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Newborn screening for spinal muscular atrophy increasingly makes sense

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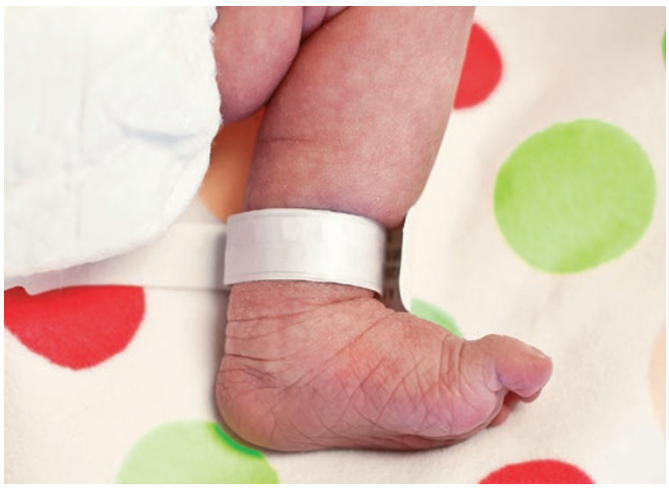
A research team from Columbia University, New York, has demonstrated the feasibility of adding newborn screening for spinal muscular atrophy (SMA) to the national uniform screening panel. SMA, the most common genetic cause of death in children under age 2, occurs in 1 in 5,000–11,000 births in the United States. The frequency of pathogenic variants (PV) is relatively common, with 1 in 54 individuals carrying the autosomal recessive PV. While population-based screening for SMA has been recommended for many years, addition of the test was denied in 2008 by the Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children owing to the lack of an available treatment. The authors say that the 2016 development of an FDA-approved treatment, nusinersen, should prompt reconsideration. If the disorder could be detected early, before the onset of symptoms, intervention to prevent motor neuron loss could promote more effective treatment, they explain. The research team's pilot study, reported in this issue, demonstrated the feasibility of newborn screening for SMA in New York. In 2016 they screened 3,826 newborns at three hospitals in New York City, testing newborns for the deletion in exon 7 of *SMN1*. One baby identified through newborn screening was successfully treated with the newly approved medication, and 59 carriers were identified. Follow-up genetic counseling was offered to families of carriers. The investigators note that more than 93% of the parents agreed to include SMA in routine newborn screening, indicating that parents would welcome the addition of this test nationally.—*Karyn Hede, News Editor*



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Maturity-onset diabetes of the young often goes undiagnosed in kids

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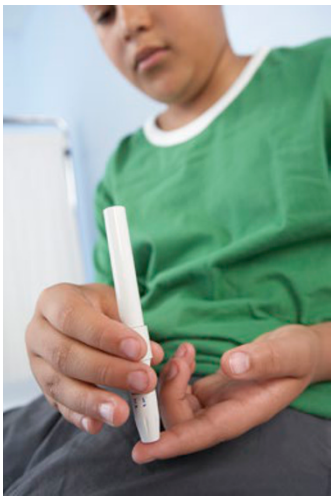

Adam Gault/SPL

Some children diagnosed with type 2 diabetes actually carry a genetic variant that causes maturity-onset diabetes of the young (MODY). A recent study of 488 overweight adolescents who had been diagnosed with type 2 diabetes revealed that more than 4% had this genetic form of diabetes instead. The number of diagnoses, 22, was high enough that the research team proposes that the disorder be considered in children not typically classifiable by current American Diabetes Association guidelines. MODY results from PV in 1 of 14 known genes, and its variable clinical presentation often results in misdiagnosis as type 1 or 2 diabetes. Researchers from several institutions participating in the Treat Options for Type 2 Diabetes in Adolescents and Youth (TODAY) clinical trial contributed to the research, the results of which suggest that many cases of diabetes in youth may be misdiagnosed. The findings are significant because the management of MODY is different from that of more common forms of diabetes. There were no observable clinical criteria to differentiate overweight or obese adolescents with MODY from those who developed type 2 diabetes. The team suggests that, despite the upfront cost of testing for monogenic diabetes, the disease provides an opportunity to implement personalized genomic medicine. They point out that under some models, genetic testing for MODY could be as cost-effective as current medical practices and potentially cost-saving. —*Karyn Hede, News Editor*