusalem, Israel. It is one of 9,000 rare diseases in the world, only five percent of which have a treatment.

About the DADA2 Foundation

The diseases' first international conference occurred in 2016 in Bethesda, Maryland, near the NIH and was hosted by the then newly created DADA2 Foundation – the only organization in the world dedicated to global collaboration around the disease. Started by Dr. and Mrs. Chip Chambers, whose two children were diagnosed with the disease following more than 15 years of a diagnostic odyssey, the Foundation has hosted two additional conferences in 2018 and virtually in 2020, which brought 500 clinical and research registrants from 30+ countries with 300 attending on any one of the three days. Convening patients and families, clinicians and researchers, biotech and pharma, the Foundation is currently planning its 4th International Conference on DADA2 on October 6th & 7th, 2023, in Washington, DC. The Foundation is also

launching the world's only Patient Registry dedicated to DADA2 so that the natural history of the disease can be studied.

"When your child is sick, and you happen to have a background in medicine, you will do anything to find the answers," says Chambers. "But the real drivers of this effort are the 3 patients and 35 clinicians and researchers who came together over the last two years to consider all of the work being done on the disease and consolidate it into something that can be used in everyday medical practice to find these patients."

The DADA2 Foundation will be disseminating patient-friendly communications about the Consensus Statement in the coming weeks and months. They will be highlighted on the DADA2 Foundation's fully revamped patient-focused website. Today, clinicians or patients who have questions about treating DADA2 can contact the Foundation, leaders of which will connect individuals to the right resources so that care can be properly considered by the patient's doctors.

For more information about the Consensus Statement, the DADA2 Foundation or DADA2 in general, please email info@dada2.org. Specific media inquiries can be routed to Kristen.Hayner@dada2.org.

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