Newborn Screening discussion taken from page 17 of the 'Screening for spinal Muscular Atrophy - External Review against appraisal criteria for the UK National Screening Committee June 2018'

Newborn dried blood spot (DBS) screening is one technique that can be used to detect SMA at this stage, with a subsequent diagnostic test required for confirmation in all cases.

Screening of newborns has the benefit of potentially identifying individuals with SMA before disease onset allowing for identification of those who would benefit from new molecular and genetic treatments.

Various ethical implications could result from the introduction of a screening programme, whether this is carrier screening, antenatal screening or newborn screening; therefore, population-based pilot studies have been conducted investigating these social issues:

One survey showed overwhelming support from expectant couples for newborn DBS screening for SMA, even considering a lack of treatment development[1].

Another survey of families affected by SMA found that, although 75% of families were in favour of screening in some form, they had concerns including carrier stigmatisation, social engineering and, for antenatal screening in particular, the risk of termination when a high quality of life could still potentially be achieved[2]. Therefore, if screening were to be implemented, the provision of genetic counselling should be carefully timed and given appropriately[3].

Evidence from the first study suggests that the majority of parents would still want to know if their child had the disease at birth, even if it would affect their child's health and shorten their lifespan.[1]

However, as there is no cure for SMA and no way to accurately predict prognosis, for example using SMN2 copy number, there is currently no clear consensus on the impact of parents receiving the news that their child is affected by SMA at birth, through newborn screening programmes, or upon diagnosis following the onset of symptoms[2,4]

One study has suggested that a diagnosis at birth hastens parental grief and allows the family to more quickly come to terms with their child's condition, and the implications it has for their family's future[5].

Other evidence has reported that a diagnosis through newborn screening, before symptom onset, may have a negative impact on parents and that the first weeks and months following their child's birth can be overshadowed by anxiety, shock and grief which can interfere with their bonding with their child[6].

An additional consideration regarding the implementation of SMA newborn screening would be whether this should be an opt-in or opt-out programme. Evidence from one study suggests that there is support for an opt-out approach considering the UK newborn screening programme already utilises this approach[7]. Additionally, there was support that, given there are finite resources within public services, an opt-in approach may require more resource

through further education and time commitments from healthcare professionals, and therefore an opt-out approach was preferred. A newborn screening programme would also not require additional blood draws or burdensome procedures for the babies and their families, as samples would already be taken for the other newborn screening tests. However, one implication could be the risk of a false positive test and the impact this may have on families and the initiation of any unnecessary treatment.

References

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