New hope for newborn screening

Newborn bloodspot screening (NBS) involves collecting a small amount of blood on day 5 of life from a heel prick which is then applied to a blood spot card. This sample is then sent to a biomedical science laboratory for testing and in exceptional circumstances, such as when the infant requires a blood transfusion, sampling can be undertaken between days 5 and 8, with all babies in the UK under one years of age eligible for screening for all 9 conditions included on the UK newborn screening panel.

One of, if not *the*, most efficient and effective of all screening programmes, NBS is utilised in the pre-symptomatic detection of infants with congenital conditions and is performed in many countries in the world, all US states and almost all European countries. In 2021 alone, 38M neonates were screened globally, leading to the identification of 38,000 positive results for rare disease or 1 baby every 15 minutes, enabling for early treatment of those affected, subsequently leading to the prevention and amelioration of long-term consequences.

The UK however, currently ranks 18th across Europe in the number of tests included on the country's NBS panel, with countries such as Italy screening for 48 conditions. This subsequently means the UK is falling behind its neighbours which is perhaps surprising, as screening newborn babies for inherited metabolic disease began in **the UK** in the late 1950s with the 'nappy test' for phenylketonuria.

Considering this, and the fact that the UK is home to an estimated 3.5M people living with rare disease, it is clear that countless rare disease diagnoses are being missed and with average diagnosis for a rare condition not picked up in NBS standing at 5-9 years, this can have dire consequences.

More must be done to ensure rare diseases are picked up earlier and that's where the Newborn Screening Collaborative comes in. This group of charities and support organisations are dedicated to improving the UK newborn screen and aim to do so through meeting three key priorities this year, that are:

Priority 1. Review of UK NBS policy and processes – Review current policies and processes including those being developed in response to the RD framework, Genomics plan etc. to identify areas that are not working, where the gaps are, whether proposal meet these gaps, what has been done internationally and would these work in the UK, make recommendations for improving or defining level of evidence required. Note: this would include the issue of the 'treatment' within the NBS 'criteria'

Priority 2. Evidence and progress review – Collate and review existing and pipeline sources of evidence, held, or planned by individual collaborative members relevant to NBS. This

could include parent / carer responses & views on NBS, clinical opinion, international evidence / consensus reports / survey outcomes / registry data. What do we have, where are the gaps, what as a group do we need to do to ensure the patient / parent view of NBS is captured if not already there? (i.e., collaborative survey)

Priority 3. Political engagement – What do different group members have in the pipeline, how can we better align, join forces. Who do we need to target, which APPG group do we need to target and how should we deliver our message (include scope of key dates from each partner organisation i.e.: rare disease day, other patient group awareness days, conferences etc.)

Even though we have a small team, we have a big impact and are continually committed to supporting people affected by all 500+ Inherited Metabolic Disorders and their communities.

Moving forward, Metabolic Support UK will do everything to support the achievement of these priorities and has taken a leading role in communicating the benefits of NBS through producing awareness campaign materials on behalf of the newborn screening collaborative, but this is just a start.

We will influence decision makers; we will achieve early diagnosis and we will be there for you. **Your rare condition. Our common fight.**