MedicalXpress

Newborn and carrier screening for spinal muscular atrophy now possible, claim scientists

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Scientists in Ohio studying Spinal Muscular Atrophy (SMA) have concluded that the technology now exists to carry out nationwide screening of newborn children and pregnant mothers. The study, published in the *American Journal of Medical Genetics*, reveals that effective screening may allow parents to find proactive treatments before the symptoms become irreversible.

SMA is the most commonly inherited lethal disease in infants, it is caused by gene mutations and affects approximately 1 in 10,000 live births. While a cure is not yet available the team, led by Dr Thomas Prior of Ohio State University, carried out pilot studies to discover if the screening of newborn babies could provide an early and definitive diagnosis, help parents to find proactive treatments earlier and allow at-risk family members to make informed reproductive choices."

"Results from pilot studies often help project outcomes and help to build a realistic national picture of what can be expected from large-scale programs," said Prior. "Given the devastating nature of SMA, our two tiered study is designed to address the clinical applicability of testing newborns and carrier screening across the population."

There has been a major expansion in newborn screening throughout the United States with an increasing consensus that newborn screening should not be limited to disorders for which there is a clear cure.

"We have demonstrated that an effective technology does exist for newborn screening of SMA," said Prior. "A newborn screening program for SMA not only allows parents to enter their children into clinical trials earlier, but would enable patients to obtain proactive treatment, for nutrition, physical therapy, and respiratory care, earlier in the disease progression."

The second pilot study dealt with carrier screening of SMA. Women or couples referred to two large perinatal centres in Columbus, Ohio were offered free-of-charge SMA carrier testing at the time of genetic counselling.

The goals of the SMA population carrier screening pilot program were to: identify couples at risk for having a child with SMA, to gain an estimate of the carrier frequency, to provide data on patients' knowledge of SMA, attitudes toward SMA screening as well as an assessment of the education material provided by the Claire Altman Heine Foundation.

"We have found that the information necessary for individuals to make an informed decision regarding carrier testing can be presented effectively and efficiently through a counselling session enhanced by printed educational material," concluded Prior.

Provided by Wiley