Most people in favour of screening for spinal muscular atrophy

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Credit: University of Warwick

Research from the University of Warwick indicates that most people are in favour of newborn screening for the potentially deadly condition spinal muscular atrophy (SMA). The study Newborn genetic screening for spinal muscular atrophy in the UK: The views of the general population has been published in the journal *Molecular Genetics & Genomic Medicine*.

SMA is an inherited neuromuscular disorder with a wide spectrum of severities and is a leading genetic cause of infant death worldwide. However, there is no routine screening programme for SMA in the UK. This is in spite of the licensing of a new therapy for SMA in December 2016, Nusinersen which is marketed as Spinraza, which has the potential

to reduce the severity of the disease when given early in life, before the onset of SMA symptoms. Up until now, however, lack of treatments and the inability of screening tests to accurately predict disease severity have been among the key reasons that the implementation of a screening programme has faltered in the UK.

The research was led by Dr Felicity Boardman of the University's Warwick Medical School. Dr Boardman said: "With the recent release of the first therapy for SMA, calls are being made internationally for a reconsideration of the current stance on screening; however, very little is known about the views of the general public. We decided to address this gap in evidence by surveying people about their views."

Dr Boardman, who worked in collaboration with Dr Philip Young in the University's School of Life Sciences, found that 84% of the 232 members of the public surveyed were in favour of newborn screening. Key reasons for support were a belief that it would lead to better healthcare and life expectancy for affected infants and facilitate informed decision-making for future pregnancies. Key reasons for non-support were a belief in the potential for significant negative impact on the family unit in terms of parent/child bonding and stress, particularly as the child's prognosis may be difficult to predict at birth.

Dr Boardman added: "Public acceptability is a key component in the evaluation of any potential screening programme in the UK. This study demonstrates that newborn screening for SMA is viewed largely positively by people unfamiliar with the condition. The perceived importance of early identification overrode all other social and ethical concerns about screening for the majority of participants."

To discover what the general public - people who have no prior relationship to SMA – think about newborn screening for SMA, the researchers used an online survey to gather the opinions of 232

individuals. The group was composed of 69 percent women, and about 40 percent of participants were younger than 25. About half the group had a university degree and 63 percent were not religious. While about half the group had children, only three percent were pregnant or trying to become pregnant at the time of the survey.

The results of the study were compared to earlier survey results from people living with SMA. Eighty-four percent of the general public favoured newborn screening - a number significantly higher than the 70 percent reported among SMA-affected families. Like the general public, the most common reason for not supporting screening amongst SMA-affected families was concern about the impact of the screen on the early experiences of the family.

The researchers also noted that for both the general public as well as SMA-affected families, levels of support for newborn screening were comparable to support for other screening methods, including preconception and prenatal screening. Indeed, for both populations, preconception screening (identifying SMA carriers before pregnancy) was considered the optimal screening approach.

More information: Felicity K. Boardman et al, Newborn genetic screening for spinal muscular atrophy in the UK: The views of the general population, *Molecular Genetics & Genomic Medicine* (2017). DOI: 10.1002/mgg3.353 , dx.doi.org/10.1002/mgg3.353

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