ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Newborn Screening Report Launch

Date: **23 July 2019**

Venue: **Room O, Portcullis House**

Parliamentarians	Guest Speakers
Catherine West MP (Chair of second half of meeting) Lord Turnberg (Chair of first half of meeting) Alex Sobel MP (Officer) Baroness Thomas of Winchester Jim Shannon MP	Nick Meade, Genetic Alliance UK Dr Rachel Carling, Viapath, Guys & St Thomas' Hospital Dr Stuart Adams, GOSH

1. Welcome from Lord Turnberg

2. Introduction from Nick Meade, Director of Policy, Genetic Alliance UK

Nick Meade outlined Genetic Alliance UK's newborn screening report, which was officially launched at the meeting and lays out recommendations for how to improve the UK's newborn screening programme.

The UK currently screens for between five and nine conditions as part of the newborn blood spot screening programme, depending on the country, with a pilot planned for one more. This is fewer than most comparable high income countries, many of which screen for between 20 and 50 conditions, and also many fewer than the current tandem mass spectrometry methods used can detect.

A workshop with Genetic Alliance UK members led to several recommendations being made around how the methodology for newborn screening should be adapted. Nick Meade stressed the importance of patients being involved in the decision-making process, especially as the UK National Screening Committee does not include patients unlike the majority of other health decision-making bodies.

Nick Meade stated that it would be important to model the value of catching conditions earlier rather than relying on long-term treatment.

3. **Guest speaker: Dr Rachel Carling, Director of Newborn Screening and Clinical Lead, Biochemical Sciences at Viapath, Guys & St Thomas' Hospital**

Dr Carling gave perspectives for context on some of the technological opportunities and challenges for expanding the UK's newborn screening programme.

4. **Guest speaker: Dr Stuart Adams, Joint Trust Lead Healthcare Scientist at Great Ormond Street Hospital**

Dr Adams talked about the opportunities for expanding the UK's newborn screening programme, particularly for Severe Combined Immuno-Deficiency (SCID)

5. **Discussion**

What the 'perfect' UK newborn screening programme would look like is not easy to define. Screening is designed to benefit the population, rather than the individual. Two babies are born every day in England with a rare disease and we have the ability to screen for many disorders, for example with mass spectrometry. Issues of false positives are significant, as a positive is dealt with as a clinical emergency. There are ways to rule out false positives, including additional testing, which helps to provide a better quality service. Tests should not be introduced just because they exist, but conversely it would not be difficult to do more testing, and provide a better service, as many tests would run easily on existing lab systems, and advances such as whole genome sequencing are being used widely elsewhere, and privately too.

The UK has 13 screening labs, which may be more than twice the number needed. Because there are so many more labs than required there are difficulties with recruiting staff, running services outside of the week. Fewer, bigger labs would deliver economies of scale benefits from accessing a critical mass of population, and would help with analytical harmonisation to raise the quality of testing. There are also now commercial kits to test bloodspots (stored on Guthrie cards) that require only tiny amounts of blood.

Babies born with Severe Combined Immuno-Deficiency (SCID) have very poor immune systems and will succumb to opportunistic infections. This will lead to death within their first year if difficult and expensive treatments such as a bone marrow transplant are not administered. It is essential to eliminate infections before treatments start. This is hard to do if the SCID diagnosis is made later, with only a 40% chance of survival at that stage. This compares with a clear cut clinical benefit from screening of 100% survival when testing is done at birth, due to the baby being the second child born when the first child had a SCID diagnosis. Sometimes a test is done post-mortem to see if a baby died of SCID, using its existing (untested) Guthrie card, in part to help inform reproductive choice.

To roll out more testing under the newborn screening programme, issues to consider include how new testing is done (which may be different for particular diseases), who owns the data, who is the custodian of the data, how and where to store samples, and who manages it all. If newborn screening can be done, someone will do it and that if not the NHS, then private companies will take ownership. It was also noted that engagement with the screening panel to add more tests is an incredibly difficult, protracted process, with no new test being added in almost five years. Current testing only considers the index case, and does not take into account impact on other family members, and the merits of cascade/sibling testing. However, regardless of how individuals and families are impacted, it is important that the choice is there.

6. Next steps

DHSC officials present at the meeting committed to providing a response on the report.

The Charity will share the report with its member charities for endorsement, and conversion into a charter. The charter will then be presented to the four UK CMOs to seek support for implementing the report's seven recommendations.

7. AOB Close

It was noted that he next meeting of the APPG will likely be in October, for the launch of the 'Resetting the Model' report.