

# Milk or no milk? Study fills long-time knowledge gap on babies with genetic disorder

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A feeding bottle filled with milk in the foreground. In the background, a mother is lying face up on the floor holding up her baby in an affectionate gesture.  
Credit: Pexels/rawpixel.com

Milk can be lethal to babies with classic galactosemia, a rare genetic disorder that severely impairs the body's ability to process a milk sugar known as galactose and is associated with a host of neurodevelopmental issues. However, the fate of children with Duarte galactosemia—a milder, more common variant—has been unclear. As a result, state-level recommendations on milk intake for children diagnosed with Duarte galactosemia have varied widely, from no limitations to banning all milk for the first year or more of a child's development.

A new study co-authored by a Washington State University researcher finally brings clarity to parents of children with Duarte galactosemia, their health care providers, and health officials. Published in a recent issue of the journal *Pediatrics*, the study found that children with Duarte galactosemia are at no greater risk of long-term developmental abnormalities than their unaffected siblings, regardless of their exposure to milk as infants.

"Therefore, we don't need to detect Duarte galactosemia, and we don't need to restrict their milk intake," said co-author Nancy Potter, a speech-language pathologist and a professor in the Department of Speech and Hearing Sciences at the WSU Elson S. Floyd College of Medicine.

Potter, who collaborated on the study with a team of researchers from Emory University, said the findings provide peace of mind to parents of children with Duarte galactosemia that they can confidently feed their child as they would any other baby.

"There has been great angst among parents of children with Duarte galactosemia," Potter said. "They're wanting to know, 'Am I harming my baby by giving him or her milk?' We know the value of breast [milk](#) to a baby—is it worth restricting that? That's the question we set out to answer."

She said the study findings also make a compelling argument for changes to newborn screening criteria for galactosemia that could provide significant cost savings while minimizing the high number of false positives associated with screening intended to detect Duarte galactosemia.

While newborns are screened for galactosemia in all 50 U.S. states, screening protocols vary from state to state—some only test for the extreme deficiencies of classic galactosemia, while others are designed to detect Duarte galactosemia as well. Newborn screening tests for galactosemia look at blood levels of an enzyme called galactose-1-phosphate uridylyltransferase (GALT), which helps convert galactose into glucose that the body can use for energy. Children with GALT enzyme levels of 1 percent or less are considered to have classic galactosemia, which affects about one out of every 50,000 babies born in the U.S. Most babies with Duarte galactosemia—which is 10 times more common—have about 25 percent of the normal level of GALT activity.

To determine long-term outcomes for children with Duarte galactosemia, the research team studied 350 children between the ages of 6 and 12 in 13 U.S. states—206 children were case patients with Duarte galactosemia; the remaining 144 were unaffected siblings who served as controls.

The researchers surveyed parents about each child's level of dairy consumption as an infant, finding that 40 percent had consumed a substantial amount of dairy, whereas 60 percent had not. They also tested all children for a wide range of outcome measures representing five developmental domains: cognitive development, physical development, speech/language and hearing, motor development, and socioemotional development. Testers were blinded to the status of their subjects, meaning they did not know whether children being tested were case patients or controls. Thorough analysis of the study data showed

that there were no significant differences in outcomes between case patients and controls.

In a [commentary published alongside the study](#), Shawn McCandless—a professor of pediatrics and genetics at the University of Colorado Denver and the president of the Society for Inherited Metabolic Disorders—said, "The field has been waiting 50 years for this study, since newborn screening (NBS) for galactosemia began. This work can be used to impact [clinical care](#) and NBS program planning. It can also be used to teach us important lessons about the public funding of research into clinical outcomes and the importance of data-driven clinical decision-making."

Potter said their study is the largest ever conducted in [children](#) with Duarte galactosemia and has contributed to clearing up conflicting findings from two earlier, smaller studies.

**More information:** Grace Carlock et al, Developmental Outcomes in Duarte Galactosemia, *Pediatrics* (2018). [DOI: 10.1542/peds.2018-2516](https://doi.org/10.1542/peds.2018-2516)

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