Ohio House of Representatives

Tim Ginter

Speaker Pro-Tempore

State Representative, 5th District

Chair Lipps, Vice-Chair Holmes, Ranking Member Liston and members of the House Health Committee. Thank you for the opportunity to speak on House Bill 749 – to Name Aromatic L-Amino Acid Decarboxylase (AADC) Deficiency Awareness Day.

AADC deficiency is an extremely rare neurometabolic disorder that leads to a severe combined deficiency of serotonin, dopamine, norepinephrine and epinephrine. AADC is the final enzyme in the biosynthesis of these neurotransmitters, so a deficiency in its production causes difficulties in the communication between neurons within the nervous system. To date, only about 120 cases of AADC deficiency have ever been reported.

The symptoms of AADC deficiency usually present within the first year of life. Children with the disorder normally have difficulty or fail to achieve physical developmental milestones such as sitting up, crawling and talking. Subsequent symptoms can include weak muscle tone and motor skills, muscle stiffness, sleep problems and difficulties with the body's unconscious functions, among other things. Unfortunately, those afflicted rarely live into adulthood, with a median life expectancy of just 7 years.

Currently, there is no cure for AADC deficiency, although there are treatments available to alleviate symptoms. Vitamin B6 supplements help the AADC enzyme to function, which can be combined with medication that mimic the functions of and strengthen existing neurotransmitters. Combined with other treatments like physical and occupational therapy, they can increase the quality-of-life for those affected. I'd also like to note the active role Ohio's hospitals have been playing in AADC research. Cincinnati Children's Hospital, Nationwide Children's Hospital and the Ohio State University Wexner Medical Center have all been engaged in research or clinical trials for AADC deficiency relief. I hope that this legislation can be used to increase awareness for AADC deficiency and support the research and treatment of those who have been affected by this extremely rare and debilitating disorder. Thank you for your time and I'd be happy to take any questions.