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Only few doctors refer children with developmental delays to genetics specialists, say researchers

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A nine-month-old boy isn't rolling over, reaching for objects or babbling as he should be. One step his doctor won't likely take right away: have him evaluated by a genetics specialist.

Almost three quarters of doctors surveyed said they would refer a child with these types of developmental delays to a specialist - but rarely a genetics specialist, University of Michigan Health System researchers report in *The Journal of Pediatrics*.

"Children who show delays in reaching two or more developmental milestones like sitting up and babbling have a greater risk of a genetic disorder. The earlier we can detect genetic diseases, the earlier we can begin treatment that can help," says lead author Beth Tarini, M.D., M.S., assistant professor of pediatrics at U-M's C.S. Mott Children's Hospital and researcher at the Child Health Evaluation and Research (CHEAR) Unit.

"We found that few primary care physicians would order genetic testing or refer a child to a genetics specialist as a first step when they see children with developmental delays. This may slow down the process to finding and treating the problem."

Genetic tests are usually done on blood and test children for specific diseases that may cause developmental delays, such as Fragile X syndrome, an abnormality on the X chromosome that causes severe mental delays. Genetic tests can also detect extra or missing pieces of genetic material that may be causing delays.

Researchers analyzed responses from nearly 450 doctors who were asked whether they would refer a boy with multiple developmental delays to a specialist or initiate testing (genetic or non-genetic), assuming insurance coverage was not a factor.

"Our findings suggest that the first doctor a child sees for development problems may not immediately think of helping the family access genetic services despite the child's increased risk of a genetic disorder," says Tarini, who is also a member of the U-M Institute for Healthcare Policy and Innovation.

"We need to understand the barriers that may prevent children with significant developmental delays from getting a genetic evaluation as soon as possible, so that we can get help to those who need it."

Source:
University of Michigan Health System
