

1 **Parents' responses to receiving sickle cell or cystic fibrosis carrier results for**
2 **their child following newborn screening**

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22 Running title: Effect of newborn carrier results on parents

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Abstract

Universal newborn screening for sickle cell disorders and cystic fibrosis aims to enable the early identification and treatment of affected babies. Screening can also identify infants who are healthy carriers but it is unclear what effect being informed of their baby’s carrier result may have on parents. Semi-structured face to face interviews were conducted with a purposeful sample of 67 family members (49 mothers, 16 fathers, 2 grandparents) of 51 infants identified by universal newborn screening as carriers of cystic fibrosis (n=27) and sickle cell (n=24), across all health regions in England. Data were analysed by thematic analysis with subsequent respondent validation.

Untoward anxiety or distress among parents appeared influenced by how results were conveyed, rather than the carrier result *per se*. Parents who had more prior awareness of carrier status or the possibility of a carrier result assimilated the information more readily. Being left in an information vacuum whilst awaiting results, or before seeing a professional, led some parents to fear their child had a serious health condition. Parental distress and anxiety appeared mostly transient, subsiding with understanding of carrier status and communication with a professional. Parents regarded carrier results as valuable information and sought to share this with their families and to inform their children in the future. However parents needed greater support after communication of results in considering and accessing cascade testing, and negotiating further communication within their families.

Keywords:

newborn screening; qualitative; communication; genetic carrier; sickle cell; cystic fibrosis.

52 **Introduction**

53 Newborn bloodspot screening is regarded as a significant public health achievement in the
54 developed world.¹ Newborn screening (NBS) models vary internationally in terms of the
55 diseases screened for, and the consent and communication models. In 2007, NBS for Cystic
56 Fibrosis (CF) and Sickle Cell Diseases (SCD) was implemented nationally across the whole of
57 England and is offered to parents of all newborns using an informed consent model.²
58 Screening guidelines and training materials stress the central role that information provision
59 plays in parents' ability to consent.^{3 4} Yet, evidence suggests several barriers to this
60 communication including parents failing to appreciate the personal relevance of screening
61 information,⁵ information overload during pregnancy,⁶ and conflicting familial or existing
62 personal knowledge.⁷ The challenge of creating antenatal communication protocols which meet
63 parents' information needs, provided when parents are able to assimilate the information and
64 appreciate the personal relevance is recognised.⁸

65 Although the rationale for informed consent is based on respecting parents' autonomy,⁹
66 information provision may influence how parents respond to NBS results. In relation to
67 metabolic disorders, NBS carrier or false positive results can trigger anxiety if misunderstood,
68 or if parents with children undergoing further testing related to false positive results are not
69 provided with timely and adequate information.^{10 11} Such anxiety has been linked to impaired
70 relationships with the baby¹¹⁻¹³ and lasting effects of stress during childhood.¹⁴ This suggests
71 that earlier fears that newborn PKU screening may trigger the *vulnerable child syndrome* for
72 some parents^{15 16} may have been realised. For these parents multiple and/or specialist health
73 professional consultations are often needed to allay such anxiety.¹⁷ Research in the US has
74 suggested that inadequate preparation for NBS results can exacerbate anxiety and shock for
75 parents informed their child is a genetic carrier of cystic fibrosis or sickle cell¹⁸ and that the
76 content of information given can affect parents' reactions.¹⁹ This paper reports a qualitative
77 exploration of the impact of receiving cystic fibrosis or sickle cell carrier results on parents as

78 part of a wider cross-sectional study of practice, methods and experience of communication
79 following newborn screening across England.⁶ addressing an identified gap in the literature
80 ²⁰specifically since informed consent is seen as a way of palliating the harms of disclosing
81 carrier status via NBS^{21 22} and England is one of the only countries that uses this model

82 **Methods**

83 *Recruitment*

84 Recruitment sites, and parents informed of their baby's carrier results were purposively
85 sampled to ensure the wide range of differing methods of communicating carrier status
86 information in England were captured.²³ Parents were sampled across all health regions in
87 England in 2007-2008. Health professionals distributed study packs to parents who had
88 received newborn carrier results. Parents who returned "consent to contact" forms were
89 contacted by a researcher to discuss the study, and arrange an interview if appropriate, with
90 informed consent undertaken at interview. Parents who returned a translation request form
91 were contacted with a professional interpreter and a three way telephone conversation was held
92 to discuss participation. Following interviews parents were informed they would receive a
93 research summary, and asked if they were willing to be contacted again for discussion and
94 validation of preliminary findings.

95 *Data generation*

96 Semi-structured interviews were conducted in participants' homes, using professional
97 interpreters where necessary for non-English speakers. Parents were encouraged to relate their
98 experiences from when they were first aware their child was to be screened and interviews
99 flexibly followed a topic guide which was modified following initial interviews. Interviews
100 were recorded and transcribed verbatim in English. Interview transcripts translated from other
101 languages were checked by independent interpreters for accuracy and equivalence of meaning.

102 *Analysis and validation*

103 Data generation and analysis were iterative, with each informing the other using techniques
104 derived from grounded theory. This was undertaken by researchers with health psychology and
105 social science backgrounds, with the wider team (clinical primary care/genetics academics)
106 contributing to development of themes from a multi-disciplinary perspective. Data were
107 managed within NVivo software. Further theoretical sampling and data collection sought
108 deviant cases to extend and challenge the analysis until saturation was achieved. This was
109 followed by a respondent validation exercise in which all participants were sent a summary of
110 the findings, translated where appropriate. Participants were then invited to participate in semi-
111 structured telephone interviews to comment on the findings, reflect further on their experience
112 and add any further views.

113 *Ethical approval:* An NHS multi-centre research ethics committee (West Midlands) granted
114 ethical approval to the study.

115 **Results**

116 Interviews were conducted with 67 family members of 51 infants identified by newborn
117 screening as carriers of CF (27) and SC (24). Interviews were conducted with parents across all
118 the nine health regions of England, including high and low prevalence areas for sickle cell
119 disorders. Eight interviews were conducted with professional interpreters (French =3; Bengali
120 = 3; Portuguese =2). Further semi-structured telephone interviews, for respondent validation,
121 were conducted with 17 parents, three to fourteen months after their initial interview.
122 Characteristics of participants and their infants are summarised in **Tables 1 and 2**.

123 *[insert tables 1 and 2 here]*

124 Parents in the sample experienced the full spectrum of methods of communication of carrier
125 results in use in England at the time of the study (see **Tables 3 and 4**).

126 *[insert tables 3 and 4 here]*

127 Four principal aspects of receiving a carrier result following newborn screening which affected
128 parents were developed from data analysis: the impact of knowing the carrier result; effect of
129 the process of communication; cascade testing within the family; and sharing of carrier
130 information with extended family.

131 ***Impact of knowing carrier result***

132 With the exception of one parent who was uncertain, all parents felt it was important they had
133 been informed of their child's carrier result. This was regarded as valuable new information
134 about, and for their child, gained fortuitously.

135 *I am happy to know it, because it didn't involve any additional tests you know*
136 *which is always nice for a little baby and it's, I think, it is valid information.[...] I*
137 *could even argue that there is a need to know...*

138 **(#33, mother of an SC carrier; low SC prevalence area)**

139 Most parents were not concerned or distressed by the carrier result *per se*. Most recognised this
140 was not "bad news" and it had no direct adverse effect on their child.

141 *I don't think [parents] should be upset about this, it is very important not just for*
142 *them, but for the child involved as well.*

143 **(#3, mother of an SC carrier; high SC prevalence area)**

144 Some parents felt positively reassured when they heard their child was a carrier, particularly
145 those parents awaiting the results of a second IRT blood test for CF (* see explanatory note below),
146 who had worried about their child being affected by the condition.

147 *When they said she didn't have it [affected with CF] I was 'away', that was it, I'd*
148 *closed down, that was fine [...] for me it was like "I can switch off now".*

149 **(#37, Mother of a CF carrier)**

150 Some parents underlined how they felt identification of carriers, and being informed of this,
151 was helpful.

152 *[...] people knowing that they've got this status is really important, because there*
153 *might be less people with the sickle cell disease in the future for people having that*
154 *knowledge. [...] hopefully the majority will take it on board and think "I don't want*
155 *my child to be ill and so therefore I'm going to be careful" you know and that's*
156 *surely important.*

157 **(#35, mother of an SC carrier; low prevalence area)**

158 When asked to reflect on the possibility that technology might ensure that carriers were
159 not identified via NBS, parents did not support this:

160 *If you stop telling people that they're carriers, that's got to be going in the wrong*
161 *direction because at least if you're a carrier you know before she has children*
162 *hopefully they'll talk about it and he'll go and get tested so they know what the*
163 *likelihood ... (#37, mother of a CF carrier, first child)*

164 Although most parents appeared to understand the 'benign' implications of their baby's carrier
165 status, a minority of parents (four families, two of SC carriers, two of CF carriers) remained
166 unduly concerned about their child's health after receiving results. They expressed negative or
167 ambivalent reactions to knowledge of their child's carrier result such as guilt or had prominent
168 concerns about the child's health.

169 *So I still worry and I just think "She's still little, anything she gets now could bring*
170 *something else out" you know you've still got it in your head even though it's been*
171 *cleared up for you, you still worry a little bit that how can something genetically*
172 *just be as black and white as that ? (#40 mother CF carrier)*

173 ***Effect of process of communication***

174 Some parents experienced distress or anxiety linked to their experience of newborn screening,
175 in particular for CF (12/27 CF carrier families, 2/24 SC carrier families). This ranged from
176 mild, short lived anxiety, to more profound distress. This could manifest in reducing the child's
177 interaction with others, arguments between couples, alteration of life plans and an inability to

178 conduct tasks of daily living such as going to work or socialising. Most parents who reported
179 such distress reflected that *how* carrier result information had been communicated to them had
180 been the cause “... *to some extent these things are often not so much about what you tell me*
181 *but how you tell me*” (#55, **mother of an SC carrier**). This was particularly salient in relation
182 to communication around the time of the repeat blood sample* in CF screening.

183 **Mother:** *I really wanted to get across just how really bad the process was for us,*
184 *but then the relief of eventually getting the results and the relief of knowing that it*
185 *wasn't cystic fibrosis and the problems of looking to know that we're a carrier and*
186 *that our baby's going to be absolutely fine really. It was more the process of the*
187 *actual results you know I think I probably had the hardest time dealing with it*
188 *because it seemed to be never-ending.*

189 **Father:** *It's the length of time, the lack of communication, lack of knowledge.*

190 **(#50, parents of a CF carrier; told would hear within 4 weeks, were contacted**
191 **at 6 weeks on a Sunday by Midwife saying “problem with heel prick test”**
192 **retested that day. Told poor sample, not enough blood. Second sample lost.)**

193 Poor communication that failed to anticipate or address concerns appeared to have a major
194 impact at this time. This included examples of parents of carriers who had become concerned
195 their child was chronically ill or perceived their child as fragile and so limited their interaction
196 with others.

197 *You think your daughter's seriously ill and could die and will need physiotherapy*
198 *all right through her life; will never be able to integrate properly at school and*
199 *you're thinking “Well, what's going to happen career-wise?” because you plan that*
200 *she'll go to full-time nursery and actually she won't be able to do that, so maybe*
201 *we'll have to give up work, [...] so your mind just goes on, and on, and on.*

202 **(#4, father of a CF carrier; parents had been told if they heard nothing in 2**
203 **weeks everything was ok; at 4th week got a phone message that “problem with**

204 **heel prick test". When called told CF test had come back positive and looks**
205 **like daughter may have CF, was unable to give parents further information)**

206 The data suggested that if parents were left in limbo regarding their child's result at any stage,
207 it could lead to rumination and catastrophic thinking. Others simply felt unable to deal with
208 people's enquiries about the results whilst waiting:

209 *[...] I mean it literally reached a point where I said "I don't want to see anybody, I want*
210 *you to leave [name of child] and me on our own and not have to " Because every*
211 *time you're speaking to someone as well, that did know our situation, it would always*
212 *come back to "Why haven't you heard anything?" and they were questions that we*
213 *couldn't answer so it became easier for me to just not deal with anybody at all. I would*
214 *probably say it was probably the worst moment of my entire life...*

215 **(#50, mother of CF carrier)**

216 During this period, some parents had felt depressed, been unable to sleep or concentrate at
217 work, or described a negative impact on personal relationships.

218 *We had two massive blow ups at each other and it was a case of me ...feeling left*
219 *out that is what it was about really. [...] you do feel that isolated, it's unbelievable.*

220 **(#41, Father of CF carrier; parents told "bad news" child is a carrier, but may**
221 **have CF, next day at hospital told child has CF before sweat test conducted, after**
222 **2 sweat tests cleared.)**

223 Fathers also appeared to struggle because at the time of waiting for the second IRT
224 result* for CF many were back at work:

225 *I still went to work but I found myself hiding in my office more often than I normally*
226 *would do. [...] I found it very difficult to manage really just the day to day disciplining*
227 *and just try to I don't know just try to keep it straight really.*

228 **(#50, father of CF carrier)**

229 However further explanation and communication of results in discussion with a well informed
230 professional appeared to have allayed these concerns in most parents.

231 *Until I actually went to the [genetics centre] and that doctor probably explained it*
232 *a lot more clearly, and to me it [feeling of guilt] sort of dampened down and from*
233 *then on .. I could feel myself much more relieved by the whole sort of thing.*

234 **(#40, mother of CF carrier; told needed second sample, but prewarned this is**
235 **common and nothing to be concerned about. Told could be carrier, but no**
236 **further information; sought information on internet and became very upset)**

237 Only three of 38 parents of CF carriers were aware of their own CF carrier status prior to
238 newborn screening of their child. In contrast, 21 of 29 parents of SC carriers were already
239 familiar with their own carrier status through previous or antenatal testing and for these parents
240 experience of distress appeared less common. Prior knowledge of personal carrier status
241 appeared to help parents assimilate the information about their child *“she is just like me, she is*
242 *AC” (#3, mother of SC carrier).*

243 Parents who had reported considerable distress regarding NBS in their original interview later
244 felt, during respondent validation, that they had now come to terms with the experience, and
245 continuing anxiety was rare. This was usually as a result of subsequent direct communication
246 with a health professional. One participant reported continuing occasional concerns when their
247 child, a carrier of CF, became unwell with a cold.

248 ***Considering ‘cascade’ testing within the family***

249 For many parents, knowing their child’s carrier status led them to question, and consider
250 establishing, which parent was a carrier if this was not already known; and to consider the
251 carrier status of their other children. In relation to parents’ own carrier status, the issue of non-
252 paternity was tangentially raised in three cases: parents’ carrier status was seen to offer
253 reassurance of paternity in two cases and in one case an oblique reference was made to the
254 child’s inheritance of a gene ‘skipping a generation’. Some parents had begun to worry if their

255 other children had not been tested and were approaching reproductive age, or were from
256 previous relationships, and how they might broach this with their children.

257 *It's just lingering because I have got two children from my previous marriage and*
258 *if I am the carrier then it would be nice to inform them and tell them, you know,*
259 *what would they like to do about it?*

260 **(#5, father of a CF carrier)**

261 While some parents felt unable to “move on” until testing had occurred for themselves or been
262 offered to other relevant family members, others varied in their desire to do so, and how it may
263 affect their future decisions.

264 *I am of the opinion of I don't want to [know] whereas you [referring to father] are*
265 *more inclined that you do want to know.*

266 **(#19, mother of a CF carrier)**

267 Some parents, however, had experienced difficulties in accessing cascade testing.

268 *My other concern was before my 3½ year old was born they weren't specifically*
269 *tested for this [before universal newborn SC/CF screening] so I wanted to know*
270 *how I got him tested and [the health visitor] had no idea.*

271 **(#13, mother of an SC carrier; sibling subsequently tested)**

272 Others were concerned that despite their newborn being identified as a carrier, they were
273 unable to access cascade testing for their other children until they were older (and had reached
274 the age of consent themselves), encountering no flexibility in the system:

275 *When we told her [sibling of newborn carrier] we said, you know, they've said you*
276 *need to have this test when you're 16 and she just turned round and went “Why*
277 *can't I have it now?”*

278 **(#12, mother of a newborn CF carrier, older sibling aged 10 years)**

279

280 This was a particular source of parents' frustration:

281 ...they won't let me have her tested. The argument is she has to wait until she is 16
282 and classed as an adult or whatever it was [...] but I feel that we are her parents
283 and should be able to be responsible (**#64, mother of CF carrier, sibling not**
284 **tested**)

285 *Sharing carrier information with extended family*

286 Most respondents felt a responsibility to share their newborn's carrier status information with
287 their extended families, but some struggled with knowing who to tell or how to raise the issue,
288 or were concerned about creating anxiety.

289 ... my cousin's girlfriend was pregnant, and [my partner] was saying "You should
290 tell them about the cystic fibrosis thing" and I was thinking "I shouldn't tell them
291 about the cystic fibrosis thing because they've got 9 months of worry and then for
292 them to think the baby will have an abnormality when its born" and I don't think
293 that's fair. (**#37, father of a CF carrier**)

294 Others had no concerns about telling family members and discussed how their extended
295 families appeared to respond positively to the news, by showing interest, getting tested, and
296 understanding that carrier status had minimal health implications.

297 [The whole family came] round for Sunday lunch, we'll sit round the table, we'll
298 discuss it. And it was really nice you know; you don't have to worry.
299 (**#12, mother of a CF carrier**)

300 However respondents also reported negative reactions from their families including relatives
301 avoiding related conversations; and refusing to believe the information in a context of stigma
302 and ignorance about the conditions.

303 ...talked to my mother about it so many times, she doesn't want to listen, it's like [...] she
304 has turned a blind eye to it and I said to her that you have to get all my younger sisters
305 to have them tested and she has turned a blind eye to it and so I have stopped talking
306 about it. (**#3, mother of SC carrier**)

307 Family members also distanced themselves from the issue by blaming the other side of the
308 family. When this occurred between parents it could exacerbate previously fractious
309 relationships.

310 [the wider family] *were just looking at it as a negative, do you know what I mean?*

311 *They were looking at it as if something that ... like really ashamed and it was like*

312 *"Oh none of us are affected". (#37, mother of a CF carrier)*

313 Some families did not wish to pursue cascade testing creating ambivalence among parents of
314 carriers who did not feel it appropriate to dictate to others, but who also had concerns they
315 should be more proactive to ensure this occurred. The lack of family members' engagement in
316 testing could cause some distress.

317 *it came out that it was his family, but you know I have told him, but none of his*

318 *family want to be tested even though his niece is trying to have a baby and... They*

319 *don't really, yeah I begged her but she is not in their family and they don't want to*

320 *be tested for it you know. (#22, mother of CF carrier)*

321 Some felt health professionals should offer advice on how to discuss the issue or become
322 involved in such communication. Where this had occurred, it was appreciated:

323 [the health professional] *was trying to be really helpful she e-mailed [husbands'*

324 *name] the letter [containing the carrier information] so that he could get it to his*

325 *family before and then she posted it out... (#58, mother of CF carrier)*

326 Discussion

327 This study suggests disclosure of CF or SC carrier results following universal newborn
328 screening can have significant effects for parents and wider family members. For many
329 parents, while being informed of their child's carrier status was positive, and this information
330 was valued, there was considerable anxiety and distress following the initial communication of
331 results. However, subsequently, parents appeared capable of understanding and assimilating

332 this information into their lives. This suggests concerns about provoking a vulnerable child
333 syndrome,¹⁵ may be avoided given appropriate communication and support.

334 The process of communicating the result, including messages from health professionals
335 which contained insufficient information, or waiting for information, was not benign and was
336 commonly reported as a key source of any distress, impacting social engagement. This echoes
337 work conducted in the US and Australia in relation to CF newborn screening^{24 25} Our data also
338 suggests sometimes detrimental impact on employment, parental relationships which may
339 already be under some strain with the arrival of a newborn, and in particular on the wider and
340 extended family. This adds to evidence that parents' information needs may not have been met
341 in a timely or adequate manner and that parents may find newborn screening distressing.^{14 26-31}
342 Ensuring parents are not left in an information vacuum at any stage of the screening process
343 may minimise the risk of harmful effects from screening.

344 Although some parents required additional specialist services, once carrier results were
345 understood this knowledge did not appear to have a lasting negative effect on most parents in
346 this study and indeed was highly valued as has been documented previously.^{20 24 32 33} Indeed
347 this study goes further by showing that parents would see changes in protocols to not identify
348 carriers as a backwards step. These findings can inform the ongoing debate about whether to
349 identify carriers or inform parents,³⁴ and conflicting findings regarding the impact of doing
350 so.^{13 25 32} The results also address an identified gap in the evidence regarding communicating
351 SC carrier results³⁵ and whether it is true that the lack of repeat testing in SC screening means
352 there is less anxiety.²⁰ Although it is certainly the case that most of the distress in CF screening
353 centred around repeat testing, there were other issues and anxiety was also found within the SC
354 sample. The issue of involving fathers more in antenatal and neonatal screening has been raised
355 previously³⁶ and the current data highlight the disempowering effects on fathers when
356 communication of results occurs solely with the mother. Although referred to, the issue of non-
357 paternity being revealed by newborn carrier results did not feature prominently with parents

358 interviewed in this study, though it is recognised they were providing ‘public’ accounts of their
359 experiences to researchers.

360 The current findings start to address the paucity of data regarding how and what parents
361 are told regarding NBS results.³⁷ In so doing they underline that communication of
362 information and support following newborn screening is required beyond the infant period, in
363 order to facilitate access to cascade testing and support the challenges of familial
364 communication. The issue of families avoiding communication about carrier information, and
365 inferring blame or stigma has been discussed in relation to antenatal screening for sickle cell,³⁶
366 as have the difficulties in working out who to tell when.³⁶ The current study highlights the
367 same for newborn screening and the current lack of a defined role for any specific professional
368 to provide such support. Given the period of time it may take for families to adapt to this
369 information, this role might, for example, be developed in primary care, as part of continuing
370 care with families, or within community based genetic counselling. The results also highlight
371 that parents’ concerns regarding communication processes for receiving carrier results are very
372 similar to parents who receive positive results,³⁷ and that such issues which have been found
373 when screening is mandatory and information provision is assumed to be minimal are also
374 found in programmes that adopt a fully informed consent model.

375 *Strengths and limitations*

376 A range of methods were used to enhance rigour, and the relevance of study results including
377 purposeful sampling, iterative data generation, analysis to saturation, involving researchers
378 from several disciplines, and use of respondent validation.³⁸ The sample is described in some
379 detail including, for example, information on parents’ own carrier status and social
380 backgrounds, and the diversity of provider methods of communication of carrier results that
381 they experienced in practice. This may aid assessment of the transferability and relevance of
382 the findings beyond the immediate study context.

383 The sample is substantial when compared to previous qualitative work with parents of
384 SC carrier,³⁹ CF carriers^{24 32 40 41} or both types of carrier.⁴² Those recruited also represent a
385 significant proportion of the very small number of newborn CF carriers identified annually in
386 England. Within the practical constraints of study recruitment and duration, the research has
387 successfully included parents' experiences across all health regions of England following the
388 introduction of universal newborn screening for SC and CF. The lack of fathers' perspectives
389 in newborn screening research has caused concern,⁴³ but the current study included fathers
390 wherever possible, ultimately forming a quarter of the sample.

391 While active inclusion of those willing to articulate their experiences is key to
392 qualitative enquiry, we recognise that the experiences and views of study participants may not
393 be typical of all receiving carrier results following newborn screening. Our sample was
394 relatively well-educated, a feature similar to other studies concerning CF screening in
395 particular^{13 44} and appeared relatively genetically literate. Their experiences may however
396 serve to reinforce the identified needs to improve information delivery and continuing support
397 for the wide range of parents who experience newborn screening.

398 Further research should investigate how best to prepare parents for NBS prior to results
399 as this appears to affect the impact of results on parents; and examine parents' attitudes
400 towards, access to and experience of cascade testing and its impact on later reproductive
401 decisions. In parallel, work is needed to develop and evaluate methods to support cascade
402 testing and communication of carrier information with children and families; and capture
403 further experience of families over time to enable greater understanding of longer term benefits
404 or harm of newborn carrier identification.

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412 *Author contributions*

413 FU and TC undertook fieldwork interviews and analysis. JK designed the study, supervised
414 data generation and analysis, and was principal investigator and grant-holder. NQ was grant
415 co-applicant and supported comment on draft revision. FU and JK wrote the paper.

416
417 * **The CF newborn screening protocol in England** aims to identify a maximum of children with CF, while
418 minimising carrier detection. This involves measuring immunoreactive trypsinogen (IRT) levels to identify babies
419 with levels exceeding the 99.5th centile, indicative of CF. Samples from newborns with raised IRT levels undergo
420 DNA analysis to establish if the child has two mutations, and is therefore likely to have CF, or has one mutation
421 and is probably a carrier. Children with suspected CF are referred for a clinical diagnosis. Those with one
422 mutation, or an initial IRT >99.9th centile and no detected mutation, require a second bloodspot test to verify if
423 IRT is still elevated at 3-4 weeks when IRT levels are more discriminatory. In those children with one mutation,
424 where the IRT remains elevated the likelihood of CF is regarded as high and triggers clinical referral, while if not
425 still elevated the likelihood of CF is low and the child is regarded a healthy carrier.

426

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541 **Table 1: Characteristics of parent respondents***

| N= 67 (%) | |
|--|---------|
| Ethnicity | |
| White British | 43 (64) |
| Black African/British/Caribbean | 14 (21) |
| Bangladeshi | 3 (4) |
| White European/Other | 3 (4) |
| Mixed | 3 (4) |
| Asian Thai | 1 (1) |
| Employment Status | |
| Employed full time | 33 (49) |
| Employed part time | 12 (18) |
| Full time parent | 11 (16) |
| Full time student | 3 (4) |
| Full time carer | 1 (1) |
| Unemployed | 1 (1) |
| Not indicated | 6 (9) |
| Highest educational attainment | |
| Degree or higher degree | 22 (33) |
| NVQ, Diploma, A-level, HND or equivalent | 17 (25) |
| GCSE or equivalent (school to 16 years) | 14 (21) |
| None | 1 (1) |
| Not indicated | 13 (19) |
| Parent carrier status | |
| Not tested | 19 (28) |
| Tested – carrier SC | 17 (25) |
| Tested – carrier CF | 11 (16) |
| Tested – not carrier CF | 11 (16) |
| Tested – not carrier SC | 9 (13) |

542 * Includes two grandparents

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|---|---------|
| N= 51 | |
| Infant carrier characteristics | |
| Carrier of CF | 27 (53) |
| Carrier of SC/other haemoglobin variant | 24 (47) |
| Gender | |
| Female | 24 (47) |
| Male | 27 (53) |
| Family Units | |
| Mother lives with baby's father | 43 (84) |
| Mother lives with partner | 1 (2) |
| Single parent household | 7 (14) |

Table 2: Characteristics of newborn infants, N (%)

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558 **Table 3: Communication of SC carrier results experienced by parent sample**

| Communication of SC carrier results experienced | Parents [n=29] (high / low SC prevalence area) |
|--|--|
| Haemoglobinopathy Counsellor | 14 (11 high /3 low) |
| <i>Letter (result), health centre consultation offered and taken up</i> | 6 (4 high /2 low) |
| <i>Letter offering appointment, health centre consultation (result)</i> | 5 (high) |
| <i>Letter alone (result)</i> | 2 (high) |
| <i>Phone call (result), advised to consult GP, second phone call from haemoglobinopathy counsellor</i> | 1 (low) |
| Health Visitor | 7 (2 high /5 low) |
| <i>Home visit (result)</i> | 3 (low) |
| <i>Home visit (result), further home visit from specialist screening counsellor</i> | 1 (low) |
| <i>Home visit (result), GP visit, phone call from haemoglobinopathy counsellor</i> | 1 (low) |
| <i>Phone call (result)</i> | 1 (high) |
| <i>Phone call (result), home visit</i> | 1 (high) |
| Screening Link Health Visitor | 4 (1 high /3 low) |
| <i>Letter (result), home visit with family health visitor</i> | 2 (low) |
| <i>Phone call (result), home visit</i> | 1 (low) |
| <i>Phone call (result)</i> | 1 (high) |
| Specialist Screening Counsellor | 3 (low) |
| <i>Letter (result), home visit</i> | 3 (low) |
| Midwife | 1 (low) |
| <i>Home visit (result), GP consultation</i> | 1 (low) |

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566 **Table 4: Communication of CF carrier results experienced by parent sample**

| Communicating need for/ performing second heel prick test | Communication of CF carrier result | Parents (n=38) |
|--|--|---------------------------|
| | Specialist screening nurse | 12 |
| Midwife- <i>phone call, home visit</i> | Specialist screening nurse- <i>phone call, home visit (result)</i> | 5 |
| Midwife- <i>home visit</i> | Specialist screening nurse- <i>phone call, home visit (result)</i> | 2 |
| Specialist screening nurse- <i>phone call, health visitor-home visit</i> | Specialist screening nurse- <i>phone call, home visit (result)</i> | 2 |
| Specialist screening nurse- <i>phone call, midwife,-home visit</i> | Specialist screening nurse- <i>phone call, home visit (result)</i> | 1 |
| GP- <i>phone call, midwife-home visit</i> | GP consultation (result), specialist screening nurse- <i>phone call & letter</i> | 1 |

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|--|---|----------|
| Interviewee cannot recall | Specialist screening nurse- <i>phone call, home visit (result)</i> | 1 |
| | CF specialist nurse | 6 |
| Midwife- <i>home visit</i> | CF specialist nurse- <i>home visit (result)</i> | 2 |
| Midwife- <i>home visit</i> | CF specialist nurse- <i>hospital consultation (result)</i> | 1 |
| Midwife- <i>answer phone, home visit</i> | CF specialist nurse & Consultant- <i>hospital consultation (result)</i> | 1 |
| No second test taken ¹ | CF specialist nurse- <i>home visit (result)</i> | 1 |
| Health Visitor- <i>home visit</i> | CF specialist nurse- <i>phone call (result)</i> | 1 |
| | Hospital Consultant | 6 |
| GP- <i>during consultation, CF nurse</i> | Hospital consultant clinic (result) | 2 |

¹ risk of CF identified antenatally, sweat test conducted

| | | |
|--|--|----------|
| & health visitor- <i>home visit</i> | | |
| Midwife- <i>phone call, home visit</i> | Hospital consultant clinic (result) | 2 |
| Midwife- <i>home visit</i> | Health visitor (child carries one gene), hospital consultant (incorrectly informed child affected, before confirming is carrier) | 2 |
| | Specialist counsellor | 5 |
| Midwife- <i>phone call, home visit</i> | Specialist counsellor- <i>phone call</i> (result), <i>home visit</i> | 2 |
| Midwife- <i>phone call, home visit</i> | Specialist counsellor- <i>phone call</i> (result), specialist counsellor & health visitor- <i>home visit</i> | 2 |
| Interviewee cannot recall | Health professional (unclear who)- <i>phone call</i> (result), specialist counsellor- <i>home visit</i> | 1 |
| | CF nurse & ‘familiar’ health professional | 4 |
| Midwife- <i>phone call, home visit</i> | GP- <i>phone call</i> (result), CF nurse & health visitor- <i>home visit</i> | 2 |
| Midwife- <i>home visit</i> | CF nurse & midwife- <i>home visit</i> (result) | 2 |
| | Genetic counsellor | 3 |
| Midwife- <i>phone call, home visit</i> | Genetic counsellor- <i>phone call</i> (result), <i>hospital consultation</i> offered but not accepted | 1 |
| Midwife- <i>home visit</i> | Genetic counsellor- <i>phone call</i> (result), <i>hospital consultation</i> offered and taken up | 2 |
| | Screening Midwife | 1 |
| Midwife- <i>phone call, home visit</i> | Screening Midwife- <i>phone call</i> (result) | 1 |
| | Screening link health visitor | 1 |
| Health visitor – <i>phone call, home visit</i> | Screening link health visitor – <i>phone call</i> (result) <i>home visit</i> | 1 |