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BRIEF REPORT

Parental awareness of newborn bloodspot screening in Ireland

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Abstract

Background There is little known regarding how familiar parents are with the newborn bloodspot screening (NBS) test or how well parents of a child with a screen-detected condition understand that condition initially.

Aim The study aim was to examine parental NBS awareness and conditions screened.

Methods Two studies were conducted: [1] Parents of children with cystic fibrosis (CF) detected via NBS and subsequently, diagnosed ($n = 124$) completed a telephone questionnaire regarding information they received at the time of NBS. [2] A cross-sectional study of women ($n = 662$ (58%) antenatal; $n = 480$ (42%) postnatal) attending three large maternity hospitals completed a questionnaire addressing NBS awareness.

Results Mothers incorrectly identified diabetes/asthma (35% postnatal; 70% antenatal) and sickle cell disease (26%) as conditions on NBS in Ireland. Phenylketonuria was correctly identified by 48/26%, CF by 82/64%, and congenital hypothyroidism by 35/13% postnatal and antenatal women respectively. Of parents of children screen-detected and subsequently, diagnosed with CF, only half ($n = 63$; 51%) reported awareness at the time of NBS that CF was included. These results should be used to improve the information provided to expectant mothers and to inform health professionals' initial discussions with parents about their child's diagnosis, building on parents' pre-existing knowledge.

Keywords Awareness · Cystic fibrosis · Newborn screening

Introduction

In Ireland at the time of this research, newborn bloodspot screening (NBS) encompassed congenital hypothyroidism, phenylketonuria (PKU), maple syrup urine disease, galactosemia, homocystinuria and most recently cystic fibrosis (CF), introduced in 2011. Little is known about parental familiarity with NBS or initial understanding of a screen-detected disease. It is recommended that the most appropriate time to provide information about NBS is the antenatal period [1–3]. The information leaflet provided by the Irish NBS programme lists the conditions being tested without providing comprehensive information; parents are directed to the NBS programme website for further information. The aim of this study was to examine parental awareness of NBS and provision of information.

Methods

The data comes from two linked studies. The Irish Comparison Outcome Study (ICOS) of CF is a national historical cohort study conducted in Ireland. Two cohorts of children were compared in this study: children clinically diagnosed with CF born in the Republic of Ireland (ROI) between July 2008 and June 2011 before screening was introduced (clinical cohort) and children identified via NBS and subsequently diagnosed with CF born in ROI between July 2011 and June 2016 following the introduction of screening (NBS cohort). Parents of children with CF detected via NBS ($n = 124$) completed a telephone questionnaire regarding the information received at the time of NBS, with the main carer completing the questionnaire. In the second study, a cross-sectional study of 1142 women attending antenatal clinics (662 (58%)) and on postnatal wards or on return to the hospital on day 5 for the NBS test for their infant (480 (42%)), were

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recruited from the National Maternity Hospital, the Coombe Women and Infants University Hospital and University Maternity Hospital, Limerick, and asked to self-complete a questionnaire. Antenatal women were eligible to participate in the study if they were ≥ 36 weeks gestation (to allow for the provision of information about NBS by health providers); aged ≥ 18 years of age and able to speak and read English. All eligible women present in the clinic were approached to participate in the study; this sample was representative of all women attending antenatal clinics. The sample did not include those women attending private antenatal clinics. For postnatal women, the questionnaire was completed immediately after their infant had NBS testing. Postnatal women were eligible if they were aged ≥ 18 years of age and able to speak and read English. Postnatal women recruited to the antenatal study were ineligible. The questionnaire used was developed using items from two previously published studies [4, 5].

Results

More postnatal than antenatal women recalled receiving a NBS information leaflet (39 vs. 14%; $p < 0.05$). Knowledge about the conditions on the NBS panel was poor, but better in the postnatal group. Women were asked to indicate from a list which conditions they thought NBS was screening for; significantly more postnatal women were aware that PKU (48 vs. 26%; $p < 0.05$), CF (82 vs. 64%; $p < 0.05$) and congenital hypothyroidism (35 vs. 13%; $p < 0.05$) are included in NBS. Over 35% of postnatal and 70% of antenatal women incorrectly believed that NBS tests for diabetes and asthma, and 26% incorrectly identified sickle cell disease as a condition on the NBS in Ireland.

Of 124 parents whose child was screen-detected with CF, 82 (66%) reported being provided with NBS information; however, only half (63; 51%) reported being aware at the time of the NBS that CF was being tested for.

Discussion

The importance of the awareness of a pre-existing knowledge when breaking bad news to parents has been previously emphasised [6]. Findings from this study show that low numbers of mothers recall receiving the information leaflet for NBS. The results also provide useful insights into women's poor pre-existing knowledge about conditions screened by NBS even though at the time of the research there were just six conditions included. For example, just 13% of antenatal women identified congenital hypothyroidism, one of I commonly detected condition on the NBS screening panel in Ireland [7]. Furthermore and most importantly, there was a lack of awareness in parents of children with a positive NBS result (for CF) that their child had been screened for this disease in the first place. This information should be used to inform health professionals' initial discussions with parents about their child's diagnosis, building on parents' pre-existing knowledge. It is important that clinicians are aware that parents may be unaware that their child was screened for the disease they have just been diagnosed with.

The very low numbers of parents who recalled receiving information regarding the NBS programme is disappointing. It would appear that significant work remains to ensure those caring for women in the perinatal period provide this important information. Equally, the lack of clarity amongst parents regarding conditions screened and not screened, is concerning. Parents sign the NBS card, consenting on behalf of their child. It is essential that this informed consent is only obtained if parents receive the appropriate information in a timely fashion to allow them to read the facts and ask questions. Our study suggests this is occurring sub-optimally for a significant proportion of parents.

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Compliance with ethical standards

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Conflict of interest Patricia Fitzpatrick declares that she has no conflict of interest. Catherine Fitzgerald declares that she has no conflict of interest. Rebecca Somerville declares that she has no conflict of interest. Barry Linnane declares that he has no conflict of interest.

Informed consent Informed consent was obtained from all individual participants included in the study.

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