



INCLUDE

(INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome)

Dr. Maria Stanley: Connecting Parents with the Best Information for Children with Down syndrome



Dr. Maria Stanley

In over 20 years of experience as a pediatrician caring for children with Down syndrome, Dr. Maria Stanley has learned a lot from the families she serves. “Part of being a clinician is never pretending to have more expertise about families’ experiences than they do,” she said. “We both want the same thing, what’s best for that child. We come together,” she said.

Stanley is a developmental-behavioral pediatrician at the University of Wisconsin-Madison’s Waisman Center which specializes in advancing knowledge of human development, developmental disabilities, and neurodegenerative diseases.

While she listens carefully to the experiences of her patients and their families, she also tries to connect patients and their families with the best information and research available to make health care decisions. Stanley helps patients’ families sort through all the information they need to navigate which established and new, or emerging, therapies are right for them. But too often, the information that she and the patients she cares for need the most, is just not available.

“As clinicians, we have our individual experiences and we make recommendations based on our own observations and experiences, based on clinical judgement. But we need research to know with greater certainty that we are making the right choices,” she said. Information about how medications work in children, in general, is lacking, she said, but for special populations with genetic differences, the absence is especially glaring.

“There are so many unanswered questions. When you look at clinical guidelines for children with Down syndrome, you see that very few topics have adequate data. This makes it really difficult to make evidence-based decisions,” Stanley said.

“There are so many unanswered questions. When you look at clinical guidelines for children with Down syndrome, you see that very few topics have adequate data. This makes it really difficult to make evidence-based decisions.” Dr. Maria Stanley



**PEDIATRIC
TRIALS NETWORK**

Making drugs safer & more effective
for use in the youngest patients

INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndromE)



Stanley is especially interested in questions about the clinical variability of children with Down syndrome. “How do we understand the enormous variation in this population? Some patients have very few long-term medical complications and challenges and others have many,” she asks. For children with Down syndrome as well as co-occurring conditions such as ADHD, anxiety, autism, or sleep apnea, doctors need better tools that address the specific needs for these conditions in this population of children.

The barriers to participation in research are similar to those of other children – travel time, lost work or school for parents and children, and procedures that might be uncomfortable or sometimes painful. But for children with Down syndrome, who have intellectual disability, there might also be difficulty in communicating assent to participate in a study. “There is also what she calls a “healthy skepticism” about potential benefits and risks. “Families want research participation to be safe, meaningful, and equitable.” Stanley said.

Down syndrome is associated with cognitive and health challenges resulting from the presence of an extra, full or partial, chromosome 21. Individuals with Down syndrome experience varying degrees of cognitive disability, as well as congenital heart defects, sleep apnea, and hearing loss. In later years, they are at dramatically increased risk for Alzheimer’s disease and dementia. Autism and ADHD are prevalent in the population, as are autoimmune conditions such as thyroid dysfunction and celiac disease. However, individuals with Down syndrome infrequently develop solid tumors, such as breast or prostate cancer, or have heart attacks despite having multiple risk factors, such as obesity and type 1 diabetes.

Read more about the [INCLUDE](#) project.

“Research is best informed by people living with Down syndrome. If research is removed from that feedback, we run the risk of incorrect assumptions about what is wanted and needed and the best way to move forward.”

Dr. Maria Stanley

When thinking about how researchers, patients and their families and clinicians can work together to further this much-needed research agenda, Stanley says it all comes back to listening to patients and families first.

“When we do what we guess is the right thing, it’s never as successful as getting input from families themselves,” she said. “Research is best informed by people living with Down syndrome. If research is removed from that feedback, we run the risk of incorrect assumptions about what is wanted and needed and the best way to move forward,” she said.

“This community is really generous. People want to participate for the good of others, even when there might not be benefit for themselves or their family member. This giving spirit is a tremendous asset.”



**PEDIATRIC
TRIALS NETWORK**

Making drugs safer & more effective
for use in the youngest patients

LEARN MORE AT:

<https://pediatrictrials.org/>

<https://www.nih.gov/include-project>