# Researchers find differences in swallowing mechanism of Rett syndrome patients

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Researchers at Wake Forest University Baptist Medical Center have found that the reflux and swallowing problems that are common symptoms in patients with Rett syndrome and other neurological impairments, may be caused by a different mechanism than they are in healthy individuals. The finding leaves researchers to wonder if these patients truly benefit from anti-reflux surgery commonly performed in these children.

In a study published in this quarter's issue of the *Journal of Applied Research*, John E. Fortunato, M.D., lead researcher and an assistant professor in the Department of Pediatrics, found that the esophagus of children with Rett syndrome demonstrates different movements than it does in patients without the neurological disorder, which may explain why so many Rett patients experience persistent reflux and swallowing issues even after undergoing surgery meant to correct those problems.

"The significance of this is for other groups of patients with neurological impairment," Fortunato said. "Do all of these patients have the same mechanism for reflux and swallowing disorders? If not, performing a fundoplication (anti-reflux surgery) may not help. In fact, it may make things worse like it did in the Rett girls."

Previous studies have shown that children with neurological impairments have increased complications after anti-reflux surgery. In this study, Fortunato found the same to be true of Rett syndrome patients who underwent fundoplication. The finding leads researchers to believe that

there may be something different causing the reflux and swallowing problems in Rett syndrome patients and possibly other patients with neurological impairments, such as cerebral palsy, brain injury and autism, than the accepted mechanism for the same problems in otherwise healthy adults and children.

Rett syndrome is a childhood neurodevelopmental disorder caused by mutations in the gene MECP2 located on the X chromosome. It is the only Autism spectrum disorder with a known genetic cause and is characterized by normal early development followed by loss of purposeful use of the hands, distinctive hand movements, slowed brain and head growth, walking abnormalities, seizures, and mental retardation. Early symptoms may also include toe walking, sleep problems, teeth grinding, difficulty chewing and breathing difficulties while awake such as hyperventilation, apnea (breath holding), and air swallowing.

Rett syndrome affects one in every 10,000 to 20,000 live female births and is associated most closely with gastroesophageal reflux disease (GERD) and difficulty and /or pain swallowing (dysphagia). Most patients affected by the mutation have trouble eating, so they often are shorter and weigh less than other children their age. To maintain proper nutrition, some children need to be fed through tubes placed in their noses or stomachs. Boys who inherit the mutated gene usually don't survive infancy, according to the National Institute of Neurological Disorders and Stroke.

The study included 32 Rett patients between the ages of 2 and 14 with prior history of feeding problems. Researchers looked at the movement (or peristalsis) of the esophagus in the girls and found unusual esophageal movement disorders.

As a result of the study's findings, Wake Forest Baptist has approved

further research to look at esophageal movement and swallowing function before and after reflux surgery, comparing children with and without neurological impairment.

"This issue is of particular interest to pediatricians who refer these patients for their 'reflux' problems," Fortunato said. "If we develop a better understanding of the mechanisms behind the problems being experienced by these children, we just might be able to find a way to make life a little more comfortable for them."

Source: Wake Forest University