Knowledge is wealth:

Investigating psychosocial distress and ameliorating knowledge deficiencies in parents of children identified, through newborn screening in Ireland, as carrying an altered cystic fibrosis gene.

Mr Stephen James Quigley

Master of Science

University of Limerick

Dr Patrick Ryan

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Abstract

Cystic Fibrosis is an inherited disease affecting mainly the lungs and digestive system. In Ireland, the incidence of Cystic Fibrosis is 1 in 1,461 births, with 1 in 19 Irish people carrying an altered cystic fibrosis gene. Newborn screening for CF was introduced in the Republic of Ireland in July 2011. A consequence of the screening process is the identification of infants who carry an altered CF gene. Significant gaps have been identified in parental understanding of CF newborn screening and the consequences of carrying an altered CF gene. Filling in these gaps in knowledge is our primary means of alleviating the psychological complications which may arise from CF carrier identification. The current intervention study provided parents with a carefully designed information pack, and established how this affected their level of stress, knowledge of CF, as well as their decision to undergo genetic testing. A total of 32 parents took part in telephone interviews. Parents who received an information pack had significantly higher CF knowledge scores than parents in the control group. 44% of parents in the control group misunderstood the health implications of carrying an altered CF gene. There was no significant difference in Parenting Stress Index scores between the groups. Parents of infants who had more than one sweat test due to insufficient sweat quantity had higher overall Parental Stress Index percentiles (50%), than parents of infants who had just one sweat test (30%), indicating greater parental stress. The study recommends six changes to CF newborn screening practices in the Republic of Ireland.
Declaration

I hereby declare that this is entirely my own work and it has not been submitted to any other university or higher education institution, or for any other academic award in this university. Where use has been made of the work of other people it has been fully acknowledged and referenced.
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Friends and family
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List of abbreviations

CF              Cystic Fibrosis
CFTR            Cystic Fibrosis Transmembrane Conductance Regulator
DC              Difficult Child
EC              European Commission
MDT             Multi-Disciplinary Team
NBS             Newborn Screening
PD              Parental Distress
P-CDI           Parent-Child Dysfunctional Interaction
PSI             Parenting Stress Index
PSI-4-SF        Parenting Stress Index Fourth Edition Short Form
QNS             Quantity Not Sufficient
Cystic Fibrosis is a life-threatening autosomal recessive genetic disease. The presence of two altered CF transmembrane conductance regulator (CFTR) genes results in abnormally thick secretions in the respiratory, digestive and reproductive systems, which may result in chronic lung infection, nutritional malabsorption and male infertility (Tluczek et al., 2011a). In Ireland, the incidence of Cystic Fibrosis (CF) is 1 in 1,461 births, with 1 in 19 Irish people carrying a single altered CFTR gene (National Centre for Medical Genetics, 2011). “Carriers” of a single mutation do not develop CF. If two people, each carrying a single mutation, have a child, there is a 1 in 4 chance of the child having CF. Survival rates for CF have improved dramatically over the last half-century. A baby diagnosed with CF fifty years ago had a 90% chance of dying before their second birthday, this compares to over a 50% chance of surviving to nearly 40 years of age for adults born in economically developed European nations in the last two decades (Mehta et al., 2010). Diagnosis of CF through Newborn Screening (NBS) was first introduced to Ireland in July 2011 and allows treatment to occur sooner than it would through clinical diagnosis.

1.1 Newborn Screening for Cystic Fibrosis

Newborn Screening (NBS) for CF is carried out on blood-spot samples taken when an infant is between three and five days old. Early diagnosis of CF through NBS has been shown to benefit for the physical health of the child (Merelle et al., 2001), the psychological health of the parents (Merelle et al., 2003), and reduce health system costs (Sims et al., 2007). The screening is broken down into two stages; immunoreactive typsinogen (IRT) testing and CFTR gene testing. If an infant has an IRT above the 99th percentile and has one or more CFTR mutations, their parents will be contacted to schedule a sweat test. Sweat testing is the gold standard for the diagnosis of CF (Farrell et al., 2008); it tests the infants sweat for an elevated concentration of chloride. Following sweat testing, the infant is found to either have CF, to have atypical CF, or to be a carrier of an altered CFTR gene. In some cases an infant cannot produce enough sweat on the day of testing, a re-test is therefore required, usually two weeks later.
1.2 Identification of infants who carry one altered cystic fibrosis gene

Following a sweat test, some infants are identified as carriers of one altered CF gene. Since the purpose of NBS for CF is to identify infants with CF, the identification of CF carriers is effectively a false-positive designation. This is defined as an out of range screening result that, after further follow-up testing, is not shown to indicate a metabolic disorder (Schulze et al., 2003). The false positive rate in the US was reported as one in every 300 infants screened (0.33%) (Hewlett and Waisbren, 2006). Infants who carry an altered CF gene do not have the condition and will not require any special medical treatment. In these cases it is certain that at least one parent is also a carrier. It may be the case that both parents are carriers. Parents are offered genetic counselling to explain the implications for their infant and themselves. They may also avail of free genetic testing to establish their own CF carrier status. Parsons and Bradley (2003) view the identification of carrier infants as an “unfortunate side effect” (p. 288) of NBS for CF. A Centre for Disease Control publication went further, identifying the potential effect on the parents of false-positive infants as the primary harm of a CF NBS programme (1997).

In order to minimize the negative ramifications of carrier identification through NBS it is firstly necessary to identify what they are. The next chapter reviews the empirical evidence for psychosocial adversity arising from false-positive newborn screening for CF.
Chapter 2: Literature Review

The systematic review of the literature was broken down into five steps (Khan et al., 2003). The first step was framing the questions under review; ‘What is the evidence for parental psychosocial distress arising from their infant’s false positive newborn screening for cystic fibrosis?’ ‘What information is currently provided to parents and how can it be improved?’, ‘What are the current rates and correlates of parental genetic testing?’. Step two involved the identification of relevant work. The databases searched were PubMed, Scopus and Web of Science. Electronic searches were not restricted to specific countries or languages. MeSH functions were used where possible, this retrieved additional citations indexed with the search terms used. Reference section of selected articles were also reviewed to ensure no relevant studies were excluded. Step three involved assessing the quality of studies. Steps four and five entailed summarizing and interpreting the results.

2.1 Psychological adversity arising from false positive NBS for CF

Eight potential causes of psychological adversity arising from false positive NBS for CF emerged from the literature review.


2. Long-term anxiety (Beucher et al., 2010).

4. Parents being presented with unwanted information that may influence their future reproductive choices (Ciske et al., 2001).


6. The burden on parents of sharing carrier information with other family members (Parsons et al., 2003)
7. Parents not facilitated by health service to have other children tested (Parsons and Bradley, 2003).


The evidence for each cause of psychosocial adversity is reviewed in turn.

2.1.1 Short-term distress

A number of studies report that parents experience short-term distress following a positive NBS for CF and prior to a diagnostic sweat-test. Tluczek et al, (2005) reported that 43.1% of parents experienced emotional distress in the clinical range during their wait for a sweat-test appointment. Distress was measured using the Center for Epidemiologic Studies—Depression Scale (CES-D). Mothers and fathers of infants with abnormal NBS results had significantly higher levels of distress (p < .05), than parents of infants with normal NBS results. These differences in distress levels were not apparent at a 6 month follow-up visit. The median wait in this study was seven days (Tluczek et al., 2005). The results of telephone interviews (n = 123) conducted by Ciske et al (2001), indicated that the wait for the diagnostic sweat-test was the worst part of the NBS experience. Tluczek et al, (1992), outlined the wide variety of emotions that such distress can create. Concern (98%), shock (76%), anger (48%), and confusion (61%), were reported by parents (n = 104). Unpublished data referred to by Dillard and Tluczek (2005), reports feelings of shock, worry, and depressive symptoms, with 77% of mothers and 38% of fathers reporting depressive symptoms in the clinical range (Dillard and Tluczek, 2005). Semi-structured interviews were conducted with 21 mothers of infants who received false-positive initial IRT test results between January 1998 and June 2002 in Leeds Teaching Hospital. The two most frequently reported categories of feelings were; “worried/ concerned/ nervous/ upset” and “devastated/ distraught/ hysterical” (Moran et al., 2007).

The degree of distress experienced by parents of infant’s diagnosed as carrying an altered CF gene, appears to vary depending on the specific screening results. Beucher et al, reported that 95% of parents whose child had one CFTR mutation identified through NBS reported experiencing feelings of anxiety at the time of the sweat test (2010). This compared to 83% of parents whose child did not have a CFTR mutation identified through NBS.
(Beucher et al., 2010). Lang et al, (2011) established that talking to a genetic counsellor at the time of scheduling the sweat test helped to significantly reduce anxiety.

There is consistent evidence therefore, that parents experience distress while waiting for a diagnostic sweat-test for their newborn baby. Cases of longer term parental anxiety are less prevalent. There is less consistency in evidence for long-term parental anxiety.

\section*{2.1.2 Long-term anxiety}

Evidence for long-term anxiety arising from false-positive NBS for CF was investigated in a prospective longitudinal study conducted in France between 2004 and 2008 (Beucher et al., 2010). The study traced parental stress and perceived child vulnerability following false-positive CF NBS. Participants were broken into two groups; parents whose child had no CFTR mutation identified and parents whose child had one CFTR mutation identified. All parents took part in a structured interview and completed a perceived stress scale and a Vulnerable Child Scale 3, 12 and 24 months after a diagnostic sweat-test. Three months after the sweat test, 17\% of parents in the one mutation group and none of the parents in the no mutation group had residual anxiety. A similar pattern was observed after 24 months; 11\% of parents in the one mutation group, and none of the parents in the no mutation group had residual anxiety. Measures on the perceived stress scale did not differ from population means at any time point. All parents said they would have the test performed on another child (Beucher et al., 2010). Group differences are indicative of greater stress associated with the discovery of a mutated gene. What’s more, this stress appears to persist over time.

Parents thinking about the NBS results may be an indication of parental anxiety. A one-year follow-up of parents of children with false positive CF NBS results in Wisconsin, showed that 7\% of parents whose child had no CFTR mutation, thought about the screening results often or constantly, this compared to 10\% of parents whose child had one CFTR mutation still (Mischler et al., 1998). Lang et al, (2010), reported that 25\% (22 of 90) of parents still think about the NBS at least once per week, with the average age of infants at the time of survey being 111 days (+/- 52 days).

Qualitative results from a 2-year follow-up study of false-positive NBS for CF provide an expanded insight into parents’ persistent anxiety. “Certain parents perceived heterozygotism to be an unresolved problem because they did not fully understand the
disease’s recessive nature or they feared for future generations” (Beucher et al., 2010, p 775). It has also been reported that a minority of parents (10.3%) can experience feelings of guilt for having passed on the altered CF gene to their child (Tluczek et al., 2010).

Evidence of long-term anxiety is not universal. Of the 21 mothers interviewed, none reported long-term psychological effects resulting from their infant’s false-positive IRT results (Moran et al., 2007). Baroni et al, reported that total parenting stress scores for parents of infants who received false positive NBS results were significantly lower than healthy comparison families (1997). They did however, have greater defensiveness responding scores, which the authors theorised, could represent “hypervigilence and emotional repression” (Baroni et al., 1997, p. 150).

Long term parental anxiety arising from false-positive NBS for a variety of biochemical disorders has also been investigated. An American mixed methods study recruited parents of children with false-positive NBS results for 1 of 20 biochemical genetic disorders between 1999 and 2004 (Gurian et al., 2006). The mean age of children at the time of evaluation was 13.3 months in the false-positive group and 6.4 months in the normal screened group. Parents in the false positive group had significantly higher Parenting Stress Index (PSI) scores, than those in the normal screened group. Although when variation in socioeconomic status was adjusted for, the difference in scores between fathers was no longer statistically significant. 11% of mothers in the false positive group scored in the clinical range on the PSI, no mothers in the normal screened group scored in the clinical range ($p = .008$). Mothers in the false positive group worried more about the child’s future, although parents did not differ significantly in worry for their child’s health. There were also more reported child hospitalizations in the first six-months of life amongst the false positive group, though this did not reach statistical significance ($p = .09$). A second study on the long-term effects of false-positive NBS for 1 of 20 biochemical disorders also reported that mothers of children with false-positive results had higher PSI scores than mothers in the normal screened group ($p < .001$, median score 67 vs. 54). However, all mothers in the normal screened group were married, whereas some mothers in the false-positive group were in single-parent families. The age of infants was significantly different between the two groups, with median score of 6 vs. 11 months in the normal and false-positive groups respectively ($p < .001$) (Waisbren et al., 2003). The difference in stress scores associated with gender may reflect the more central role of mothers during infancy.
2.1.3 Vulnerable Child Syndrome in parents

The Vulnerable Child Syndrome was first outlined by Green and Solnit (1964). They hypothesized that the threatened loss of a child early in life may significantly disturb the parent-child relationship. This disturbance manifests in anxiety fuelled preoccupation with a child’s health, and over-use of medical services on parents’ behalf, as well as separation anxiety, infantile behaviour, bodily over concerns, and school underachievement on children’s behalf (Green and Solnit, 1964).

Some parents may also continue with the grieving process, mourning the loss of the perfect child. Anxiety and grief reactions associated with carrier-state diagnosis are thought to place families at risk for impaired parent-child bonding, disrupted relationships, personality problems, and the development of psychogenic symptoms or some variant of the vulnerable child syndrome (Farrell and Farrell, 2003, p. 710).

The studies which Farrell and Farrell cite to support this statement pertain to genetic testing in general (Ross and Moon, 2000), and risk perception amongst relatives of a child with CF (Grosfeld et al., 1997). A retrospective study carried out in Australia investigated parents’ attitudes toward the identification of their child as a CF carrier through NBS. 17% of parents in a 1996/1997 cohort and 12% of parents in a 2001 cohort indicated that they worried more about their CF carrier child than with their siblings (Lewis et al., 2006). Perceived vulnerability may be entwined with a parental misunderstanding of test results. A long-term qualitative study included 39 semi-structured interviews with parents of infants with false-positive NBS for CF. One couple remained highly sceptical about the test results and believed their child actually had CF “When she had pneumonia my first thought was back to the CF” (Tluczek et al., 2010, p. 180).

Further evidence in favour of a VCS has been reported following false-positive NBS for disorders other than CF. A study conducted in China that focused on the psychosocial effects of false-positive NBS for a variety of disorders also reported several findings indicative of vulnerable child syndrome. This included the fact that children in the false-positive group were three times as likely to be hospitalised in the first six months of life compared to children in the normal group (Tu et al., 2012). Waisbren et al, (2003) reported
that mothers of children with false-positive NBS results for 1 of 20 biochemical genetic disorders had significantly higher scores in the parent-child dysfunction subscale of the Parenting Stress Index compared to mothers in a normal screened group \( p < .001 \). 21% of children in the false-positive group had been hospitalized, compared to 10% of children in the normal screened group, however this trend was above the threshold of significance \( p = .06 \). These figures are in keeping with those reported by Morrison and Clayton (2011). They outlined that 10% of False positive parents reported that their child required extra care and worried about their child’s future health.

Despite evidence for a VCS following false-positive newborn screening for certain disorders, several studies focused on CF have not supported its presence. There was no difference in mother-baby relationship between a carrier and a control group six months after a diagnostic sweat test for CF (Parsons et al., 2003). No parental withdrawal or distancing from babies was evident in a group of 57 parents (Pollitt et al., 1997). Contrary to reports of increased levels of hospitalization, Lipstein et al, reported no significant association between false-positive NBS results and health care utilization after controlling for possible extraneous variables such as age, race, and socioeconomic status (2009). Finally, a long-term follow-up study which evaluated the effects of genetic counselling following false-positive CF NBS, found no evidence of parental perceptions of child vulnerability 11-14 years after NBS (Cavanagh et al., 2010). The authors reported that four parents with high Child Vulnerability Scale scores reported a time when they feared for their child’s life.

In short, the evidence for a VCS is far from certain. In their review of the subject, Parsons and Bradley (2003) conclude that “There is no evidence from the literature that the mother–baby relationship is significantly affected” (p. 289). The evidence that supports a vulnerable child syndrome tends to be derived from general studies of false-positive NBS, as opposed to specific studies relating to false-positive CF NBS.

2.1.4 Parents being presented with unwanted information that may influence their future reproductive choices

The knowledge that at least one parent is a carrier of an altered CFTR gene is not actively sought by parents, nor is this the purpose of a newborn screening programme. Instead parents could view this as unwanted information, which may affect their future reproductive choices (Wilfond et al., 2005). Ciske et al, reported that 16.2% of parents responded “yes” to the
questions “Has the knowledge of your child’s CF carrier status caused you to change your reproductive plans?”. 31.8% of these parents chose to have no more children (Ciske et al, 2001, p. 702). Similar results were reported by Dillard and Tluczek, who found that 22% of parents were uncertain about their future reproductive decisions as a result of their experiences of newborn screening (2005). Lewis et al, (2006) reported that 13% of respondent in a 1996/1997 cohort and 12% of respondents in a 2001 cohort decided to have no more children due to the carrier status of their infant. A one-year follow-up study conducted by Mischler et al, differentiated between parents whose child simply had elevated IRT and those who had elevated IRT and a CFTR mutation identified through NBS (1998). 17% of parents in the IRT/DNA group and 4% of parents in the IRT group, believed that NBS did influence, and may have altered their attitudes toward subsequent pregnancies ($p < 0.01$). The literature indicates that a small but consistent portion of parents alter their future reproductive plans as a result of false-positive NBS for CF. The effect on parental reproductive plans “further demonstrates the need for accurate counselling about heterozygote status” (Ciske et al., 2001, p. 704).

If both parents are identified as CF carriers some countries offer prenatal testing. Data from Australia suggests that parents are willing to terminate a pregnancy if prenatal screening identifies that their foetus is affected by a severe condition (Massie et al., 2007, p. 722). In the state of Victoria, Australia, 70% of parents who had a child with CF, availed of prenatal testing in subsequent pregnancies (Sawyer et al., 2006). It has been reported that prenatal screening for CF in Edinburgh has resulted in a lower incidence of the condition there (Cunningham and Marshall, 1998). It is therefore apparent that the newfound knowledge of a CF carrier status may influence parents’ reproductive planning and behaviour.

2.1.5 Stigmatisation of the child

Farrell and Farrell outline that stigmatisation, community ostracization, and even devaluing a CF carrier as a potential marriage partner, may affect a child who carries an altered CF gene, in the long term (2003). Examples of these pertain to other genetic diseases and tend to arise from a belief that a child may develop the disorder in time (Farrell and Farrell, 2003). If stigmatisation did arise from CF carrier identification, it would be reasonable to assume that parents of these children would object to the practice of CF NBS. However, this is not the case. A one year follow-up of such parents found that 90% of them
thought that NBS for CF should be done (Mischler et al., 1998). As Parson and Bradley (2003) note, studies from Wales (Parsons et al., 2003) and Wisconsin (Ciske et al., 2001), do not indicate that parents view information about their child’s carrier status as problematic (p. 289). Dillard and Tluczek note that “little is known about what influence this information might have on the individual’s psychosocial development” (2005, p. 96). A long-term follow-up study of parents of infants with false-positive CF NBS results, reported that some parents expressed concerns about the potential stigmatizing effects of being a CF carrier (Tluczek et al., 2010). Long-term follow-up studies involving CF carriers themselves are necessary to investigate if CF carrier identification through NBS leads to stigmatisation.

2.1.6 Burden of sharing carrier information with other family members

“Information available to the index family becomes a potential burden that they have to share with other family members” (Parsons and Bradley, 2003, p. 288). The authors conclude that “It would seem that some discomfort is caused because the new information has the potential to bring to the surface dysfunctional family relationships and create new tensions” (p 289). A long term follow-up of carrier parents found that a minority of parents viewed sharing genetic information with family members as a burden, with more parents viewing this as an opportunity. Parents who viewed this as a burden felt that the information communication aggravated already strained or estranged relationships (Tluczek et al., 2010). In a long-term study of CF carrier testing of non-parent relatives of children diagnosed with CF through NBS, it was reported that, on average, 2.7 non-parent relatives availed of carrier testing (McClaren et al., 2010). It should be noted that participants in this study were provided with a letter for distribution within their family to aid them in discussing carrier testing (McClaren et al., 2010). It may be the case that such a letter would benefit family members of children identified as CF carriers also. Parents may be facilitated in communicating information to relatives, though the decision to communicate or not ultimately lies with parents.
2.1.7 Parents not being facilitated by health service to have other children tested

Following the discovery that a newborn infant carries an altered CF gene, parents may wish to avail of carrier testing for other children (Parsons and Bradley, 2003). This facility is not open to parents in the majority of UK CF centres (Parsons and Bradley, 2003), or in CF centres here in Ireland. It has also been suggested that parents will go elsewhere to get CF testing for other children if it is not available in CF centres (Balfour-Lynn et al., 1995). Tluczek et al, reported that some parents of carrier infants wondered if their older children might have CF (2010). This adds a further layer of potential psychosocial stress, which may arise from false-positive CF NBS. A 1994 report from the working party of the Clinical Genetics Society in the UK did not recommend CF carrier testing of other children until they are old enough to consent to such testing of their own volition (Clarke, 1994). The situation is similar in the US, the American Society of Human Genetics Board of Directors outline that “If the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult onset diseases, genetic testing generally should be deferred” (1995, p. 1233).

2.1.8 Discovering non-paternity

It may be the case that questions about paternity arise if the parents genetic testing does not reveal the genetic mutation found in the child. In one case, a father’s misunderstanding of CF genetics led him to question his paternity, however this proved unfounded and the negative consequences on the spousal relationship were resolved at a 12 month follow-up visit (Tluczek et al., 2010). This case further highlights the need for accurate communication of information for both parents. One population based CF screening programme uncovered instances of non-paternity of grandparents, though not parents (Super et al., 1994). Nevertheless it is possible that genetic testing actually confirms non-paternity. In their long-term study of cascade testing, McLaren et al, outline that “although this potential harm is not routinely discussed in a genetic counselling session, genetic counsellors have skills to support parents for whom this possibility is a reality” (McClaren et al., 2010, p. 1088).
2.1.9 Conclusion

There is significant evidence of short-term distress in advance of a diagnostic sweat-test. There is also evidence of long-term residual anxiety amongst a minority of parents, particularly among mothers, when a CF mutation has been found. Evidence for a Vulnerable Child Syndrome has been derived from studies of false-positive NBS for a range of disorders, as opposed to purely false-positive NBS for CF. Therefore, the burden of proof seems to have shifted to those who argue for its existence amongst parents of children with false-positive NBS for CF. There is consistent evidence that learning of a child’s CF carrier status will lead to some parents having no more children. Currently there is no evidence of stigmatization of CF carrier children later on in life, long-term follow-up studies are required to investigate this phenomenon. It may be the case that sharing carrier information with other family members is a burden on parents. Providing parents with a carrier information letter has yielded good carrier testing rates amongst relatives of children with CF, the same strategy may be helpful for parents of carriers. This type of strategy may help address the observations of Tluczek et al., (1992);

Although most families with false-positive results handle the newborn screening process with only short-term concerns, our observations suggest that the potential psychological risks of misunderstanding test results have significant consequences for the patient and family (p 185)

The majority of screening programmes do not provide the facility for parents to have other children tested before the age of 18. More research is required to establish if this is a source of anxiety for parents. Finally, non-paternity is a risk if parents choose to undergo genetic testing, though no instances have been reported.

2.2 Provision of information to parents

The type of information currently provided to parents and the manner of its provision, will firstly be outlined. Secondly, the shortcomings of the current system, which are evidenced by parents’ lack of CF knowledge, will be detailed. Three areas of parental knowledge deficiency are evident; general knowledge of CF, genetics of CF, and health implications of
being a CF carrier. Correlates of parental CF knowledge are then detailed. This section will conclude with a review of suggested changes to the provision of information in newborn screening programmes.

### 2.2.1 Information currently provided to parents

Tluczek et al, (2006) reported five categories of information required by parents during genetic counselling following NBS for CF. These areas were;

1. The probability of a CF diagnosis
2. How infants were identified through NBS
3. Information about the sweat test procedure
4. Facts about CF genetics
5. Implications for their children’s future

The timing, manner and content of information provided to parents vary from country to country, and in some cases it varies within countries. Some of this difference can be attributed to the screening strategy adopted in a country. There are approximately 26 different screening strategies present across Europe (Southern et al., 2007). European best practice guidelines for neonatal screening outline a three-stage schema for parent’s communication needs (Castellani et al., 2009);

1. Before screening
2. Between bloodspot screening and sweat testing
3. After final diagnosis

What follows is an overview of the communication system present in the state of Wisconsin in the United States of America as detailed by Dillard and Tluczek (2005). In Wisconsin, parents’ pre-screening communication needs are met by verbally informing them about newborn screening and providing them with an information booklet. The booklet describes the mandatory nature of screening, the screening method, the nature of results and how parents can obtain further information (Dillard and Tluczek, 2005). The second stage of parent’s communication needs commences when a primary care provider contacts the family
to inform them of the positive NBS result, usually one to four weeks after the birth of the child. In some cases the doctor then schedules a sweat-test for the infant and in other cases this is left to the family. The sweat test itself takes place between one and six weeks after the child’s birth. There is no other formal provision of information to parents until their child’s sweat test appointment and this has been identified as a time of significant emotional distress for parents who typically turn to other information resources for additional information (Dillard and Tluczek, 2005). On the day of the sweat-test, parents receive in-clinic genetic counselling from a nurse or genetic counsellor while they await their child’s test result. The third stage of information provision consists of genetic counselling at the time of the sweat test. A nurse or genetic counsellor informs the family about CF, how it is transmitted, and possible implications for the family. There is no further formal communication with parents following this genetic counselling (Dillard and Tluczek, 2005).

The second stage of parent’s communication-needs, between blood-spot screening and sweat-testing, received further attention from Tluczek et al, (2005). They reported that parents engaged in a search for additional CF related information while they were awaiting their infant’s sweat test. The NBS brochure received at their infant’s birth, textbooks, Internet, friends and family members were all consulted as potential sources of information. Parents placed additional value on information sourced from medical fields. The authors concluded that knowledge had “Reduced fear of the unknown. At the same time this information seemed to increase emotional distress for parents who perceived the probability of their infant having CF as high”(p. 1700).

2.2.2 Parent’s knowledge of CF

Deficit’s in parents knowledge of CF, following the identification of a child as carrying an altered CF gene, can be broken into three broad categories;

1. General knowledge of CF
2. CF genetics
3. Health implications of being a CF carrier

Each of these categories will be reviewed in turn.
General knowledge of CF

Ciske et al, (2001) investigated parental CF NBS experiences and knowledge of CF using posted questionnaires (n = 138) and a follow-up telephone interview (n = 123). 88.3% of parents understood that their child was a CF carrier, but 12.4% of parents were unsure that at least one of them was a CF carrier. This study may have suffered a response bias as only 138 of the 483 parents contacted (28.6%) actually completed the questionnaires. Respondents had achieved a higher level of education than a comparable population of Wisconsin young adults; 58.8% of study respondents had graduated from college or vocational school, compared to 23.1% of a comparable Wisconsin population. Consequently the study may offer a positively biased view of parental knowledge of CF, researchers acknowledged that education was positively correlated with knowledge, though they outlined that this did not reach statistical significance (Ciske et al., 2001). Lang et al, (2011) concluded that parent’s had a reasonable grasp of the overall nature of CF. 94% of parents surveyed understood that their child did not have CF. However, a sizeable minority still had gaps in their knowledge, particularly surrounding the nature of the CF carrier status. 21% of parents did not understand that their child was definitely a carrier of CF, and 26% of parents thought that over time, carriers of CF can develop CF (Lang et al., 2011).

CF genetics

Parents’ understanding of the genetics of CF is the second area where deficits have been identified. A crucial piece of information for parents to be aware of is that there is a one in four chance of having a child with CF if both parents are carriers of an altered CF gene. Three studies found that there is still a significant lack of understanding around this point amongst parents of CF carrier children. A postal questionnaire administered in Australia between 1996 and 1997 found that 33% of parents did not know that there was a one in four chance of having a child with CF if both parents are carriers of an altered CF gene (Mischler et al., 1998). A similar study reported that 40% of participants held this misunderstanding (Lewis et al., 2006). In a long-term follow-up study of parental knowledge of CF, Cavanagh et al, found that 50% of parents did not understand the one in four risk of two CF carrier parents having a child with CF (2010). This was despite the fact that all three parents
populations sampled in these studies had received genetic counselling for CF at the time of their child’s sweat-test.

Other genetic misconceptions have also been identified; Lang et al, found that 38% of parents believed that CF could be inherited if just one parent was a carrier of CF (2011). One of the questions used by Ciske et al., assessed parents understanding of their child’s risk of having a child with CF (2001). The researchers found that 43% of parents did not know that there was a one in four risk of their child having a child with CF if he/she reproduced with another CF carrier. Similar misconceptions regarding the genetics of CF were reported by Mischler et al, (1998). In a one year follow-up of parents of infants with false positive NBS for CF, 69% of parents did not know that their risk of having a child with CF is greater than the general population, despite parents receiving both genetic counselling and genetic testing. Overall, these results indicate significant misconceptions regarding even the most fundamental concepts in CF genetics.

A potential effect of parents’ low levels of CF knowledge was outlined by Cavanagh et al, (2010). In a long-term follow-up study evaluating genetic counselling following false-positive NBS for CF, one of the three main reasons that parents (n = 13) had not yet informed their children about their carrier status was “Lacking knowledge about CF genetics” (Cavanagh et al., 2010, p. 206).

Health consequences of being a CF carrier

The final area of deficiency in parental CF knowledge relates to health consequences of being a CF carrier. Lang et al, reported that 26% of parents believed that carriers of CF could develop CF over time (2011). In their 2001 review of the communication process in neonatal screening Ciske et al, reported similar findings, with 15.4% of parents unsure whether being a carrier of CF could result in illness. Indeed, one year after their child’s false-positive NBS, 5% of parents believed that their child might actually have CF (Tluczek et al., 1992). DeLuca et al, conducted 48 interviews with parents of children who had abnormal NBS test results (2011). They reported that a number of parents of infants with negative test results thought there might be long-term health implications for their children.

A mixed methods long-term follow-up study of parental CF knowledge carried out in Wisconsin, highlighted other significant misunderstandings on behalf of parents (Cavanagh et al., 2010). The qualitative methods utilised in this study allowed the researchers to delve
deeper into parental misconceptions. For example, one parent believed that her child’s altered CF gene was a consequence of one of the child’s parents having been exposed to toxins whilst serving overseas with the military. Another parent believed that the altered CF gene was responsible for the child having chronic nasal congestion (Cavanagh et al., 2010). In both cases, the parents in question had received genetic counselling. A similar long-term follow-up study reported that some parents (9 parents of 39 interviews conducted), expressed doubt as to the accuracy of the diagnostic sweat-test when their children developed respiratory illnesses (Tluczek et al., 2010).

2.2.3 Factors correlated with parental knowledge of CF

DeLuca et al (2011), explored parent’s experiences of newborn screening for a variety of disorders via structured interviews (n = 48). Results indicated that parents who were younger, had less formal education, or were from underserved minorities, had inaccurate information about the disorder and results. The variables of education and genetic counselling have received further attention in the CF NBS literature.

**Genetic Counselling**

“Genetic counselling is the process of helping people to understand contributions to disease” (Resta et al., 2006, p. 77). Cavanagh et al, reported that parents who received genetic counselling had significantly higher long-term retention of genetic knowledge compared to those who did not receive genetic counselling (2010). 65% of parents who had received genetic counselling and 22% who had not, knew that if only one parent is a CF carrier there is no chance of having a child with CF. 70% of study participants had received genetic counselling at the time of their infant’s sweat-test (Cavanagh et al., 2010). Ciske et al,’ 2001 publication also highlighted the advantage of genetic counselling to parents knowledge of CF. They reported that parents who received genetic counselling had significantly more correct answers to CF related questions than parents who did not receive genetic counselling. However, Mischler et al, reported the opposite trend; “people who had DNA testing for carrier status and more genetic counselling had significantly more incorrect responses” (1998, p. 48).
Despite the deficits in knowledge and potential for anxiety amongst parents of children with false-positive NBS results, some NBS programmes do not provide genetic counselling for this group. A 2001 survey of US NBS programmes found that only 1 of the 22 programmes surveyed provided genetic counselling for parents of children with false positive CF results. This compares with 13 of 22 programmes providing genetic counselling for parents of children with false-positive phenylketonuria results (Farrell et al., 2001). The majority of respondents from the NBS programs indicated that in the case of false-positive results, genetic counselling was the responsibility of primary care physicians, though they acknowledged that the quality of this counselling was lower than other professionals. The Irish CF NBS system provides free genetic counselling to all parents of infants with false-positive CF NBS results.

**Education**

Tluczek et al (1992), reported that parents’ level of education was positively correlated with knowledge of neonatal screening ($p = .0001$, $n = 98$). This is in keeping with a 1991 study by Tluczek et al, which included the same correlates ($p = .0001$, $n = 104$). Ciske et al, reported that parents with a college education had a higher percentage of correct answers but this trend was not statistically significant (2001). A similar non-significant but positive relationship was reported by Cavanagh et al (2010), in a long-term follow-up of parents of infants with false positive NBS for CF. The small sample size ($n = 37$) may have contributed to the non-significant relationship (Cavanagh et al., 2010). The correlation between CF knowledge and education was significant in a study of population CF carrier testing conducted by Clayton et al, (1995). 60% of participants with less than a high school education had a sufficient understanding of the health consequences of having CF or being a carrier, as compared to 94% of college graduates (Clayton et al., 1995). Overall recall of the NBS experience was correlated with younger better educated parents in a long-term follow-up (Tluczek et al., 2011b). Similar results have been reported in relation to other conditions. A study of maternal anxiety following NBS for Type 1 diabetes reported that mothers with a high school education or less, had less understanding of their child’s risk of diabetes and had higher levels of anxiety 3.5 weeks after screening (Bennett Johnson et al., 2004).

Results from a recent qualitative study also point toward a link between education and parents retention of CF knowledge. Tluczek et al, carried out a qualitative review of interview
data collected from five couples whose infants had received equivocal diagnostic test results following positive genetic NBS for CF. One couple misinterpreted the results which they received in the post several weeks after their infants testing; “A bunch of pages with high-tech words that we didn’t know” (p. 213). The couple believed their child carried only one CF mutation when in fact the child had two. The authors noted that a couple’s level of education is likely to influence their ability to understand genetic test results; the aforementioned couple had less than a college level of education (Tluczek et al., 2010).

A mechanism by which education may influence retention of CF knowledge was explored by Dillard et al., (2010). The Wisconsin based study compared parents of children with a positive newborn screen for CF across a number of variables. Parents with a higher level of formal education sought CF information from a greater number of sources prior to a clinic visit and asked a greater number of collaborative questions than parents with a lower level of formal education. In turn, asking more collaborative questions was correlated with a greater retention of CF information six weeks after a clinic visit (Dillard et al., 2010).

### 2.2.4 Recommendations for changes to information provision

Various recommendations to the provision of NBS information have been made by authors who have conducted studies and reviews in this area. These recommendations can be broken into How, What, and When;

- How is NBS information best communicated.
- What information should be provided to parents.
- When this information is best communicated.

**How is NBS information best communicated**

Rock et al, emphasised the importance of effective communication with carrier parents following their child's sweat test. “Understanding this complex information can be challenging, and researchers are continuing to search for improved methods of genetic risk communication” (Rock et al., 2005, p. 67). Individual publications have made various recommendations with regard to communication of NBS results.
In designing a tailored approach to family-centred genetic counselling for CF NBS, Tluczek et al, recommended that counsellors use data from multiple sources to formulate an educational plan unique to each family (2011). The authors outline that “Effective communication is the foundation of any psychoeducational intervention” (Tluczek et al, 2011, p. 118). Indeed, it has been found that most parents prefer a combination of reading, viewing and listening (Jedlicka-Kohler et al., 1996). Wilfrond et al, noted that a strategy to reduce the psychological risk associated with false-positive CF NBS, is to ensure effective communication of results coupled with counselling and educational resources which facilitate an accurate understanding of the meaning of results (2005). Farrell et al, cite two measures to ensure optimal outcomes after neonatal screening. The first of these is “effective communication of test results and its implications” (2003, p. 710). They outline that good communication has been shown to increase patient satisfaction (Hall et al., 1994), improve compliance (Maiman et al., 1988), and enhance health outcomes (Kaplan et al., 1989). Unfortunately, several studies have raised concerns relating to the communication skills of physicians (Farrell et al, 2001, and Ciske et al, 2001). Farrell et al, note that the state screening bodies could conceivably take responsibility for the consistent communication of CF NBS results, but at the time of screening no such state-wide best practice was in existence in the US (2003). Thankfully, NBS information guidelines have recently been published for Europe.

European best practice guidelines for neonatal screening were published by Castellani et al, in 2009. The authors acknowledge that the many national variations in European NBS programmes presents obvious challenges to developing standardised guidelines for the provision of information. Nevertheless, the authors’ outline that parents everywhere have basic communication needs which can be addressed through sound communication principles. The authors provide a series of questions and sample answers for the parents of children identified as carriers of an altered CF gene (Castellani et al, 2009). The guidelines go on to outline that the “Psychosocial complications of identifying carrier status are so severe and common that it has been argued that for such parents , effective communication may be the only way to “ensure more good than harm”(Farrell and Farrell, 2003)” (Castellani et al, 2009, p. 169). Cavanagh et al, called for more investigation into the communication of risk to parents, with an emphasis on understanding and retention of genetic facts (2010).
What information should be provided to parents

One of the key conclusions of Lang et al, 2011 study on parental understanding of a negative diagnostic sweat test for CF, was that parents want more information about CF. This desire for more information is also reflected in China. A 2012 study of the psychological effect of false-positive NBS results for a range of disorders conducted in China found that 56% of parents (n = 72) expressed a desire for more information about NBS and false-positive results (Tu et al.).

A review of information provided to parents by newborn screening programmes in the US was conducted by Fant et al., in 2005. Researchers compared the information currently provided to parents with information recommended in a blueprint from the Academy of Paediatrics (2000) task force on NBS. None of the 51 programmes reviewed contained all the information recommended by the task-force (Fant et al., 2005). Unfortunately, it may be the case that parents are being provided with a sufficient quantity of CF NBS information, the problem may lie in the quality of that information. “Authors are particularly concerned about the nature of the information given to carrier families and the potential for misunderstanding about the health of their child” (Parsons and Bradley, 2003, p. 290).

When this information is best communicated

It may be the case that the conclusion of the emotionally charged sweat-test procedure is not a good time for parents to absorb new CF information. In her review of parents’ experiences of NBS, Clayton (2005) states that “A major problem with current efforts to educate parents about NBS is that they typically take place in the peripartum period, reflecting the origin of NBS in pediatric practice. This, however, is a terrible time for education because of the many transitions that occur. As a result, parents generally know little about NBS” (Clayton, 2005, p. S27). Indeed, other authors have come to similar conclusions with regard to parents’ hindered capacity for information absorption following NBS. “Parents’ psychological state and perceptions about the likelihood of their child having CF represent critical contexts that may affect their capacities to absorb information and the meaning they ascribe to the genetic counselling at the time of the clinic visit for a sweat test” (Dillard and Tluczek, 2005, p. S95).
Mischler et al. (1998) came to a similar conclusion; “The anxiety at the time of the sweat test may be an obstacle to understanding this information” (p. 50).

A further hindrance to parents’ absorption of information was alluded to by Tluczek et al., in their 2011 overview of a tailored approach to genetic counselling for CF NBS. The authors speculated that the sleep deprivation associated with night time feeding of infants which was documented by Tluczek et al., (2005), “may contribute to parents inability to retain NBS information presented during counselling” (p. 117). These recommendations are in keeping with those of Fant and colleagues (2005), following their review of the information provided by NBS programmes; “The postpartum period is a challenging time for parents and it is unclear how much information they can retain” (p. 1270). The authors make reference to the task force on NBS which recommended that prospective parents should ideally receive NBS information during a routine third-trimester prenatal care visit (Task Force on Newborn Screening, , 2000). Based on their review of the psychosocial effects of false-positive NBS results, Hewlett and Waisbren recommended that a follow-up information brochure should be provided by physicians to parents (2006). The authors went on to recommend follow-up studies of parental stress and repeat screening to track potential improvements (Hewlett and Waisbren, 2006).

2.2.5 Summary

This review of parental understanding of CF NBS has identified consistent knowledge deficiencies on parents’ behalf. These deficiencies include parents’ general knowledge of CF, CF genetics and health implication of being a CF carrier. In their brief review of the subject, Tluczek et al., (2011), state that “In short, the empirical literature documents significant parental distress and knowledge deficits associated with genetic NBS for CF” (p. 119).

The manner in which parents are made aware of their child’s NBS results can be improved on several fronts. What information is communicated to parents is a crucial question for any NBS programme. When the information is communicated has come under consistent criticism in the NBS literature, with the bulk of information being imparted at a highly stressful time of parents’ lives. Finally the question of how communication takes place
requires review – in a time of ever expanding means of communication, other mediums may provide a welcome supplement to face-to-face communication. DeLuca et al, suggest that “Different methods for notifying parents of abnormal screening results could be examined to determine the effects on parents’ distress” (2011, p. 59). DeLuca et al, further emphasised that future research, focused on “improving parents’ exposure to and retention of newborn screening education information is needed” (2011, p. 59). In short, NBS programmes must re-evaluate their education systems. Albert Einstein referred to insanity as “doing the same thing over and over again and expecting different results”.

One of the key decisions which parents face once their child has been identified as a CF carrier is whether or not to undergo genetic testing. Testing may benefit parents by clarifying their own CF carrier status. Since the question of genetic testing arises as a direct result of false-positive NBS for CF, it behoves the screening programmes to ensure that parents receive adequate genetic testing information.

### 2.3 Parental genetic testing

This section will begin by providing a background to genetic testing and its development in the area of CF. The various factors which may motivate parents to undergo testing following their child’s CF newborn screening (NBS) results will then be examined. This is followed by an overview of genetic testing rates and correlates which have been reported in the literature. The section will conclude by reviewing the consequences of genetic testing, with a focus on parents’ future reproductive choices and cascade testing.

#### 2.3.1 Overview of genetic testing

Genetic testing for susceptibility to medical disorders and disease is a relatively new phenomenon in the history of medical screening. Genetic testing for CF only became possible with the discovery of the Cystic Fibrosis Transmembrane Conductance Regulator Gene (CFTR) in 1989 (Kerem et al., 1989). Since then, an expanding list of alterations to the CFTR gene has been identified. As of 1998, a little over 800 pathogenic mutations had been identified (Wildhagen et al, 1998), this number had risen to over 1300 by 2005 (Parad and
Comeau, 2005), and is currently estimated as 1964 (Cystic Fibrosis Mutation Database, 2014). Despite the high number of alterations identified, the vast majority of CF cases are caused by a relatively small number of genetic alterations. Internationally, less than 20 CFTR alterations have a population frequency greater than 0.1% (Babadilla et al., 2002). Since 1991, DNA testing has been an element of the Wisconsin Newborn Screening Program as a research protocol, and since 1994 as standard practice (Farrell et al., 1997). The specific mutations to which an individual is susceptible is largely dependent on that individual’s geographic, religious or ethnic grouping (Culling and Ogle, 2010). For example, 57.5% of the Caucasian CF population in the UK are F508 homozygotes, whereas only 24.7% of the UK CF population from the Indian Subcontinent are F508 homozygotes (McCormick et al., 2002). Ameliorating the adverse psychosocial consequences of genetic testing becomes ever more relevant as screening protocols expand with the discovery of new genetic mutations.

### 2.3.2 Motivation to undergo genetic testing

The knowledge that a newborn baby is a carrier of an altered CFTR gene presents relatives with a new question with regards to their own carrier status. Wildhagen et al, outline that the “identification of newborn carriers provides an opportunity to test both parents with a view to ascertaining previously unrecognized high risk couples and extend their future reproductive choices” (1998, p. 863). The authors go on to outline that contact with a known CF carrier relative may engender greater familiarity with CF and therefore facilitate more informed choices about screening and reproduction than would be possible for the general population (Wildhagen et al., 1998). Parental genetic testing may also alleviate parents’ anxiety in relation to the health of their newborn baby. Parents’ who establish their own CF carrier status may make a connection between their own good health and that of their CF carrier child (Parsons et al., 2003).

### 2.3.3 Genetic testing rates

A broad spectrum of parental carrier testing rates have been reported in the literature. In October 1998, Ciske et al, conducted a retrospective study of parents of infants who received false positive NBS for CF in Wisconsin (US) between July 1994 and December
1997. 41.9% of mothers and 33.8% of fathers surveyed had undergone genetic testing. 41 of the 138 respondents (29.7%) stated that both parents had undergone carrier testing (Ciske et al., 2001). A 2011 study based in Chicago reported that 40 of the 90 parents surveyed (44%) had undergone carrier testing, but only 13 (14%) reported that both parents had been tested (Lang et al., 2011). It should be noted that at the time of the interview, a further 9% of parents surveyed had a genetic testing appointment, giving a potential overall testing rate of 53% (Lang et al, 2011). Mischler et al, (1998) conducted a one-year follow-up survey of parents of children who had false positive CF NBS results following IRT/DNA testing. The response rate was 58% (n = 62), 58% of whom had received genetic testing for CF at the time of the survey (Mischler et al., 1998). Finally, a study based in Australia reported testing rates of 53% in a 1996-1997 cohort and a rate of 85% in a 2001 cohort. The authors offered no explanation for this increase in testing rates (Lewis et al, 2006). These studies also outlined variables that were correlated with parents’ decision to undergo genetic testing.

2.3.4 Genetic testing correlates

A number of variables have been correlated with parents’ decision to undergo genetic testing for CF. Lang et al,’ (2011) reported that being married, white, a college graduate, and having private health insurance were all positively correlated with undergoing genetic testing. Lewis et al, Australia based study also found that more mothers than Fathers chose to undergo genetic testing in both the 1996-1997 and the 2001 cohort (2006). This pattern was also present in Ciske et al,’ (2001) study which found that in cases where only one parent was tested, mothers (41.9%) were more likely to undergo genetic testing than fathers (33.8%). The decision to undergo genetic testing was also correlated $(p < .001)$ with parents having received genetic counselling. Ciske et al, (2001) also reported that parents who received genetic counselling had a better general knowledge of CF. This raises the question of whether parents’ knowledge of CF directly influence their decision to undergo genetic testing, or if such knowledge is a separate variable which increases as a result of genetic counselling but does not influence their propensity to undergo genetic testing?

Mischler et al, (1998) investigated parents’ motivation to undergo CF carrier testing. Parents were asked to rate eight potential factors on a 5-point likert scale. The most influential factor was “I was interested in accurate recurrence risks for future pregnancies”,
with the least important factor being “Time and travel to the CF centre (Mischler et al., 1998, p. 49).

2.3.5 Population Screening

It is interesting to note that similar rates of genetic testing are evident among population wide screening programmes and testing on foot of positive NBS result. The two populations are also similar in terms of the demographic variables of individuals who avail of carrier testing. Population screening offered in Western Australia had an uptake of 43.5% (Honnor et al., 2000). The correlates for undergoing genetic testing, ordered in decreasing magnitude of effect were as follows: people planning to have children, women, people without children, young people and people with higher levels of education. Individuals who declined genetic testing and those who tested negative were less well informed about CF and the implications of test results than individuals identified as carriers. The authors speculated that this difference might arise from the fact that counselling is offered to test-positive individuals but not test negative individuals (Honnor et al., 2000). A 2010 study based in Sweden found that 47% of parents of children with CF wished to know their CF carrier status. The question was also put to two control groups; 45% of parents of a child with diabetes and 50% of matched population parents also wished to know their CF carrier status (de Monestrol et al., 2011).

The results from these population screening studies are interesting for two reasons. Firstly, there is similar rate of genetic testing among the parents of children identified as CF carriers as control populations. Secondly, the profile of those who avail of testing following their child’s NBS is similar to those who avail of population based testing. An individual availing of genetic testing is more likely to be female, have a higher level of education and plan to have children in the future.

One population screening study reported on the potential emotional consequences of CF carrier testing. Gordon et al, (2003) surveyed individuals 18 months after availing of population wide testing in Australia. Those identified as CF carriers and those with negative results, believed that carriers would experience more negative feelings than non-carriers. However carriers felt more positive about their results than they believed most carriers would feel (Gordon et al., 2003). The authors concluded that there was minimal adverse psychological effects arising from CF carrier identification.
2.3.6 Influence of CF carrier status on reproductive plans

The reproductive options open to parents vary from jurisdiction to jurisdiction. Pre-implantation genetic diagnosis is now available in Ireland (Roche, 2013). Prenatal testing is made available to carrier couples in many countries, but parents’ options after a positive test vary greatly. A foetus that is diagnosed with CF cannot be aborted in Ireland (Citizens-Information-Board, 2013), but may be in Australia (Sawyer et al., 2006). In short, the effect of carrier identification on reproductive plans varies widely depending on the options open to parents.

In Australia, if both individuals’ are CF carriers, or if one individual has CF and the other is a CF carrier, they may avail of prenatal genetic testing. “If the foetus has both parental CFTR mutations termination of pregnancy may be sought” (Culling and Ogle, 2010, p. 78). The rate of CF prenatal diagnosis of CF in Victoria, Australia, increased by a magnitude of 2.25 since the introduction of NBS (Balnaves et al., 1995). Prenatal testing in Victoria identified 32 cases of CF between 1989 and 1991, all but one of these pregnancies was terminated (Balnaves et al., 1995).

A longitudinal study based in Australia compared hypothetical and actual reproductive behaviour in mothers of children with CF (Sawyer et al, 2006). Baseline attitudes were taken when the child with CF was 2 years old, behaviours were assessed five years later. 82% of participants reported that they would be likely to use prenatal testing in a subsequent pregnancy, 56% reported that they would likely terminate an affected pregnancy. One third of mothers changed their mind about prenatal testing in the interim (both for and against testing). Prenatal testing was used in 33 of the 55 pregnancies reported. Five of the 33 pregnancies were affected by CF, all were terminated (Sawyer et al, 2006).

The effect on future reproductive plans is not just limited to carrier couples. Due to the high number of pathogenic CF mutations in existence, false-negative carrier screening results are possible. If the partner of a known carrier is found not to be a carrier of CF through genetic testing, the chance that they would have a child with CF is reduced but not eliminated. An individual’s ethnic background and the type of genetic testing carried out will determine the level of risk (Culling and Ogle, 2010).

Another important consequence of both NBS and parental genetic testing is the potential for a cascade of genetic testing among extended family members. Cascade testing allows the relatives of CF carriers to determine their carrier status. Ideally if this occurs pre-
conceptually, at-risk couples will have time to consider all reproductive options available to them (Culling and Ogle, 2010)

2.3.7 Cascade Testing rates

“The advantage of cascade testing is that the relatives of offspring of the affected individual have a higher than average risk of being carriers” (Wildhagen et al., 1998, p.863). A Belgian study conducted in 1990 indicated that 80% of parents of a child diagnosed with CF, informed their siblings about the genetics of CF (Denayer et al., 1990). Similar results were reported in Wisconsin, where 88% of parents informed other family members of their infants CF carrier status and the potential that they may also be carriers (Mischler et al., 1998, p 48). This figure is remarkably similar to the figure reported in a Chicago based study (89%), with 75% of participants also informing their partner’s siblings (Lang et al, 2011). In their 2005 review of information flow following positive NBS for CF, Dillard and Tluczek wrote that “Knowledge of the carrier status of one individual has the potential to motivate carrier testing in other individuals, presumably because of the implications for family planning” (p. S96).

Uptake of CF carrier testing amongst non-parent relatives was investigated in Australia between 2000 and 2009. 716 relatives of children diagnosed with CF through NBS were identified as eligible for carrier testing. 82 of these underwent carrier testing by the end of the study. On average, 2.7 non-parent relatives availed of carrier testing per affected family. Women were more likely to undergo testing than men. Relatives who were more closely related to the affected child were also more likely to undergo carrier testing (McClaren et al, 2010). The prevalence of cascade testing highlights the need for parents to have a clear understanding of CF. They are not simply passive recipients of genetic information, parents actively inform those close to them of the nature of CF.

2.3.8 Summary

Genetic testing is a relatively recent development in healthcare services. The parents of children identified as CF carriers are offered free genetic testing to establish their carrier status. Establishing the future reproductive risk of having a child with CF is a key motivating factor for parents. Being female, having a high level of education, and planning to have
children in the future are all correlated with genetic testing among both parent and population-wide groups. The impact of testing on reproductive behaviour varies depending on the options open to parents. If prenatal testing diagnoses a foetus with CF, the pregnancy is frequently terminated, where this is an option. The prevalence of cascade testing amongst extended family members casts parents in the role of educators and communicators of genetic and CF information. This role underscores the importance of delivering adequate newborn screening education to parents.

2.4 Study aims

The study is designed to generate both qualitative and quantitative data. Quantitative data aims to test the following three hypotheses. Hypothesis one: The provision of an information pack to the parents of children identified as carriers of an altered cystic fibrosis gene will increase their levels of knowledge of CF. Hypothesis two: The provision of an information pack to the parents of children identified as carriers of an altered cystic fibrosis gene will decrease their level of stress. Hypothesis three: The provision of an information pack to the parents of children identified as carriers of an altered cystic fibrosis gene will increase the likelihood that they will undergo genetic testing for cystic fibrosis. Parents’ experience of the NBS system, the effect of their infant’s result, any changes they may suggest, along with any emerging issues, will be explored using qualitative data. It is hoped that the fusion of qualitative and quantitative data will allow us to establish the effect of accurate information provision as well as reflecting parents’ lived experiences.
3.1 Method overview

The parents of infants newly diagnosed as carrying an altered CF gene were informed of the research study by a CF consultant after their infants sweat test. If they chose to participate, parents received a letter in the post one week later, this provided further information on the study and gave parents several means of opting out of the study. Participating parents were alternatively assigned to a control or experimental group, with those in the experimental group receiving an information pack two weeks later.

Five weeks after their infants sweat test, parents were contacted by telephone to confirm their consent and complete a questionnaire. The questions established parent’s level knowledge of CF, level of stress, and several other demographic variables. The purpose of this telephone call was purely to collect data and under no circumstances did the researcher impart information to the parents.

Figure 1. Study timeline
3.2 Recruitment of participants

Following receipt of their infant’s sweat test results from a CF Consultant, the parents of infants newly diagnosed as carrying an altered CF gene were informed of the on-going research exploring peoples’ experience of the CF newborn screening programme. Parents filled in a brief information form (Appendix 1) which included their contact details and a preferred time and day for completing the telephone interview. Parents signed this form to consent to being sent further study information.

If they chose to participate, parents received a letter in the post one week later (Appendix 2). This gave further information about the study and what would be required from participants. Parents were given several means of opting out of the study; including a telephone number, email address and a stamped addressed envelope with the information letter. If these parents wished to take part in the study, they did not have to take any further action.

The same protocol was used for four of the six CF centres. AMNCH and Temple St hospitals required signed consent for study participation from each participant. These participants received the same initial briefing from their CF consultant but were required to fill in and return the information letter which was sent to them one week after their infant’s sweat test.

3.2.1 Participant inclusion and exclusion criteria

Study participants had to be a parent of a child identified as carrying an altered Cystic Fibrosis gene through CF newborn screening in the Republic of Ireland. Parents of children who; a) have CF, b) are unaffected by CF, c) have ambiguous sweat-test results, or d) are under 18 years of age, were excluded from the study. Only one parent was included per household. If two parents were listed on the contact form, the first name listed was the parent contacted.
3.2.2 Survey method

The two primary survey methods considered for the current study were telephone survey and postal survey. Previous CF studies have shown that there is a relatively poor response rate to posted requests; in a 2001 study based in Wisconsin the researchers received 138 completed questionnaires from the pool of 483 parents who were initially contacted, a 29% response rate (Ciske et al., 2001). A similar postal survey conducted in Australia had response rates of 45% and 64% for the two cohorts used (Lewis et al., 2006). In contrast the results of a telephone survey published in 2011 by Lang et al, into parents’ understanding of negative sweat test results had 90 respondents of the 111 eligible participants, a response rate of 81%. Though it should be noted that parents were offered $10 for participation. Given the small size of the population being surveyed and the importance of a high response rate, it was felt that a telephone survey was the best option.

3.3 Questionnaire design

This section will give an overview of how the current questionnaire was designed. A copy of the questionnaire is included in Appendix 3. A questionnaire used in a previous study (Lang et al., 2011), was used as a template for the current questionnaire, a copy of that questionnaire is included in Appendix 4. Explanations are provided for questions which were excluded or changed. Changes made to the 20 True/False questions in Lang et al 2011, are then outlined. Additional questions used in the current questionnaire are explained, followed by an overview of the Parenting Stress Index. This section concludes with a justification for the survey method chosen.

3.3.1 Changes made to Lang et al, (2011) questionnaire

A 93 item questionnaire utilised by Lang et al, (2011), which examined parental understanding of newborn screening for CF after a negative sweat-test, was used as the starting template for the current questionnaire. A number of questions which appeared on the Post Sweat test survey used by Lang et al, were excluded from the current survey. The excluded questions fall into the following 12 categories;
1. **Questions relating to the method by which parents were informed of the positive blood-spot test and scheduling of a sweat test.**

   The system for informing parents of a positive blood-spot test for CF is standardised in Ireland. Parents are contacted via telephone by a CF nurse and a sweat test is scheduled, generally within 24 hours.

2. **Questions relating to prenatal discussions of newborn screening and CF.**

   These questions were not relevant to the hypothesis under investigation.

3. **Questions relating to parents’ understanding of the bloodspot screen and sweat test results.**

   These topics were addressed sufficiently in the questions relating to a child’s CF and CF carrier status, questions 13 and 14 respectively.

4. **Questions relating to parents’ current levels of stress.**

   This was assessed using the Parenting Stress Index – Short Form, rendering other stress-related questions unnecessary.

5. **Question relating to parents understanding of the ethnicity of CF.**

   Parents’ knowledge of CF was assessed sufficiently in the ten specific CF knowledge questions.

6. **Questions relating to the timing of CF carrier testing for both parents.**

   At the time of the survey, parents will not have been afforded an opportunity to avail of CF carrier testing within the health service. Carrier testing is only offered after a genetic counselling session. Parent’s intention to undergo carrier testing is assessed in the current questionnaire.

7. **Question 30 in Lang et al, (2011).**

   The authors of the study outlined that this question may have been difficult for parents to understand as it conflated both knowledge and value (Lang et al, 2011).

8. **Questions relating to parental knowledge of other genetic diseases were excluded, i.e. Questions 51 – 53 in Lang et al, 2011.**
The current study is purely focused on Cystic Fibrosis and so these questions were unnecessary.

9. **Question relating to who should be made aware of a child’s CF carrier status.**

This question was not relevant to the hypotheses under discussion.

10. **Lang et al, (2011) listed relatives to establish if they were aware of the positive NBS result, their knowledge of the CF gene and their interest in carrier testing (questions 76 -81).**

    The current study phrased these questions in an open, non-prescriptive manner (questions 24 – 26).

11. **Questions relating to parents’ marital status, health insurance, religion and the number of children they have.**

    These questions were unnecessary.

12. **Questions 91 and 92 in Lang et al, 2011, provide an exhaustive list of options relating to ethnicity and race.**

    Instead parents were asked an open question regarding their Nationality (question 9 in the current study).

### 3.3.2 Changes made to Lang et al (2011) true/false questions.

Lang et al, included two 10-item true or false sections. These assessed parents’ knowledge of genetics and the health conditions associated with CF. The questions were changed in a number of ways.

1. In keeping with the CF information pack which was designed as part of this study, the term “CF carrier” was not used. This was to avoid individual’s being defined by their carrier status. The terminology used in the questionnaire was an individual who “carries CF”, or who “carries an altered CF gene”.

- 34 -
2. Some of the questions listed under the Knowledge of Genetics section of Lang and colleagues (2011) were deemed to be assessing knowledge of the health implications of CF and not CF genetics, e.g.

   29. Over time, carriers of CF can develop CF

   31. There are things a person with CF can do to avoid some of the complications

   32. There is no cure for CF

These were replaced with genetic questions.

3. Questions relating to the health conditions associated with CF were significantly reduced. Some of this information is not relevant to the parents of a child identified as carrying an altered CF gene, e.g.

   44. Men with CF often have fertility problems

   47. Playing sports will worsen the symptoms of cystic fibrosis

4. The current questionnaire included more questions which assessed parents understanding of the health implications of carrying an altered CF gene.

   15. If you carry one altered CF gene you are more likely to get chest infections

Certain questions which were not broached in the Lang et al (2011) questionnaire, were included in the current questionnaire. Question 18 assesses parents overall support for the newborn screening, in the case that a parent is not supportive, subsection (18. (b)), assesses why this is. Question 27 “Do you think that your child’s newborn screening result is a benefit or a harm?” was included as it had not heretofore been addressed in the literature.

3.3.3 Parenting Stress Index

The Parenting Stress Index Fourth Edition Short Form (PSI-4-SF), was used to assess parental stress. It consists of 36 items which constitute three sub-scales and a total score. The three subscales are Parental Distress (PD), Parent-Child Dysfunctional Interaction (P-CDI) and the Difficult Child (DC). The PSI-4-SF can be administered in less than ten minutes (Abidin, 2012). The items used include 33 statements to which the respondent indicates their
degree of agreement; For example, in the case of item 26, parents will indicate whether they strongly agree, agree, not sure, disagree, or strongly disagree with the statement; “My child generally wakes up in a bad mood”. A further three items require parents to choose one of five responses to a question. Alpha-reliabilities of the PSI-SF subscales have been reported between .78 (for Difficult Child) and .90 (for Total Stress) (Roggman et al., 1994). Average cronbach’s alpha for the overall scale has been reported as 0.85 (Abidin, 1995). The PSI was used in previous false-positive NBS studies (Baroni et al., 1997, Gurian et al., 2006), including at least one telephone interview study (Morrison and Clayton, 2011).

3.4 Ethics

The psycho-social stressors affecting this study population raise a variety of ethical issues. Following a comprehensive literature review, it was apparent that these parents of newborn infants were likely to be experiencing higher than average levels of stress. One of the goals of the current study was to establish if this stress could be reduced through the provision of timely and appropriate information. Nevertheless, the control group did not receive any additional information. It was therefore paramount that participation in this study not become a source of stress to these parents. This was a guiding principal for developing each facet of the study. Great care and consideration was taken in designing the information pack. It had to strike a balance between scientific accuracy and comprehensibility, and crucially, not be an additional stressor for parents.

3.4.1 Ethics Committees

Ethical approval was required for this study from the following ethics committees; 1) University of Limerick, 2) Mid-Western Regional Hospital, Limerick, 3) Our Lady’s Children’s Hospital Crumlin, 4) Adelaide and Meath Hospital incorporating the National Children’s Hospital / St. James’s Hospital, 5) Temple Street Children’s Hospital, 6) University College Hospital Cork, and 7) Galway University Hospital.

A presentation was made to the Research Ethics Committee of the Mid-Western Regional Hospital Limerick on 21st November 2012. The Committee approved of the study and ethics submissions were made to all remaining committees. Approval was received from
the Clinical Research Ethics Committee of University College Hospital Cork on 14th January 2013. The Ethics Committee of Our Lady’s Children’s Hospital Crumlin required a full ethics submission and presentation. A presentation was given to the committee on 15th January 2013. The ethics committee expressed concerns in relation to the study design and the contents of the information pack. These concerns and how they were addressed are outlined below. Changes were then made to both the study design and the contents of the information pack. A full submission and presentation was once again made to the Ethics Committee of Our lady’s Children’s Hospital, Crumlin on 12th March 2013. The committee approved of the study at that meeting.

Due to the changes made to the study design, it was necessary to submit amended applications and explanatory letters to all other committees. Approval of the amended application was received on 8th April from University College Hospital Cork and 11th April 2013 from Galway University Hospital. Temple Street Children’s Hospital would not accept the Standard Ethics Application form which was accepted by all other Hospitals and so a new ethics application was made to Temple Street using their ethics application form. Due in part to a lack of ethics board meetings over the Summer period, ethical approval was not received from Temple Street Children’s Hospital until 11th September 2013.

3.4.2 Issues raised by Our Lady’s Children’s Hospital Crumlin Ethics Committee

The Ethics Committee Chairperson expressed concerns in relation to the study design and the contents of the information pack. A meeting with the Chairperson, Professor Andrew Green, and the CF Genetic Counsellor for Ireland, Dr Alana Ward and Mr Stephen Quigley took place in the National Centre for Medical Genetics on 5th March 2013. The aim of the meeting was to ascertain further details of the issues raised and develop a plan to address them. With regard to the study design, Prof Green was concerned that CF related information could be inadvertently given to a participating parent by a researcher during data collection. He also felt that conducting a second telephone survey would be unnecessary. With regard to the information pack, Prof Green requested a revision of the diagrams used and the wording of certain segments. In particular he outlined that a person should not be defined by their carrier status, i.e. the information leaflet should refer to a ‘person who carries an altered CF gene’ as opposed to a ‘CF carrier’. The Information film was reviewed and Prof Green recommended that a short segment relating to equivocal results of Newborn Screening for CF
be removed from the film. The issues raised were addressed and the study design was edited accordingly.

3.4.3 Hospital briefings

Briefings took place in each of the six participating Hospitals to ensure standardised practices and comprehension of the research. These briefings took place during Paediatric Cystic Fibrosis Multi-Disciplinary Team (MDT) meetings and tended to last approximately 25 minutes. Those present received a condensed study overview. A detailed study overview was also supplied to each team along with the necessary paperwork for recruitment of participants. It was hoped that these meetings would not only inform MDT members about the study, but would generate interest among MDT members, thus increasing its chances of success. The MDT in Our Lady’s Children’s Hospital Crumlin was briefed on 2nd April 2013, Cork University Hospital was briefed on 23rd April 2013, University College Hospital Galway was briefed on 24th April 2013, Children’s University Hospital, Adelaide and Meath National Children’s Hospital, Mid-Western Regional Hospital, Limerick on 16th May 2013, Tallaght 23rd May 2013, and Temple Street on 24th November 2013.

3.5 Overview of information pack

The information pack sent to parents consisted of an information leaflet and DVD. The information leaflet included frequently asked questions for parents of children identified as carrying an altered CF gene through newborn screening, a summary of important information and two diagrams. The template for the information leaflet was taken from the “European best practice guidelines for cystic fibrosis neonatal screening” (Castellani et al., 2009, p. 169). Several changes were made to this template, these are discussed in detail in Appendix 3. The DVD included in the information pack contained filmed interviews with a CF consultant and CF genetic counsellor. Each one answered a series of frequently asked questions. A detailed overview of the interviews is contained in Appendix 4. Problems with existing information leaflets and justification for using audio-visual information are outlined in the next sections.
There is clearly a need to identify the most effective counseling methods to improve parents’ understanding of genetic information.

3.5.1 Problems with existing patient information leaflets

Many patient information leaflets fail to fulfil their goals. A review of head injury information estimated that less 30% of the population would understand more than 90% of leaflets (Macdonald et al., 2010). A review of written information given to patients and families by palliative care units reported that 64% of leaflets could be understood by approximately 40% of the British population (Payne et al., 2000). A quotation from a 2007 evaluation of written medicines information for Type 2 diabetes gives a voice to patients frustrations:

Too complicated... the words they use, the actual way the documents are written people would tend to look at it and say ‘I don’t understand this’!

(Lee et al., 2007, p. 922)

In their review of 26 orthodontic patient information leaflets, Harwood and Harrison, reported that an IQ of greater than 104 would be needed to understand 42.3% of leaflets. This means that 24-40% of the UK population would not be able to read them (Harwood and Harrison, 2004). It has also been shown that individuals with low levels of health literacy are more likely to be hospitalized, to have inaccurate understandings of their health problems and treatment, and to generally report poor health (Baker et al., 1997).

In all areas of medicine there is increasing awareness that patients need information that is clear, relevant and appropriately timed… Doctors have, historically, been poor communicators of such information.. there are a surprising number of practical problems to be addressed before such communication can become a reality in daily practice.

(Timms et al., 2008, p. 442).

Best practice guidelines were reviewed and incorporated to ensure patient centred communication for the information leaflet. A detailed discussion of the information used in the leaflet is contained in Appendix 3.
3.5.2  Optimal presentation of information

Plain English

Plain English is clearly necessary to communicate with a wide audience. Various guidelines on Plain English were consulted to create the current information leaflet. These included UK Department of Health (2003), Plain English Ireland (2013) and Timms et al, (2008). A questions and answers format was chosen for the leaflet as recommended by UK Department of Health (2003).

Several drafts of the information leaflet were reviewed by a CF consultant (Dr Barry Linnane), a Senior Psychologist (Dr Sorcha Connellan) and a Genetic Counsellor (Dr Alana Ward) to ensure the accuracy of information. This was then reviewed by an official from the Irish Health Service Executive Communication Department, which led to significant improvements from a plain English perspective. Following an initial ethics board hearing in Our Lady’s Children’s Hospital, Crumlin, the leaflet was edited once again. It was felt that in over-emphasising the need for Plain English, the information leaflet had lost some of the necessary factual accuracy. For example, the original leaflet had referred to the passing on of the “CF gene”, however, all humans carry the CF gene, it is in fact the “mutation” or “alteration” to the CF gene which, when passed on, can cause CF. The leaflet was therefore reviewed once again and the necessary changes were made. This episode highlights the difficult balance which must be struck between providing information in a plain and simple manner, whilst ensuring its technical accuracy.

Font size

The European Commission Guideline on the readability of the labelling and package of medical products for human use, outlines that Times New Roman type size 9 should be considered a minimum (2009). In their Clear Print Standard, the Royal National Institute of Blind People recommend type size 12 for general readers and size 14 to 16 for partially sighted readers (Walter, 2011). A minimum of size 12 font has also been recommended for creating written patient education materials (Ivnik and Jett, 2008). The current leaflet uses size 12 in the body of the text and size 14 for headings.
Font Style

European Commission guidelines outline that italics and underlining should not be used and the widespread use of capitals should be avoided (2009). Using groups of capitals is not recommended as they are more difficult to read since there is less distinction between each group of space (Hartley, 1994). Italics should also be avoided as they may reduce the speed at which a reader comprehends text (Kitching, 1990). There is some disagreement about the use of Serif or Sans-Serif typeface. Serifs are the small finishing strokes at the end of a letter, Times New Roman is an example of Serif type, Arial is an example of a Sans-Serif type (Sans from the French word meaning ‘without’). The UK Department of Health recommend the use of Sans Serif, such as Frutiger Roman or Arial (2003). However, it has also been argued that Serif font facilitates readability (Ivnik and Jett, 2008). According to Hanyaloglu of Adobe Acrobat, Serif fonts are more commonly used for printed text in North America, whereas Sans-Serif fonts are more commonly used in web text and European printed text (Hanyaloglu, 2008). Italics, CAPITALISATION and underlining were not used in the current information leaflet. Sans-Serif typeface was used throughout the information leaflet.

Headings

A bold typeface or a different colour may help headings stand out. More than two levels of headings should be avoided as readers may find it more difficult to navigate (European Commission, 2009). Headings were typed in a larger font size and placed in bold to draw attention. Only one level of headings was used in the body of the information leaflet, these were the Frequently Asked Questions.

Colour

“As a general rule dark text should be printed on a light background” (European Commission, 2009, p. 9). There is nearly universal agreement that written information should be highly contrasted with its background (Walsh and Shaw, 2000). Dark text should be framed by white space (Albert and Chadwick, 1992). This allows the eye to be drawn to text and avoids a crowded look (Ivnik and Jett, 2008). In an experiment on web text-background colour combinations, 136 participants rated various colour combinations in terms of
readability and recall. Black text on a white background had the highest participant readability ratings, though recall did not differ significantly (Hall and Hanna, 2004).

*Sentence length*

“Long sentences should not be used. It is better to use a couple of sentences rather than one longer sentence, especially for new information” (European Commission, 2009, p.9). In their review of the literature, Walsh and Shaw (2000), cite Hartley (1994) who states that sentences of less than 20 words are preferable and Maher (1996) who also emphasises short sentence length. A recent study attempted to improve the usability of patient information leaflets by reducing sentence length (from 13.5 words to 11 words on average), and increasing lexical simplification (Matt and Lentz, 2010). The revisions resulted in a higher literacy score, to patients being able to locate information with greater ease, and to the information itself being comprehended and appreciated better (Matt and Lentz, 2010). Attempts were made to minimise sentence lengths at each revision of the current information leaflet.

*Bullet points*

It has been noted that bullet points will gain a reader’s attention more successfully than solid text (Dickinson et al., 2001). Information presented in bullet points is remembered and understood better than information presented in paragraphs (Myers and Midence, 1998). European Commission guidelines state that no more than five or six bullet points in a list are recommended (2009). Ivnik and Jett concluded that “Bulleted items draw attention to specific/key messages” (p. 1040). They also recommended placing key messages in a box in the text. The ‘quick facts’ in the current CF information leaflet consists of 5 bullet points contained in a separate box to the body of text.
Paper weight and finish

EC guidelines outline that Paper should be sufficiently heavy to avoid transparency (2009). Glossy paper should not be used as it reflects light and can impair reading. The current leaflet uses 250grm weight paper with a matt finish.

Authors

In her review of the literature, Shaddock recommends the inclusion of authors' names and credentials in preparing health information for the public (2002). Authors and their credentials are listed at the end of the information leaflet.

Illustrations

A review of 55 studies compared learning from illustrated text with learning from text alone (Levie and Lentz, 1982). The authors concluded that illustrations are more beneficial for people with lower literacy levels. Illustrations may also act as a mnemonic aid for information that is easy to understand but difficult to remember (Levie and Lentz, 1982). In their guide for providing patient health information, Tang and Newcomb report that patients frequently report valuing illustrations (1998). The appropriate use of illustrations is important; Smith and Alford outline that illustrations should be placed next to the text to which they refer (1988). The Adult Literacy and Basic Skills Unit outline that illustrations should relate to the surrounding text and be located at the end of a sentence or paragraph (1994). Ivnik and Jett also concluded that relevant illustrations should be used, but only where they enhance the text (2008). In a study of 637 patients with osteoarthritis, Moll reported that those who received illustrated booklets had greater comprehension as measured on a Multiple Choice Questionnaire, as compared to those who received un-illustrated booklets (1986). Two illustrations were used in the current information leaflet. The first illustrates the inheritance of alterations to the CF gene when both parents carry an altered CF gene. The second illustrates the inheritance of the alterations to the CF gene when one parent carries an altered CF gene. Both illustrations are accompanied by explanatory bullet points.
3.5.3 Justification of written information

The need for clear and concise written information in a healthcare setting has been well established. Gauld (1981) compared the recall of medical information between individuals who received verbal information and those that received both written and verbal information. 67% of those who received written information could recall 90-100% of information, compared to just 28% of those who received only verbal information. George, Waters and Nicholas (1983), compared patient groups who were given information leaflets at the time of their drug prescription with those who had not. Patients who received an information leaflet had more knowledge of the drug and were more likely to report being completely satisfied with their treatment.

In his review of the subject, Weinman concluded that patients want more written information and that this will increase their knowledge and adherence to treatment (1990). However, the efficacy of any written information depends on its adequacy and the extent to which it meets patients’ needs (Weinman, 1990).

There has been some debate on the efficacy of written versus video information for patients. In a study of asthma patients information recall, Wilson et al, reported that primary care patients given take-home print information had better recall than those who viewed video information or controls (2010). Wilson et al, reviewed the literature comparing the efficacy of print and multimedia health materials (2012). 54% of studies reported no difference between the two in terms of health outcomes. Multimedia material led to better patient outcomes in 38% of studies, whereas 9% of studies reported in favour of print material (Wilson et al., 2012). The next section provides further justification for the use of audio-visual information.

3.5.4 Justification of audio-visual information

Barkhordar et al, compared information presented in the form of a leaflet to that presented in a multimedia format (2000). They concluded that both mediums are effective at informing patients but that multimedia information has a more positive short-term effect than an equivalent leaflet. In his review of the literature Kessels recommended that spoken information should be supported by written or visual material (2003). Weiss et al, highlighted the importance of non-written methods of communication for individuals with below-average
literacy (1995). 97% of 177 older adults indicated that they primarily received their information from the television. The authors concluded that clinicians and administrators should therefore consider transmitting healthcare information through non-written means (Weiss et al., 1995). It has been outlined that parents with lower levels of CF knowledge also had lower level of education (Tluczek et al., 1992). Kessels concluded that “Visual communication aids are especially effective in low-literacy patients” (2003, p. 221).

Several recent studies have supported the use of video based information for patients. The results of an online video-based sunscreen information intervention also supports the use of video materials (Armstrong et al., 2011). 94 participants were randomly assigned to video or pamphlet based education. Participants in the video group had greater knowledge and adherence than those in the pamphlet group (Armstrong et al., 2011). Video information has also been shown to increase treatment compliance and attendance at a follow-up appointment in patients newly diagnosed with Obstructive Sleep Apnea Hypopnea Syndrome (OSAHS) (Wiese et al., 2005). A Personal Data Assistant (PDA) based video intervention was conducted with 51 adults who were taking medication for the treatment of HIV/AIDS (Brock and Smith, 2007). The quasi-experimental study reported significant increases in patient knowledge of disease, medications and adherence behaviours. However, it should be noted that no control group was used in this study.

3.6 Statistical analysis

Parents level of stress and their level of knowledge of CF: ‘Between groups t-test’ will be used to assess differences.

Parents decision to undergo genetic testing: ‘Fisher’s exact test for the differences between proportions’ will be used.

Statistical advice was provided by Dr Helen Purtill from the Statistical Consultancy Unit of the University of Limerick.
3.7 Qualitative Analysis

Qualitative data from telephone interviews was conducted using Content Analysis, in accordance with established guidelines (Elo and Kyngas, 2008). Based on the existing research on parental experiences of false-positive newborn screening for CF, a deductive content analysis approach was utilised (Burns and Grove, 2005). This was as opposed to an inductive approach, which is best utilised when there is little or no knowledge available on the phenomenon being studied. The unit of analysis was sentences or shorter meaning units where applicable.

3.8 Methodological strengths and weaknesses

The study initially included a pre and post intervention survey. This would have bolstered the quantitative findings of the study. The pre intervention survey was excluded from the final study design on the request of the ethics committee of Our Lady’s Children’s Hospital, Crumlin.
Chapter 4: Results

4.1  Table 1: Demographic information of parents in intervention and control groups

<table>
<thead>
<tr>
<th></th>
<th>Intervention group</th>
<th>Control group</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total number n</td>
<td>16</td>
<td>16</td>
</tr>
<tr>
<td>Age, mean(SD)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Parent (Years)</td>
<td>31.19 (4.39)</td>
<td>31.00 (5.31)</td>
</tr>
<tr>
<td>Child (Days)</td>
<td>82.81 (19.64)</td>
<td>98.25 (36.47)</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female/Male</td>
<td>14/2</td>
<td>12/4</td>
</tr>
<tr>
<td>Education, n (%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Junior Certificate</td>
<td>1 (3%)</td>
<td>1 (3%)</td>
</tr>
<tr>
<td>Leaving Certificate</td>
<td>3 (9%)</td>
<td>1 (3%)</td>
</tr>
<tr>
<td>Some College</td>
<td>1 (3%)</td>
<td>3 (9%)</td>
</tr>
<tr>
<td>Diploma</td>
<td>3 (9%)</td>
<td>2 (6%)</td>
</tr>
<tr>
<td>Degree</td>
<td>6 (19%)</td>
<td>8 (25%)</td>
</tr>
<tr>
<td>Masters or higher</td>
<td>2 (6%)</td>
<td>1 (3%)</td>
</tr>
<tr>
<td>Sweat test location, n (%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Limerick</td>
<td>3 (9%)</td>
<td>3 (9%)</td>
</tr>
<tr>
<td>Cork</td>
<td>1 (3%)</td>
<td>1 (3%)</td>
</tr>
<tr>
<td>Galway</td>
<td>1 (3%)</td>
<td>4 (13%)</td>
</tr>
<tr>
<td>Crumlin</td>
<td>5 (16%)</td>
<td>2 (6%)</td>
</tr>
<tr>
<td>Temple Street</td>
<td>1 (3%)</td>
<td>2 (6%)</td>
</tr>
<tr>
<td>Tallaght</td>
<td>5 (16%)</td>
<td>4 (13%)</td>
</tr>
</tbody>
</table>

4.1.1 Hypothesis One: Parents’ CF knowledge

Independent t-tests were conducted to compare the CF genetic knowledge as well as CF and CF carrier status knowledge. Parents in the intervention group demonstrated significantly higher CF genetic knowledge scores ($M = 9.5, SD = .63$) than parents in the control group ($M = 8.56, SD = .96$), as measured on an independent samples t-test; $t(30) = 3.25, p = .003$). Levene’s test of equality of variance was not significant ($p = .11$) so equal variance was assumed. The magnitude of the difference between the means was large ($eta squared = 0.26$) as per guidelines of Cohen, 1988.

Parents in the intervention group also demonstrated significantly higher CF and CF carrier status knowledge scores ($M = 9.88, SD = .342$), compared to parents in the control group ($M = 8.75, SD = .931$), as measured on an independent samples t-test; $t(30) = 4.54, p < .001$). Levene’s test of equality of variance was significant ($p = .003$) so equal variance was not assumed. The magnitude of the difference between the means was large ($eta squared = .407$).
4.1.2 *Table 2: CF Knowledge percentages*

<table>
<thead>
<tr>
<th></th>
<th>Intervention Group</th>
<th>Control Group</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Total Knowledge</strong></td>
<td>97%</td>
<td>87%</td>
</tr>
<tr>
<td><strong>CF genetics</strong></td>
<td>95%</td>
<td>85%</td>
</tr>
<tr>
<td><strong>CF and CF carrier status</strong></td>
<td>99%</td>
<td>89%</td>
</tr>
</tbody>
</table>

Mean percentage correct responses

4.1.3 *Table 3: Number of parents with correct CF knowledge answers in each group*

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Answer</th>
<th>Intervention Group</th>
<th>Control Group</th>
</tr>
</thead>
<tbody>
<tr>
<td>CF is a genetic condition</td>
<td>True</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>To inherit CF, both parents must be carriers of CF</td>
<td>True</td>
<td>13 (81%)</td>
<td>14 (88%)</td>
</tr>
<tr>
<td>Genetic testing can establish if a person carries an altered CF gene</td>
<td>True</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>If two parents carry altered CF genes there is a one in four chance of any child of theirs having CF</td>
<td>True</td>
<td>15 (94%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>If neither parent is a carrier or affected by CF they can have a child who carries an altered CF gene</td>
<td>False</td>
<td>13 (81%)</td>
<td>11 (69%)</td>
</tr>
<tr>
<td>There is a vaccine that stops people from getting CF</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>1 in 150 people in Ireland carry an altered CF gene</td>
<td>False</td>
<td>16 (100%)</td>
<td>8 (50%)</td>
</tr>
<tr>
<td>If one parent carries an altered CF gene and the other parent does not, there is a 50:50 chance of their children carrying an altered CF gene</td>
<td>True</td>
<td>15 (94%)</td>
<td>13 (81%)</td>
</tr>
<tr>
<td>1 in 19 people in Ireland carry an altered CF gene</td>
<td>True</td>
<td>16 (100%)</td>
<td>12 (75%)</td>
</tr>
<tr>
<td>If you carry an altered CF gene there is an increased chance of your biological family also carrying an altered CF gene</td>
<td>True</td>
<td>16 (100%)</td>
<td>15 (94%)</td>
</tr>
<tr>
<td>People with CF often suffer from chest infections</td>
<td>True</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>You can catch CF through physical contact with someone with CF</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>People with CF can experience difficulties digesting food</td>
<td>True</td>
<td>16 (100%)</td>
<td>14 (88%)</td>
</tr>
<tr>
<td>To have CF you must have two altered CF genes</td>
<td>True</td>
<td>15 (94%)</td>
<td>14 (88%)</td>
</tr>
<tr>
<td>A person with an altered CF gene will not need special medical treatment</td>
<td>True</td>
<td>15 (94%)</td>
<td>10 (63%)</td>
</tr>
<tr>
<td>Over time, people who carry one altered CF gene can develop CF</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>If you carry one altered CF gene you are more likely to get chest infections</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>A child who carries one altered CF gene will need to be cared for more than normal</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>People who are carriers of CF have a mild form of CF</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>People who carry an altered CF gene become sick more easily than others</td>
<td>False</td>
<td>16 (100%)</td>
<td>16 (100%)</td>
</tr>
<tr>
<td>A person will not suffer health problems because of having one altered CF gene</td>
<td>True</td>
<td>16 (100%)</td>
<td>7 (44%)</td>
</tr>
</tbody>
</table>

n (%) correct responses
4.2 Hypothesis Two: Parent Stress

An independent t-test was conducted to compare the Parenting Stress Index percentiles of the intervention and control groups. There was no significant difference in total Parenting Stress percentiles between parents in the intervention group ($M = 34.63, SD = 24.8$), and control group ($M = 30.13, SD = 24.61$; $t(30) = .52, p = .610$). Levene’s test of equality of variance was not significant ($p = .974$) so equal variance was assumed.

4.2.1 Table 4: Parenting Stress Index percentiles

<table>
<thead>
<tr>
<th>Stress domain</th>
<th>Intervention Group</th>
<th>Control Group</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total Stress</td>
<td>35 (24.8)</td>
<td>30 (24.61)</td>
</tr>
<tr>
<td>Parental Distress</td>
<td>47 (27.91)</td>
<td>42 (28.01)</td>
</tr>
<tr>
<td>Parent-Child Dysfunctional Interaction</td>
<td>33 (20.51)</td>
<td>33 (23.83)</td>
</tr>
<tr>
<td>Difficult Child</td>
<td>34 (27.3)</td>
<td>25 (20.4)</td>
</tr>
</tbody>
</table>

Defensive Responding (n) 2 4

Values: Percentile (Standard Deviation)

4.3 Hypothesis Three: Intention to avail of genetic testing

94% (n = 30) of parents interviewed, intended availing of genetic testing. One couple had already undergone genetic testing due to the use of in vitro fertilisation (IVF), and so were excluded from hypothesis testing. There was no significant difference in intention to avail of genetic testing between the intervention and control group ($p = .484$, Fisher’s exact test). Only one parent and partner definitely did not intend being tested for altered CF genes. All parents intended attending a genetic counselling appointment.

4.3.1 Table 5: Intention to avail of genetic testing

<table>
<thead>
<tr>
<th>Genetic testing</th>
<th>Intervention Group</th>
<th>Control Group*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parent</td>
<td>16 (100%)</td>
<td>14 (93%)</td>
</tr>
<tr>
<td>Partner</td>
<td>14 (88%)</td>
<td>14 (93%)</td>
</tr>
</tbody>
</table>

Values: n (percentage of group). *excluding those previously tested
4.4 Consequence of newborn screening result

The study evaluated parents stress using the PSI-SF, summarised in Table 2. No Parents’ PSI total stress percentile was in the clinical range. Six participant responses were defensive (defensive responding score ≤ 10). Parents gave details of the highly stressful experience between receiving the phone call to book the sweat test and receiving a diagnosis (see 4.3.1). There were several other consequences of the newborn screening result, outlined below. The study established how often parents think about the results as well as their attitude toward the results. A possible effect on family planning also arose as a potential consequence.

4.4.1 Stress in advance of successful diagnostic sweat test

Parents outlined their experiences in advance of the sweat test.

“The waiting was terrible, like waiting for a death sentence” P1.

“I was extremely upset for 24 hours and didn’t look up the consequences” P2.

“It was a huge blast” P8.

“I was panicked because it was the day before, it was a shock” P18.

“it was a shock. I thought the heal-prick was something yea just did, I didn’t expect anything to come back from it” P30.
Most parents had to wait approximately 24 hours for a diagnostic sweat test, however others had a much longer wait (see para 4.9).

“Spent the first month worrying, until my step mother told me to chill out” P12.

“Our child couldn’t get enough sweat, we had a three week wait for another sweat test, me and my partner could barely sleep and eat” P14.

“There were three attempts to get the sweat test done, every day I was so worried about it” P19.

“Sometimes you’re looking for things that are not there.. you’re questioning absolutely everything like. I know we do that anyway, but I guess we just do it in finer detail because of what happened” P29.

4.4.2 Figure 4. Frequency with which parents think about their infants NBS result

<table>
<thead>
<tr>
<th>Frequency of thoughts</th>
<th>% Parents</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not at all</td>
<td>9%</td>
</tr>
<tr>
<td>Seldomly (once or twice)</td>
<td>13%</td>
</tr>
<tr>
<td>Occasionally (once a month)</td>
<td>19%</td>
</tr>
<tr>
<td>Often (once a week)</td>
<td>34%</td>
</tr>
<tr>
<td>Constantly (daily)</td>
<td>25%</td>
</tr>
</tbody>
</table>

4.4.3 Do parents view screening result as a benefit or a harm?

When asked whether they considered their child’s newborn screening result to be a benefit or a harm, all parents indicated that they considered it a benefit.

“knowledge is wealth” P2.

“At the time I was alarmed, but in the long-run it’s better to know” P8.
“It’s a benefit, you know where you stand” P 11.

“It’ll give her better choices in the future when she has children” P 25.

4.4.4 Effect of screening result on family planning

Two parents raised the topic of future family planning;

“I don’t think we would have any more kids if both me and my boyfriend are carriers” P17.

“If me and [partner’s name] are carriers, it could be just lucky that he [newborn infant] doesn’t have CF, that he didn’t take the two genes, we would have to consider if we’d have further kids”.

This was not a subject intentionally broached by the interviewer but was voluntarily raised by parents.

4.4.5 Information seeking

88% of parents sought additional information about CF, prior to their child’s sweat test. 78% of parents used the internet, 22% consulted with a health care professional. Google was used to guide internet searches. Parents specifically mentioned visiting the following websites; Wikipedia, CF Ireland, Munster region CF website and WebMD.

The information which parents uncovered did not always relieve their distress;

“We used google, which made us panic more” P14.

“First thing after the phone call was to google it. We got CF information from google that just made us more anxious” P20.

“There was so much information online, it made me anxious, I thought it was a handicap, I wasn’t sure about the implications for my child” P22.

“I started reading a lot on the internet, and the more I got education about cystic fibrosis, the more I started to feel down, low, and at one stage I tried to puke” P31.

Whilst other parents deliberately avoided seeking additional information;

“I refused to google CF before the sweat test” P15.

Parents experience of accessing information online in advance of the sweat test was not all negative;

“I did a google search so I knew what to expect from the test” P 26.
“We spoke to our own GP on it, but really it was just the internet… you kind of have to do the research yourself to be honest” P32.

4.4.6 Informing others

Informing others of the sweat test result proved to be an emotive issue for parents. The study established if parents intended to inform their child of the NBS result and if so, at what age. The study also established if parents intended informing other relatives, and if so, if the relatives would be interested in genetic testing. The results are outlined below.

4.4.7 Informing child of CF carrier status

All parents intend informing their child of her/his carrier status when their child is in her/his teens/ early adulthood. Some parents specifically mentioned intending to tell their child before he/she becomes sexually active.

“I plan to tell [child’s name] when he is around 14, I think he might meet a girl who is a carrier of the gene as well so I am thinking about the future and about him having his own children so I will tell him definitely” P19.

“I’d like for him, if he’s marrying somebody, to know does that person need to be tested as well, it’s for them to make a decision themselves” P32.

4.4.8 Figure 5: Parents informing relations of CF NBS result

![Bar chart showing communication of carrier information to relatives](image)
Parents had diverse attitudes toward informing their relatives of their infant’s CF carrier status. The majority of parents informed only immediate family of their infant’s CF carrier status. Many parents intended informing extended family if they are found to carry an altered CF gene. Parents who did not share their child’s carrier status with other family members, cited concerns regarding other peoples’ misunderstandings of CF. 19% of parents indicated that sharing carrier information with relatives would be a burden for them. Two parents (6%) were concerned that their child’s carrier status could result in stigmatisation.

“I didn’t tell the relatives (pause) people jump to conclusions” P21.

“I wouldn’t talk about it in my family, if [child’s name] was sick, it would be a big deal” P12.

“I didn’t want everyone to know or treat her differently, I didn’t want friends to treat her differently, my friend has a son and when it comes to dating I wouldn’t want her to be treated differently” P22.

76% of parents shared the information with immediate family members only;

“I kept it to my parents and siblings, I was worried about misconceptions” P 8.

“I told my close family, but my boyfriend told all his relatives, that really upset me, I didn’t want pity, people looking at me, poor you poor you, I wanted to get a routine, I didn’t want pity” P 17.

Motivations to share information included closeness to other family members and siblings who were planning to have children of their own.

“like from [partner]’s side, his brother is getting married” P29.

“We did because my sister is pregnant at the moment, so that was my worry to start with, would she get the same kind of result” P30.

“If I’m a carrier I’ll tell my side, we’re very close anyway” P25.

4.4.9 Relatives’ interest in genetic testing

54% of the parents who informed their relatives of their infant’s CF carrier status, believed their relatives would be interested in genetic testing. 32% indicated that their relations were waiting to see their own results first, before deciding whether to be tested themselves. This indicates that up to 86% of relatives may seek genetic testing. Finally, 14% of parents believed their relatives would not be interested in CF genetic testing.
4.5 **CF awareness**

This section summarises parents’ understanding of their infants’ carrier status and the degree to which they were aware of CF before their infant’s birth.

4.5.1 **Prior awareness of CF**

88% of parents had heard of CF before their infant was born, with 40% of parents actually having known someone who had CF. Only 9% parents knew someone who was a carrier of CF, prior to their child being born.

4.5.2 **Parents’ understanding of sweat-test results**

Reassuringly, all parents said that their infant definitively does not have CF. 94% of parents were aware that their infant definitively carries and altered CF gene. One parent was not sure, and one parent said their infant *may* be a carrier. At the time of the study, the majority (66%) of parents rated their infant’s health as Excellent. 28% of parents rated their infant’s health as Very Good, with a final 6% rating their infant’s health as Good. No parents rated their infant’s health as Fair or Poor.

4.6 **Changes to CF NBS suggested by parents**

The question which evoked the largest response from parents, was question 19; “Is there anything you would change about the process of newborn screening for CF?” It should be noted that 47% of parents did not suggest any changes to the newborn screening process.

“I thought about it and googled things, I’m still supportive, I wouldn’t change anything” P17.

The changes which parents did suggest are divided into several categories;

4.6.1 **Timing of sweat-test**

The time between the telephone call from the CF centre and the confirmatory sweat test was highlighted as being very stressful for all parents. 9% of parents suggested that there should be less of a delay between the phone call and the sweat test

“process should be immediate, it was totally unnecessary” P1.

Whilst two (6%) parents suggested that more time would be beneficial;
“I only got the phone call the day before about the sweat test. It was so last minute. I would prefer three days or so notice. I was panicked because it was the day before, it was a shock” P18.

The majority of parents (84%) did not suggest changing the wait time for a sweat-test one way or another.

“I don’t think so, I don’t think it could be done quicker, only one evening to dangle the nerves” P26.

4.6.2 Mode of communicating heal-prick results

Some parents suggested using a different mode of communicating heal-prick results and booking the sweat-test.

“The process seemed cold, I just woke up from a sleep and got the phone call.. perhaps a script would be useful.. I rang back the CF centre but the phone wasn’t manned, there was no one at the other end of the line, it was tough, I rang my GP’s personal number and he rang the CF centre” P2.

“The phonecall was from some people I had never heard of. A standard call-back from my GP would have been preferable” P8.

“I would change the way you are told.. if maybe GP could call it would be a better option” P31.

4.6.3 Manner of communicating heal-prick results

Parents had contrasting experiences of the manner in which they were informed of their infants heal prick result and the need for a sweat test. 19% of parents criticised the manner of communication. 13% of parents felt that the call was cold or impersonal.

“[the phone call] could have been done with more sensitivity, there was limited information, no statistics, she just told me not to go on the internet. In the hospital I was given more statistics. You need stats with the initial phone call, hearing the statistics was greatly relieving” P4.

“The whole thing was handled really well, the people couldn’t have been nicer.. the lady who phoned was lovely, everyone was lovely and reassuring.” P8.

“Staff were excellent. If at all possible parents should have the appointment (sweat test) very soon. First phone call was very reassuring and calm” P15.

“I would like a little bit more information would have been preferable. I think they should ring and make them feel that it’s not kind of a big thing you know” P31.
“It was a poor phone call, and it was cold as well, I was really upset, it was poorly handled. The initial one was shocking I have to say” P32.

4.6.4 Type of information communicated

Some parents made suggestions for changes to the information communicated. Parents requested more information on the nature of CF and the screening process. Some suggested that the type of disorders being screened with the heal-prick test, should be made clearer from the outset.

“I would’ve liked a better explanation of the screening, let people know the number of people with carriers” P3.

“Maybe let people know they screen for CF before it’s done. People aren’t aware that it’s in the testing. I got a fright before the test, if people were given information before the heal prick they could prepare” P10.

“I would have been more relieved if I’d known that the chances were so low. If I had known there was a possibility of needing a repeat I would have been more anxious” P15.

“When I got the phone call for further testing you need more information, we were told to just come to the hospital, the call was out of the blue” P20.

“We went to the hospital and there was not much information given by the hospital to us regarding CF, what it is or what it’s not” P31.

“I had no knowledge of CF, and after the phone call, my understanding was that he more than likely had CF” P32.

4.6.5 Parents desire for genetic testing of their older children

One parent expressed a wish to have her older children undergo CF carrier testing;

“It’s weird that my other children can’t be tested until after they’re 16, I’d like it for my three year old as the heal prick didn’t test for it at the time” P22.

Whilst another expressed her relief that her first child had already been tested;

“The screening was in place when our first child was born, that would have been horrific, to realize that she hadn’t been tested” P30.
4.7. Multiple sweat tests

In some instances an infant could not produce enough sweat during the initial sweat test (n = 5), or the sweat-chloride result was borderline (n = 1). In these cases, a follow-up sweat test was scheduled. This was a source of considerable stress to parents.

“Our child couldn’t get enough sweat, we had a three week wait for another sweat test, me and my partner could barely sleep and eat” P14.

“We got the phone call when (child’s name) was two weeks old to do the test, but she didn’t make enough sweat. Then we were told that they usually do it at 6 weeks, I would have preferred to just get the test at 6 weeks” P21.

“We thought if she didn’t sweat enough there must be something wrong. It let fears build up, I convinced myself she was sick because she had a little cough or regurgitated food” P25.

“The salt level has to come back between 1 and 30 and [child’s name] scored 41, which put him into a borderline category, so they couldn’t either diagnose it or rule it out, that meant it went for further testing, and his blood went off to Manchester, to test for the full 1500 mutations” P29.

“There were three attempts to get the sweat test done, every day I was so worried about it.. I tried everything to make [child’s name] you know, sweat, you know, so I put on basically all the clothes he had on him, he was so miserable, because you know he was so hot, he was crying, so I was not only worried about the result of the test but you know [child’s name] was crying, so I was stressed, it was tough experience” P19.

“There was one month between the first test and second test and the same between second and third” P19.

“I was 5 weeks early, I came out from hospital with him after 2 weeks and the day after I came out, I got a phone call to say he could possibly have CF. I think it was the following Friday we had the sweat test, and we had one again 2 weeks after that and again two weeks after that” P32.

Parenting Stress Index percentiles for the single sweat test and insufficient sweat groups are outlined in Table 4. Higher PSI percentiles are indicative of more parental stress. Mean total Parental Stress was 20% higher in parents of infants who had more than one sweat test due to insufficient sweat quantity, with the Difficult Child subscale having the largest difference in scores (24% higher in insufficient sweat group). “The Difficult Child subscale focuses on some of the basic behavioural characteristics of children that make them either easy or difficult to manage” (Abidin, 2012, p. 60). PSI scores for the parent of the borderline infant are remarkably low, (PSI percentile = 12). Two parents had infants who underwent three
sweat-tests. Their total PSI percentiles were both 76%, their Parental Distress (PD) subscale percentiles were both in the clinical range; 94% and 86%. “The Parental Distress subscale score determines the level of distress a parent is experiencing in his or her role as a parent as a function of personal factors related to parenting” (Abidin, 2012, p. 60).

4.7.1 Table 6: Multiple sweat test Parenting Stress Index percentiles

<table>
<thead>
<tr>
<th>Stress domain</th>
<th>Single sweat-test (n = 26)</th>
<th>Multiple sweat-test Insufficient sweat (n=5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total Stress</td>
<td>30 (22.8)</td>
<td>50 (29.3)</td>
</tr>
<tr>
<td>Parental Distress</td>
<td>42 (27.6)</td>
<td>58 (29.8)</td>
</tr>
<tr>
<td>Parent-Child</td>
<td>32 (19.2)</td>
<td>43 (34.2)</td>
</tr>
<tr>
<td>Dysfunctional Interaction</td>
<td>26 (22.5)</td>
<td>50 (24.8)</td>
</tr>
</tbody>
</table>

Values: Percentile (Standard Deviation)

4.8 Parents’ opinion of newborn screening

During the interview, parents expressed their views of the CF staff they encountered and of the CF newborn screening process in general.

4.8.1 Professionalism of staff

The vast majority of parents praised the professionalism and care provided to them by staff in CF centres.

“The whole thing was handled really well, the people couldn’t have been nicer” P8.

“The nurse and doctor were absolutely fantastic, brilliant, absolutely brilliant, they made sure I understood everything” P25.

“There’s nothing more you can do really I think. The doctors were so supportive” P29.
4.8.2 Support for newborn screening

All but one parent said they were ‘very supportive’ of CF NBS;

“We had one sleepless night, but I’d go through that 100 times over for the sake of newborn screening” P15.

“110%, I think like previous to that, it would have been at least four before they were diagnosed, the damage could be done, 110%” P29.

The parent who was not supportive of NBS for CF cited the experience as being extremely stressful. She was unwilling to share the test results with her family for fear of misunderstandings; in particular the association between genetic abnormalities and Down Syndrome;

“I was very sensitive and emotional, I lost the ability to make breast milk from the stress and had headaches for a few days afterwards” P1.
Chapter 5: Discussion

5.1 Summary

Parents who received the information pack had significantly more knowledge of the CF carrier status and CF genetics than parents in the control group. All parents understood that their child did not have CF, however, 44% of parents in the control group misunderstood the health implications of carrying an altered CF gene. There was no significant difference in parental stress between parents who received the intervention pack and those who did not. None of the study participants were in the clinical range for stress as measured on the Parenting Stress Index – Short Form. The results therefore do not indicate persistent parental stress arises from false positive CF NBS. However, one in four parents still thought about their infant’s NBS result on a daily basis. Parents of infants who had more than one sweat test due to insufficient sweat quantity had higher overall PSI percentiles (50%), than parents of infants who had just one sweat test (30%), indicating greater parental stress. Two parents of infants who underwent three sweat tests had total PSI percentiles of 76%. The current study indicates that parents of infants identified as carrying an altered CF gene, are overwhelmingly supportive (97%) of newborn screening for CF.

5.2 Evidence for psychosocial adversity

The literature review identified eight potential causes of psychosocial adversity arising from false positive NBS for CF. The evidence from the current study for and/or against each of these is outlined below.

5.2.1 Creating short-term distress in advance of a diagnostic test

Significant parental distress was reported between receiving the phone call to book the sweat-test, and a successful diagnostic test. This high stress period lasted approximately 24 hours for the majority of parents. However, in cases where a repeat sweat-test was required, this high stress period lasted up to two months. This finding is in keeping with other studies which indicate that parents stress levels decrease as soon as they receive the confirmatory test

5.2.2 Long-term anxiety

There is no universally accepted definition of what time period constitutes ‘long-term’. However, according to the DSM 5, the symptoms of Generalised Anxiety Disorder (GAD) need to be present for at least six months in order to be diagnosed as having GAD. In this light, the results of the current study should not be taken as indications of ‘long-term’ anxiety, since the mean age of infants in the study was 90 days. No participating parents had total stress scores in the clinical range at the time of the interview. However, despite the normal range of PSI scores, 59% of parents still thought about the results at least once a week, with 25% of parents thinking about the results daily. Lang et al (2011) reported that just 25% of parents thought about the results at least once a week. However, the lower population incidence of CF in the US and the fact that infants were an average of 21 days older in Lang et al’ study, may account for some of this difference. Two parents of infants who underwent three sweat tests, had Parental Distress subscale scores in the clinical range. Further investigation is required to establish if multiple sweat-tests arising from insufficient sweat quantity results are correlated with long term anxiety in parents. Despite parents’ persisting thoughts about the result, the current study does not indicate that long-term anxiety arises from false-positive CF NBS followed by a single sweat-test.

5.2.3 Vulnerable Child Syndrome

One of the true/false questions in the CF knowledge questionnaire was “A child who carries one altered CF gene will need to be cared for more than normal”. A ‘true’ response to this question may be indicative of perceived child vulnerability. All parents correctly responded ‘false’ to this item, a clear indication of a lack of VCS traits among participating parents. This is in keeping with evidence from Cavanagh et al (2010) who reported no long-term evidence of child vulnerability following false-positive CF NBS. The Parent-Child Dysfunction Interaction (P-CDI) sub-scale of the Parenting Stress Index assesses parents’ perception that a
child does not meet expectations and that interactions with the child are not reinforcing, and includes items such as “My child is not able to do as much as I expected” (Abidin, 1995). P-CDI scores in the current study were in the normal range, with an overall mean percentile of 22%. Finally, 94% of parents rated their child’s health as excellent or very good, no parents rated their child’s health as fair or poor, this is in keeping with Lang et al (2011), who reported that 98% of parents rated their infants’ health as good to excellent. In short, despite evidence of possible VCS following false-positive NBS results for other disorders (Tu et al, 2012, Waisbren et al, 2003), these results are not indicative of VCS arising from false-positive NBS for CF. The burden of proof now lies with those who argue for the existence of VCS in parents of infants identified as carrying an altered CF gene.

5.2.4 Unwanted information that may influence future reproductive choices

The current study did not seek to assess the influence of false-positive CF NBS on future reproductive decisions. However, one parent did outline that if she and her partner are found to carry altered CF genes, they would choose not to have any more children. Another parent indicated that if she and her partner are found to be carriers they would “have to consider if we’d have further kids”. These statements appear to support the existence of this unintended consequence of CF newborn screening, as outlined by Ciske et al, (2001) and Wilfrond et al, (2005).

5.2.5 Stigmatisation of the child

This study did not explicitly seek to assess the potential for child stigmatisation. Two parents (6%), did cite concern for how their child would be treated as a result of carrying an altered CF gene. This concern regarding stigmatisation is in keeping with a long-term follow-up of parents of infants with false-positive CF NBS results, in which a minority of parents expressed this concern (Tluczek et al, 2010). However, all parents in the study viewed their infant’s CF NBS result as a benefit, rather than a harm. A concern regarding stigmatisation in a minority of parents does not equate to actual stigmatisation. Longitudinal studies would be required to assess this potential phenomenon. However, if parents are in a position to discuss the meaning of being a CF carrier with their child, they may be better equipped to negate any stigma. Thus, further emphasising the need for adequate CF parental education.
5.2.6 Burden of sharing carrier information with other family members

Parents expressed different attitudes to sharing their infants CF NBS result with family members. 91% of parents shared the carrier information with family members, and 9% did not. 19% of parents made some indication that sharing carrier information would be a burden for them. This is in keeping with a review by Parson and Bradley (2003), who concluded that for a minority of parents, sharing carrier information is viewed as a burden. An information letter for distribution within a family has previously been used to facilitate such communication (McClaren et al., 2010), and may be useful to parents. In their tailored approach to genetic counselling, Tluczek et al (2010) outlined that parents may appreciate being able to re-contact genetic counsellors, when sharing genetic information with relatives. Given the six-month wait time for an initial genetic counselling appointment in the Republic of Ireland, re-contacting genetic counsellors may not be practicable, under the current system.

5.2.7 Parents not facilitated by health service in having other children tested

The current study did not explicitly seek to assess parents’ attitude toward having other children tested. Giving parents unsought genetic information regarding their newborn infant, is a consequence of false-positive newborn screening for CF. Having made attempts to facilitate parents’ comprehension and acceptance of this information, the health service does not provide genetic testing for the child’s older siblings. One parent expressed a desire to have another child tested, and frustration that this could not be done until the child is aged 16, another parent expressed her relief that CF NBS was in place when her older child was born. Since the benefit of testing other siblings would not accrue until adulthood, it is standard practice in the US and UK that siblings can only be tested when they are old enough to legally consent (Clarke, 1994, American Society of Human Genetics, 1995).

5.2.8 Non-paternity

Since parents had not yet undergone genetic testing at the time of the study, it was not possible to have encountered an instance of non-paternity.
5.3. Parents CF knowledge

A tailored information pack was developed to maximise the chance of increasing parents understanding of the CF carrier status, whilst minimising stress. Best practice guidelines and countless drafts were needed to create the CF information document. Scripted interviews with healthcare professionals were recorded and edited to create the information film on DVD. Several studies indicated that audio-visual information can aid comprehension, particularly for individuals with lower levels of education (Kessels, 2003). Some individuals have a preference for one means of communication over another (Wilson et al, 2012). Information presented verbally in a healthcare setting has very low recall rates (Gauld, 1981). There is an onus on health services to proactively develop means of multimedia communication. This is particularly the case for information relating to genetic diseases, where the implication of adequate comprehension, is far wider than a single patient.

Parents in the current study had a good understanding of the overall implications of being a CF carrier. All parents correctly indicated that their child definitely does not have CF, and 94% indicated that their child was definitely a CF carrier. Tluczek et al, (1992) reported that 5% of parents believed that their child might still have CF. 6% of parents in Lang et al’s 2011 study, were not sure that their child did not have CF. However, a significant minority of parents in the current study misunderstood the health implications of being a CF carrier. The CF and CF carrier status question with the highest rate of incorrect answers was Question 10; “A person will not suffer health problems because of having one altered CF gene”. 44% of parents in the control group (N = 7) incorrectly answered ‘false’ to the question. No parents in the intervention group incorrectly answered this question. The CF genetics question with the highest rate of incorrect answers was Question 7; “1 in 150 people in Ireland carry an altered CF gene”, with 50% of parents in the control group incorrectly answering “True” to the question. Once again, no parents in the intervention group incorrectly answered this question. Parents’ misunderstanding of the health implications of being a CF carrier is the most troubling misconception. Thankfully, the intervention pack appears to address these and other misconceptions.

Parents’ CF knowledge scores are high when compared to similar measures from other countries. 33% of parents in one Australian study (Mischler et al, 1998) and 40% of parents in second Australian study (Lewis et al, 2001), did not know that there is a one in four chance of having a child with CF if both parents are carriers of an altered CF gene. This
compares to just one parent (3%) in the current study. Using their true/false questionnaire, Lang et al, (2011) reported that 26% of parents believed that carriers of CF can develop CF over time. No parents in the present study held that mistaken belief.

The study also established if parents knew someone with CF or who carries an altered CF gene prior to their child’s birth. More parents knew someone who had CF (n = 13), than who carried an altered CF gene (n = 3), prior to their child being born. This is despite it being far more common to carry an altered CF gene than to actually have CF (1 in 19 vs 1 in 1,461 respectively in Ireland – National Centre for Medical Genetics, 2011). The number of people who know someone who carries an altered CF gene is likely to rise as NBS continues to identify them. It therefore becomes all the more important that people who carry an altered CF gene, understand the carrier status.

5.4 Parents information seeking and communication

Parents are not passive recipients of information, they actively seek out information from various sources. On successful completion of the sweat test, parents themselves become the primary means of communicating CF/genetic information to their relatives. Parents may communicate their infant’s CF NBS result with relatives, and in time, with their child.

5.4.1 Parents information seeking

88% of parents sought additional information in the time between receiving the phone call and their infant’s sweat test. 78% of parents sought information online, 22% of parents sought additional information from health care professionals, such as their GP or qualified friends and family. Dillard and Tluczek (2005), also reported multiple information resources being accessed by parents. This is in keeping with a finding that parents seek information from the internet to relieve their anxiety, sometimes with the opposite effect (Schmidt et al, 2012). Some parents found the information helpful, whilst others felt it increased their anxiety. Some parents expressed their limited capacity to absorb the information being given to them during the call. The variation in parents willingness and capacity to absorb information, coupled with differing reactions to that information, show that there is probably no catch-all solution to parents information needs. Rather, it appears that the health service
should enable parents to actively determine the medium and quantity of information they require, in keeping with Tluczek et al (2011) tailored approach to genetic counselling.

Rock et al (2005) called for improved methods of communicating this complex genetic information to parents. One parent suggested that follow-up information should be provided to them via email. Given the short wait for a sweat test, it would not be possible to post information to parents in advance of the test, nor would it be practical to ask parents to call to a CF centre for additional information. Email may be a timely and cost effective means of providing accurate information to parents before the sweat test. An alternative would be to guide parents towards a specific website, such as CF Ireland, which could host approved multimedia information. Tluczek et al (2006), outlined five categories of information required by parents following NBS for CF, these may for the basis for the content of this information. The current practice of advising parents not to search for information online appears to be a futile exercise. Active engagement between the health service and online resources can ensure that clear and concise information is accessible to parents online.

5.4.2 Parents communication of CF NBS information with relatives and child

76% of parents had informed their immediate family of their child’s carrier status, with 15% informing extended family. A minority of parents (9%) chose not to tell their relatives. This decision appeared to be predicated on the fear that relatives may misunderstand the implications of being CF carrier. Similar figures have been reported in Wisconsin (Mischler et al, 1998) and Chicago (Lang et al, 2011), where 88% and 89% of parents informed family members of their infants carrier status, respectively. Content analysis of interviews identified several motivations for sharing or withholding this information. Parents who did not share information expressed a desire for privacy or worried that their child may be treated differently as a result of carrying an altered gene. Those who shared the information cited closeness to other family members and outlined that it might be of benefit for siblings who are starting families. Many parents expressed the view that they would share the carrier information with extended family if they were found to carry an altered CF gene.

All parents said that they would inform their child of his/her CF carrier status when their child is a teenager/ young adult. Parents’ high levels of CF knowledge may have
influenced this; Cavanagh et al (2010) outline that one of the main reasons cited by parents for not informing their children, was a lack of CF knowledge.

5.5 Parents’ attitude toward screening result

97% of parents in the current study were “very supportive” of NBS for CF. That is slightly higher than the 90% support rate reported in France by Mischler et al (1998). A longitudinal study conducted in France, reported that all parents of children identified as carrying an altered CF gene would have the test performed on another child (Beucher et al, 2010). Previous studies have evaluated the potential negative psychosocial consequences of false-positive NBS and postulated the balance of benefit and harm to parents (Farrell & Farrell, 2003). However, at the time of publishing, no research had asked parents whether they view their child’s newborn screening result as a benefit or a harm. The results of the current study show that parents unanimously viewed their infants’ newborn screening test result as a benefit. This positive view of the newborn screening result was independent of parents’ opinion of the screening process. Several parents wished to change the process, one did not even support NBS for CF, but all viewed the screening result as a benefit rather than a harm.

5.6 Genetic testing

As testing becomes more advanced, people will be presented with more personalised genetic information. Crucially, some genetic knowledge is not actively sought by people, but is presented out of the blue, such is the case with CF newborn screening. By understanding how best to communicate genetic information to parents, we can both facilitate their own understanding as well as enable them to accurately inform family members. However, the potential risks of CF genetic testing, such as stigmatisation, must also be examined. The present study shows that all parents considered the knowledge of their infant’s carrier status to be a benefit.
5.6.1 Genetic testing rates

If parents receive genetic testing it facilitates them in making more informed family planning decisions in the future. The vast majority of parents (94%) and their partners (88%) were interested in receiving CF genetic testing. Genetic testing rates from other countries have been reported as low as 34% of fathers in Wisconsin (Ciske et al., 2001), and as high as high as 85% in a group of Australian parents (Lewis et al, 2006). However, parents stated intention in an interview scenario may not lead to them actually undergoing genetic testing. Genetic testing figures for the first year of CF NBS operation were requested from the National Centre for Medical Genetics. They reported that for the period 01 July 2011 to 30 June 2012, 50 couples were referred for genetic counselling and offered appointments. Eight couples cancelled or failed to attend the appointments, two of these couples arranged genetic testing, for both parents, through their GP. 41 of the 42 couples who attended genetic counselling proceeded with carrier testing (Ward, 2014). In the case of one couple, only the mother received genetic testing. This gives a national carrier testing rate of 88% for mothers and 86% for fathers. These testing rates compare very favourably to reported rates from other countries.

5.6.2 Genetic testing of relatives

Those identified as carrying altered CF genes may inform their relatives of the increased likelihood that they too may carry an altered CF gene. This enables a cascade effect of genetic testing and awareness of the CF carrier status. 86% of parents believed their relatives would be interested in receiving CF genetic testing, if they are identified as carrying an altered CF gene. McClaren et al (2010) reported that an average of 2.7 non-parent relatives of infants diagnosed with CF, availed of CF carrier screening. Given the growing waiting period for parental genetic counselling and testing, now approximately six months, and the expanding cascade effect of genetic screening, it may be necessary for the health service to provide additional resources to provide testing to parents and their relatives.
5.7 Multiple sweat tests

In some cases the sweat-test was unable to diagnose the infant and so a repeat test had to be scheduled. Five infants (16%) could not produce enough sweat, one infant (3%) had a borderline sweat-chloride result. Parents found the wait for a repeat sweat test very stressful. Mean PSI percentiles for parents (n = 5) whose infants could not produce sufficient sweat was 50% as compared to 30% in the single sweat test group. In two cases, two repeat tests were needed, resulting in a prolonged diagnostic delay. These parents’ total PSI percentiles were both 76%, their Parental Distress (PD) sub-scale percentiles were both in the clinical range; 94% and 86%.

There is a balance between the benefits of early diagnosis of CF and the harm of putting parents through undue distress. A 24 hour period of stress for parents has, for the most part, been accepted as a necessary evil of CF NBS. If screening programmes are to continue to ensure more good than harm, the risk of a protracted diagnostic delay should be minimised. Preterm birth, low birth weight, CF care center, and race have been associated with insufficient sweat production during CF NBS confirmatory testing (Kleyn, 2011). The US CF foundation released a standard that the sweat quantity not sufficient (QNS) rate, should be ≤10% for patients 3 months old or younger (CFFC, 2009). However, QNS rates as high as 26% have been reported in testing of infants less than six weeks old (Farrell et al, 2008).

An infant’s corrected age, that is gestational age plus delivery age, appears to account for a significant amount of QNS. Kleyn et al reported that infants tested less than 39 weeks of corrected age, were 7.4 times more likely to have a QNS result (2011). One infant in the current study who was born one month premature, underwent three sweat tests. This finding is in keeping with a Canadian study which reported a sweat test QNS result in 18.3% of infants <3 months old, compared to 4.5% in infants >3 months old (Beachamp, 2005). The Canadian study went on to recommend that centres should conduct a minimum of 50 sweat tests per year, 10 per operator (Beachamp, 2005). These testing rates would prove difficult to achieve under the current system in the Republic of Ireland, with only 52 sweat tests being conducted between 1st July 2011 and 30th June 2012 in 6 CF centres (Ward, 2014). An infant’s corrected age and other predictors may be used to minimise the risk of QNS results.
5.8 Changes to CF NBS suggested by parents

Parents were very satisfied with the professionalism and care given to them by CF staff in all hospitals. Several potential changes to CF NBS were nevertheless communicated by parents. These are divided into the following categories; initial communication, information communicated, and timing of sweat-test.

5.8.1 Initial communication.

In keeping with European best practice guidelines, the time between the positive screening result and the diagnostic sweat test is kept as short as possible, to minimise parental distress (Castellani et al, 2009). Informing parents via telephone call from the CF centre is the preferred method in many countries (Nahrlich and Zimmer 2013). The means of communicating heel prick results were criticised by 19% of parents. Some parents suggested a more personal approach to the communication of this stressful information. The information provided to parents is currently standardised in Ireland. Some parents (13%), felt that the telephone call was cold or impersonal. Given the limited information available, and the realities of the risk of CF, this will always be a difficult phone call for parents and staff. Providing this information through local health care providers or general practitioners, as suggested by some parents, may lead to a more personal approach but could also increase the chance of misinformation and the monetary cost of screening. Staff in CF centres were given communication skills training on the initiation of CF NBS in 2011. However, they have not been afforded an opportunity to take part in follow-up training or workshops. Follow-up workshops and communication skills training for staff that make these difficult phone calls, may be of benefit to both parents and staff.

5.8.2 Information communicated

The type of information communicated in advance of the sweat test was criticised by some parents. Three parents (9%) outlined that people should be made aware of the fact that CF is included in NBS. Two parents (6%) recommended that more information be provided during the initial phone call. It is worth noting that research on communication in a health care setting identifies divergent attitudes on patients’ desire for information. Namely, some people
desire more information than others (Borracci et al, 2012). It may be the case that staff should be afforded some leeway in the information they communicate to parents during the phone call. As previously outlined, a follow-up email or website containing approved information may also be of benefit to parents.

5.8.3 Timing of sweat test

The timing of the sweat test was criticised by 16% of parents. However, whilst some wanted to expedite the sweat testing process (9%), others wanted more time prior to the sweat test (6%). From the literature review it is clear that a 24 hour wait is current European best practice (Castellani et al, 2009).

5.9 Recommended changes to CF NBS in Ireland

In reviewing the CF information being provided to parents, Clayton (2005) concluded that the majority of information is provided in the peripartum period, reflecting the origins of NBS in paediatric practice. Clayton concluded that this is a terrible time for communicating information. The health service needs to tailor CF NBS programmes for the needs of its clients, and not simply replicate existing practices. The following are six recommended changes to CF NBS based on the findings of the current study;

1. Follow-up communications skills training for staff who make the sweat-test phone call.

2. Email or online multimedia information made available to parents after receiving the phone call.

3. Information pack given to parents of children diagnosed as having one altered CF gene.

4. Adoption of international best practice to minimise the number of quantity not sufficient (QNS) sweat-chloride results.
5. Providing parents with an information letter to facilitate discussing their infant’s NBS result with relatives.

6. Greater resources for genetic testing of parents and relatives in order to minimise wait time.

5.10 Study limitations

The study aimed to recruit a total of 40 participants but succeeded in recruiting only 32. A larger sample would have increased the power of quantitative data, and may have given more depth to qualitative data. Unfortunately, the data collection was constrained by ethics boards to 12 months. Attempts were made throughout the data collection period, to ensure a high participation rate. These included phoning and emailing CF nurses and consultants, to give study updates and to reiterate the deadline for data collection. Collecting study data via telephone interview may have led to some defensive responding. Six study participants were deemed to be defensive in their PSI responses. Defensive responding has been reported in a previous study of infants found to carry an altered CF gene (Baroni et al, 1997), and may be a trade-off for the high participation rate associated with telephone interviews (Lang et al, 2011). Ideally the study would have established the genetic testing rates in the intervention and control groups. It may be that the significant increase in CF knowledge in the intervention group, led to an increase in genetic testing. However, the wait time for a genetic counselling appointment increased from an average of three months at the time of planning the study (early 2012) to an average of six months by the study’s conclusion (mid-2014). This, coupled with assessing parents’ experiences of genetic counselling, may be topics for future research.
5.11 Conclusion

It has been stated that policy makers must balance potential benefits and risks when introducing CF into newborn screening programmes (Wilfond et al, 2005). False-positive results has been identified as the primary harm of CF newborn screening (Centre of Disease Control, 1997). A review of genetic counselling recommended the use of data from multiple sources for parents (Tluczek et al, 2011). The current study developed a tailored information pack for provision to parents which significantly increased their knowledge of the genetics of CF and the implications of carrying an altered CF gene. All parents were aware that their child carried an altered CF gene, but 44% of parents in the control group misunderstood the health implications of being a carrier. Parents recounted considerable distress between receiving the CF centre phone call and their infant successfully completing a diagnostic sweat test. This took approximately 24 hours in most cases but lasted up to two months in cases where an infant could not produce enough sweat, or had a borderline result. At the time of interview, the vast majority of parents had stress scores in the normal range as measured by the Parenting Stress index. Parents suggested various changes to the newborn screening process, some more feasible than others. Parents unanimously viewed their infant’s newborn screening result as a benefit rather than a harm. Despite their infant’s false positive result, parents were overwhelmingly supportive of newborn screening for CF. Based on these findings, the study recommends six changes to CF NBS practices in the Republic of Ireland.
Appendix 1: Study information provided to parents by CF consultant

Cystic Fibrosis Newborn Screening Study,
Psychology Services,
St. Joseph’s Hospital,
Mulgrave Street,
Limerick.

Part 1: Study information to be read to participating parents by CF consultant on receipt of sweat-test results.

1. There is an on-going Irish study which hopes to learn about parents’ experiences of newborn screening for Cystic Fibrosis. It is hoped that by better understanding parents’ experiences, it may be possible to improve the screening process.

2. Participation in the study is entirely voluntary; you will receive the same level of care whether or not you choose to take part in the study.

3. If you agree to take part, a researcher will contact you by telephone in about a month and to ask you some questions. The questions cover a range of topics; your experiences of the newborn screening programme, your view of Cystic Fibrosis, any changes you would make to the screening, your levels of stress, and some general questions such as your gender and age. There will be a total of 63 questions and the survey will take about 20 minutes to complete. As part of the study you may be posted some information about CF and the newborn screening process.

4. If you take part in the study and choose to receive genetic counselling, the researchers will be informed. The researchers are also interested in your experience of genetic counselling and whether or not you choose to have genetic testing. If you take part in the study, a researcher will call to your house in about 8 months to learn about your experience of the screening process and genetic counselling. This interview will last about 30 minutes.

5. If you agree to take part, I will pass on your contact details to the researchers. In about two weeks you will get a letter in the post that tells you more about the research.

6. You can stop taking part in the study at any time.

7. We really appreciate your time in helping us to improve newborn screening for cystic fibrosis in Ireland.

8. If you are happy to take part, is there a particular day of the week that would suit you for a researcher to call? And would morning, afternoon, or evening suit you best to talk?
Part 2: Parent contact Information, to be returned by CF centre.

Please fill in the necessary information and send to:

Cystic Fibrosis Newborn Screening Study, Psychology Services, St. Joseph’s Hospital, Mulgrave Street, Co. Limerick.

Or email:
stephen.quigley@ul.ie

Parent name: ________________________________

Child name: ________________________________

Contact Telephone Number: ________________________________

Contact Address: __________________________________________

Parent’s preference for day and time of phone call:

Suitabl □ □ □
Dear Parent,

We are writing to you in relation to the Cystic Fibrosis (CF) Newborn Screening Study. Your CF consultant will have told you about the study when you received your baby’s newborn screening results. Newborn screening for CF has been in Ireland since July 2011. This study aims to learn about parents’ experiences of the screening. It is hoped that by learning about your experience, it may be possible to improve the screening process.

If you agree to take part in the study, a researcher will phone you in about four weeks to ask you some questions. The questions cover a range of topics; your experience of the newborn screening programme, your view of Cystic Fibrosis, changes you would make to the screening, your levels of stress, and some general questions such as your gender and age. There will be a total of 63 questions and the survey will take about 20 minutes to complete. You may also be posted some information about CF and the newborn screening process. We are also interested in your experience of genetic counselling. If you take part in the study, a researcher will arrange to meet you in about 8 months to learn about your experience of the screening process and genetic counselling. This interview will last about 30 minutes. If you feel under stress, or if you have any other health concerns, please don’t hesitate to contact your local doctor for advice.

If for any reason you do not wish to take part in the study, you can simply complete the cutaway portion below and return it in the envelope provided, or you can email stephen.quigley@ul.ie or telephone 087 6783300. As outlined above, if you want to take part in the study you do not have to do anything at this stage. We really appreciate the work and the time you are giving to help improve newborn screening for CF in Ireland.

Sincere Thanks,

Patrick Ryan, head of department of education and professional studies, University of Limerick.

Barry Linnane, Pediatric respiratory consultant, Midwest Regional Hospital.

Sorcha Connellan, senior clinical psychologist, Midwest Regional Hospital.

Stephen Quigley, psychology research student, University of Limerick.

I do NOT wish to partake in the Cystic Fibrosis Newborn Screening Study.

Name (Please Print): _______________________________

Reason for not taking part (optional): ____________________________________________________________
Appendix 3: Changes made to European best practice information leaflet

The initial template for the information leaflet was taken from the “European best practice guidelines for cystic fibrosis neonatal screening” (Castelanni et al, 2009, p. 169). Some additional questions and answers were added. In all cases identifying a child as a ‘carrier of CF’ was avoided, rather they are identified as a ‘child who carries an altered CF gene’. A breakdown of the changes made to individual questions, and an explanation for each change is provided here;

1 Original Question and answer:

What is my baby's screening result?

The screening result suggests that your baby is a carrier of CF. Approximately 1 in every 20 to 37 healthy people are carriers of a mutation in the CF gene.

1 New Question and answer:

What was my Child's screening result?

The screening result shows that your child carries an altered CF gene. That makes them similar to 1 in 19 people in Ireland who carry CF.

1 Explanation:

The carrier rate was changed to reflect the rate in here in Ireland.

2 Original questions and answer:

What does it mean to be a carrier of the CF gene?

Your baby is just like one of his or her parents and has only one copy of a mutated CF gene. To have CF you need two copies of a mutated CF gene passed from each of the baby's carrier parents.
Why is my child a carrier of Cystic Fibrosis?

A child with an altered CF gene must have inherited it from one parent. This means that the child’s mother or father also carries an altered CF gene. There is a chance that both parents carry an altered CF gene. The following two diagrams show how couples with one or two altered CF genes can pass on the altered gene to their children.

Explanation:

The term ‘mutated’ was avoided due to its negative connotations. The word count of sentences was reduced. The original sentences for this answer were on average 20.5 words long, the revised sentences are on average 14.2 words long. Each sentence makes a clear point. Providing diagrams and other visual aids where possible is in keeping with best practice in this area. In keeping with the NHS Toolkit for producing patient information (2003) “Diagrams and pictures are very effective and should be in line with our communication principles. Where appropriate, use them to illustrate the text..” (p. 6). Since the concept of genetic inheritance is complex. However, this concept is crucial for parents to understand. It increases their knowledge of their own genetics and that of their child.

How will being a carrier affect my child?

Your child will not be affected by the condition and will not need any special treatment. ‘Carriers’ can pass on the altered gene to their children and you may wish to tell your child this when they are older.
How does being a carrier of Cystic Fibrosis affect my child?

Although your child carries CF, he or she will not have any health problems because of this. Your child will not need any special treatment. People who carry an altered CF gene may pass it on to their children. You may want to tell your child later in life that he or she carries an altered CF gene.

3 Explanation:

The sentences were broken down to increase comprehension. Each sentence delivers one clear message. Although there are more sentences, the word count of sentences is smaller.

4 Original questions and answer:

What is CF?

Children with CF are susceptible to chest infections and may not put on weight like they should.

4 New question and answer:

What is Cystic Fibrosis?

Cystic Fibrosis (CF) is an inherited condition, affecting mainly the lungs and digestive system. To have CF you must have two altered CF genes. People with CF can suffer from chest infections and experience difficulties digesting their food.

4 Explanation:

It was felt that it was necessary to expand on the answer. Some of the structure was taken from the Newborn Screening Cystic Fibrosis Association of Ireland information leaflet (2011).
Original questions and answer:

Is it possible that my child does have CF?

The answer to this question is determined to some extent by the CF NBS programme. However, the screening test is not perfect and there is a very small risk that your baby has CF. There are uncommon mutations in the CF gene that are not recognised by the screening test. It is therefore possible that a baby with this result will have a second, uncommon CF gene mutation and will have CF. If you are worried about the result you should discuss this issue with your family doctor.

New question and answer:

Does my child have CF?

No. Your child’s sweat test indicates that he or she does not have CF.

Explanation:

It was felt that giving a definitive answer to this question is crucial in alleviating anxiety. This is the message that parents are given following their infant’s sweat test. This message should remain plain and simple. In his review of the literature, Kessels outlines that “Simple and specific instructions are better recalled than general statements” (2003, p. 221).

Original questions and answer:

If we have children in the future, could they have CF?

Your baby has been recognised to be a carrier of CF, but there is an increased risk that if you have children in the future they may have CF. Accessing genetic counselling is preferable and could be important before planning further pregnancies.
New question and answer:

If I have children again in the future, could they have Cystic Fibrosis?

Since you and / or your partner have an altered CF gene, there is a increased chance that if you have children in the future they may have CF. This will vary depending on you and your partner’s genetic family history and ethnicity. A genetic counsellor can give you much more detailed information on the chance of having a child with CF.

Explanation:

A short explanation of risk variance is included. The word “risk” is replaced by “chance” due to negative connotations of “risk”.

Original questions and answers:

Who else can I talk to about my baby's screening result?

You can discuss this with your health care professional.

Where can I find more information or support?

[INSERT Local CF Association address, phone and website].

New questions and answers:

How do I find out more about Genetic Testing?

The best person to talk to about genetic testing is a Genetic Counsellor. You may already have an appointment to see a Genetic Counsellor. All parents of children who are shown to carry a CF alteration by newborn screening should be offered a referral to a Genetic Counsellor. If you are unsure about this you could contact your local Cystic Fibrosis centre to ensure a referral has been sent. Contact details for your local centre are provided here. Since the
introduction of newborn screening for CF to Ireland, the majority of parents whose children were identified as carrying an altered CF gene, have received genetic testing.

How do I find out more about Cystic Fibrosis?

There are some useful websites which give detailed information about CF. These include the Cystic Fibrosis Association of Ireland (www.cfireland.ie), and the Cystic Fibrosis Trust (www.cftrust.org.uk). However, remember your child does not have CF, they simply carry a single altered CF gene.

Explanation:

Genetic testing was added to the FAQ's as parents understanding and acceptance of genetic testing is a key variable of interest in the present study. Websites of interest are added and it is re-emphasised to parents that their child does not have CF. Contact details for local CF centres are included at the end of the leaflet. The Theory of Planned Behaviour was utilised in the phrasing of this question.

The Theory of Planned Behaviour is a cognitive model for understanding and predicting human behaviour. In their 2001 meta-analytical review, Armitage and Conner identified 185 independent studies which had utilised the Theory of Planned Behaviour up to the end of 1997. The review found that the Theory of Planned Behaviour accounted for 27% of behaviour, and 39% of intention to carry out a behaviour (Armitage and Conner, 2001). The relative success of the TPB led to it being selected as the cognitive model for creating the CF information leaflet and film. The current version of the Theory of Planned Behaviour was outlined by Icek Ajzen in 1991. In this article, Ajzen outlines that the domains of Attitudes toward a behaviour, subjective norms with respect to a behaviour, and perceived control over a behaviour, can accurately predict the intention to carry out a given behaviour (Ajzen, 1991). By clearly outlining how genetic counselling can be availed of, we may increase an individual’s perceived control over the behaviour i.e. how easily the behaviour can be completed. Perceived behavioural control was the
The strongest predictor of intention to undergo a mammography; it explained 24% of variance in intention (Steele and Porche, 2005). Question 7 above outlines that “Since the introduction of newborn screening for CF to Ireland, the majority of parents whose children were identified as carriers of an altered CF gene, have received genetic testing”. This establishes that genetic testing is ‘the norm’, potentially having a positive influence on an individual’s subjective norm and increasing their likelihood of undergoing genetic counselling.

The topic of parental carrier screening was then added to the frequently asked questions.

What does genetic testing involve?

After you have spoken to your genetic counsellor it is usually possible to get a genetic test. A sample of blood is drawn to be tested. It takes between six and eight weeks to get the test results. Genetic testing is free of charge.

What do the results of genetic testing mean?

Since your child carries an altered CF gene, either you or your partner must also carry an altered CF gene. However, it may be the case that both of you do. Genetic testing can check you and your partner for the most common CF gene alterations. If genetic testing does not find any of the common CF alterations, this would be reassuring and there would be a much smaller chance that both of you carry an altered CF gene. Finding out that you carry an altered CF gene means that extended family members are at an increased risk of carrying the family CF alteration.

Explanation:

Parental genetic testing is a key variable of interest in the current study. The knowledge that a newborn baby is a carrier of an altered CF gene presents relatives with the choice to undergo genetic testing. Wildhagen, Kate and Habbema outline that the “identification of newborn carriers provides an opportunity to test both parents with a view to ascertaining previously unrecognized high risk couples and
extend their future reproductive choices” (1998, p. 863). The authors go on to outline that contact with a known CF carrier relative may engender greater familiarity with CF and therefore facilitate more informed choices about screening and reproduction than would be possible for the general population (Wildhagen et al, 1998). Parents’ who establish their own CF carrier status may make a connection between their own good health and that of their CF carrier child. By engendering a more positive view of genetic testing and individual’s establishing specific genetic susceptibility, we may be able to increase the rate of genetic testing.

Finally, one of the early FAQ’s addresses the NBS process itself;

How did the screening identify my child?

A small prick of blood was taken from your child’s heel when he or she was a few days old. This blood sample was tested for various conditions, including CF. Your child was found to have high level of an enzyme called IRT. The blood sample was then tested for alterations to the CF gene. An alteration was found, and so your child had a sweat test. The sweat test measured the amount of salt in your child’s sweat. People with CF have a lot of salt in their sweat. Your child was found to have normal levels of salt. Therefore the testing results are reassuring that your child does not have CF but carries an altered CF gene.

Explanation:

There is a high degree of variance in how screening programmes are run across Europe. Therefore it would not be possible for this question to be addressed in the European best practice document. This paragraph brings together the NBS and sweat testing process in a clear manner. It is important for this to be addressed since this is the process that has led to the identification of parent’s newborn as a carrier of CF.

A list of quick facts was included at the end of the information leaflet. These re-emphasise key points of information from the leaflet itself. Contact details of local CF centres, the authors names and the date of publication were also included.
Introduction

“The film you are watching was created for the parents of children who were identified as carrying an altered cystic fibrosis gene through newborn screening here in Ireland. It aims to give parents a better understanding of cystic fibrosis, the newborn screening system here in Ireland, as well as the genetic of cystic fibrosis. To do that I will be talking to two experts in the field of cystic fibrosis. The first is Doctor Barry Linnane, he is the pediatric respiratory consultant here in the Regional Hospital, Limerick, and he was a member of the steering group which oversaw the introduction of newborn screening for cystic fibrosis in Ireland. So he is going to be able to tell me a lot of things in general about Cystic Fibrosis and specifically about the newborn screening system. The second person I am going to be talking to is Dr Alana Ward, she is the genetic counsellor for cystic fibrosis in Ireland, so I am going to be asking her questions specifically about the genetics of cystic fibrosis”.

Interview with Dr Barry Linnane

The interview with Dr Linnane consists of the following questions;
1. Would you describe Cystic Fibrosis in your own words?

2. Can you talk us through the newborn screening process?

3. What is the final sweat test?

4. What does it mean for an infant to be identified as a carrier of CF?

5. Does being a carrier affect a persons’ health or lifespan?

Three key points will be summarised in writing at the end of the interview;

• CF is a serious condition which affects the lungs and digestive system.

• Being a carrier of an altered CF gene will not make your child ill or require special treatment.

• 1 in 19 people in Ireland carries an altered CF gene.

Interview with Dr Alana Ward

1. What is the difference between a genetic disease and any other type of disease?

2. How is CF passed on?

3. If an infant is identified as a carrier of Cystic Fibrosis, what implications does it have for the infant’s health?

4. If a child is identified as a carrier of Cystic Fibrosis, it means that at least one of the parents is a carrier and both of the parents could be carriers of an altered CF gene?
5. What are the benefits of genetic testing for parents?

6. Are there any final points you would like to make in relation to Cystic Fibrosis?

Three key points will be summarised in writing at the end of the interview;

• The alteration to the CF gene is inherited from a parent.

• It may be the case that both parents’ carry an altered CF gene.

• Free genetic testing is available for parents of children identified as carriers of an altered CF gene.

Conclusion

“This brings us to the end of the information film. In this, you’ve seen interviews with Dr Barry Linnane, who told us about newborn screening here in Ireland. He described that being a carrier of Cystic Fibrosis does not itself cause illness or require treatment. Secondly we talked to Dr Alana Ward who told us about the genetics of Cystic Fibrosis, and described that carrier testing is available for free for the parents of children identified as carriers of Cystic Fibrosis. If you would like further information about Cystic Fibrosis, there are two useful websites, the first of these is the website of the Cystic Fibrosis Association of Ireland (website address appears on screen - www.cfireland.ie), and the second is the website of the Cystic Fibrosis Trust in the UK, (Website address appears on screen - www.cftrust.org.uk). I would like to thank all of those who contributed to the film for their time in making it. Thank you”.
Appendix 5: Study questionnaire

Parent Survey

1. Date of interview:

2. Interviewee: Mother
   Father
   Other:

3. Interviewee Date of Birth: ________________________________

4. Infant Name? ________________________________

5. Infant Date of Birth? ________________________________

6. Where did your infant have their sweat test? ________________________________

7. Infant's gender: Male
   Female

8. What is your highest level of education?
   Junior Certificate
   Leaving Certificate
   Some college
   Diploma
   Degree
   Masters
   PhD

9. What is your Nationality? ________________________________

10. Had you ever heard of cystic fibrosis (CF) before your child was born?
    Yes
    No
    Not sure

11. Do you know someone with CF?
    Yes (WHO?: ____________)
    No

12. Do you know someone who is a carrier of CF?
    Yes (WHO?: ____________)
    No

13. Which of the following statements best describes your child with regard to CF?
    He or she definitely does not have CF.
    He or she definitely has CF
    He or she still might have CF; we have to undergo further testing.
    He or she might have a mild case of CF.
    I am unsure

14. Which of the following statements best describe your child with regard to carrier status?
    He or she is definitely a carrier for CF
    He or she is definitely NOT a carrier for CF
    He or she may be a carrier for CF
    He or she may actually have CF
    I am not sure
The next part consists of some true or false questions. I’m going to read some statements and I would like to know if you believe they are true or false.

(KNOWLEDGE OF GENETICS)

<table>
<thead>
<tr>
<th>Statement</th>
<th>True</th>
<th>False</th>
</tr>
</thead>
<tbody>
<tr>
<td>CF is a genetic condition</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>To inherit CF, both parents must be carriers of CF</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>Genetic testing can establish if a person carries an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>If two parents carry altered CF genes there is a one in four chance of any child of theirs having CF</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>If neither parent is a carrier or affected by CF they can have a child who carries an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>There is a vaccine that stops people from getting CF</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>1 in 150 people in Ireland carry an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>If one parent carries an altered CF gene and the other parent does not, there is a 50:50 chance of their children carrying an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>1 in 19 people in Ireland carry an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
<tr>
<td>If you carry an altered CF gene there is an increased chance of your biological family also carrying an altered CF gene</td>
<td>True</td>
<td>False</td>
</tr>
</tbody>
</table>

15. Are you a carrier of CF?
   - I actually have CF disease
   - Yes
   - No; definitely not a carrier
   - No; no mutations were found
   - Not sure

14. (b) Do you plan to be tested to learn if you are a carrier of CF?
   - Yes
   - No
   - Not Sure

15. Is your partner a carrier of CF?
   - Actually my partner has CF
   - Yes
   - No; definitely not a carrier
   - Not; no mutations were found
   - Not sure

15. (b) Does your partner plan to be tested to learn if he/she is a CF carrier?
   - Yes
   - No
   - Not Sure

The next part again consists of some true or false questions. I’m going to read some statements about health conditions associated with CF and I would like to know if you believe they are true or false.
16. Since your child had a sweat test, how often have you thought about the results?
   - Not at all
   - Seldom (once or twice)
   - Occasionally (once a month)
   - Often (once a week)
   - Constantly (daily)

17. How do you rate your child’s health now?
   - Excellent
   - Very Good
   - Good
   - Fair
   - Poor

17. (b) [If fair or poor]: do you think your child’s health is related to their CF carrier status?

18. Overall do you support newborn screening for CF? (do you think NBS for CF is a good idea)
   - Yes very supportive
   - Yes moderately supportive
   - Yes minimally supportive
   - No, not supportive (see next question)

18. (b) [If not Supportive] : Why are you not supportive of newborn screening for CF?
19. Is there anything that you would you change about the process of newborn screening for CF? 

20. Do you have a follow-up appointment for additional genetic counseling and testing? 
   - Yes 
   - No 
   - Still deciding about it 

21. Have you sought out additional information about Cystic Fibrosis? 
   - Yes 
     - before sweat test 
     - after sweat test 
   - No (skip next question) 

22. What sources did you use? (check all that apply) 
   - Internet – (Which websites?) 
   - Support group 
   - Health care professionals 
   - Media (books, magazines, journals) 
   - Other: 

23. At what age will you tell your child that he or she is a carrier for cystic fibrosis? 
   - When they are young (<10yrs) 
   - When they are a teenager / young adult 
   - I don’t plan to tell him/her 
   - Specific Age: 

24. Did you tell any relations that your child was identified as a carrier of cystic fibrosis? 

25. Prior to your child’s birth, were your relatives aware that the CF gene runs in the family? 

26. Are any of your relatives interested in receiving CF genetic testing? 

27. Do you think that your child’s newborn screening result is a benefit or a harm?
The final part of the survey contains 36 statements about stress and family relationships. Please listen to each statement carefully. Focus on your newborn infant, and give the response that best represents your opinion. For each statement I will ask you whether you Strongly Agree with the statement, Agree with the statement, Not Sure if you agree with the statement, Disagree with the statement or Strongly Disagree with the statement.

While you may not find a response that exactly states your feelings, please give the response that comes closest to describing how you feel. YOUR FIRST REACTION TO EACH QUESTION SHOULD BE YOUR ANSWER.

Please give only one response to each statement and respond to all statements.

1. I often have the feeling that I cannot handle things very well.     SA  A  NS  D   SD
2. I find myself giving up more of my life to meet my children’s needs than I ever expected.     SA  A  NS  D   SD
3. I feel trapped by my responsibilities as a parent.     SA  A  NS  D   SD
4. Since having this child, I have been unable to do new and different things.     SA  A  NS  D   SD
5. Since having a child, I feel that I am almost never able to do things that I like to do.     SA  A  NS  D   SD
6. I am unhappy with the last purchase of clothing I made for myself.     SA  A  NS  D   SD
7. There are quite a few things that bother me about my life.     SA  A  NS  D   SD
8. Having a child has caused more problems than I expected in my relationship with my spouse (or male/female friend).     SA  A  NS  D   SD
9. I feel alone and without friends.     SA  A  NS  D   SD
10. When I go to a party, I usually expect not to enjoy myself.     SA  A  NS  D   SD
11. I am not as interested in people as I used to be.     SA  A  NS  D   SD
12. I don’t enjoy things as I used to.     SA  A  NS  D   SD
13. My child rarely does things for me that make me feel good.     SA  A  NS  D   SD
14. Sometimes I feel my child doesn’t like me and doesn’t want to be close to me.     SA  A  NS  D   SD
15. My child smiles at me much less than I expected.     SA  A  NS  D   SD
16. When I do things for my child, I get the feeling that my efforts are not appreciated very much.     SA  A  NS  D   SD
17. When playing, my child doesn’t often giggle or laugh.     SA  A  NS  D   SD
18. My child doesn’t seem to learn as quickly as most children.     SA  A  NS  D   SD
19. My child doesn’t seem to smile as much as most children.     SA  A  NS  D   SD
20. My child is not able to do as much as I expected.     SA  A  NS  D   SD
21. It takes a long time and it is very hard for my child to get used to new things.     SA  A  NS  D   SD

For the next statement, choose your response from the choices 1 to 5 below.

22. I feel that I am:  
   - not very good at being a parent 1 
   - a person who has some trouble being a parent 2 
   - an average parent 3 
   - a better than average parent 4 
   - a very good parent 5
23. I expected to have closer and warmer feelings for my child than I do and this bothers me.  
24. Sometimes my child does things that bother me just to be mean.  
25. My child seems to cry or fuss more often than most children.  
26. My child generally wakes up in a bad mood  
27. I feel that my child is very moody and easily upset.  
28. My child does a few things which bother me a great deal.  
29. My child reacts very strongly when something happens that my child doesn't like.  
30. My child gets upset easily over the smallest thing.  
31. My child's sleeping or eating schedule was much harder to establish than I expected.  
For the next statement, choose your response from the choices 1 to 5 below.  
32. I have found that getting my child to do something or stop doing something is:  
   much harder than I expected 1  
   somewhat harder than I expected 2  
   about as hard as I expected 3  
   somewhat easier than I expected 4  
   much easier than I expected 5  
For the next statement, choose your response from the choices 10+ to 1-3.  
33. Think carefully and count the number of things which your child does that bother you. For example: dawdles, refuses to listen, overactive, cries, interrupts, fights, whines, etc.  
   10+  
   8-9  
   6-7  
   4-5  
   1-3  
34. There are some things my child does that really bother me a lot.  
35. My child turned out to be more of a problem than I had expected.  
36. My child makes more demands on me than most children.  
If you feel it would be useful to have support in coping with any emotional issues raised as a result of taking part in this study please don’t hesitate to contact your GP. That’s the end of the survey, thank you very much for your time. Your help will give us a much better understanding of peoples experiences of the CF Newborn Screening programme in Ireland.
Appendix 6: Lang et al, 2011 questionnaire

POST SWEAT TEST SURVEY

Subject ID #:

Date of interview: Was a mutation found? 01 YES 02 NO

Interview Location: 01 Phone 02 Friend Date of Birth:

Sweat Test Location: 01 U of C 02 Children’s Date Sweat Test:

Hospital where child was born: 01 U of C 02 Prentiss 03 Other _______________

1) Had you ever heard of cystic fibrosis (CF) before your child was born?
   01 Yes
   02 No
   55 Not sure

2) Do you know someone with CF?
   01 Yes (WHO?)
   02 No
   03 Carrier for CF (WHO?)

3) Did you have any prenatal discussions about newborn screening (in general) with your OB/midwife or prenatal care provider?
   01 Yes
   02 No (skip next question)
   66 Don’t recall (skip next question)

4) If yes, what did they tell about newborn screening? 88 SKIP

5) Did you have any prenatal discussions, counseling or testing specifically for Cystic Fibrosis with your OB/midwife or prenatal care provider?
   01 Yes
   02 No
   66 Don’t recall

6) Your child had a positive CF screening blood test when he/she was a newborn and needed a sweat test. Do you know why your child needed a sweat test?
   01 High IRT screen (high blood screen)
   02 High IRT screen and they found my child had a gene for CF
   55 Don’t know

7) Your child had a sweat test. What was the result of the sweat test?
   01 It was normal
   02 It was ambiguous (neither normal nor abnormal); we need to undergo a repeat sweat test
   03 It was abnormal
   66 I don’t recall

8) Which of the following statements best describes your child with regard to CF?
   01 He or she definitely does not have CF.
   02 He or she definitely has CF
   03 He or she still might have CF; we have to undergo further testing.
   04 He or she might have a mild case of CF.
   55 I am unsure
9) Which of the following statements best describe your child with regard to carrier status?
   01 He or she is definitely a carrier for CF
   02 He or she is definitely NOT a carrier for CF
   03 He or she may be a carrier for CF
   04 He or she may actually have CF
   55 I am not sure

THE NEXT set of questions refers to information you were provided about the NBS for CF and your initial reactions to learning about your child’s positive newborn screen

10) Who initially called to tell you that your child had a positive NBS for CF.
   01 No call, I received a letter (skip to question 15)
   02 child’s pediatrician
   03 nurse or nurse practitioner
   04 Illinois department of public health staff
   05 Genetic counselor
   06 Other (fill in the blank ________________________)
   66 Don’t know, don’t remember

Note: if no call or letter, but was informed in by person in #10 check this box… □

11) Did the individual who called describe what CF is?
   01 Yes
   02 No
   55 Not sure

12) Did the individual who called provide you with information regarding what a positive CF NBS means?
   01 Yes,
   02 No
   55 Not sure

13) Did the individual who called provide you with information regarding the sweat test procedure?
   01 Yes
   02 No
   55 Not sure

14) Rate your level of anxiety when you first learned that your child had a positive CF NBS.
   01 Very worried
   02 Modestly worried
   03 Minimally worried (a little bit worried)
   04 Not worried at all
   66 Don’t remember

Now Skip to question 17 (NEXT PAGE)

15) What did you do when you received the letter?
16) see question #14
THE NEXT set of questions refers to your experience with scheduling the sweat test.

17) Did you call to make a sweat test appointment or did someone call you?
   01 I called
   02 The University of Chicago called me to inform me of the positive screen and scheduled the sweat test at that time
   03 Other (ELABORATE)

18) When you scheduled your sweat test, did you speak with a genetic counselor?
   01 Yes
   02 No
   55 Not sure / Don’t remember

19) Did the person who scheduled your sweat test describe what CF is?
   01 Yes
   02 No
   55 Not sure

20) Did the person who scheduled your sweat test provide you with information regarding what a positive CF NBS means?
   01 Yes
   02 No
   55 Not sure

21) Did the person who scheduled your sweat test provide you with information regarding the sweat test procedure?
   01 Yes
   02 No
   55 Not sure

22) Rate your level of anxiety after scheduling the sweat test.
   01 Very worried
   02 Modestly worried
   03 Minimally worried
   04 Not worried at all
   66 Don’t remember

22a) Rate you level of anxiety NOW (at time of interview) after finding out sweat test results and speaking to the genetic counselor:
   01 Very worried
   02 Modestly worried
   03 Minimally worried
   04 Not worried at all
   66 Don’t remember
### PART I: KNOWLEDGE OF GENETICS

<table>
<thead>
<tr>
<th></th>
<th>True</th>
<th>False</th>
</tr>
</thead>
<tbody>
<tr>
<td>23. CF is a genetic condition</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>24. To inherit CF, both parents must have at least one CF gene.</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>25. CF can be inherited if one parent has CF, even if the other parent is not a carrier and does not have the disease.</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>26. People who are carriers of CF have a mild form of CF</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>27. You can be a carrier for CF even if neither parent is a carrier or affected with CF</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>28. CF can be transmitted by physical contact with an affected person</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>29. Over time, carriers of CF can develop CF</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>30. It is important to know if you are a carrier for CF even if you don’t have any symptoms because of possible health risks associated with being a CF carrier</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>31. There are things a person with CF can do to avoid some of the complications.</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>32. There is NO cure for CF</td>
<td>1</td>
<td>2</td>
</tr>
</tbody>
</table>

33) In what ethnic groups is CF found?
   01 Mostly in individuals of African ancestry (Blacks)
   02 Mostly in individuals of European ancestry (Caucasians)
   03 Equally in all races and ethnicities

34) Are you a carrier for CF?
   01 I actually have CF disease
   02 Yes [**did you have counseling on what it means to be a carrier prior to your child’s birth?**]
   03 No; definitely not a carrier
   04 No; no mutations were found
   05 Not sure

35) If don’t know, do you plan to be tested to learn if you are a carrier of CF?
   01 Yes
   02 No
   05 Not Sure
   77 Not applicable, I already know my status

36) When were you **tested** for CF carrier status?
   01 Prenatal visit
   02 After my child’s positive NBS
   03 After my child’s sweat test
   04 After a relative was diagnosed with cystic fibrosis.
   05 After a relative was diagnosed as a carrier
   06 During a community screening event (at my synagogue or Church or college)
   77 Not applicable; I’ve never been tested
   08 Other (ELABORATE)
37) Is your partner a carrier for CF?
   01 Actually my partner has CF did you have counseling on what it means to be a carrier prior to your child’s birth?
   02 Yes
   03 No; definitely not a carrier
   04 Not; no mutations were found
   05 Not sure

38) If don’t know, does your partner plan to be tested to learn if he/she is a CF carrier?
   01 Yes
   02 No
   05 Not Sure
   77 Not applicable; partner already knows he/her status

39) When was your partner tested for CF carrier status?
   01 Prenatal visit
   02 After my child’s positive NBS
   03 After my child’s sweat test
   04 After a relative was diagnosed with CF
   05 After a relative was diagnosed as a carrier, I went for genetic testing
   06 During a community screening event (at my synagogue or Church or college)
   77 Not Applicable, S/He’s never been tested
   08 Other (ELABORATE)

PART II: KNOWLEDGE OF THE HEALTH CONDITIONS ASSOCIATED WITH CF and severity of CF relative to other diseases

<table>
<thead>
<tr>
<th>Statement</th>
<th>True</th>
<th>False</th>
</tr>
</thead>
<tbody>
<tr>
<td>40. Individuals with CF often fail to grow at a normal rate</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>41. Individuals with CF are usually of normal intelligence</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>42. Individuals with CF usually have problems with hearing.</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>43. Individuals with CF usually have breathing problems</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>44. Men with CF often have fertility problems</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>45. Women with CF often have fertility problems</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>46. Individuals with CF must eat a special diet</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>47. Playing sports will worsen the symptoms of cystic fibrosis</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>48. Individuals with CF often have muscle weakness</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>49. Individuals with CF need to take special vitamins</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>50. Individuals with CF get frequent respiratory infections</td>
<td>1</td>
<td>2</td>
</tr>
</tbody>
</table>
### 51) How familiar are you with the following conditions?

<table>
<thead>
<tr>
<th></th>
<th>NEVER heard of it (skip to next condition)</th>
<th>NOT familiar 1</th>
<th>minimally familiar 2</th>
<th>moderately familiar 3</th>
<th>VERY familiar 4</th>
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<tbody>
<tr>
<td>A</td>
<td>Sickle cell disease</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>B</td>
<td>Cystic fibrosis</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
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<td>C</td>
<td>Diabetes</td>
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<td>Autism</td>
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</tr>
<tr>
<td>F</td>
<td>Depression</td>
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<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>G</td>
<td>Seizures (epilepsy)</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>H</td>
<td>Down syndrome (Trisomy 21)</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
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</table>

### 52) Do you know someone with the following conditions?

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>NO</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
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</tr>
<tr>
<td>B</td>
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<tr>
<td>C</td>
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<tr>
<td>D</td>
<td>I</td>
<td>2</td>
</tr>
<tr>
<td>E</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>F</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>G</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>H</td>
<td></td>
<td>1</td>
</tr>
</tbody>
</table>

### 53) How would you rate severity of the following conditions?

<table>
<thead>
<tr>
<th></th>
<th>NEVER heard of it (skip to next condition)</th>
<th>NOT serious 1</th>
<th>slightly serious 2</th>
<th>moderately serious 3</th>
<th>Serious 4</th>
<th>VERY serious 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Sickle cell disease</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>B</td>
<td>Cystic fibrosis</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>C</td>
<td>Diabetes</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>D</td>
<td>Kidney failure on dialysis</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>E</td>
<td>Autism</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>F</td>
<td>Depression</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>G</td>
<td>Seizures (epilepsy)</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>H</td>
<td>Down syndrome (Trisomy 21)</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
</tbody>
</table>

### 54) Since your child had a sweat test, how often have you thought about the results?

- 01 Not at all
- 02 Seldom (once or twice)
- 03 Occasionally (once a month)
- 04 Often (once a week)
- 05 Constantly (daily)
55) How do your rate your child’s health now?
   01 Excellent
   02 Very Good
   03 Good
   04 Fair
   05 Poor

56) Overall do you support newborn screening for CF? (do you think NBS for CF is a good idea)
   01 Yes very supportive
   02 Yes moderately supportive
   03 Yes minimally supportive
   04 No, not supportive

57) What would you change about the process of newborn screening for CF and sweat testing that you
   experienced
   DESCRIBE:

58) Do you have a follow-up appointment for additional genetic counseling and testing?
   01 Yes
   02 No
   03 Still deciding about it

59) Have you sought out additional information about Cystic Fibrosis?
   01 Yes
      (1) before sweat test
      (2) since sweat test
   02 No (skip next question)

60) What sources (check all that apply)
   01 Internet
   02 Support group
   03 Health care professionals
   04 Media (books, magazines, journals)
   05 Other

THE FOLLOWING SECTION IS ONLY IF PARENT IDENTIFIES CHILD AS A CARRIER (Otherwise skip to question 82)

DISCLOSURE: Now we will be asking you about sharing information within your families.

61) How would you rate how private you are regarding medical information and decision making
   01 Very private
   02 Somewhat private
   03 Somewhat open
   04 Very open
   99 No answer
### 62) Should [name] know that your child is a CF carrier?

<table>
<thead>
<tr>
<th></th>
<th>Yes, they <strong>should</strong> know</th>
<th>NO, it <strong>doesn’t matter</strong> if they know</th>
<th>NO, I do <strong>not</strong> want them to find out</th>
<th>Not applicable</th>
</tr>
</thead>
<tbody>
<tr>
<td>A Your partner</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>B Your Siblings (child’s aunts and uncles)</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>C Your Parents (child’s grandparents)</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>D Your child’s siblings</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>E Other relatives</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>F Your child’s pediatrician</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>G Emergency room physicians</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>H Dentists</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>I Insurance company</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>J Teachers</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>K Classmates</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>L Church leader</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
<tr>
<td>M You child’s future partner</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>77</td>
</tr>
</tbody>
</table>

### 75) At what age will you tell you child that he or she is a carrier for cystic fibrosis

- **01** When they are young (<10yrs)
- **02** When they are a teenager / young adult
- **03** I don’t plan to tell him/her

### 76) Your parents

<table>
<thead>
<tr>
<th>A. Did you tell them that your child had a positive NBS for cystic fibrosis?</th>
<th>Yes</th>
<th>No</th>
<th>B) Prior to your child’s birth, were they aware that the CF gene runs in the family?</th>
<th>Yes, they <strong>are interested</strong></th>
<th>No, they <strong>already know</strong> their CF status</th>
<th>No, they <strong>are not interested</strong> in being tested</th>
<th>We <strong>didn’t discuss</strong> CF testing</th>
</tr>
</thead>
<tbody>
<tr>
<td>76) Your parents</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>77) Your siblings</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>78) Your spouse/ partner’s parents</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>79) Your spouse/ partner’s siblings</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>80) Other relatives of yours</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>81) Other relatives of your spouse/ partner</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
</tbody>
</table>
ALL RESPONDENTS’ DEMOGRAPHICS

82) Does CF carrier or cystic fibrosis run in your family
   01 Yes, mine
   02 No
   04 Not in mine, but in my partner’s
   14 Yes mine and partner
   55 Don’t know

83) Does sickle cell disease or sickle cell trait run in your family
   01 Yes
   02 No
   04 Not in mine, but in my partner’s
   14 Yes mine and partner
   55 Don’t know

84) How old is your infant (calculate in weeks)

85) Gender
   01 Male
   02 Female

86) How old are you? ___________ years

87) Are you married?
   01 Yes
   02 No

88) How many children do you have? # __________
    (age and gender of each child)

89) What is your highest level of education?
   01 Did not graduate high school
   02 High school graduate
   03 GED
   04 Some college
   05 College graduate
   06 Some graduate school
   07 Graduate degrees
90) What type of (health) insurance do you have?
   01 No insurance
   02 Private Insurance
   03 Public Aid

90a. Do you have a personal doctor other than an OB or emergency room physician?
   01. Yes
   02. No

90b. Where does your child get his or her well child care and immunizations?
   01 Friend Center
   02 University of Chicago DCAM (skip to question 23)
   03 Other (skip to question 23)
   77 N/A child does not have a routine clinic

91) What is your ethnicity?
   01 Hispanic or Latino
   02 Non-Hispanic or non- Latino

92) What is your race? (Select all that apply)
   01 Black or African-American
   02 Asian / Pacific Islander
   03 Caucasian / European
   04 Other ________________________________

93) What is your religion?
   01 Non-denominational Christian
   02 Baptist
   03 Methodist
   04 Catholic
   05 Jewish
   06 Muslim
   07 Buddhist
   08 Other __________
   09 No
References


WARD, A. 10 June 2014 2014. *RE: CF genetic testing rates in the Republic of Ireland*. Type to QUIGLEY, S.


