Government's glacial speed in expanding newborn screening leaves infants at risk of life-threatening rare diseases

- **New poll finds 86% of parents want to know if their newborn has a rare condition**

- **65% of parents in favour of screening newborns for as many conditions as possible**

23rd May 2022: At least 150 newborn children have not been diagnosed or treated for life threatening rare diseases over the past three years because of the Government's failure to expand the National Newborn Bloodspot Screening Programme.

Rare Diseases Ireland has called for greater action and accountability. It has criticised the Government for failing to start screening for the rare condition ADA-SCID that it added to the national screening programme in July 2020 and for moving at glacial speed to identify and save children with treatable conditions.

The organisation has today published a new research study which reveals that 86% of Irish parents want to know if their newborn has a rare condition, even if it is not yet treatable. 65% of parents are in favour of screening newborns for as many rare conditions as possible.

The study, which is based on a national poll of 1,000 respondents conducted by iReach Insights, has also found that 83% of parents think that families with newborn babies with an increased risk of a rare condition should be able to ask for specific screening tests to be included on the screening panel to ensure early diagnosis.

"Ireland was one of the first countries in the world to introduce a national newborn screening programme in 1966. We are now languishing amongst the lowest ranked countries in the EU. Italy screens for 45 conditions while we currently screen for only 8 conditions. Our failure to keep pace is costing Irish children their lives," said Vicky McGrath, CEO of Rare Diseases Ireland.

"The Minister for Health, Stephen Donnelly, T.D made an emphatic commitment to the Oireachtas and all stakeholders in 2020 '*to provide the public health and scientific resources that are needed to save as many children as can be saved and to identify every treatable condition that can be identified and treated in this country*'. Not one change has been delivered since," she added.

Rare Diseases Ireland is questioning why so little progress has been made by the National Screening Advisory Committee (NSAC) since it was established as an independent expert group in 2019 to prioritise the expansion of the National Newborn Bloodspot Screening Programme and to make recommendations to the Minister.

The Committee has only made one recommendation to expand the Programme to date. Its recommendation to add ADA-SCID, which is a rare, inherited immune system disorder, to the list of conditions screened for was approved by Minister Donnelly in July, 2020. It has yet to be implemented by the Health Service Executive (HSE).

Rare Diseases Ireland is concerned that the NSAC is not fully accountable because it is operating on a non-statutory footing following deferral of the National Screening Advisory

Committee Bill 2020. The Bill was at Second Stage when the Seanad agreed to an amendment from Minister Donnelly to defer consideration for a 12-month period, commencing 30th September 2020. Had the Bill been enacted, NSAC would have been required to prepare a report and make recommendations to the Minister in relation to expanding the conditions tested for by the national newborn screening programme within 6 months.

Proposing the amendment, Minister Donnelly told the Seanad debate that the real risk of putting the Committee onto a statutory footing was that it "*would simply slow down what is vital and life-saving work*" and that it "*needed to be left to build up some pace and a head of steam*". He gave an assurance that changing the 12-month extension would require a Cabinet decision.

The HSE National Newborn Bloodspot Screening Programme Governance Group wrote to NSAC last year regarding conditions which may be considered for addition to the screening programme. The list of conditions is based on expert clinical opinions of the Group members, a high level of understanding of whether something could be screened for, and the likely impact of earlier screening on outcomes for children with those conditions.

The Department of Health is now progressing the development of a methodology for prioritisation of conditions on the list for more extensive evidence assessment and evaluation by the Health Information and Quality Authority (HIQA).

"The Government is moving at glacial speed and the process of adding new conditions is mired in administrative bureaucracy, even though this work should be led by the National Screening Advisory Committee as an independent expert group. We do not have the luxury of time. We need action right now and we are calling on Minister Donnelly to deliver upon the promises he made to all stakeholders in 2020", Ms. McGrath said.

"Parents are hugely supportive of expanding screening. 65% of parents believe we should be screening newborns for as many conditions as possible. The fact that 86% of parents want to be informed if their newborn has a rare condition, even if it is not yet treatable, shows that families recognise the valuable role that screening and early diagnosis has to play in helping them to plan for the future and to manage their child's healthcare needs," she added.

The national survey also found that only 39% of respondents are aware of the existence of NSAC and that only 14% were aware of its first Annual Call for submissions for changes to screening programmes in Ireland.

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For interviews with Vicky McGrath, CEO of Rare Diseases Ireland and further information on its National Newborn Screening Research Study contact:

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About Rare Diseases Ireland

Rare Diseases Ireland is a non-governmental organisation and patient advocacy national alliance for voluntary groups representing people affected by or at risk of developing a rare disease. RDI is committed to identification, treatment, and cure of rare diseases through programmes of education, advocacy and patient engagement.