Exploring Parents' & Health Professionals' Experiences of the Introduction of Severe Combined Immunodeficiency (SCID) into the Newborn Screening Programme

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NBS for Severe Combined Immunodeficiency (SCID)

Funded by DHSC /NHSE

- From September 2021, an evaluation took place to determine if NBS for SCID works in practice - Lasting 2+ years and including a 5 year follow up

- Thirteen sites were approached for inclusion in the project at various points during the evaluation. This included sites (n=8) where this new form of screening was being evaluated as well as additional sites (n=4) where clinicians were involved in the care of these babies and a comparator site (n=1) to recruit parents of children who had received an inconclusive result from other areas of the screening programme e.g., CFSPID

- Parents could choose whether they wanted to be involved in the evaluation

Clinical Pathway for NBS for SCID

Ascertain eligible population

Provide information to parents with STFYYB and explain SCID evaluation

Obtain consent

Collect dried blood spot (Day 5) and screen TREC levels

Are TRECS adequate?

YES → SCID not suspected
Provide results to NBFS and CHIS and then to GP and parents (the latter by 6 weeks)
Results sent to family & healthcare team

NO → Urgent immunology referral

Specialist service contacts parents urgently, explains result an arranges OPD (usually the next day)
Email relevant information to parents.

Baby and parents seen in OPD
Baby assessed
Flowcytometry and other relevant investigations performed
Result given to parents usually the same day

Results compatible with SCID

Explain results to parents.
Arrange urgent referral to national specialist services
Initiate any urgent management
Write to GP and provide a copy for parents.

Parents and child seen by specialist team
Results of previous assessment explained
Management plan including investigations explained and initiated
Write to GP and provide a copy for parents.

Results **not** compatible with SCID

Explain results to parents.
Discuss treatment plan
Write to GP and provide a copy for parents.

Investigations and assessment **normal**

Explain results to parents.
Write to the GP and provide a copy for parents.

At each contact, assess general health and immunological state and review treatment options
Ensure appropriate support is in place

CHIS Child Health Information system
NBFS Newborn Failsafe System
STFYYB Screening tests for you and your baby
OPD Outpatient Department
TRECS T cell Receptor Excision Circles

SCREENING	CONFIRMATORY TESTING	ONGOING CARE

PARENT STUDY

Parent Study

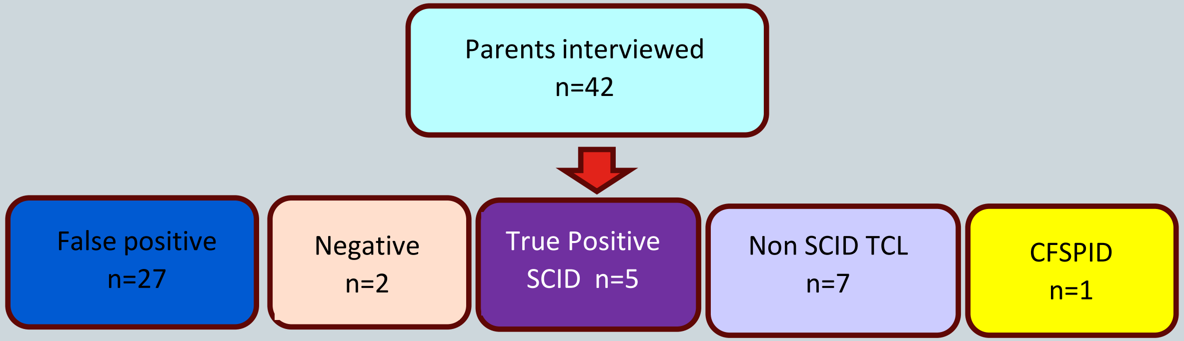
- **Work Package 1: Parents**

A ***mixed-methods study***: Postcards, interviews including journey mapping and questionnaires.

Timeframe: ***Point of screening*** results being returned to parents until the ***child's fifth birthday***.

Data analysis: Simple descriptive statistics and content analysis (postcards), thematic analysis (interviews) and official procedures (questionnaires).

Interview timing	EQ-5D-5L	GAD-7	ITQOL-47	EQ-5D-5L-Y
0-12 months	■	■	■	
12, 24, 36, 48 months	■	■		
60 months	■	■	■	■



Parents interviewed
n=42

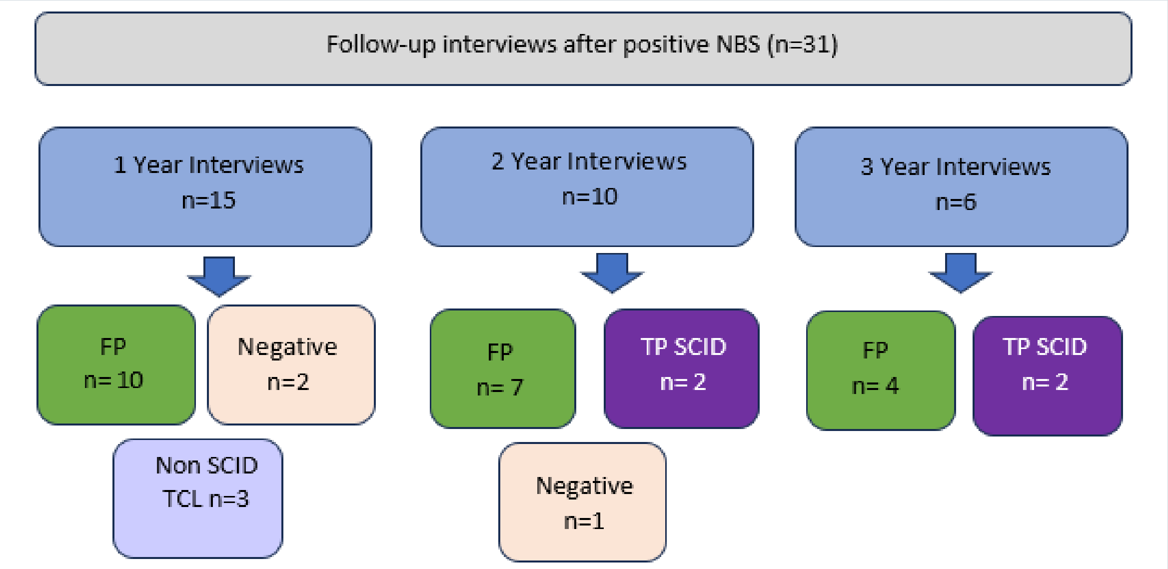
False positive
n=27

Negative
n=2

True Positive
SCID n=5

Non SCID TCL
n=7

CFSPID
n=1



Follow-up interviews after positive NBS (n=31)

1 Year Interviews
n=15

FP
n= 10

Negative
n=2

Non SCID
TCL n=3

2 Year Interviews
n=10

FP
n= 7

TP SCID
n= 2

Negative
n=1

3 Year Interviews
n=6

FP
n= 4

TP SCID
n= 2

The heel prick test

Screening information

- ***Information provision*** prior to NBS and at the time of the ‘heel prick’ was considered ***suboptimal*** and ***impacted*** the ***informed consent*** processes.

‘I didn't realise there was a choice, I thought they were mandatory *“…you just don’t even think about what could happen as a consequence of those test results and* ***you’re probably not even listening, because you don’t expect anything to be wrong****…” Mother, SCID, P18*

- Parents recollection of the provision of pre-screening information was variable
  - Only a few recalled receiving information about screening/ conditions being screened for
  - None recalled the SCID evaluation being discussed
  - Parents described being unaware of the possibility or likelihood of a positive or false positive result
  - Parents reported feeling falsely reassured about the likelihood of receiving a negative NBS result

- Some parents described a sense of optimism bias i.e., that a positive NBS result happened to other people rather than themselves.

*“I was aware that these illnesses and conditions existed, but you always live in a state of…****you're aware of them but I think you always believe they're never going to happen to me****.” Mother, SCID, P45*

Communication of the initial positive NBS result

Communication of the positive screening result for SCID - Inpatient

- Inpatient
  - Distress associated with being informed of their child's positive NBS result for SCID was exacerbated for parents of babies who were inpatients in the hospital setting (e.g., in the neonatal intensive care unit).

*"**I was on my own**…very scary, because **you're miles away from home** really [admitted to neonatal unit due to extreme prematurity] and **you've got nobody to talk to** and, you know, share your concerns and your worries with and that's really hard, because she was very ill anyway, so touch and go at the time. So, to have that on top was worrying…shock, I think, because we felt, you know, really unlucky because, **'How can she have that when she's already fighting with what she's got?'**" Mother, False Positive, P7*

Communication of the positive screening result for SCID - Outpatient

- Outpatient
  - Parents described the communication as unexpected, being from a private/withheld number, sometimes being received while in a location/position that was not amenable to answering the call, frequently being communicated to one parent while they were on their own meaning the onus was placed on them to relay this information to their partner/significant other.

  - Parents described finding it hard to reconcile how, with no known family history of genetic variants, their seemingly well baby could be at risk of a potentially life-threatening condition if not detected and treated promptly.

"I didn't take it seriously enough 'cause ***my default assumption was everything is going to be fine. Like, she looks healthy, [mum] and I are both healthy****." Father, Non-SCID TCL, P68*

Communication of the positive screening result for SCID – The immunology team

- Receiving the initial positive NBS result for SCID from a member of the immunology team could highlight the potential seriousness of the NBS outcome

*"I realised that **it must be something fairly serious for the doctor to ring me from the hospital to need to see him the next day.**" Mother, SCID, P45*

- Speaking to a member of the immunology team was viewed favourably in terms of providing reassurance, being able to answer any immediate questions and discussing the possibility of a false positive result.

*"**The lady at immunology in [hospital] explained the process from end to end** of what we would be expecting, why we needed to come in the next day, why it was so serious, and that we would also be getting results the next day…we should know by the time we left the hospital…**she was really clear on what the next steps would be**." Father, Non-SCID TCL, P68*

Confirmatory Testing

Waiting for the immunology appointment

- Parents reported that waiting for the immunology appointment, although this was less than 24 hours, was extremely distressing

*"…**it was only 24 hours but at the same time it felt like an eternity** just waiting to hear if she was okay." Mother, Non-SCID TCL, P74*

- Despite being advised not to seek further information online about the positive NBS SCID result, all parents did so

*"…you go on and google, **you get all the worst-case information**, and I think that was the scary part because it tells you all the worst things." Mother, False Positive, P26*

- Parents suggested the use of leaflets to provide up to date and accurate information that they could trust
  - *"…**leaflets would be a massive help**. Because sometimes, I can go to appointments…**you're using all these big words, it goes in one ear and out the other**…leaflets or websites-, somewhere you can reread the information that you've just been told…it's a lot of information to obtain." Mother, Non-SCID TCL, P73*

False Positive Outcome – immediate impacts

- For most parents, a false positive confirmatory result was met with an immense sense of relief and gratitude that their child had been screened despite the additional anxiety the process had caused.

*"**Just so relieved**…you feel like you could collapse 'cause you're just so relieved. You know, **you think your life could be completely different, to then hear it's going to be alright and everything's gone back to normal**… just so relieved, so happy…just go home and pick up from where you left off." Mother, False Positive, P34*

- Despite this, parents frequently commented on the impact of their experience on their mental health

*"…there were occasions I think where, **I motor on and I'd be fine, a couple of months after [baby] was discharged, and then there would be something triggering like a triggering piece of music we'd heard or something, and I would just burst into tears…follow up would have been good**. Just because I think, obviously the false positives, they're not seen as important" Mother, False Positive, P14*

False Positive Outcome – Residual concerns

- Some parents described residual concerns which led them to taking additional steps to isolate their child for fear of them acquiring an infection

*"We'd never to take her to soft play or, baby groups while she's this young. I think **we're just too scared**... In case she gets an illness or something, and she never bounces back from it... I'd never take her to a shop on, like, Saturday afternoon. I would only go of, like, a night-time if it was a bit quieter." Mother, False Positive, P4*

- Other parents were reassured by the fact that their child had had numerous infections and remained well which they understood to mean that their child did not have SCID.

*"**He's in great health…his immunity is really good**…He goes to nursery four days a week and **a couple of occasions there's been, like, a cold that's wiped people and he still keeps going** because he seems to be fine with it. Yes, he seems really happy and healthy." Mother, False Positive, P14*

- Most parents felt that as time went on, they thought about the SCID NBS experience less and less.

*"…maybe for a few weeks after…we'd thought about it a lot. But **I don't think I've had any lasting…it's not something I really [think about now]."** Mother, False Positive, P9*

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Article

Parental Views on the Psychosocial Impact of False-Positive Results Following Newborn Screening for Severe Combined Immunodeficiency in England

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Abstract

The project aimed to explore the psychosocial impact on parents of receiving a false-positive outcome following a positive newborn bloodspot screening (NBS) result for SCID for their child. A mixed-methods design was employed using semi-structured interviews and standardised health-related questionnaires (EQ-5D-5L, ITQOL-47, and GAD-7). The participants were recruited from six National Health Service hospital trusts in England involved in the NHS England In-Service Evaluation of Screening for SCID. A total of 22 interviews were conducted with 28 parents. Health-related questionnaire data were collected from 26 of these parents. The interviews were analysed using a reflexive deductive approach to thematic analysis. For the health-related questionnaire data, a comparison of group means against population norms was undertaken using *t*-tests with unequal variances. The findings from the interviews showed that receiving a false-positive outcome following a positive NBS SCID result could cause parents to have an enhanced view of their child's vulnerability in the short term. However, negative sequelae were largely mitigated as parents viewed their child's exposure to 'normal' infections as evidence of a functional immune system. The health-related questionnaire data showed that the parents had significantly worse health than the population norm (as indicated by EQ-VAS: $p = 0.0296$); however, all the other measures were non-significant. More research is needed to explore the potential longer-term psychosocial impact of a false-positive screening result for SCID on parents beyond their child's first year of life.

Keywords: severe combined immunodeficiency; SCID; newborn screening; false positive; parents; psychosocial; quality of life; mixed methods

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True SCID Outcome – Immediate impacts

- Parents described an immediate sense of shock and emotional paralysis upon receiving the news, with some recalling feeling as though their life had abruptly stopped. This initial reaction was often accompanied by deep fear and uncertainty about how to interact with their baby and fear for the future.

“I couldn't believe it. **It was like my life had just stopped**…I just thought, oh my gosh, **I cannot touch you**, **I can't go near you**, I don't know what to do…it was a bad moment.” Mother, SCID, P45

- Parents reported adopting defensive behaviors, including social withdrawal and restricting daily activities, as they sought to minimize any potential risk to their baby. This had the potential to impact on several areas of their lives including work, social, and family life.

“…**we just went straight on the defensive. Barricaded ourselves in** …so obviously **I can’t go into work**…we pretty much **battened the hatches down**, **didn’t let anybody come and see her**…we didn’t want…to be near anybody, really, just in case we caught anything…” Father, SCID, P19

True SCID Outcome – Residual concerns

- Parents reflected on the challenges of leaving the security of the clinicians they had come to trust following treatment.

*"…then we got back to [home location], I found that a little bit tricky because **I'd got so used to the team in [site offering transplant] being there, [baby] being protected on the ward**, that then to come back and take him back to [home location hospital]." Mother, SCID, P45*

- This was exacerbated by a perception that health care professionals outside the immunology team did not have the knowledge required to care for their baby in the same way.

*"…**the knowledge of SCID amongst [other HCPs] isn't there.** When we've come back to see GPs or doctors at [other site not in evaluation], it's like they are just not aware of this condition, which is like frightening isn't it?." Father, SCID, P46*

- One parent reflected on the impact of their SCID journey

"**You kind of wish that that would be recognised about what you've been through and not just be classified as normal**. Some doctors would walk in the room and just be like it's normal now. What are you doing here? Is he sick? He'll get sick and he'll get better…**everyone's extremely positive about him, but there is always a clause** of, but you know he has got SCID. We've not changed, [son], we've given him a transplant and that transplant hopefully will live with [son] for a very long time. But **my understanding is we're not cured**." Mother, SCID, P45

Non-SCID TCL – Immediate impacts

- Similar to SCID, parents with a non-SCID TCL outcome for their child, described feelings of fear and distress

*"**I panicked** then… we were sort of **shell-shocked**… this is **absolutely horrific**, like what are we going to do? **We might lose our daughter,** it was just **horrendous**." Mother, Non-SCID TCL, P59*

- Parents assessed the severity of the outcome based on their perception that, although their baby had an immunological condition, it was not as serious as a SCID diagnosis.

*"…the overriding feeling was just **pure relief that it was not SCID**…. **not as serious as if it was someone with SCID**." Father, Non-SCID TCL, P68*

- Parents described struggling to enjoy their initial experience of parenthood due to persistent uncertainty about their baby's health. This also had the potential to affect their mental health

*"I've not enjoyed it [maternity leave] at all. **I've not been able to enjoy it because I've not known since she's been born if she's okay…it's definitely affected my mental health 100%.** I would say **more so me than my partner's because…I'm here 24/7 just me and my thoughts, you know what I mean, on my own**…[my] mental health I would say is poor at the minute…it does weigh you down eventually." Mother, Non-SCID TCL, P74*

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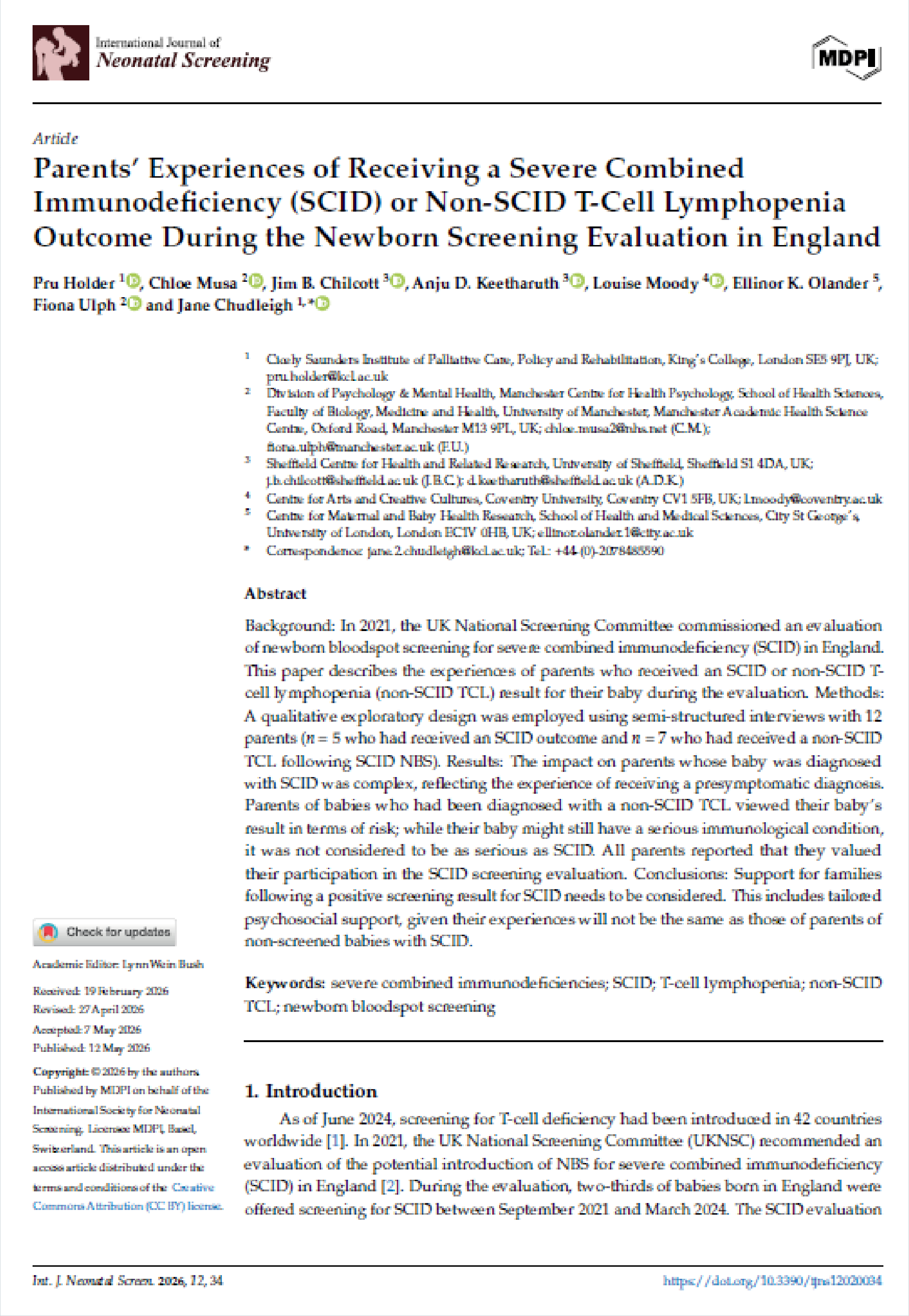

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MDPI

Article

Parents' Experiences of Receiving a Severe Combined Immunodeficiency (SCID) or Non-SCID T-Cell Lymphopenia Outcome During the Newborn Screening Evaluation in England

Pru Holder [1], Chloe Musa [2], Jim B. Chilcott [3], Anju D. Keetharuth [3], Louise Moody [4], Ellinor K. Olander [5], Fiona Ulph [2] and Jane Chudleigh [1,*]

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Abstract

Background: In 2021, the UK National Screening Committee commissioned an evaluation of newborn bloodspot screening for severe combined immunodeficiency (SCID) in England. This paper describes the experiences of parents who received an SCID or non-SCID T-cell lymphopenia (non-SCID TCL) result for their baby during the evaluation. Methods: A qualitative exploratory design was employed using semi-structured interviews with 12 parents ($n = 5$ who had received an SCID outcome and $n = 7$ who had received a non-SCID TCL following SCID NBS). Results: The impact on parents whose baby was diagnosed with SCID was complex, reflecting the experience of receiving a presymptomatic diagnosis. Parents of babies who had been diagnosed with a non-SCID TCL viewed their baby's result in terms of risk; while their baby might still have a serious immunological condition, it was not considered to be as serious as SCID. All parents reported that they valued their participation in the SCID screening evaluation. Conclusions: Support for families following a positive screening result for SCID needs to be considered. This includes tailored psychosocial support, given their experiences will not be the same as those of parents of non-screened babies with SCID.

Keywords: severe combined immunodeficiencies; SCID; T-cell lymphopenia; non-SCID TCL; newborn bloodspot screening

Check for updates

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1. Introduction

As of June 2024, screening for T-cell deficiency had been introduced in 42 countries worldwide [1]. In 2021, the UK National Screening Committee (UKNSC) recommended an evaluation of the potential introduction of NBS for severe combined immunodeficiency (SCID) in England [2]. During the evaluation, two-thirds of babies born in England were offered screening for SCID between September 2021 and March 2024. The SCID evaluation

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Impact on relationships, resources and support needs

Impact on families – relationships and resources

- Parents reported mixed feelings about the impact on their relationship

“…**I remember feeling, like, 'This is your fault.' Even though I knew it wasn't his fault**, and it was something way beyond our control, **I remember-, not hating [husband], but essentially hating [husband], thinking, 'This is your fault.'…it was a lot of hatred**.” Mother, Non-SCID TCL, P73

“**We united as parents for the first time** in a time of, like, anxiousness and nervousness; my husband and I have obviously never been through anything like that. He’s our first baby; **it was the first piece of kind of scary news that you get as a parent**. So, I guess **it kind of united us in something we’d never been united in before… you realise, you’re an absolute team**” Mother, Non-SCID TCL, P64

- Potential impacts on siblings were also discussed

“…**leaving [sibling] was heartbreaking**. There was no way [dad] wasn't going to not be with [baby] and me…but **that meant sacrificing [sibling] for we didn't really know how long at that point**.” Mother, SCID, P45

- Isolation also had the potential to affect extended family members, who missed the opportunity to share early moments with the baby.

“…that **lack of contact for them was probably really hard** too because **they’re missing out on those very first moments with their granddaughter** that they wish they could have had.” Mother, SCID, P18

- Parents also discussed the impact of their experience on future reproductive decisions

*“…[husband] said, **'I don't want any more children…because I don't want them to end up like [baby], it's like we dodged a bullet.’** He goes, **'With the next one, we might not be so lucky**.’…” Mother, Non-SCID TCL, P73*

- Parents of babies diagnosed with SCID and non-SCID TCL also discussed the financial impact associated with their SCID NBS experience

“…**we took on a rental property which we never used, but lost money on… and instead of one year off, she had two years off work**.” Father, SCID, P46

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Health-related questionnaires

Health-related questionnaires – false positive

Standardised health related questionnaires were completed with 26 parents (EQ-5D-5L, EQ-VAS, and GAD-7) at different time points.

Mean EQ-5D-5L scores for the study population were below the population norms, (indicating worse parental health compared with the general population), although this difference was not statistically significant at the 5% level (p>0.05).

EQ-VAS scores were significantly lower than the population norms (p=0.03); indicating worse health compared with the general population.

For the GAD-7, parental anxiety levels were higher than population norms (indicating higher levels of anxiety), however this difference was not statistically significant at the 5% level (p>0.05).

Parental Instrument	Sample mean, (SD), [n]	Population norm mean, (SD), [n]	Mean difference (observed minus norm)	p-value
EQ-5D-5L[1]	0.81 (0.20) [19]	0.86 (0.18) [493]	-0.05	0.318
EQ- VAS[1]	79* (15) [20]	87 (14) [423]	-8.00	0.030
GAD-7[2]	4.79 (4.66) [19]	3.20 (3.96) [283]	1.59	0.163

Health related questionnaires cont'd – false positive

There was good evidence of reduced HRQoL within the first year for parents and infants compared to population norms.

There was also strong evidence of reduced general health perceptions at one year post birth compared with population norms (p<0.01).

There was also strong evidence of lower parental emotional impact scores compared with population norms (p<0.05).

Differences within all other domains were not statistically significant at the 5% level (p>0.05).

ITQOl-47 domain	Sample mean, (SD), [n=26]	Population norm mean, (SD), [n=493]	Difference (observed minus norm)	p-value
Physical abilities[1]	90.39 (22.80)	96.06 (15.88)	5.67	0.241
Growth and development	89.62 (18.77)	96.02 (11.57)	6.40	0.080
Pain	74.52 (20.46)	76.7 (18.23)	2.18	0.599
Temperament and moods	80.29 (12.05)	83.25 (12.30)	2.96	0.233
Combined behavior[2]	79.58 (13.33)	79.42 (12.04)	-0.16	0.977
General health perceptions	64.10** (19.29)	81.21 (13.26)	17.11	<0.001
Parental emotional impact	82.21* (18.26)	92.02 (15.56)	9.81	0.012
Parental time spent	94.23 (12.20)	91.41 (19.06)	-2.82	0.275
Family cohesion	87.12 (18.50)	81.69 (20.62)	-5.43	0.158

Health care usage

Questionnaires

- n = 84 completed questionnaires with data from 30 babies

- n = 30 babies included n = 20 who had received a false positive screening result, n= 2 with a negative screening result, n= 3 with a positive NBS result that led to a diagnosis of SCID, n= 4 with a positive NBS result that led to a diagnosis of non-SCID TCL and n= 1 with a diagnosis of CFSPID

- There was no clear pattern relating to the use of hospital and primary care services between participant groups of over time[1]
- There was no pattern for the number of contacts with all health services and parent-initiated health service use[1]

- The highest mean costs for all services were incurred by babies with a diagnosis of non-SCID TCL

[1] The number of observations was very low for all groups except the false positive group

Parental views of NBS for SCID

Parents views of NBS for SCID

- Parents were overwhelming in favour of NBS for SCID

*"…although it was stressful and we were worried and really anxious about it, **I wouldn't have changed it**, I wouldn't have had it any other way. I think it's really important because if he would have had SCID and, you know, this trial didn't exist, it wouldn't have been picked up." Mother, False Positive, P38*

*"**I'm just so grateful** that when I fell pregnant, it came in that month and so she was then picked up obviously when she had the heel prick… I'm so grateful that the [SCID evaluation] was in place when she was born." Mother, SCID, P18*

*"**I got given a golden ticket when it came…[Baby] got given a golden ticket when it came to the NHS because there wasn't a single day where [Baby] was not put at the centre**…" Father, SCID, P46*

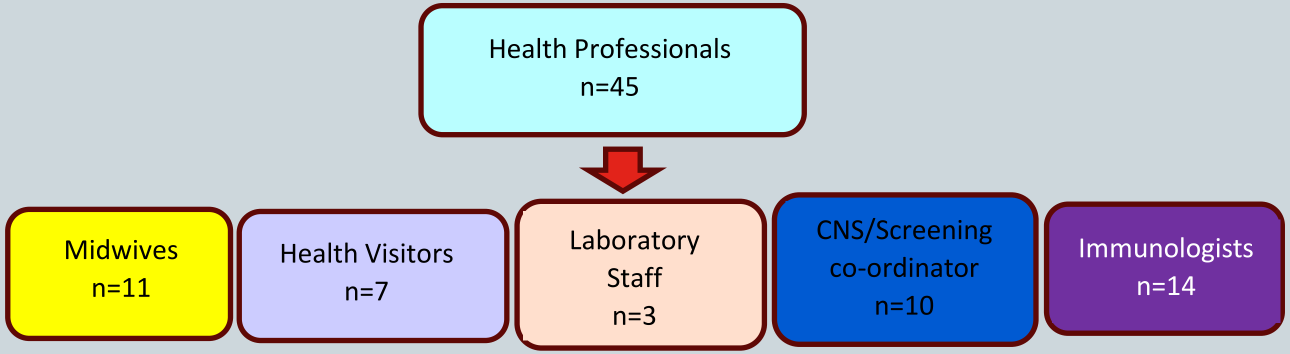
HEALTH PROFESSIONAL STUDY

Health Professional Study

- **Work Package 2: Health Professionals**

A ***qualitative interview study***

Data analysis: Thematic analysis.



Health Professionals
n=45

Midwives
n=11

Health Visitors
n=7

Laboratory
Staff
n=3

CNS/Screening
co-ordinator
n=10

Immunologists
n=14

Obtaining the NBS sample

- Midwives and health visitors were familiar with providing information about NBS and were confident in obtaining informed consent for NBS and for the SCID evaluation.

*"…**we give as much information as we can**, and when we do go to the house, we also again, discuss again, **are you consenting for everything including SCID**, **have you read the information, do you understand what you're consenting**? And if they say, yes, then we go ahead." HV03*

- Midwives and health visitors talked about providing additional information specifically about the SCID evaluation to families to ensure informed consent had been obtained prior to the sample being taken.

*"So, **we would just explain to parents that, you know, it was a research project**, it was an additional blood test, it was being added onto the routine screening if they were happy for us to include that, explaining was SCID was, et cetera." MW05*

- Other clinicians expressed concerns that while they believed parents were able to consent to the procedure being performed (i.e., obtaining blood from the child), this did not necessarily constitute informed consent due to a lack of understanding about what was being screened for.

*"I think they have it but **I don't think they know what they're consenting to, no**. And that's been a concern for me as a midwife when I was completing the test as well. Like I said **they proffer their baby's heel up because they know that it's happening because that often is embedded. But whether or not they truly understand the implications**." HV07*

SCID via NBS

- The introduction of NBS for SCID also led to changes in the clinical presentation of affected babies; most children would be asymptomatic at the point of presentation to the clinical team instead of parents meeting the immunology team to gain answers for their sick child.

“**They all come to us very upset, very confused**, yes, find it very difficult… **one of them sat and cried for four hours, like, really angry, 'You're carrying research out on my baby**.' She just was so confused and overwhelmed…**All of a sudden they think their child's got a condition they might die from, they don't see the positives of, you know, if we know what's wrong with the baby we can make it better soon** but I think **it's what everyone's said all along in working up to the newborn screening was, 'You've got a really healthy well-looking baby,'** unless they're in NICU, **and then you're about to tell them they've got this devastating disease**...**The newborn screening ones just think it's a complete shock because the child's not been sick**…But also **if you have a baby in your arms that looks well and tell them they might have to have chemotherapy and a bone marrow transplant, it doesn't tie in**. What they're seeing doesn't tell them that's what is needed.” CNS06

Communicating the positive NBS result

- The unexpected contact with families made it vital that the content and delivery of the message did not cause unnecessary distress to the family. Clinicians discussed steps they took to mitigate this:

*"…**say congratulations** to, first, on the birth of the baby. **I tell them who I am, I tell them where I work, I explain who [paediatric immunologist] is,** and then **I talk to them about the heel prick** that they've had. And I ask them if they remember what that's for, and then we talk about what that's for and then **I'll say to them unfortunately that's thrown a positive result, so we would like to invite you to come in**, you know, either in the afternoon if it's a morning phone call, but often it's been the day after…and then **I am going to send you some information now from the NHS website via an email**. And **then I go through a whole lot of contact details with them**, so I'd get the parents' telephone, names. I take the parents' names, address, telephone numbers, date of birth et cetera, et cetera, and then we meet them the day after." CNS05*

- Many clinicians spoke about being keen to discuss the possibility of a false positive result as early as possible to ensure parents were aware that the screening result was not definitive.

*"**I explain the concept of screening in general. It's not a diagnostic test**…**I normally give them what I think are the likely outcomes of a flow test**, which is either, one, it needs repeating because it's not worked, which thankfully hasn't happened that much actually, and, two, **they've got SCID**, three, **they've definitely not got SCID** and it's all fine, or, four, **it's not SCID but we need to repeat it in a few months' time**, we might have picked up another immune disorder by accident which would be a good thing again to pick up something else by accident…" IMM06*

Challenges

- Communication between clinical departments (e.g., neonatal units and immunology departments) within each hospital and between different hospitals could also be challenging until new ways of working had been adopted.

*“The **biggest thing to work out is whether or not they're at home or they're in hospital**. For me, what I do in that case is **if they're in hospital, I will ring the hospital, neonatal unit, and try to get some clinical details about the patient and also work out whether or not they're still in and work out whether or not they may have died**.” IMM14*

- Communicating with the primary care team when the baby may not be registered with a GP could be challenging as it would not be clear who the clinic letters should be sent to.

*“I suppose, is **who do you send the letters to because a lot of these patients haven't been registered with a GP** and clearly, they need to be sent out. And my secretary's lovely but sometimes…she's sent it to unnamed GP, doesn't go very far, does it? And literally, you've got letters that were addressed to the parents with an unknown GP.” IMM01*

- Language and communication barriers could also pose an issue particularly in relation to medical terminology. In addition, clinicians felt that consideration needed to be given to parents who may have a learning difference.

*“I mean, **learning difficulties** as well. We have some parents who need lots of support with just doing the basics...So, **getting that information across in a simple way that they are going to understand and be able to make that choice** whether they want their baby to have the test or not can be a bit of a challenge……with some of the **language barriers**, I know **it's sometimes really difficult to know that they're not just nodding and agreeing because that's the easiest thing to do**. So, trying to work out, 'Have you understood this?’” MW09*

Support for families

- Support following the positive NBS for SCID was seen as vital to the success of the programme and was approached in various ways. This included both financial as well as psychosocial support.

*"…**what I'd do if someone said, 'I can't afford to come up to London,'** and **whether or not we can get funding** for them to doing that, which I think we probably can, but that's a bit of-, say clarity around that probably, but that must be part of the newborn screening programme and protocol anyway." IMM13*

- In terms of psychological support, setting up a peer support programme was considered to be advantageous since there would be a limited number of families with similar experiences given NBS for SCID was still in its infancy.

*"…**we're able to set up a little network for those new families as well and put them in touch with the families that have already been through**…we've developed this network of parents and families as well that have been through it. So, we're able, to newborn babies that we're receiving, **set them up with sort of a buddy, if you like, of families so they can talk them through their experience**." CNS05*

Impact on health professionals

- Midwives and health visitors indicated that the SCID evaluation had not significantly affected their workload and would therefore be feasible if it were to be introduced nationally. This contrasted with other members of the multidisciplinary team such as laboratory staff and members of the immunology teams involved in NBS for SCID, who all felt SCID screening had impacted quite significantly on their workload.

*"…**to do the SCID test essentially is probably two-thirds of a day**. So, you know, you have to punch, you have to prep, that takes a few hours. Then you have to run it, and that takes an hour. Then you have to look at the results, and report it, and whatnot. So…**it means that you actually need a person on it every day.**" LAB01*

*"**I didn't realise how much it would impact on the amount of staff requirement and the time needed**… the fact that **we have to pick up additional work,** so to be **available at the drop of a hat** to do the initial telephone consultation and then **see the family on the day after**." CNS04*

- This additional workload was primarily associated with the number of false positive results that had been detected during the screening evaluation that would not have formed part of the immunology teams' workload prior to the evaluation.

- Aside from the additional workload, many clinicians spoke about the impact communicating the positive NBS result for SCID had on them due to the distress caused to parents because of the unexpected nature of the initial telephone call.

*"…**it's not an easy call and I don't look forward to it**, but it is part of my job, and I always have to keep in focus the reason that we're doing it. And I think that's probably the same, we've discussed it as a team, and **I think we've all felt the same because it's horrible making these calls**…" CNS09*

- Clinical teams acknowledged the importance of ensuring anyone who spoke with families about the SCID NBS result were experienced and able to answer questions as they arose. This included anyone who was involved in the SCID screening process such as midwives, health visitors and GPs as well as members of immunology teams.

*"…you just need to be **an experienced professional** that's **used to, I suppose, delivering bad news** in some respects, **whilst it's not delivering bad news, I think it's probably initially perceived in that way**. So, I think as a health professional, no matter what your designation is, **I think you need to have lots of experience**" CNS05*

Health professionals' views of SCID NBS

- Although the purpose of the evaluation was to identify children with SCID before they became symptomatic, children with non SCID TCLs were also identified, and clinicians could also see the benefit of identifying some of these children.

*"…we've got a few DiGeorge patients who've had thymic problems who've been picked up, who-, in fact, we had one with Downs Syndrome as well…**they've got into the system to be reviewed and look at their immune system a bit quicker than normal**. Normally, they're picked up because they've got a heart problem or they do some testing, in a few months, a few years down the line, we get-, **so hopefully that can stop them getting problems. So, I think it's helping in some of those patients**." IMM01*

- All health professionals felt there introducing SCID into the NBS Programme would reduce morbidity and mortality in children with SCID but also reflect what other screening programmes were doing internationally.

*"Well, **I'm hoping it's going to reduce morbidity and mortality in my patients**…It gives people a better understanding of the progression of immune deficiencies…But **the principal thing is to reduce mortality**. "**It's worked well in the US. It works well in other European countries, and I don't see why it can't work well in the UK**." IMM02*

- For some this was because they had first-hand experience of caring for babies with SCID or Non SCID TCLs who they felt would have greatly benefited from NBS for SCID.

*"You know, **I just think it's such a horrible disease that if it can be picked up, why not**, yes? **If you can make the outcome better-**, because that's the problem, **we've had other children where, you know, they've been picked up at six months old, when their mum's antibodies have dropped off. They've ended up with significant illnesses**." CNS03*

Key Recommendations

- Based on their experiences, ***parents would recommend that screening for SCID should be included*** in the national NBS programme.
- Consideration needs to be given to the ***management of screening for SCID in pre-term infants*** due to the additional anxiety they experience and the high false positive rate.
- Screening information resources provided antenatally and at the time of screening and ***consent processes*** need to be revisited so that the potential outcomes of screening and the meaning of these outcomes are clearer.
- ***Support (financial and psychosocial) for parents*** following a positive screening result for SCID requires careful consideration.
- ***Funding for laboratory and immunological teams*** needs revisiting if screening for SCID is added to the national NBS programme.
- Consideration should be given to the ***provision of information to parents*** following screening in relation to parent-initiated care that is specific to their child's designation/diagnosis.
- The ***impact of screening on parental health-related quality of life***, should be recognised in any decision relating to SCID screening. Whether this could be incorporated into economic modelling is less certain, as is specific data that would be used to parameterize such an analysis.
- Consideration should be given to ***using electronic record linkage to assess the extent and cost of patient-initiated care relating to SCID screening results***. Such an approach should also be considered for any future expansions of NBS to other conditions.

Thank you

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