PE1823/C
Genetic Alliance UK submission of 29 September 2020

Genetic Alliance UK welcomes the opportunity to provide a submission to the Public Petitions Committee of the Scottish Parliament in response to Public Petition PE01823 "calling on the Scottish Parliament to urge the Scottish Government to offer full body scans for neonates in Scotland with the aim of detecting and hopefully treating rare and hidden conditions".

Background

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by genetic, rare and undiagnosed conditions. We are an alliance of over 200 patient organisations. We are home to Rare Disease UK – the national campaign for people with rare diseases and all who support them – and SWAN UK (syndromes without a name), the only dedicated support network available for families of children and young adults with undiagnosed genetic conditions in the UK.

A rare condition is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases and one in 17 people will be affected by a rare condition at some point in their lives. This equates to approximately 420,000 people in Scotland.

The long and difficult journey many patients with rare conditions have to go through to secure a rare disease diagnosis has been widely acknowledged across the healthcare sector and is known as the 'diagnostic odyssey'. Delayed diagnosis can have a significant impact on a patient's health, treatment options and even life expectancy. Early diagnosis and intervention can enable patients to access the most effective treatments in a timely manner, and importantly, help them find answers about their condition.

Response to PE01823

Genetic Alliance UK agrees with the view put forward in response PE01823/A provided by the Scottish Government that full body scans for all neonates would 'carry an element of risk' and does not recommend routine scanning of newborns.

However, Genetic Alliance UK fully supports the aim, put forward by the petitioner, of improving the early detection of rare conditions, and the opportunities for treatment. One way to achieve this would be for the Scottish Government to review and expand the existing Newborn Screening Programme.

Newborn screening plays an important role in aiding the identification of babies at risk of developing rare or genetic conditions, allowing diagnosis before symptoms develop. Such early detection and intervention is particularly important for rare conditions of early childhood, many of which are progressive and irreversible. Early detection provides an opportunity for stabilising treatment before the deterioration in health or development of a child occurs, potentially preventing the most serious effects of these conditions, which can include severe disability and death. Early diagnosis of an affected child also offers the added benefit of supporting family

planning, providing couples with the opportunity to exercise reproductive choices if they wish to.

Genetic Alliance UK believes that the Newborn Screening Programme in Scotland is not fulfilling its potential. Scotland currently screens for just nine conditions. This is far fewer than the current tandem mass spectrometry methods used can detect and far fewer than other comparable high income countries. Iceland, for example, screens for 47 conditions, the Netherlands for 34 and Norway for 28.

Decisions on what conditions are screened for in Scotland are based on the recommendations of the UK National Screening Committee (UKNSC), although it is important to note that Scotland is not bound by these recommendations and could choose to go further.

In July 2019, Genetic Alliance UK published the Patient Charter 'Fixing the Present, Building for the Future: Newborn Screening for Rare Conditions', endorsed by over 50 patient organisations. The Charter examines how the UKNSC has made changes to the criteria used to make decisions, which have disadvantaged rare conditions; and exposes how membership of the committee excludes members of our community.

The Charter comprehensively explores opportunities for improvement and has set forth a series of recommendations including:

- The methodology for decision making on newborn screening should be adapted in recognition that the conditions being screened for are rare and thus present specific challenges
- Decisions on newborn screening should be made by a body with specific and relevant expertise
- Benefits to the patient, family and broader society other than preventative interventions should also be considered
- Newborn screening should be recognised as a mechanism for earlier diagnosis as part of a broader care pathway, keeping step with progress in disease identification and diagnosis in symptomatic patients
- The newborn screening programme should be 'opportunity based' based on categories of conditions it is possible to detect through screening, not condition by condition.
- Measures should be taken to address out of date infrastructure and technologies
- A pilot of genome sequencing in newborn screening should be planned.

A comprehensive screening programme in Scotland, with the high identification and diagnosis rates it would have the potential to deliver, could contribute vital information to the Congenital Anomalies and Rare Disease Registration and Information Service for Scotland (CARDRISS). This will allow our understanding of rare conditions to grow, facilitating research that can lead to future treatments. Some of the newest innovative treatments could not have been developed in Scotland, because a screening programme is necessary to recruit children before symptoms arise, (an example of this is the newest gene therapy for spinal muscular atrophy).

It is for the Scottish Government to make decisions on how the Newborn Screening Programme is delivered in Scotland. Now is the time for fresh thinking and this petition presents an opportunity to consider how the Newborn Screening Programme can be developed and expanded in Scotland to achieve the aim put forward by the petitioner – to improve the detection of rare conditions.

Genetic Alliance UK urges the Public Petitions Committee to take further evidence on the role of newborn screening in Scotland and to consider a review of the existing Newborn Screening Programme to build a system that will be fairer to the rare disease community.