Exploring the Impact of Living with Fanconi Anaemia & Diamond-Blackfan Anaemia: A Systemic Consideration.

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ABSTRACT

Aims: Amidst a growing body of medical research, little is yet known about the psychological impact of living with two rare life-limiting conditions, Fanconi Anaemia (FA) and Diamond Blackfan Anaemia (DBA). One might expect some level of impact on well-being as indicated by wider literature on other childhood illnesses. At present, understanding of this impact is limited in being generalised from other research, as therefore are the supports available. This research seeks to begin a consideration of the psychological impact, and to consider how support has been deemed.

Method: Using semi-structured interviews, parents and individuals living with FA or DBA were interviewed regarding their experiences. Thematic analysis was used to achieve an overview of these experiences.

Results: Themes identified for individuals included: 1. 'Knowledge', which outlined the impact of lack of knowledge and 2. 'Illness Concept' relating to one's relationship to FA/DBA and how this is socially mediated. For parents, themes identified were: 1. 'Knowledge', which centered on uncertainty and 2. 'Social Responses' which related to areas of support and strain. These themes indicated an impact on individual well-being. Two joint themes were identified: 1. 'Family Dynamics' which outlined how relationships and family well-being may be impacted and 2. 'Mental Health Care as Necessary but Inadequate'.

Conclusion: There was a clear impact articulated regarding the well-being of family members, individually and collectively in concerns regarding relational dynamics. There was also clear demand for psychological support which, in its current form, was generally considered inadequate. Research must be responsive in furthering efforts to establish clear and adequate pathways and a standard of care for the well-being of all families living with FA/DBA.

ABBREVIATIONS

BMF	Bone Marrow Failure
CBT	Cognitive Behavioural Therapy
DBA	Diamond-Blackfan Anaemia
FA	Fanconi Anaemia
NHS	National Health Service
PGD	Preimplantation Genetic Diagnosis
PPN	Psychological Professions Network
TA	Thematic Analysis
US	United States

LIST OF TABLES

Contents

CHAPTER ONE: INTRODUCTION

Amidst a growing body of medical research, little is yet known about the psychological impact of living with two rare life-limiting conditions, Fanconi Anaemia (FA) and Diamond Blackfan Anaemia (DBA). This chapter will firstly provide a narrative review of the literature base and research context. This will consider how illness is understood with emphasis on the separation of physical and mental health in illness research. Drawing out the social elements of illness, this chapter presents the rationale for a systemic focus, before presenting a scoping review of relevant literature regarding FA and DBA. The chapter will conclude by outlining the research questions.

1.1 Literature Search Strategy & Presentation

The databases EBSCO, SCOPUS and Google Scholar were used to conduct a literature search. A combination of research terms was used, starting broadly and refining as familiarity with the literature grew. Terms used included 'Fanconi anaemia', 'Diamond Blackfan Anaemia', as well as the US spelling 'anemia', 'FA', and 'DBA'. These terms were searched with the addition of 'psychosocial', 'psychological', 'mental health', 'well-being', 'family', and 'UK'. The titles and abstracts of results were reviewed, and selection of papers was based on relevance to the topic. Reference lists of identified papers were also consulted. On account of the limited results this uncovered, a decision was made to first provide a narrative review, followed by a scoping review of more focused literature. To build a narrative review chronic childhood illness and the family was searched more generally. Terms used included: 'chronic childhood illness', and 'chronic illness + family'. Additions included 'well-being', 'mental health', and 'psychological impact'. Finally,

government websites were used to search for relevant policy papers.

1.2 Defining Terms

Diamond-Blackfan Anaemia (DBA) is an inherited bone marrow failure (BMF) condition characterised by reduced red blood cell production (Bartels & Bierings, 2018). Its incidence rate is recorded as 1 in 360,000 births (Zen et al., 2011). Fanconi Anaemia (FA) is an inherited BMF condition marked by decreased production in all blood cells, red, white, and platelets (Nalepa & Clapp, 2018). Its incidence rate is estimated as 5-7 births per 1 million (Pospisilova et al., 2012). Whilst both are inherited and typically arise in childhood, a diagnosis may not be made immediately on account of relative wellness masking symptoms, or those symptoms being overlooked. Both conditions may be associated with physical characteristics such as short stature, low weight, and atypical limb formation (Dufour, 2017; Bartels & Bierings, 2019). Both involve impaired bone marrow function. In FA this typically leads to the development of aplastic anaemia where the body stops or fails to produce a sufficient supply of new blood cells, also referred to as BMF. In DBA this may lead to red cell aplasia where the body fails to produce a sufficient supply of red blood cells, leading to severe anaemia. These differences mean different treatment options and thereby differences in the lived experiences of individuals living with either condition. Whilst DBA impairs bone marrow function in the failure to produce red blood cells, it does not usually lead to full BMF, and from a medical perspective many individuals live relatively well into adulthood treated with steroids and iron chelation. In contrast, FA marked by failure across all blood cells, typically leads to BMF and the subsequent necessity of stem cell transplant as a life-saving intervention. The complications of this procedure may impact on the prognosis for FA patients. No treatments can offer a cure at this time but are geared toward prolonging life and symptom management. These conditions differ from other

10

forms of aplastic anaemia, which may arise across the life span and range from mild to severe; FA and DBA arise with added complexities.

FA and DBA are linked with many other health related issues including fatigue and a predisposition to other illnesses, including a higher risk of developing leukaemia and other cancers (Nalepa & Clapp, 2018; Vlachos et al., 2012). Both are therein linked with a reduced lifespan and are therefore considered life-long and life-limiting. This picture is changing; with medical advances in treatment, people with either condition are living into their 30s and 40s (Ristano et al., 2016; Mangla & Hamad, 2019). Whilst this is so, both remain largely uncertain prognoses. In addition, the reality of many individuals living for longer raises new issues of adjustment across the lifespan and the pressing need to consider quality of life.

1.3 Medical Conceptualisation of FA & DBA

As is evident from the above, there is a wealth of medical research on both conditions, a research base that continues to be updated. It is worth then noting that both conditions are currently conceptualised almost entirely in regard to their medically defined symptoms, with definitions outlining the serious physical health implications. Indeed, the present introduction defined FA and DBA according to these same criteria. Whilst this focus is necessarily crucial, there is a longstanding literature delineating illness in its medical form from the illness experience constructed socially. Kleinmann (1988) argues that in making a medical diagnosis, a listening clinician hears the nuanced account of their patient and reconfigures this information into a set of symptoms, which are used to label this experience as a medical diagnosis. In this way something of the experience is lost, and the result is a desocialised picture of an individual's physical health. This picture misses out the relationships within which illness is lived, relationships which may improve or make worse the experience of living with illness (Stiell et al., 2007). This diagnostic recasting may indicate to an individual what has been used from their telling and

what has not. Perhaps in this way certain elements are signaled as not of relevance or concern and are thereby delegitimised as important for discussion. As a result, interventions, as for both conditions here, take the form of medical treatments geared toward symptom management or reduction. These are of course necessary and life sustaining but are not necessarily sufficient to support individuals with the impact of the wider illness experience. Whilst prolonging life may be the main concern, research has yet to consider the quality of those lives and how the current provision of support impacts on this.

1.4 Illness Experience: The Social Elements

Research around FA and DBA has not yet incorporated this thinking about illness experience. Thereby the research base that exists is almost entirely void of any wider consideration of the impact of living with FA/DBA. To consider this illness experience is to begin thinking more relationally about illness and disease. Doing so uncovers how individuals often live within socially constructed ways of understanding illness. Kleinman (1988) points out for instance that ideas of illness are bound up in ideas of recovery, of a discrete period of unwellness which will subside. Indeed, medical treatment is largely aligned with this same rhetoric, with level of recovery from illness often a measure of treatment success (Stam, 2000). Where though does this rhetoric leave those living with chronic health conditions, who will not benefit from a cure or recovery? Individuals living with FA or DBA may have periods of relative physical wellness, but recovery in the sense of cure is not possible. Without asking those concerned, our present understanding of how it is to live with FA/DBA is applied to these individuals from a more general understanding of illness that may not map onto the reality of a chronic, life-limiting condition.

Other research has added to these ideas, highlighting how culture may influence

how we understand illness too. Jørgensen et al. (2020) link the concept of recovery in illness to Western neoliberalist ideology, which attributes value to one's ability to work and thereby maintain the economic status quo. There is a strong focus on work as a marker of worth and value and therein an emphasis on recovering from illness to ensure one can continue working. There is a binary distinction here between healthy as good and illness as bad, the latter a marker of one's inability to contribute. Whilst many individuals living with long term conditions are capable of and do maintain long-term employment, where do they fall on this categorical distinction? Arguably this is largely influenced by how the individual is positioned socially. Given these representations of illness that circulate within society it is clear that despite relative wellness, individual's living with FA or DBA may well be positioned as otherwise by those around them, according to general understandings of illness. Western culture is also widely acknowledged to be individualistic (Buss, 2000). This may also play out in healthcare, with treatment and interventions targeted at individuals, who are held as individually responsible for maintaining their level of wellness (National Health Service [NHS], 2015; Brown & Savulescu, 2019).

1.5 Meaning Making

Given this context, one may question how individuals make meaning regarding their diagnosis of FA or DBA. It is widely held in the field of social psychology that meaning making occurs socially according to and in communication about the local knowledges and understandings available (Moscovici, 1984). Willig (2012) references the interpretative act that occurs in medical diagnosis, firstly by the clinician who assigns a diagnosis, secondly by the individual and their understanding of the diagnosis, and thirdly, by those with knowledge of the person's illness who position that person accordingly. If we consider the common cold for example, there is a widely held understanding of what this is as well as a set of expectations around how one should behave when experiencing a cold. Both conditions here though are rare and therefore much less known. This means there is much less information to

13

draw upon in making sense of either diagnosis. Lexically speaking, titles are important too (Fleischmann, 1999). In this case both are titled with reference to anaemia, a well-known and relatively mild condition. This complicates the social process of receiving this diagnosis and of re-telling it socially on account of our tendency to understand based on the commonsense knowledge we have access to. On hearing the labels many individuals may well think of this common condition. Medical professionals as the initial tellers and (often) parents as the re-tellers must navigate these complexities in the sharing process.

This picture is complicated further in that those individuals diagnosed with FA or DBA are medically vulnerable to a host of other illnesses, most notably to forms of cancer. Though uncertain as to when and what type, the statistics mark this as a significantly elevated likelihood (Nalepa & Clapp, 2018; Vlachos et al., 2012). Individuals and families must live with the uncertain certainty of this diagnosis. Willig (2012) outlines the cultural interpretations of cancer that circulate within Western society. These involve for instance war metaphors often used to conceive of cancer as a 'battle' to be won, as well as expectations that individuals must remain positive in the face of cancer. Death is taboo and largely not discussed as an option at all. It is amidst these social ways of understanding that individuals must come to terms with and make sense of their health condition as one predisposing to cancer. Beyond the experience of physical symptoms then, individuals must find ways of understanding a relatively rare diagnosis. This is likely influenced by the social knowledge available regarding illness. Research must address these complexities in order to best support those needing to make meaning and navigate the telling socially.

1.6 Integration of Physical and Mental Health

Thus far the chapter has explored the separation of medical understandings of illness from the wider illness experience. Following this it is worth considering how theory and practice have begun attempts to bridge this separation within the field of

illness.

1.6.1 Biopsychosocial model

The biopsychosocial model (Engel, 1977) made some effort to bring together the biological and medical with the psychological and social, and paved the way for the rise of health psychology as a discipline. Whilst this model popularised a consideration of the interplay between these domains, it has been argued that the model has in some ways upheld the subservience of psychology as bound to medicine (Stam, 2000). It is further claimed that psychology has not been as astute at exploring power in the area of health as it has in other domains (Stam, 2000). Healthcare systems can change at the whim of shifting political and economic forces. Data indicates that a period of austerity implemented following the 2008 economic crash meant that while that Department of Health and Social Care budget did continue to grow, it did so more slowly than in previous years (The Kings Fund, 2021). Indeed, there has in recent years been reference to a political crisis within the NHS, with governments of all stripes criticised for the lack of sufficient NHS funding (Ham, 2017). Budgetary changes change what is available as treatment at the point of access. What is on offer is not just based on need but based on economic and political decisions. This is important to consider in thinking about the arena in which research is funded, and from this what supports are made available to families living with FA/DBA. Research itself is carried out in social contexts, where personal and institutional values and interests are at play, and therein where status and connection can make the difference in ideas that are advanced and those which are not (Ziman, 1996; Morgan et al., 2018). Holding this context in mind, it is worth noting that anecdotal evidence highlights the presence of psychologists in certain health settings; oncology, sickle cell disease, chronic pain, cystic fibrosis; it is timely to question how psychology comes to sit within these domains, and conversely how psychology has remained largely absent from the conditions under consideration here.

1.6.2 Integrated healthcare

Nonetheless the Biopsychosocial model did encourage a growth in research. This research has consistently evidenced a link between mental and physical health, as well as a reciprocity to this interaction (Prince et al., 2007). It has been estimated that those living with long-term health conditions are 2-3 times more likely to experience a mental health difficulty (Naylor et al., 2012). These may be psychological difficulties relating directly to the experience of living with a long-term health condition. Individuals may experience distress related to their symptoms, treatment or side effects, as well as in adjusting to their diagnosis and its potential impact on social roles and day-to-day functioning. Naylor et al. (2016) hold that healthcare systems have not been sufficiently responsive to this data, that identification of co-morbid issues is not routine, and that support for the psychological aspects of living with a health condition is inconsistent nationally. Coventry et al. (2011) support this highlighting that consultations typically focus on physical symptoms (by both practitioners and patients) overlooking mental health concerns. Again, this may relate to expectations arising from the diagnostic process about what is signalled as relevant or not, and thereby what a patient feels able to voice in medical consultation. It may also relate to social expectations regarding illness and the metaphors of fighting, which add to the stigma around the admission of experiencing psychological distress (Corrigan et al., 2014).

The result of this is a direct impact on the health and well-being of those individuals concerned, including reduced quality of life and poorer clinical outcomes (Katon et al., 2004; Moussavi et al., 2007). Research has for example indicated higher mortality rates when depression is experienced following heart attack or diagnosis of diabetes (Blumenthal et al., 2003; Park et al., 2013). Thus, there is a clear and growing rationale for a more integrated approach in addressing physical and mental health. To this end research has a responsibility to give voice to the wider illness experience more generally, and to the wider illness experience of FA/DBA in this research specifically.

Taking an economic perspective can support this further by considering the cost incurred by health care systems operating in their present form. It is estimated that the NHS spends between £8 - £13 billion in how it currently supports those with co-morbid mental and physical health conditions (Naylor et al., 2012). Analysis by the same authors average that of the total NHS expenditure on long-term health conditions, 12-18% is linked to supporting mental health concerns that are most commonly understood as depression and anxiety disorders. One could bolster this argument further by considering the estimated annual £22 billion cost incurred by employers and taxpayers linked with work related absence and sickness (NHS, 2014). The researcher includes this for the persuasion of those interested in the economic forecast but contests the idea of employment as a measure of social worth. The overriding message here is that separately treating the mental health of those experiencing chronic illness is inefficient and costly.

1.6.3 Policy landscape

In recent years there have been several government and policy initiatives designed to respond to this data. They attempt to address the gap between this growing evidence of the relationship between mental and physical health and the reality of the separate care provided. A government strategy in 2011 coined the now popular phrase 'no health without mental health' calling for coordinated action to improve mental health outcomes (HM Government, 2011). This paved the way for a demand on government to work toward a parity of esteem between mental and physical health care. The Health and Social Care Act 2012 made this a legal responsibility for the NHS to work toward, whilst the government at the time committed to achieving this by 2020.

The NHS Five Year Forward plan (NHS, 2014) set out the task of achieving integration across three levels, between health and social care, primary and specialist care, and physical and mental health care. Naylor et al. (2016) argue that integration has not yet been sufficiently achieved in physical and mental health care. Whilst parity of esteem commonly came to be held as a call for mental health care to

be as good as physical health care, the authors call instead for mental health care to be delivered as part of physical health care. In this way they advocate for a level of integration that equips services to take a whole person approach in offering support for both medical and mental health needs.

1.7 Epistemic Injustice

Beyond the policy there is an ethical argument to be made in seeking a more integrative approach too. Given the situation as outlined in which the research base is almost entirely medically oriented, it may be argued that an epistemic injustice has developed and is maintained by current systems of research and practice. Fricker (2007) coined the term epistemic injustice to highlight the ways in which injustice is done onto others in how knowledge is claimed and processed, often by those in power. Hermeneutical injustice is referred to specifically here whereby individuals are disadvantaged by a gap in collective knowledge. In this case the gap pertains to understanding the wider experience of living with FA or DBA including the psychological impact. This is a gap the research has thus far failed to close.

Research also has a very practical impact. Healthcare professionals are required to conduct practice and intervention according to evidence-based practice which arises from research (Akobeng, 2005). Available supports and treatments naturally stem from this research base. As outlined, this research base currently lacks consideration of the wider illness experience of FA/DBA. Moreover, research practices are designed around a hierarchy of evidence. Whilst many variations on the hierarchy exist, there is undoubtedly a level of subjectivity in judgements about which types of evidence are considered best. Indeed, the hierarchy has been critiqued as privileging the needs of service providers and funders in how the ranking is constructed (Every-Palmer & Howick, 2014; Borgerson, 2009). Personal accounts rarely make the hierarchy in terms of medical research and this is presently the case with research on FA/DBA.

Accordingly, Carel & Kidd (2014) further the consideration of epistemic injustice highlighting how modern healthcare practice continues to privilege third person accounts of illness, without reference to the personal testimony or experience of those individuals the research is conducted about. Indeed, critical research has cautioned that a focus only on disease or illness is one that risks overlooking the person and therein being led only by what is effective for the illness, which is not necessarily what is effective for the person in their wider context (Miles, 2009). The research base currently lacks the voice of those living with FA or DBA. It is only by addressing this that we might ensure the supports available are as good as they can be.

1.8. System Focus

1.8.1 Theoretical Research

So far it has been considered that illness is conceptualised and understood socially. In addition, it is generally held across the research that the impact of illness is social too, affecting not just the person, but their closest system, most often the family. Thinking systemically about illness is not new. An early and comprehensive consideration is provided in Rolland's (1984) Family Illness Model, which seeks to consider how chronic illness might affect the family as a unit, as well as each member individually. The model outlines four characteristics by which chronic illnesses may vary: onset, course, outcome, and severity. It is proposed that clinicians seeking to support families must consider the interplay of these factors with the specific characteristics of each family, including the family life cycle, values, and transgenerational beliefs. Rolland (1990) later used the same theory to consider the impact of anticipatory loss on families who manage a chronic illness. In doing so a greater focus was applied to family life cycle transitions and how these might produce periods of more intense grief or feelings of loss, as developmental tasks are missed or need to be revised. The model holds that it is only in seeking to explore family's beliefs and feelings around loss, that they might be supported to

explore options for alternative versions of the future.

Eiser (1985,1993) has also written comprehensively on growing up with chronic illness, taking a developmental view from infancy onward and considering relationships within the family. She writes of unique stressors attached to each developmental stage of the child growing up with an illness, ranging from supporting a child to understand a diagnosis to navigating appropriate levels of independence. Even at this early stage in the development of health psychology, Eiser (1993) makes an argument for the role of clinical psychologists in the promotion of health in working preventatively to minimise the impact of chronic illness on families. Social Ecological Theory (Bronfenbrenner, 1979) has also been used to theorise about the child living with chronic illness and stresses the importance of social context in how the child and family will respond to diagnosis and treatment. Theories such as this are crucial in supporting thought around wider geo-political factors such as socio-economic status or race that may intersect to impact how healthcare is accessed, delivered and received, as structural competency increasingly supports (Metzl & Hansen, 2014).

Thinking systemically then allows us to account for how illness is experienced by the person in their wider context and family system. With multiple medical teams often involved, one can readily point to these teams as well as the family as multiple systems individuals hold membership of. Relationally this may mean competing demands and expectations placed upon individuals**.** Alexander (1981) likens the positions imposed by medical treatment to Bateson's (1956) Double Bind. At any one time an individual is asked to be an active agent involved in their treatment, and at other times, particularly those of acute illness, are treated as passive patients unto whom care is given. The conflict is clear in a pull between active and passive positions. This plays out in a family too in which children naturally must navigate a move from a position of dependence to independence with age. Consider the complexities of this transition when having to factor in supporting a child's growing understanding of and responsibility in managing the medical demands of a rare

health condition, including frequent appointments, medical decisions, and treatment itself. This serves to highlight the relational and social elements and pressures of living with FA and DBA, and therein the need to consider these conditions beyond the experience of medical symptoms alone.

Whilst this literature is fairly old, research has continued to lend support to using a systemic focus in considering the impact of living with illness across a system. Stiell et al. (2007) have written of how illness is situated within a context. This context involves dynamic relationships and so it is pertinent to consider how health conditions unfold in these relational settings. A reciprocity may be seen to exist in which illness and relationships may impact on one other. Coyne et al. (2001) has evidenced a link between spousal relationships and survival rates in partners recovering from heart failure, whilst Baider et al. (2001) reported similar rates of stress and low mood in both members of couples where one member had received a cancer diagnosis. As such it seems pertinent to consider the impact of living with FA or DBA within a system context in which all members may be affected, a context which may well hold the potential to support or hinder an individual living with these conditions. In holding that illness occurs within a social context, so too must we accept that coping and treatment strategies may well also be considered interpersonal processes (Stiell et al., 2007). It is crucial to explore whether systems can be better supported in living with FA/DBA.

1.9 Empirical Research

1.9.1 Chronic Illness and the Family

Whilst much research has been theoretical, there has been increasing focus across health literature on the impact of chronic illness on the family. The summative message is that chronic illness can and does impact on the emotional well-being of the family and family quality of life (Golics et al., 2013). At the same time, there has

been some interest in resilience or protective factors. A Risk and Resistance Model seeks to consider the factors that help families adjust adaptively to a diagnosis of child chronic illness and to maintain wellbeing (Wallander & Varni, 1998). Whilst levels of family cohesion are noted to positively impact on adjustment to diagnosis, this is also mediated by the amount of support received by parents and the family. It has also been reported that how families appraise the threat of a diagnosis will impact on how they adjust and cope following diagnosis (Folkman, 1999). This research highlights the importance of effective and sufficient support for family well-being. It presents further impetus to consider the impact on families in order to better understand the supports desired and needed.

Other research has focused more on specific family roles in considering how family members are impacted by chronic illness. An overview of this research is provided here.

1.9.2 Parents

The literature is agreed that the parental relationship is affected by chronic illness of a child, and in turn impacts on the experience of the child. Research notes that in managing a child's chronic health condition one parent often oversees the bulk of the child's care, whilst if there are other children the partner will oversee their care, creating subunits within the family as parents try to manage the multiple demands of healthcare and family life (Ray, 2002). This need not be regarded as a negative outcome. Whilst this may lead to strained relationships (Kratz et al., 2009) other research indicates that this role divide may in fact boost cohesion in the close communication required to effectively manage this sharing of family tasks (Fawcett et al., 2005). Further it has been reported that parents may perceive their relationship as strengthened in a sense of mutual commitment and shared experience in caring for a child with health needs (Ray, 2002). Parents are noted to experience worry and uncertainty about the future, about prognosis, about medical procedures, and finances (Gannoni & Shute, 2010). Parents often need to take time

off work to attend medical appointments which may exacerbate financial strain and add to parental stress.

Parental stress has also been associated, positively and negatively, with access to information. Research notes a positive impact in having timely information about a child's condition as well as an opportunity for contact with other parents in similar situations (Hartmen et al., 1992). This is supported by more recent work looking at adolescent idiopathic scoliosis, which highlights the stress parents experience in having to be involved in medical decision making whilst at the same time facing obstacles in accessing appropriate information (Motyer et al., 2020). Whilst health policy increasingly advocates for individuals to be active collaborators in their care (Le Var, 2002), for children this role will be filled by parents. It is therefore crucial to consider the experience of parents receiving a diagnosis and how best to support them with timely and appropriate access to information. Whilst research has indicated that parental perception regarding the impact of the specific health condition may influence parental and family adjustment (Swift et al., 1967) this is further evidence still of the importance of supporting parents with timely access to information.

Research also indicates that parents may continue to experience times of heightened emotional distress across the child's life. Writing on chronic sorrow, Coughlin & Sethares (2017) report that health crises or new developmental milestones may act as trigger points for a renewal of feelings related to sorrow, sadness or loss. These findings are reminiscent of Rolland's (1984) model indicating how the findings of practical research map onto and support the theory.

It has also been documented that social support networks are an important aspect in coping with the emotional and practical demands of caring for a child with chronic illness (Lauver, 2008). It has been noted, however, that information about and therefore access to support groups appears to happen by chance, rather than their being shared as part of formal care delivery (Ray, 2003).

1.9.3 Individuals

Research has indicated a greater risk that individuals with chronic illness may experience psychological distress, with Meleski (2002) documenting that young people living with chronic illness experience a reduced quality of life in comparison with their peers. Yeo & Sawyer (2005) note that the emotional impact of chronic illness may be substantial and wide ranging. Depending on the medical demands, school and education may be disrupted. This may also present difficulty in the maintenance of friendships and thereby engender a sense of separation from one's peer group. Children have reported difficulty handling questions from peers regarding their absence on return to school after time off relating to their illness, as well as difficulty re-initiating relationships upon their return (Gannoni & Shute, 2010). Academically, children may also have difficulty maintaining academic work, which may impact on short-term factors such as self-esteem, and long-term factors such as choice of career.

The literature to date has been largely focused on understanding the experience of individuals and may often appear negative in outlining the difficulties experienced. Research focused on strengths or positive experiences has been lacking, but there has been some extrapolation of positive or helpful factors as experienced by children and their parents. Berg et al. (2007) note that the parent-child relationship can act as a buffer for negative emotions when experienced as collaborative, noting more positive mood and fewer markers of depression for adolescents when this is the case. This indicates how positive and protective the parent-child relationship can be, and thereby further rationale for supporting parents and families with help to maintain these relationships.

1.9.4 Siblings

Historically research looking at the experience of siblings of children with chronic illness was considered lacking (Wallander, 1998). Whilst there has been greater attention paid to these experiences, the results remain complex to interpret due to

issues regarding the heterogeneity of samples, illnesses and interventions (Smith et al., 2018). Some research has reported on aspects of sibling guilt regarding their status as healthy (Yeo & Sawyer, 2005). It has also been reported that siblings may feel they receive less attention from parents (Knecht et al., 2015). A meta-analysis by Vermaes et al. (2012) reported a small but significant overall negative impact in areas of psychological functioning for siblings, whilst Fullerton et al. (2016) reported raised levels of emotional and behavioural difficulties. In the latter case difficulties were mediated by family socioeconomic position as well as time since diagnosis, and so by no means represent a straightforward relationship. This indicates a need for research that is sophisticated and sensitive to these nuanced variables. Chudleigh et al. (2019), writing of cystic fibrosis specifically, raise the valid point that lots of research will use parents as proxies for sibling experiences and advocate for more that gives voice to the experience of siblings directly.

1.10 One Condition or Many?

In reviewing the research an issue that arose concerned the area of focus. There appear to be two schools of thought on whether the focus of research should be condition specific or on chronic illness more generally. On the one hand a critique of the generalised approach is that to focus on chronic illness generally is to consider those individuals and families as one homogenous group, which of course is incredibly misleading given the variables already outlined, both in terms of illness trajectory and family related factors. On the other hand, to focus only on specific conditions has, as in the present case, led to a situation where there are gaps across the literature where some conditions are widely published on, and others are not recognised at all. This may impact on eligibility for and relevance of support in that specific groups may receive an intervention or support generalised from other research. It has also been noted that there is a bias in illness research according to factors such as clinician interest in particular conditions, and even how sympathetic medical clinicians might be to psychological ways of thinking (Eiser,1985). This has

produced a research landscape that is largely unequal.

On a more practical level it is perhaps more difficult clinically to justify funding for small groups. Delivery of healthcare continues to operate according to utilitarian principles with provision aligned to majority need (Garbutt & Davies, 2011). This may be exacerbated by the economic appetite of pharmaceutical companies which operate as businesses (Goldacre, 2013) in that producing a cure for rare diseases will not be as profitable a venture, which may in turn mean that such research efforts receive less funding. In this case funding bids for rare conditions may be less successful. From this perspective one can see benefit in referring to children with chronic illness more generally, as this is likely to make a more persuasive case for funding, as well as more cost-efficient support provision.

Indeed, there is a growing body of literature which advocates for research focused on the shared experiences of living with rare conditions, holding that there may in fact be more commonalities in these experiences than there are differences (von der Lippe et al., 2017). This area has been largely under-researched. Whilst this is an emerging topic within the literature, comparisons have only been possible in studies which has compared multiple conditions, of which there are few, or by comparing the results from single condition research. In either case, both conditions here are not represented within this pool of research. Whilst accepting the likelihood of commonalities across the experiences of living with rare conditions, we cannot possibly appreciate important points of difference if certain conditions are not represented; for this reason, it is argued here that both strands of research continue to be necessary, and that neither focus is sufficient in isolation, but that this tension necessitates both. The two could enjoy a transactional relationship in that one may be used to justify funding for support, and the other to ensure clinicians are able to tailor the provision of that support to the specific health experience of the child or family.

1.11 Focused Review

A scoping review was conducted with the aim of establishing how research to date may have considered the psychological impact of living with FA and DBA for individuals and/or their families. Whilst limited, the following research base does indicate a relational element to living with either diagnosis and therein a need to consider the impact at a wider system level.

1.11.1 UK Research

Following the literature search criteria previously set out, no relevant papers were identified within the UK. One paper was identified which retrospectively followed a cohort of 80 individuals diagnosed with DBA across a period of 20 years (Ball et al., 1996). The focus of the paper was medical, outlining rates of incidence and response to available treatments. The paper had used data from the DBA UK registry which was developed to establish a research database on aetiology, pathophysiology and treatment of DBA. The paper in question is not directly relevant to the present research terms. It is mentioned, however, to highlight that in this early consideration of DBA, psychological distress, psychosocial issues, and well-being were not considered part of the clinical picture. The current landscape of research within the UK would suggest that this largely remains unchanged for both FA and DBA. One further UK paper was identified which referred to the 'psychological strain' which may be placed on the individual living with FA and their family (Tishkowitz & Hodgson, 2003). The authors noted problems in early life, stem cell transplant and susceptibility to cancer as contributors to this strain. The authors advocate for psychosocial support and the input of a multi-disciplinary team as central to providing care. Whilst this recognition is undoubtedly progress, it received the coverage of only two sentences within a larger review article. Moreover, without reference to any published research, one questions what this recognition is based

on beyond anecdotal understanding. Whilst it does well to introduce the idea of well-being as part of the experience of FA, it highlights further the lack of current research which gives voice to those living with either condition.

1.11.2 International Research

For the purposes of exploring this potential research gap, the literature search was widened to consider international research. The majority of papers considered for inclusion originate from the United States (US). Some of these have focused on the parental experience of raising a child with a diagnosis of FA or DBA. This has highlighted the difficulty experienced by parents living with prolonged uncertainty (Zierhut & Bartels, 2012). The focus of the authors was on the waiting period between diagnosis and treatment and to understand the factors considered helpful or otherwise by a total of seven parents interviewed. The analysis presented themes relating to parent's emotional responses on receiving a diagnosis, which included grief and shock, daily thoughts concerning FA, and supportive sources during the wait period (medics familiar with FA, religion, and websites or blogs). Parents also touched on coping mechanisms, and reflections on how family dynamics had been impacted. Medical professionals were cited as sources of both most and least support depending upon their level of knowledge and empathy, with parents recommending medics receive more education from FA specialists and families.

Information and uncertainty appear across other studies. Zierhut et al. (2013) considered parents experiences of preimplantation genetic diagnosis (PGD) in FA, a process that profiles embryos preimplantation to facilitate genetic matching for transplant. They report that less than 35% of parents had been offered the procedure, and only 70% were aware of it. This indicates how experience will vary depending on knowledge made available to families.

Other studies broadened their focus to include parents and children. Hamilton et al. (2015) considered the relationship between knowledge and genetic information seeking behaviours in families supporting a member with an inherited BMF syndrome. The authors report a link between knowledge and the information opportunities individuals have accessed such as genetic counselling or internet information. For those families living with FA, greater FA knowledge was linked with having sought information from other families affected as well as support groups. This might indicate a role for peer support in the sharing of information and thereby in mediating uncertainty. Decision making has elsewhere been linked with perceived lack of choice from medics (Hutson et al., 2013) and as mediated by parent emotional reactions (Hamilton et al., 2013).

A publication by US Fanconi Anemia Research Fund (Hays et al., 2014) covers impressive ground in documenting possible areas of psychosocial distress or difficulty across the family affected by FA. Many of these are symptom specific, with reference to nausea, obesity, transplant, and head and neck surgeries. There is, however, a whole chapter dedicated to psychosocial issues more generally in the family, which are considered according to developmental stage, for example navigating parental protectiveness regarding health and a child's seeking independence in adolescence. However, the publication itself notes that these issues have not been assessed from the perspective of children and families living with FA, and so the theory and ideas are generalised from research specific to other conditions. Elsewhere the content is likely based on the years of experience the organisation has in working with FA families. Whilst this experience may be sound foundation for the content of the publication, it may, albeit inadvertently, help maintain a situation in which specialised knowledge accumulates in a few areas and has not filtered outward into the medical sphere. This may help explain why, as previously noted, parents may encounter obstacles in access to information and support. Research could bolster the work of these supporting organisations by formalising their specialist knowledge and establishing it within the research sphere. Therein medics may be better placed to offer information and signposting to specific

organisations.

1.11.3 Individuals

Five papers were identified as relevant in thinking about individuals and the psychological impact of their diagnosis. One paper considered the efficacy of electronic monitoring feedback at improving participation in treatment in an adolescent diagnosed with FA and experiencing anxiety and depression (Hilliard et al., 2011). Although this paper does not consider the context in which anxiety and depression was experienced, it does highlight the importance of integrative approaches. Another, a scoping review, explored how Cognitive Behavioural Therapy (CBT) had been adapted for use with adolescents with chronic illness who experience depression (Morey & Loades, 2020). Of the 12 studies selected for inclusion, 1 concerned FA. Common adaptations made to CBT are outlined including behavioural activation that balances illness-related activities with enjoyable ones, and cognitive restructuring specifically of illness-related thoughts. The paper concludes that CBT is commonly adapted for chronic illness, and that how an illness presents may impact the cognitions and behaviours targeted. It concludes that more evidence is yet required. This paper helpfully begins to question the efficacy of current psychological interventions for use in chronic illness. In this case however, the illnesses considered varied widely from diabetes to polycystic ovary syndrome to FA. It is noted that life limiting conditions was an exclusion criterion indicating some misunderstanding of FA and that this paper was perhaps included in error. Even still, these conditions vary hugely in terms of physical symptoms and thereby one might expect in terms of psychological impact too. Given also the small sample number overall and for each condition, it is difficult to draw any meaningful conclusion here and indeed the paper refrains from making any conclusion about how well CBT is adapted. The papers selected are cases presented by researchers and do not include the direct experience of those receiving the intervention. Moreover, the sessions received ranged from 6-35 sessions which, again, makes comparison very difficult. What the paper does make clear is that condition specific knowledge may

30

be beneficial if one is to adapt CBT.

Three other papers noted have touched on psychological well-being in the context of specific symptoms: piebaldism (Janjua & Guldbakke, 2007), stem cell transplant procedures (Packman et al., 2010), and obsessive compulsive disorder documented in one patient with DBA (Pallanti, 2008). Each conclude a possible role for psychological support within the aspect under consideration. Though fragmented, the literature is beginning to present a wider picture of how individuals may be psychologically impacted by aspects of living with FA/DBA.

1.11.4 Siblings

One paper recounted the experience of a sibling donor indicating the complex emotional processes involved in this experience (Parmar, 2003). Research exploring the impact on siblings has noted themes of invisibility and worry (Hutson & Alter, 2007), and moreover highlighted the complexity of decision making within families regarding harvesting tissue from siblings (Rubeis & Steger, 2019). There is research questioning the ethics behind conceiving a child for donor matching as well as preimplantation genetic diagnosis testing (PGD), (Bernal, 2004). Research also advocates that haematologists be aware of the psychological implications of these procedures (Wagner, 2005). Whilst Bernal (2004) concludes that these are in fact morally sound actions, it is interesting to note that many families live in regions such as Switzerland and Ireland where PGD is banned (Bellavia et al., 2010). This raises issues around politics, judgement and access that are a real part of the experience for some families. This indicates that as research continues to grow in considering psychological well-being, issues such as the country and medical system must be taken into consideration.

1.11.5 Predominance of medical focus

Another paper selected in the search made reference to the importance of providing psychological support and information for parents (Korthoff et al., 2013). Whilst this

31

may be a welcome suggestion, it was made amid a wider medical discussion without any real consideration of why this might be necessary. This raises an ontological concern whereby the experience of parents may be assumed, and recommendations are made based on this. This is particularly problematic to see in medically focused papers which robustly support and reference their scientific claims. This indicates a disparity in how evidence is managed, as well as the predominance of a medical focus even where research is beginning to broaden its focus. Indeed, whilst one paper was scanned on account of the multi-disciplinary team it advocated for in the management of DBA, this multi-disciplinary team appeared to include medical professionals only (Vlachos & Muir, 2010). This may be improving, with a recent paper noting that a constant need for medical treatment may impact negatively on quality of life and therein noting a role for an inter-professional team which did include those from psychology backgrounds and other professions (Gadhiya & Budh, 2021). This indicates a small move in the condition-specific literature at recognising and documenting the potential interplay between physical and mental health in those with FA or DBA.

1.12 Research Rationale, Aims & Questions

1.12.1 Rationale

The research overview indicates a need to consider more comprehensively how individuals and systems are impacted upon by living with FA and DBA. Whilst there are beginnings of this focus in US literature, differences in healthcare provision warrant a consideration in a UK context specifically. It is only in exploring the impact that we can ascertain a need for and identify relevant supports. The existing research would also seem to indicate that there may be some benefit in providing access to specialised psychological support for these families. Zierhut & Bartels (2012) note a frustration encountered by parents in having to explain repeatedly, including to health professionals, about their child's diagnosis. Extrapolating from this one can assume that psychologists working in general mental health settings may not be well versed on these conditions further exacerbating this frustration for

32

individuals seeking support. Moreover, providing this support without knowledge of the condition in question risks perpetrating a hermeneutical injustice (Fricker, 2007) in that a clinician's lack of knowledge may deprive an individual of a way in which to conceptualise and understand their distress. Whilst oncology services and those for sickle cell disease are staffed by psychologists with specialised knowledge it seems timely to consider whether similar psychological offers of support would be beneficial to the families concerned here.

1.12.2 Aims

This chapter has outlined that illness is marked by more than physical symptoms alone, and thereby identified a research gap regarding the wider illness experience of individuals living with FA and DBA. Moreover, it has demonstrated that this illness experience is one that happens within a system or family context, and thereby all members may be impacted in some way. Drawing on research from other chronic illnesses indicates that this impact is multifactorial: self-esteem, self-image, stress, grief, worry, relationships, quality of life. All of these factors have been referenced throughout this chapter as experienced across the family. To help capture this multitude, the term psychological impact will be used to facilitate a thorough consideration of how individuals and families may be affected in living with FA/DBA. This will consider any aspects relating to cognitive, behavioural or affective changes.

The proposed research then aims to build upon a limited research base. In doing so it seeks to capture the experience of individuals and their system across the lifespan. It will seek first to establish the psychological impact of living with FA/DBA on the individual and their family. Secondly it will seek to establish whether there is demand for psychological support, whether this has been accessed, and where so whether this has been deemed appropriate.

1.12.3 Research Questions

1. What is the psychological impact on a family system of living with Fanconi Anaemia or Diamond Blackfan Anaemia?

2. What is the current demand for psychological support and how suitable has provision been deemed?

CHAPTER TWO: METHODOLOGY

This chapter will outline the epistemological stance that has informed this research. It will then describe the methodological approach used including details relating to the sample, recruitment, and analysis of data.

2.1 Epistemology

The research will be approached through the lens of critical realism. The researcher accepts the biological reality of the health conditions that are FA and DBA. At the same time, however, the research also seeks to understand the psychological impact of these conditions, an altogether more abstract and relative concept. On this the researcher is drawn to systemic ideas pertaining to how one's context and system may impact and shape experience, in this case of living with a lifelong health condition. Whilst systemic practice is often linked with constructionism, Pocock (2013) has argued for a move toward critical realism within systemic thought and practice. In accepting an underlying reality (biological symptoms of an illness), critical realism tempers the relativism of social constructionism. This allows one to anchor the present research on a biological reality whilst still providing space to consider how social factors may influence one's experience of living with FA or DBA. Critical realism then is not only in keeping with the systemic frame used to approach this research, but it also offers a way of accommodating the complex interplay of the natural and social sciences inherent in the topic.

A caveat must be added, however, for whilst critical realism accepts a social aspect to the production of knowledge, it has been argued that it does not go far enough to consider the position of the scientist, researcher, or knowledge producer (Albert et al., 2020). It is widely theorised elsewhere that science is anything but value free,

and that its processes are rife with systemic inequalities pertaining to power (Harding, 1993). In short, the knowledge produced is shaped by those with the power to do the producing. Our current knowledge of FA and DBA, largely medical, will have been shaped by these processes, which in turn impacts on the provision of supports and services for the families concerned. The researcher wishes to draw upon the work of Albert et al. (2020) who, in using critical realism, seek to consider the power processes beneath science and its knowledge production. A *critical* critical realism then, will imbue the present research with a consideration of which voices have been supported and which have been marginalised in producing our current knowledge base concerning FA and DBA. It will allow consideration of how the prevalence of a scientific paradigm may have impacted on our understanding.

2.2 Thematic Analysis

In line with this, it is important to situate the present approach to Thematic Analysis (TA) as a critical one too, in which language is seen as creating rather than reflective of reality. Thematic Analysis has been selected on account of the flexibility it affords in approaching qualitative research. Kidder and Fine (1987) make a useful distinction between small q TA and big Q TA. Small q TA seeks to conduct qualitative research in a way that is acceptable to quantitative research standards. It upholds traditions of positivism not in fitting with the epistemological or systemic frame used here. Small q notions of reliability depend on the assumption that the truth exists waiting to be uncovered in the data and may be reached by the agreement of several researchers. This approach to coding and analysis risks overlooking nuances deemed irrelevant or anomalous by a coding frame designed in advance. Big Q TA calls for a greater flexibility, particularly regarding the process of developing codes and themes. It permits the researcher an active role in analysis that is fluid and flexible as their engagement with the data deepens. Big Q TA also rejects the possibility of uncovering a universal truth. This is important for the present research, which seeks to explore the unique psychological impact on

families living with FA and DBA. Whilst a similar set of biological symptoms may mark an external or biological reality that can be uncovered by medical diagnosis, the research hopes to understand the lived 'reality' of individuals. It is theorised here that lived experiences of the same biological reality will vary in how they are mediated by different familial and systemic contexts and meanings. As such the accounts recorded will be regarded as constructed by each individual, and as being informed by their particular familial and sociocultural context. It is accepted that these accounts will shed light not on an objective reality, but on individual lived experience in a UK context. These accounts will also be interpreted by the researcher, who will thereby exert some influence over the final analysis presented. This subjectivity is considered a strength, not a weakness of the approach (Braun & Clarke, 2020). An individual account may not to refer to the wider socio-political context that has shaped its experience, but the researcher holds freedom and flexibility to consider this wider scope. She is thereby enabled to look for the presence of wider patterns and themes that may underlie individual experiences. Analysis will be inductive rather than data driven.

2.3 Design

2.3.1 Participants

In seeking to meet the systemic frame of the research, two sets of participants were required: individuals living with each condition (referred to as individuals throughout), and family members of those individuals. Led by recommendations from Braun & Clarke (2013) recruitment was capped when 15 individuals had been reached. On account of one parent interviewing alongside their partner the total number of participants was 16: 6 individuals, 10 parents. The requirement for inclusion was that individuals had received a diagnosis of either condition or were a family member of someone who had. Additionally, all participants were required to speak English for practical reasons, reside within the UK, and be above the age of 18 years to meet ethical approval. The age requirement was stipulated on account of an ethical

concern regarding interviewing children; mindful that parents may share different amounts and at different times, it was felt that to involve children might introduce worries or concerns unnecessarily. Whilst this risk could be managed with an appropriate research design, the scope of such a piece was beyond the timeframe and practicalities of the current research. Given the lack of research on this area at the present time, a small sample should not be regarded as a drawback; the present research seeks not to generalise, but to initiate some insight into how families may be psychologically affected in living with either condition.

A demographic table of participant information is included in Table 1. The specific age of individuals now and at diagnosis was not included to protect anonymity in what is a small community. Individuals ranged in age from 20-50 years, and in age at diagnosis from 2.5 months – 38 years. Parents spoke of their children who have a diagnosis of FA/DBA; children ranged in age from 2-20 and in age at diagnosis from 3 months – 13 years.

Table 1

Demographic Information

Participant No.	Gender	Diagnosis	Years since diagnosis
Individual 1	Female	DBA	8
Individual 2	Female	FA	17
Individual 3	Female	DBA	50
Individual 4	Female	DBA	29
Individual 5	Female	DBA	15
Individual 6	Female	FA	14
Parent 1	Female	FA	14

Parent 2	Female	DBA	5
Parent 3	Female	FA	3
Parent 4	Male	DBA	6
Parent 5	Female	DBA	1
Parent 6 & 7	Female & Male	DBA	2
Parent 8	Female	FA	1
Parent 9	Female	DBA	3
Parent 10	Female	DBA	7

2.3.2 Recruitment

The researcher drew upon the links of charitable organisations specific to each condition to reach those the research was relevant to. Individuals were invited to participate via social media platform Facebook. There are well established online communities for both FA and DBA which were utilised with permission from group administrators. After receiving an email of interest, an invitation letter was sent (Appendix A) and following this a consent form was required (Appendix B). Initially recruitment sought to reach individuals living with either condition with a view to these people self-selecting those within their immediate system (parents, siblings, and/or partners) who would then be contacted to seek consent. In practice individuals did not readily select family members. To avoid producing only an individual focus, a decision was made to recruit family members of individuals living with FA/DBA directly. In this way there remains presented here a wider consideration of the impact on the family.

2.3.3 Interviews & focus groups

Given the sensitive nature of the content for discussion, it was envisioned that 1:1 interviews with individuals would be ethically most appropriate. It was initially

planned that focus groups would then be held separately with groups of parents/siblings/partners to gather further perspectives regarding impact at a system level. In practice, all family members seeking to participate were parents. Due to this being a small number and based on the rich detail of the 1:1 interviews which had begun, a decision was made to offer 1:1 interviews to parents in place of focus groups. In both cases a semi-structured approach was taken allowing the interview to be guided by the conversation.

Due to the government guidelines around COVID-19, interviews were held remotely via Microsoft Teams. Interviews lasted for up to one hour with an average time of forty-five minutes.

2.3.4 Developing semi-structured interviews

The interview schedule (Appendix C) was developed in conjunction with the research supervisor. Given the lack of research in this area, questions were largely guided by the research aims of seeking to capture the experience of living with either condition and the possible psychological impact. In order to consider lifespan factors at an individual and system level, the schedule explicitly sought to cover the period prior to diagnosis, receiving a diagnosis, and life after. Whilst it was envisioned that many of the individuals who had received a diagnosis in childhood may not remember this clearly first-hand, it was included nonetheless to give space to consider how the story had been shared and received with age and growing understanding.

2.3.5 Transcriptions

The content of interviews was typed up verbatim. Microsoft Teams did provide a written transcript of the recordings, but due to its automated nature this was error prone. All transcripts were thus corrected independently by the researcher against the audio recordings to ensure accuracy. Those sections used in the write up of this research were altered only where clarity was necessary (Willig,2013). Long passages of text were shortened based on transcribing conventions (Banister et al.,

1994), (Appendix D). Care was taken to ensure no changes to the content of the data. Analyses of data focused on the content of discussion only; for this reason, non-linguistic elements of speech were not transcribed.

2.4 Ethical Approval

Ethical approval was agreed by the University of East London Psychology School Ethics Committee (Appendix E). Written consent was obtained from all those who elected to participate, as well as verbal consent to audio recording of interviews, their transcription and anonymised use in write up. Anonymity was achieved by removing identifying details and assigning a number to each person.

2.5 Analysis

The process outlined by Braun & Clarke (2006) was used to inform the procedure of analysis. This began with familiarisation during which the transcripts were read, re-read and re-listened to in order to achieve a level of immersion with the data. A curious stance was maintained in which the data was questioned and reflected upon during these readings, which was helped by using margin notes and a reflective log. This helped in forming early analytic ideas. Moving on to initial codes, data was then labelled where of relevance or interest to the research questions (sample in Appendix F). In this way the data began to be organised around patterns and themes generated by the researcher. Construction of themes followed, an active process (Braun & Clarke, 2019) during which codes were reviewed and collated into groups around a core of shared meaning. These were elevated to provisional themes (Charmaz, 2000). Whilst the research question was used to guide this process, ultimately the researcher selected which pieces of information were most salient in terms of their relevance. Thematic maps were employed at this stage to

support in understanding the connections between themes and the wider data (Appendix G). As in line with systemic theory around hypothesising (Cecchin, 1987), these initial themes were held lightly at this stage to avoid restricting further thought and revision. The penultimate stage involved review and refinement of initial themes, to ensure that they maintained the meaning in the data segments previously coded, whilst also capturing and fitting with the story provided by the dataset as a whole. Theme names were generated and refined to ensure clarity regarding the content of each theme. The final process was that of producing the report and choosing quotations from the data to present. These were selected with the aim of blending an illustrative and analytic style.

2.5.1 Process of analysis

The process involved moving from descriptive summaries to interpretation (Langridge, 2004) with the intent that analysis would provide an understanding of latent meaning. Whilst seeking to respect and give space to participant meaning and semantic content, the analysis sought to build on this by exploring what the researcher saw as underlying patterns and meanings across the individual accounts. A systemic theoretical framework was used in thinking about family systems; this will inevitably have shaped the analysis provided. In upholding a critical realist approach interpretation was undertaken to theorise how participant views may be mediated and informed by underlying socially driven factors, ideas and understandings. Consideration was given to how the participants oriented towards the questions, as well as the world view and assumptions that might underlie their responses (Terry et al., 2017).

2.5.2 Quality Considerations

Consideration was given to quality as part of the ongoing process of the research. Consistent with the epistemological stance, this was approached by sustaining reflexivity as opposed to employing traditional positivist measures of quality. Terry et al. (2017) holds that researcher subjectivity is part of this quality assurance. Rather

than relying on a second coder, which would evidence no more than two people trained or socialised to see the same meaning in the data, they highlight that a better measure of quality lies in deep immersion with the data and an iterative process of ongoing code and theme refinement. Whilst endeavouring to work in line with this view, the researcher sought to remain mindful of her own world view and personal beliefs or biases that may form part of her interpretation.

2.5.3 Ethics of Interpretation

Whilst accepting that interpretation is a necessary part of data analysis, the researcher was mindful of ensuring interpretations were made ethically. To this end efforts were made during interviews to check where a statement was unclear or open to interpretation, in the same way as would be used therapeutically to ensure understanding. It has been suggested that the utility or helpfulness of interpretations can only be evaluated by their consequences for those they concern (Willig, 2012). To this end the interview questions were explicitly aligned with the research questions. This will ensure that any recommendations made by the research stem directly from the voice of those involved, and not from a detached analysis.

2.6 Relationship to the Research

In seeking to maintain a reflexive stance throughout the research process it was necessary to consider my own relationship to the topic. Moreover, having approached the research using a systemic frame it was useful to consider my own family of origin and position within. Starks & Trinidad (2007), writing of the researcher role, note their part as instrument and interpreter; that is, they actively decontextualise data in some way to rearrange, re-present and thereby recontextualise - or interpret - it in write up. My family of origin is membered by an individual who lives with a lifelong and life-limiting condition. Whilst different from both conditions under consideration here, this experience lends me some insider status in appreciating the interplay of physical health and well-being within a family

43

system. This is perhaps what spurred me to consider the experience of FA and DBA systemically. Based on this experience it is my view that living with chronic illness affects the whole system; that the resources available to and relationships within a system may vastly change the lived experience of what is medically diagnosed as a common set of symptoms. It is also my view of mental health and the profession I have elected to enter, that the role of a clinical psychologist is to promote well-being and its maintenance even where it already exists, not just to be involved in ameliorating distress. My stance therefore is one that promotes prevention and support. Time was spent reflecting on this before embarking on the research. Cognisant of my own views, I approached the research then intentionally primed to look out for alternative perspectives and ideas. This is in line with the epistemological position and systemic frame used, which seek to uncover the multiple perspectives. I cannot claim to have successfully separated all that is mine from what is presented here, but drawing upon the clinical aspects of my training, I have sought to hold lightly to my own hypotheses throughout the process (Cecchin,1987).

CHAPTER THREE: RESULTS

Themes identified during analysis will be outlined in this chapter. For clarity, themes will be outlined for individuals first and then for parents. As outlined in Table 1, two main themes and four sub-themes were identified for individuals. Two main themes and four subthemes were also identified for parents. Two further themes with three subthemes each were identified as arising with strong overlap for both individuals and parents and will therefore be presented together. Quotes will be used to evidence and support themes.

Table 2

Summary Themes & Sub-themes

	Themes	Subthemes
INDIVIDUALS	Knowledge	Medical Uncertainty
		Knowledge Acquisition
	Illness Concept	Relationship to Condition
		Social Evaluation
PARENTS	Knowledge	Medical Uncertainty
		Questioning Knowledge

	Social Responses	Losses & Gains
		Social Judgement
JOINT	Family Dynamics	Parental Relationship Strain
		Protective Communication Patterns
		Concerns Regarding Siblings
	Mental Health Care Necessary but Inadequate	Impact Across Family
		Unequal Access
		One Size Does Not Fit All

3.1 INDIVIDUALS

3.1.1 Theme One: Knowledge

The idea of knowledge about FA and DBA arose across all interviews. It was particularly clear that a lack of knowledge was encountered in many aspects of living life with either condition. Individuals articulated links between lack of knowledge and the impact this had on their emotional well-being, as well as touching on the ways in which knowledge is gained.

3.1.2 Subtheme One: Medical Uncertainty

All individuals outlined some level of uncertainty surrounding their diagnosis. For some this meant a diagnosis was not made until adulthood, but even for those diagnosed as children, who had grown up aware of their health condition, the uncertainty regarding medical knowledge was still known. There was a sense that diagnosis relied on chance or luck that the medical team had experience of working with the conditions.

46

It was actually quite easy mainly because I was under the care of [Hospital Name] and there was actually a specialist, well, someone who'd heard of my illness before and so was able to diagnose quite quickly and I was able to get the right treatment from there.

Individual 4

If I think about it too much I do get little bit angry because when you look at my presentation for DBA, I'm a classic presentation with all the abnormalities. But not one doctor thought to connect all the dots.

Individual 1

When we saw the specialist, he said your medical history is a classic case, all of those things would be a classic childhood case that just wasn't picked up when I was a baby.

Individual 5

The medical uncertainty does not end with diagnosis, and participants described an ongoing sense that their treatment and health prognosis were uncertain too.

It could be that tomorrow my medication doesn't work, and I need a transplant and I just don't know.

Individual 4

 For some there was a sense that this uncertainty was not well supported, that the aim of medicine was to make a diagnosis and there is a gap in support following this.

It was just a bit like here we go, and the diagnosis for them was the endpoint, whereas for me it was just the start. I think helping people navigate that would be massively helpful.

Individual 5

If I have any questions about DBA then [hospital] have been amazing but from the sort of processing everything that's happened I've kind of just been left myself to get on with it.

Individual 1

There was a tension noted between the perceived certainty of developing additional illnesses in the future but an uncertainty as to when.

It's like an MOT to just check for these things every so often just to make sure. So my original problem – solved, that's fine, that's a done deal, but now it's just the waiting on other things.

Individual 2

And then sometimes I'm like well the statistics will be like, right well you're [age] now so prepare for cancer that's on the way sort of thing, and so obviously that is on my mind.

Individual 6

In this same certain-uncertain tension individuals touched on death and mortality. There was a sense of feeling pressure in this knowledge.

And it's known, or it's widely thought of in our community that we're not all going to live very long, and we're gonna die at a really young age, so just that itself is a lot of pressure for everyone to put on themselves.

Individual 4

For many participants a combination of knowledge about their condition and its uncertainty had impacted on their life decisions, most notably around family planning, but also career.

> *I guess the median age thing is like between 20 and 30 something, and that's like a lifespan in general. So I think about that quite a bit. Like I feel like I need to consider that. I could have done a foundation year and knowing that kind of made me not do a foundation year.*
>
> *Individual 6*

> *I did always want children and it kind of changed my view. Because I thought, even if we can have the IVF so that the child doesn't have DBA, I'll still have it so that child will still grow up in an environment where hospital is normal, where we have to plan around these things. And I just thought my life is fine, I don't want to add in an extra complication.*
>
> *Individual 5*

3.1.3 Subtheme Two: Knowledge Acquisition

Participants described three main sources of knowledge acquisition regarding their diagnosis: medics, parents, and the internet. They described how it could be overwhelming to acquire information and make sense of their diagnosis. Level of involvement in medical appointments appears to be an important factor.

> *I was always included in what was going on. You know, discussions weren't really had without me in the room as far as I am aware […] I think this is really important because that gave me an understanding of what was actually happening instead of just being a sample or specimen or something.*
>
> *Individual 3*

49

I hate people, even at that age, I don't like people hiding things from me. […] I know you're trying to protect me and everything and not trying to scare a child, but if I'm dying, I need to know. And obviously you have those questions as a child you know. […] That was the really annoying thing, is that people treat you like it's nothing to do with you, that they're trying to fix your body, not you.

Individual 1

One person touched on developmental issues in wishing to have a separate space to discuss with medics but being bound by the convention of age and parental presence.

I think like the person I would [discuss with] is the doctor, like the consultant. Then again, my parents were always in the room, so I didn't want to upset them constantly about that. And if I had somebody to talk to, like just alone, then I think that would have been nicer for me.

Individual 6

All individuals cited the internet as a source used to access knowledge. For some internet research actually led to their diagnosis, highlighting further the above theme of medical uncertainty, and indicating a level of agency required in accessing a diagnosis. These accounts highlight the central role of condition-specific charities in supporting individuals with access to specialised knowledge.

So I just started researching the problem I had with my hands, the problem I had my heart, everything I have wrong with me. And ended up contacting the Fanconi Hope charity. They said why don't you try contacting the DBA UK charity because it sounds quite similar to DBA. So I got hold of the DBA charity. I then went back to my GP and said look you've got to refer me.

Individual 1

So I felt I was doing a lot of the legwork, as in I've spoken to charity and they've told me this, and then the doctor would be like oh OK. And at times I felt I was guiding it, and that was hard because I thought, well if I get this wrong then I don't know where else to turn because the doctor doesn't know and I've said the wrong thing.

Individual 5

Elements of internet use were noted as less helpful. One participant described having accessed the internet as a child in search for knowledge about their condition. This highlights the importance of thinking about how knowledge is accessed and shared, particularly with children.

I think it was overwhelming at the time, and also because no one else my age knew or just any age really knew what it was either […] so I think it felt quite isolating to know or find this information and not really know what to do with it either.

Individual 6

In addition, the use of social media can be difficult when confronted with the death of peers.

I've seen what's happened to other people and the age, I think has really impacted me in my head. I don't always admit to it, and I think I don't always like to acknowledge it, but you know, every time one of us drops off, there's an RIP notice on our pages and you think argh.

Individual 2

Yeah so there was, there's always things coming out about - this is why I try and stay off the Facebook groups - because it's like this one's passed away or that one's passed away really young.

Individual 4

Some people raised the idea that one can have too much knowledge though, and that this may be disabling in the level of parental protection.

> *Now I think back I think I am glad I had a relatively blissful childhood of not knowing. I just think of the children now and I just think, oh I know my mum would be like no, she can't do that because of this.*
>
> *Individual 5*

> *I do wonder whether there's sometimes, the whole social media thing, it kind of makes you think about things too much. So from our point of view as a family we just got on with things because they didn't know any better. Sometimes, maybe knowing too much maybe has a negative impact.*
>
> *Individual 3*

3.1.4 Theme Two: Illness Concept

There was variation across individuals regarding how they thought of their health condition. There was, however, a general concern about how one might be viewed by others regarding health status.

3.1.5 Subtheme One: Relationship to Condition

Some individuals spoke about how their parents had viewed their health condition, indicating that this may contribute to forming a relationship toward one's diagnosis. Levels of restriction in place may mediate perceptions of how the condition is impacting everyday life.

> *Whereas I've always kind of been like, this is just something that I have. So I'm really lucky in the sense that my parents were like that and let me do what I wanted to do instead of kind of holding me back.*
>
> *Individual 4*

You are extremely ill and we need to wrap you up in cotton wool until we can fix you.

<div align="right">

Individual 5
</div>

It was indicated that parents may be supported in how to think about this by the advice of medics.

I think it was, something that one of the haematologists always said to my mum was to remember that I was a child first, and that I just happened to have this blood problem.

<div align="right">

Individual 3
</div>

Some people spoke of initial difficulty understanding the diagnosis according to common conceptualisations of illness which their experience did not match, particularly understandings of illness as discrete with an ending or cure.

People telling you you are ill and you not feeling ill, or ill in the, the sort of typical sense that you would recognise.

<div align="right">

Individual 2
</div>

It's just, yeah that was my initial reaction, was to just let's just get rid of this thing.

<div align="right">

Individual 5
</div>

One participant captured the in-between space occupied by individuals. They highlight the difficulties experienced in trying to relate to other ideas of illness or other people experiencing illness.

> *Some of them are like recovered and we can kind of talk about it, but not really in any, there's some people who have more like terminal things, and obviously it's a weird thing because I'm in like an in-between part, so I can't relate to people who are terminal, but at the same time I can't relate to people who are complete cured of something.*
>
> *Individual 6*

Relationship to one's condition is not static but open to change. Participants spoke of how an initial mindset may become outdated when faced with the reality of physical limitations, particularly as one ages. There were aspects of emotional difficulty and grief attached to this.

> *So what I'm really struggling with at the moment is actually coming to terms with the fact that maybe I do need to slow down and knowing that I've spent 30 years thinking one way, which is I'm not gonna let it beat me, I'm gonna kind of just go fast, like live fast and hard if you like.*
>
> *Individual 4*

> *I'm sort of grieving because all of a sudden I had to stop my former life, […] I had to start thinking about everything that was wrong with me where I'd never really paid attention before, I kind of just ignored it all and if I wanted to go out I went out even if I felt awful.*
>
> *Individual 1*

3.1.6 Subtheme Two: Social Evaluation

Whilst there is a sense that both conditions are poorly understood, there was reference to negative social evaluations that might be attached to illness more generally, and concern about being viewed according to these.

I don't want to be one of these parents that always sleeps on the sofa and you have a child going into school going, but Mummy's really ill, she's asleep. I refuse to do that.

Individual 1

I push more, I push through because I don't want to be seen as like someone that's weak and that as someone who has those limitations.

Individual 4

This included concern regarding disclosure of diagnosis on new partner relationships.

Now I've got to try and find somebody else who is going to just be like, OK, yeah this is something that we can just deal with.

Individual 5

Individuals touched on how difficult it could be for others to understand FA/DBA raising ideas of visibility and stigma regarding illness. Whilst Individual 1 speaks of sharing as an adult, Individual 5 speaks of sharing as a child.

I do think that if I chopped my arm off, they might get it a bit more because they can see that there's physically something wrong.

Individual 1

And then […] a few years after I told my friends, you know I tried to explain it to them very simply like my blood doesn't work, but I think like a lot of people, I mean some people thought I was infectious and things like that, and so like some people would not go near me after I told that.

<div align="right">*Individual 5*</div>

The idea of discomfort arose in feeling that other people were avoidant of discussing aspects of FA/DBA openly. It was noted that people often respond with positivity, raising the idea that positivity is expected from others.

I did once [talk about worries regarding death]. But I don't like to. People are so like, they try and be positive about it.

<div align="right">*Individual 2*</div>

Discomfort and negative judgement were also noted to occur in interactions with medical clinicians too.

One of the first appointments I went to with my mum, I don't know whether this is very ethical, but the doctor said don't get pregnant because your children could get it.

<div align="right">*Individual 5*</div>

Like even now I still have doctors that will avoid what I'm asking or else they'll just put it very nicey and well that's not honest, I want an honest answer.

<div align="right">*Individual 6*</div>

In worrying about the financial consequences of disclosure, one individual touched on wider concerns about how illness may be penalised.

And I just thought, well, is it classed as a disability? Do I need to tell them? How much do I tell them before the interview? What do I tell them when I get the job? […] if we were ever to get a mortgage, would I need life insurance, would we be able to get a mortgage?

<div align="right">

Individual 5

</div>

3.2 PARENTS

3.2.1 Theme One: Knowledge

As with individuals, reference to knowledge and the lack thereof was prevalent throughout interviews with parents. The departure point is one of perspective with parents recounting the uncertainty of seeking a diagnosis for a child. Reflections from parents gave rise to ideas about both obstacles and supports in this period of waiting.

3.2.2 Subtheme One: Medical Uncertainty

All parents indicated how difficult the period of uncertainty was whilst waiting for a diagnosis, with indication that this impacts negatively upon parental well-being. Parents touched on how this challenged their usual expectations of medicine.

It was just really hard to cope with because on a day-to-day basis she's well […] but you've just got this underlying condition that potentially is incredibly serious. […] And, yeah, you want to find an answer and a solution, but you're not getting that, so it's still quite hard.

<div align="right">

Parent 5

</div>

And when asked about the life expectancy and she said reasonable and I thought, what does that mean? And it just sort of plunged me into a bit of a spiral of sadness, guilt.

Parent 2

And I think that the hardest part, not being dramatic, but the hardest part is that the doctors didn't know what was wrong with him and it was like they're going oh, I don't know what it is and you're like, well, you know, you're meant to know this.

Parent 9

Whilst some parents indicated that it would be difficult to envision any helpful support in this period of uncertainty, there was an indication that this was not helped by delays in medical communication.

But it was still there, and it wasn't until the October that we got the diagnosis and we found, like people were very reluctant to answer any questions.

Parent 10

So you know, they [symptoms] were pointing in the direction of Diamond Blackfan, but still nobody had said Diamond Blackfan to us.

Parent 6

At the same time, one parent who had been offered differential diagnoses indicated that this was not altogether helpful either. Another spoke of being reassured by medics even without information indicating perhaps a need to be guided by parental preference.

I can remember her talking me through both the possibilities in quite excessive detail […]it was really upsetting. And then after I thought why did she do that when she wasn't able to say it definitely was that?

<div align="right">

Parent 10
</div>

And then he said, you know, don't worry, don't get worried about anything, don't go Googling anything, and the way he put our minds at ease was fantastic.

<div align="right">

Parent 3
</div>

There were ideas that structural issues within medical systems may impede access to information and diagnosis. This included issues around how expert knowledge may be distributed geographically, as well as internal politics around referrals and interprofessional communication.

And we were asking for a referral so we could just go and see this specialist directly, but they were really resistant to it, they really didn't want that to happen. And we've subsequently learned [from specialist] that this is a theme that quite a lot of smaller hospitals are reluctant to let go of these unusual rare cases because they're interesting.

<div align="right">

Parent 6
</div>

Why did it take another mum telling me about [specialist Centre] for example, why didn't you know, she [local consultant] could have asked who's the expert in this? Like just I felt that she was a barrier really and the lack of lack of knowledge down here [region].

<div align="right">

Parent 10
</div>

One parent highlighted further structural issues when seeking support for a child with complex and additional needs.

So that was always the thing, because he was too complex and a lot of the systems we came up against we always broke because he was so complex.

Parent 4

In contrast it appears that where medical knowledge does exist, access to information and referrals for additional supports are prompt, mediating the sense of uncertainty. This was the exception for this sample of parents.

I've been really lucky I think with the support we've had and even to the point where our doctor referred us to Click Sergeant straight away.

Parent 8

I do think our experience has been maybe exceptional in respect of if we've needed something or wanted something, it's either already been - we've had information about it, or it's been very straightforward to find that information.

Parent 5

And whilst there was consensus that diagnosis brought with it some relief, the relative lack of knowledge means that uncertainty continues with parents initially feeling overwhelmed.

Yeah, I think for us or for me, in some ways it was a relief because obviously we had a diagnosis, and we then knew what we were dealing with.

Parent 2

And the implications just felt huge and overwhelming and that we just were not up to that challenge. It's like being asked to do something without any preparation.

Parent 7

One parent reflected on the knowledge acquired in finding a specialist dietician. This highlights that following diagnosis medical uncertainty continues to present an obstacle in accessing effective care for children.

> *And you have the dietician not knowing his condition, not knowing that he can't like, his tummy doesn't absorb things as quickly as a normal child and all those little things.*
>
> *Parent 9*

3.2.3 Subtheme Two: Questioning Knowledge

In discussing medical uncertainty, a second theme arose pertaining to how parents began to question sources of knowledge, particularly whilst having to make decisions about medical treatment.

> *And then also what we were finding was two different doctors were telling us completely different things. I'm like well where's the rule book here? Where's what we follow? And it's like, no, it's your decision, you need to make it. And I was like I don't know. And it's learning that.*
>
> *Parent 9*

This appears to coincide with parents seeking out and becoming aware of other sources of information. Charities specific to either condition were cited as a main source of knowledge, support, and hope, particularly in being able to connect with other parents caring for children with FA/DBA.

> *It was amazing and to have them kind of guide us through the medical side of things and what to ask for, on top of the emotional side of things was very helpful. And just being able to have the forum where you can ask a question without any judgment and know, you know, if someone knows they're going to tell you.*
>
> *Parent 2*

It was really nice to meet other parents and to see teenagers and adults with Fanconi and kind of, just be able to see that this, you know wasn't the death sentence I thought it was.

<div align="right">

Parent 8

</div>

One parent spoke of the practical support and knowledge shared by other parents, indicating that this support is not always clearly labelled or offered elsewhere.

I had one parent who helped me with my Disability Living Allowance application because, you know, someone says oh, have you had this? And you're like oh, I don't know what that is. And I'm paying hundreds of pounds in car parking fees every week.

<div align="right">

Parent 9

</div>

In growing awareness of other knowledge sources, parents appeared to grow more confident at challenging medical opinion and advocating for their child, which many regarded as necessary.

And they've also, especially [play therapist] she's the one who said to me you can advocate for her. And nobody'd kind of told me that […] how many goes at doing a cannula is reasonable before you can say no, or you can say I want to take a break.

<div align="right">

Parent 10

</div>

And I firmly believe that now, that it is OK to say to doctor, I don't think that's right. And because it's, you know before this you don't know any of that, and you just think they're God and what they say is given.

<div align="right">

Parent 9

</div>

There was some indication that this questioning leads to an increased confidence in handling and choosing which information to be guided by, and a sense of placing the child before the medical statistics.

> *I think we were led by her, a lot actually. You know she's growing up a very spirited, very happy, funny person and continues to do so […] and that's the thing, is that for her it's not a tragedy, for her it's an inconvenience.*
>
> > *Parent 6*

> *I don't want to look at things like life expectancy statistics and all of that, I just think that's out there, but I don't need to know that, and that's not her, and her journey will be whatever it turns out to be.*
>
> > *Parent 10*

3.2.4 Theme Two: The Impact of Social Responses

Parents repeatedly touched on interaction socially and with wider systems including friends, work, and school. It appears that social responses regarding news of the diagnosis or to care needs may impact upon parental wellbeing.

3.2.5 Subtheme One: Losses & Gains

Parents spoke of both losing and gaining friendships across the process of receiving and sharing a diagnosis. While many parents were clear that the support of friends had been 'amazing' (Parent 8), parents also indicated a sense of their social world becoming smaller indicating feelings of isolation.

> *You know your whole social circle, everything just contracts and your world becomes very small, very quickly.*
>
> > *Parent 9*

So they've been very supportive in those ways but yeah, I think it becomes a bit of a barrier to relationships beyond, like, with wider family and with friends in some ways because I think it's difficult for people to empathise.

Parent 2

There was reference to social processes at play in sharing a diagnosis, with people becoming uncomfortable or unsure what to say. There is also, however, a sense of friends and supports gained, with parents referencing the charities and links to other parents.

People that we have hardly known have become incredibly close and advocates for [child] and are in touch constantly and people we've known our whole lives are barely in touch at all […] you do start to see these kind of communication flaws in people when, when things become a bit real and a bit uncomfortable […] and it becomes, I think, challenging when something like this happens and people don't really know what to say.

Parent 7

Again, with Fanconi Hope there are people within the UK that have become my friends. And so, they know when I need to moan about something that another friend won't understand, they would 100% get where I'm coming from.

Parent 8

In line with the above, other accounts highlight a role for peer support toward improving well-being.

Maybe buddying up with somebody who has been through it fairly recently […] because sometimes you just need to talk to somebody who's in the same sort of boat as you.

Parent 1

3.2.6 Subtheme Two: Social Judgements

Parents noted that social interactions may involve a sense of judgement or evaluation regarding health status.

There were lots of you know, people kind of asking is it a good idea to have another baby, you know that it's definitely not going to have DBA?

Parent 2

And she said to me I'm really sorry to hear about [child] […] And she said, oh I just thank God my boys are healthy.

Parent 8

Judgements were encountered by wider systems in place too. Parents of a child with disabilities and additional needs expressed concerns regarding how difference and disability are viewed by education and care systems.

We kind of fought with them [nursery] about that, saying well this is discrimination and basically, they spoke to their solicitors and then they came back with a list of reasons why they couldn't look after [child] anymore.

Parent 4

This same parent noted how these care structures may overlook disability and bar access.

So we asked them about well could we do remote appointments? They said, well, the way the [NHS] funding system works, there has to be someone here face to face […] So he said we'll try and fix it, but now COVID has transformed that.

Parent 4

There were also concerns about how disability or difference might be judged in terms of a child's future.

> *And you think about like, if I think about secondary school and two boys walk down the corridor and ping his ears or something.*
>
> *Parent 9*

It was indicated that the school system could be helpful or unhelpful depending on how they engaged with the child's care needs.

> *And sort of they put into place care plans and little things for the teachers to know like oh if this happens, let mum know.*
>
> *Parent 8*

A parent touched on the inflexibility of standard school measures applied to those with more complex situations.

> *I suppose one thing that annoyed me somewhat was on their end of year report, it said attendance, you know needs improvement.* And I thought I wish my child could attend, I wish my child didn't have to go to hospital and miss days of school.
>
> *Parent 3*

The response of work regarding care needs was also reported to be a factor which could impact parental stress and well-being.

> *They were as accommodating as possible. They made sure that I always had at least one full day off on my timetable so that I could go to the hospital.*
>
> *Parent 10*

So at that point I thought no, I'm not going to let her do that because she [line manager] was putting me under so much stress I thought I'm gonna end up being ill because of what she's doing so I can't, I'm not having that.

Parent 1

3.3 Joint Themes

3.3.1 Theme One: Family Dynamics

Both individuals and parents reflected on family relationships and dynamics in the context of living with FA or DBA. Although there was variation, responses indicated common patterns and concerns arising across the families represented.

3.3.2 Subtheme One: Parental Relationship Strain

Every parent interviewed reflected on the initial strain accessing a diagnosis placed on their relationship. For most there was a sense that both individuals were initially on different pages regarding diagnosis and thereby the pace at which they came to terms with it. In all but one of the interviews mothers recalled feeling sure the diagnosis would be made and began researching accordingly, whilst fathers typically preferred to wait on the official outcome. For many couples this posed an obstacle in feeling understood and supported.

He's always been far more of the let's just wait and see what the doctors say and then deal with what they tell us, whereas I'm far more of the proactive rather than reactive let's just go and research everything about everything […] There have been a lot of arguments, there's been a lot of arguments.

Parent 8

Some couples indicated that it was difficult to process or grieve whilst the uncertainty continued, and whilst their partner had not yet reached the same conclusion.

But then my partner was on the other side and he was like no, the genetic doctor said it might not be it and at that point it almost made me worse because I was, I couldn't grieve because I didn't know if he had it or not.

Parent 9

Because I was, we were in different places erm because I was kind of, I knew what she was going to come back and say. And none of them were any, they were all terrible and he was going you're just being a pessimist, you shouldn't be pessimistic about these things. This is probably fine. And I'm going, I'm kind of like really stressed and grieving, and he's going, I don't understand why you're being so, sort of negative about things.

Parent 5

There was an indication then that both individuals ended up moving through stages at different paces, with one parent noting how isolating this felt.

I was already in the process of thinking OK, researching, and getting my head around it, whereas he was in very much a limbo situation of thinking well, it might not be.

Parent 8

And I think like for me I felt very isolated with the whole thing. I felt like my experience of it was very different and that my husband didn't really understand that experience, he wasn't sharing the same experience, he wasn't really talking about any of it. So I think it definitely put a strain on us.

Parent 10

Parents also reflected on different coping styles and needs. This typically centred around the need of one member to talk whilst the other was less inclined to.

Yeah, I think it's at times put a bit of strain on things. My husband isn't, I think, like a lot of men doesn't openly talk about things and wants answers, wants it's sorted.

<div align="right">*Parent 5*</div>

Relational dynamics were noted to change in role divisions that evolved around caring for their child, with mums often reducing their hours or giving up work completely. One parent summed this up noting both the financial impact and that on well-being.

You know, in terms of finance and stuff like that too, I had to quit work. So then going down to one income was really difficult. I think also my mental health, just you know, having not been able to go out and work and do the job that I really loved.

<div align="right">*Parent 8*</div>

Parents reflected on structural elements that contribute to this role division happening in quite a gendered way, touching on underlying social expectations and assumptions. Parent 10 reflected on how this leads to a distribution of knowledge that maintains the division.

My partner, she got really frustrated with the lack of support from the council, and saying well I don't want to have to give up work and their attitude was, but why not, you're his mother? So kind of very old fashioned.

<div align="right">*Parent 4*</div>

I think partly it just sort of evolved in some ways, because obviously when she was a baby and there were all the appointments I was on maternity leave, so obviously I was going to take her to all of those, and there was that pressure you know for my husband that he couldn't necessarily take time off work. So obviously I [...] I've got a sort of knowledge and experience he doesn't have therefore he's kind of happy to let me have that and kind of run the show. Whereas sometimes I would like some of the pressure taken off.

Parent 10

For many a joint focus or action mediated some of these tensions, with parents highlighting the unity found in researching and making plans. Some parents also reflected on having to reconnect with their partner as part of the process of adjusting.

We need all the information, and we need the best treatments and all this. So then we kind of became a team working together and that really bonded us.

Parent 8

There were various things that we were able to do together that kind of brought us, you know it reminded us of all the good things in life, and what we share, the passions that we share.

Parent 6

3.3.3 Subtheme Two: Protective Communication Patterns

A repeated pattern concerned family not talking about the condition, which appeared to be a protective stance. Some touched on an idea of this being a learned coping method.

You know we just need to help her cope with this, because obviously she's not showing any of it outside, but it's also like me, internalising it.

Parent 5

Sometimes I talk about things sometimes I don't. Because I was so ill as a child and in and out of hospital so much I'm very used to being on my own and processing things on my own.

<div align="right">

Individual 1

</div>

Others indicated that not talking was employed to protect family members from upset or worry.

And I didn't want them to worry about me, 'cause I knew they were worried about me, 'cause obviously if I went to my appointment, my dad would get a bit teary afterwards […] so I didn't really want to bring it up.

<div align="right">

Individual 6

</div>

We didn't tell my sister because she was pregnant. I didn't want to worry her about it.

<div align="right">

Individual 5

</div>

There was also a sense that individuals were sometimes treated as younger by family members who are protective, and conversely that independence was something parents must navigate.

Well, I'll tell you that my brother and sister, they're younger than me, but they're are a lot more protective over me then I think they would be normally.

<div align="right">

Individual 4

</div>

I think moving out from home is a good step for me, because I feel like my mum babies me, you know like treats me like I'm younger […] But I think it's to do with the fact that my mum spent a lot of time in hospital with me, my dad not so much and he treats me fine, it's not really an issue. But my mum treats me in a certain way, and I guess she's not changed in the ways that she treats me since I was a kid.

Individual 6

Because then I was married, they feel comfortable enough to kind of let go of the apron strings a bit, you know and then be like yeah, she's fine now.

Individual 5

3.3.4 Subtheme Three: Concerns Regarding Siblings

Participants reflected on their relationship to siblings of individuals diagnosed with FA/DBA, touching on concerns around sibling well-being, and the possibility of resentment. It is worth documenting that parents and siblings spoke of supportive and positive sibling relationships too, but that concerns exist, and families feel it important to think about these.

We have a lot of issues with our older daughter as in, so if [individual] gets a reward toy for having blood taken, [sibling] feels that she should have something.

Parent 8

And then my sister went through the genetic testing to see whether she was a match for the bone marrow transplant, and she wasn't, so that was massive. That was awful for her because she felt like she couldn't fix it.

Individual 5

Parents reflected on the difficulty of juggling the needs of multiple children, particularly during periods of treatment.

I was basically in hospital all the time with [child] and then coming home at, most weekends I tried to get away for a day for one night and then we would go straight back up again. So not great, and I think yeah, [other child's] and my relationship's never been the same since. Yeah it was sad, it makes me sad.

Parent 5

I mean we do have to devote an awful lot of time to [individual]. Which means [sibling] has lost out.

Parent 4

There were also concerns expressed regarding the relationship between siblings.

But I've always been aware that I don't want him to feel resentful of her, or to feel that she is more important than him, so I've always been very conscious of making sure I'm giving him time, quality time in doing nice things with him.

Parent 10

She said that at times she felt [Individual] was more important, and I said, well, her needs sometimes are more important, but she's not more important. You both are important, and it was just, there were times that we had to go for medical treatment, she had to have this done.

Parent 5

My youngest sister – I have quite a complex relationship with her. I think she feels. Erm, she has quite bad anxiety and she blames that on my mum not giving her enough attention.

Individual 1

3.3.5 Theme Two: Mental Health Care Necessary but Inadequate

Across the interviews there was a clear need and demand for support around well-being and mental health. The previous themes outlined will doubtless already have highlighted how well-being is impacted, nonetheless participants spoke explicitly of mental health and an overview is offered here. Emphasis is placed here on noting that the researcher presents this theme not to suggest medicalising or therapising the experience of all parents, but to highlight the possible impact on mental health. This will be discussed further in the Discussion chapter.

3.3.6 Subtheme One: Impacted Across Family

In total 6 of 9 parents and 4 of 6 individuals interviewed had sought out some form of psychological support. For parents, emotional difficulties relating to mental health were experienced from the outset of seeking a diagnosis.

> *I was really struggling, and I had a breakdown, a full breakdown when we had [child's] diagnosis and I couldn't do anything. I had to finish work for a period of time and there was no support at all.*
>
> *Parent 9*

> *I don't think it's a very of the moment way of putting it, but I definitely had a breakdown. I lost myself. And I wouldn't say I was ever suicidal, but I didn't want to be here.*
>
> *Parent 6*

> *It all felt so difficult and traumatic, I'm trying to think what even kept me going at that point. […] He wouldn't sleep at all. He basically cried 24/7. Nothing you could do would comfort him he was just deeply distressed at all times.*
>
> *Parent 2*

For many parents, their newborn was removed following or soon after birth prompting worry and concern around attachment and bonding. Infancy is remembered as a stressful time.

> *And when I think of [child's] babyhood, it's just this barrage of appointments and stress and worry, and I find it really hard to remember the nice things I did with her.*
>
> *Parent 10*

> *He was born by emergency C-section 'cause his heart rate kept dropping and when they lifted him up […] he just looked so poorly he was completely grey, he wasn't really doing anything and that's when they checked his haemoglobin and whisked him off at to neonatal and he was there for 12 days.*
>
> *Parent 9*

Parents also reflected on the emotional difficulty of supporting the child's growing understanding of the condition.

> *And it just kind of made me think, oh my God, she thinks she's going to, that it's not forever. And that's really hard thinking at some point she will realise it is.*
>
> *Parent 10*

Individuals reflected on mental health growing up. For some, difficulties first arose around peer relationships and negative self-evaluation.

And then teenage years, I didn't know why I was so tired all the time obviously then. And PE - I hated PE because I hated the pressure of you've got to race. I hated it so I always took the, I was always a bit of a joker, and they said, oh, you can be the ref 'cause you're the lazy one. And I just kind of went yeah yeah yeah I'm the lazy one.

Individual 5

Things I'd normally get excited about like getting a pizza, simple things […] I'd just feel nothing, and I'd feel very neutral all the time, so I wasn't happy but I wasn't sad so I was in this kind of in-between state for a long time. I think my friendships at the time didn't help it because I was being left out a lot 'cause people […] they just kind of assumed that I was too tired to do anything.

Individual 6

Individuals noted distress and difficulty later in adulthood too, with one reflecting on illness and work as attached to worth.

The nine months I spent off work […] I kind of thought well if I'm not doing, if I'm not working what use am I? What can I do?

Individual 5

In the middle of last year I actually had a mental breakdown and I have been diagnosed with OCD, anxiety, and depression.

Individual 4

3.3.7 Subtheme Two: Unequal Access

The majority of parents and individuals reported having encountered difficulty in accessing mental health support.

> But I think in terms of what would have been helpful, I was shouting from day one […] I said OK where's the psychological support? What do we have on offer to us? Only because luckily, I know to ask and I know what to ask for. But even based on that there was nothing, absolutely nothing.
>
> *Parent 7*

> And it was at that point I realised like this is a massive thing, I've never been offered - apart from when we went to the IVF appointment - I've never been offered any psychological support, nothing like that. I don't know whether that was because I was diagnosed as an adult and it's kind of you have to get on with it.
>
> *Individual 5*

Whilst this was so, there were a couple of families who had not encountered this difficulty indicating some inequality of access to psychological support. Accounts suggest this is perhaps mediated by knowledge and geography.

> And, ah, the paediatric psychology service at [Hospital] got in touch with us and said this is what we can offer [individual], this is what we've got for you, this is what we can offer for any siblings.
>
> *Parent 5*

> And because we're so far, we were 100 miles away at home, so trying to arrange stuff I think would have been really difficult anyway.
>
> *Parent 8*

One parent indicated that stigma rather than structural issues may also present a barrier to accessing support, particularly when one must seek rather than be offered support.

You sort of don't get that question about how are you? And actually, are you comfortable enough to say, well actually I'm not coping because then are you seen to be failing in looking after your child and you sort of have that thing that you've got to be, look like you're strong and look like you're coping otherwise are you putting your child into jeopardy?

Parent 9

3.3.8 Subtheme Three: One Size Does Not Fit All

When individuals did manage to access support, there were mixed feelings about how this was received indicating that an element of choice may be necessary. For many individuals the standard NHS offer was deemed unsuitable or lacking in relevance. There was also an idea that physical health is regarded as separate from mental health.

I went to my GP a couple of times and I didn't find him very helpful really. He sort of said oh you need to sit down with a cup of tea, have piece of cake kind of thing. And then at one point he said we can look at antidepressants if you want. And I said I want to talk to somebody. And then he said, oh there's a couple of self-referral things you can do. So I did that, and that was like waiting for months. […] And when I went to it, it was like a kind of, I don't know, anxiety by numbers thing. I was given a booklet […] and it was things like, you know, write your worries down, put them in a box and then open the box at 7 o'clock each evening.[...] So I phoned the provider of the thing and I said look, I need some help, my baby is really poorly, I don't know if she's going to be OK, I need some proper counselling about this I don't need a booklet to fill in.

Parent 10

I had CAMHS and to be honest it really wasn't relevant for stuff, like medical stuff. It was kind of like, they would talk about friendships a lot, I guess because that was kind of the problem at the time, but they ignore the whole health side of things […] they were just treating it like any other kid who had low mood. And it's like well you need to consider that I'm not just going through a rough time at school and hormones, there's a lot more to it but they kind of just ignored that part.

<div align="right">

Individual 6

</div>

I think it was just having somebody that didn't know who I was, didn't know my family, didn't know anything about DBA actually. Just having someone different to talk to and have them say yes, your feelings are valid. That was enough.

<div align="right">

Individual 5

</div>

There was a sense that the support offered could be inflexible or limited.

Yeah, and it was kind of like even though I was feeling depressed, the sheet didn't say I was depressed so we didn't do that.

<div align="right">

Individual 4

</div>

There was also a feeling that support is often reactive rather than being provided in advance of difficulty or distress.

Why *couldn't you teach me to deal with it earlier? 'Cause that would have been a lot easier for my family as well.*

<div align="right">

Individual 1

</div>

I was asking him what support have you got for the children? And he said what do you mean, nothing. I said I want some coping strategies for me, but also for my children. […] You know if we know something's very likely to happen, why don't we give some coping strategies upfront rather than trying to deal with it afterwards?

Parent 9

Individuals also spoke about the lack of mental health talk in their medical care.

Also I think, I at the time, like I had no idea that maybe my DBA was causing any problems mentally, erm so it would have been helpful just to have that option there and the consultant, even though he's obviously medically trained, to maybe have known, maybe have spoke about it as well.

Individual 4

Common elements considered helpful were being able to have an ongoing relationship to a supporting person, which was most often achieved only in having sought out private therapy, with one couple introducing the aspect of privilege.

I got really anxious and stressed again, and I thought right, I'm gonna go see her now, I'm not gonna wait till it gets worse. And I just went for that, almost like a little one off, and that was enough to kind of remind me of strategies and things. And I know I would go to her again if I felt I needed to.

Parent 10

There's not words for the trauma, but we're so aware now, particularly in hindsight, of our privilege. There were modest funds for some private therapy. There was a big family home that we could move into. And it's left me thinking a lot about what is there really for people that don't have the advantages that we do because it was about enough for us.

Parent 7

80

There was also a desire for specialised knowledge, though not necessarily condition specific.

It would have been nice I guess to have someone that had worked with people with long term illness to be able to talk to about it and understand.

Individual 4

And I think the issues and the fears are probably universal whatever the condition. […] And actually, I think locally they could set up support groups for people with children with rare conditions. And it wouldn't matter that you were talking to somebody whose child had a different condition to your own, because you'd be empathising and understanding.

Parent 10

CHAPTER FOUR: DISCUSSION

This research sought to explore the psychological impact on a family system of living with FA or DBA, which was identified as a gap in the UK literature base. Based on systemic theory, the research considered it important to investigate the impact on both the individual and their family. By psychological impact the research was interested in all areas pertaining to mental health and well-being. It also hoped to better understand any demand for mental health support, as well as how current provision is experienced.

Presented here are the main findings situated within a broader discussion of current relevant literature, and which will be used to address the research questions posed at the outset. A critical appraisal will be offered to acknowledge the limits of this research. Finally, consideration is given to the possible implications and applications of these findings.

4.1 Main Findings

4.1.1 Research Question 1: What is the psychological impact on a family system of living with FA/DBA?

For clarity, findings will be discussed as presented in the results, with reference to individuals first, followed by parents, and then by a consideration of the whole family with reference to interviews from both.

Individuals: The first theme presented for individuals pertained to knowledge; this touched on lack of knowledge, acquisition of knowledge, and the impact of social knowledge, or lack of. Individuals touched on complex ways in which knowledge may impact on well-being. Medical uncertainty and lack of knowledge were

associated with feelings of pressure and stress for many. Regarding medical uncertainty some individuals diagnosed as adults expressed feelings of anger or frustration that their symptoms had been missed in childhood. This is perhaps indicative of having had to navigate an adjustment phase on receiving a diagnosis as reported elsewhere in the literature on chronic illness (Stanton et al., 2007). Lack of knowledge meant that individuals sometimes felt unsupported through this phase, and in some cases reported feeling pressure to guide their own medical treatment. Motyer et al. (2020) has written of the importance of ensuring access to adequate information if people are to be expected and enabled to collaborate in their medical care. In this case the rarity of both presentations and the resultant lack of knowledge necessitates a level of agency to source information not usually required in medical care, and therein stress and worry experienced by individuals. Recalling Alexander's (1981) application of the Double Bind (Bateson, 1956) to health conditions, it is clear that the necessity of having to lead on one's healthcare, particularly without adequate information, may well exacerbate the distress experienced when individuals are then expected to inhabit a passive position when receiving care.

Individuals spoke of living with uncertainty regarding their prognosis. Some individuals spoke of reaching an age where they felt they were waiting for cancer or other complications associated with FA/DBA, raising ongoing awareness of mortality. In line with other literature, we can extrapolate that this will invariably mean periods of sustained stress, worry, or low mood experienced in living with the uncertainty of chronic illness, with a potential impact on overall quality of life (Fortier et al., 2013; Maikranz et al., 2007). Awareness of FA/DBA also impacted on life decisions, including career considerations and the decision not to have children, both significant choices shaped by living with FA/DBA.

Regarding knowledge acquisition, there was a clear idea that being involved in appointments helped individuals to feel informed and avoided worry that things were being kept secret. This speaks to other literature which documents the importance of

providing children with sufficient information to offset associations with depression and anxiety (Last et al., 1996). On the other hand, issues around the internet and instant unfiltered access to information raise important considerations around how to navigate and satisfy a child's growing awareness without this being overwhelming or frightening, as described by some. This indicates a need for ongoing reflection and flexibility regarding the information sharing process navigated by medics and parents (Bahrami et al., 2017). Individual 6 shared recalling a desire to have a separate space to discuss with medics separately from parents. This indicates the need for developmental considerations individualised to each person. It is clear that consideration of these issues is necessary to better understand and maintain emotional well-being in sharing information and understanding between children, parents and clinicians. These findings recall systemic writings on childhood chronic health conditions (Rolland, 1984; Eiser, 1986, 1983), in which developmental perspectives are used to consider the nuances of age and developmental stage of both individuals and the family generally. These provide useful theoretical lenses through which to consider family transitions and well-being in the context of health. A promising bridge between research and practice might involve using such theories to inform practice in supporting families to navigate these changes.

The second theme outlined was illness conceptualisations. Again here, individuals indicated a link with emotional well-being. Individuals indicated that how parents were perceived to respond to FA/DBA was linked with how one views oneself as an ill person with limitations or otherwise. Parental restriction has elsewhere been linked with negative adjustment to life with chronic illness (Swift et al., 1967). It must be stressed that no blame is directed at parents here, particularly given the lack of knowledge many face on receiving a diagnosis. Rather this link is made to highlight the complex ways in which lack of information and support for parents early on may impact on a child's sense of self. It highlights the importance of supporting parents well at this early stage.

There were nuanced issues raised around being perceived as ill, and an indication

that stigma or negative judgement may be associated with this. Several people touched on the disjuncture between illness as generally understood and their experience, which did not match up. This raises ideas around how one makes sense of their diagnosis amidst social interactions which may position one as ill according to common conceptualisations of illness, which may not feel fitting for the individual. Whilst those diagnosed as children grew up aware of their condition, most described a process of growing awareness as marked by questions around prognosis and death. This highlights the ongoing process of making sense of one's diagnosis and the potential distress associated with increasing understanding. This may be complicated further in parental or clinician decisions to omit information considered developmentally inappropriate. As was the case for some individuals interviewed, their knowledge was sourced independently via the internet which was experienced as overwhelming in volume and content. This combined with patterns of protective not talking, mean that children may navigate meaning making alone, with the potential for increased worry and distress.

There was also a sense of difficulty encountered in seeking identification with others. Those living with FA/DBA occupy an in-between space, with no access to a cure, but also not considered terminally ill. Individuals described difficulty in being understood socially and sometimes experienced a sense of isolation or difference because of this. The developmental literature indicates the importance of peer identification for well-being (Jetten et al., 2017). For many this identification was eventually found in meeting peers via charities that bring individuals together. This should highlight the importance of the work of charitable organisations in supporting well-being.

Accounts also indicate that periods of adjustment in one's relationship to diagnosis may occur across the lifespan. Some individuals described an initial mindset of not letting their diagnosis stop them. They described emotional difficulty and grief when these mindsets grew outdated when faced with age and a greater physical impact.

This indicates that support needs may change with time.

In the social evaluation sub-theme individuals referenced experiences pertaining to stigma and negative social evaluation. There were recollections of classroom bullying with fears of contagion referenced by one person, and attributions of laziness referenced by another. One adult described how their health status has been discredited or questioned by some on account of its being invisible. It is clear that social perceptions and actions may negatively impact self-esteem. Research has also linked perceptions of illness stigma as negatively impacting on depressive symptoms and social belongingness (Gamwell et al., 2018). One individual expressed association of weakness and limitations to being considered ill, suggesting elements of internalised stigma. Another individual touched on worry regarding how disclosure might impact on relationships, given the associated infertility and life expectancy of FA/DBA. There were also some concerns regarding the repercussions of disclosure as an adult, in terms of financial impact in access to jobs, and around mortgages or travel insurance. These concerns speak to wider issues regarding illness stigma and the exclusion that can be enacted in the interactions and structures of neoliberalism (Chamaz, 2020]. It is timely to consider not only how individuals may be supported in navigating this stigma, but on a wider level to begin addressing this stigma directly.

In returning to the systemic frame of the research, it is useful to consider how the findings here are supported by the systemic literature base, and in turn add support to the ongoing relevance of this literature. The findings for individuals highlight well the reciprocal influence of relationships on health conditions and vice versa as theorised in systemic writings (Stiell et al., 2007). In this case how parents view either condition impacts on how the child experiences life growing up, and eventually perhaps on how they view their health too. On a larger scale, social views of illness and subsequent evaluations understandably impact on how individuals may be positioned socially and thereby directly impacts on their experience as an individual living with a life-long and life limiting condition.

Furthermore, the findings from individuals speak to the ongoing and lifelong nature of adjustments in living with a health condition. It is proposed here that the Family Illness Model (Rolland, 1984) is a useful framework for applied use in supporting families to understand and navigate such adjustments.

Parents: Parents indicated an impact on emotional well-being and mental health too. The first theme outlined for parents also concerned knowledge. All parents described how difficult the initial period was of waiting for a diagnosis, with descriptions of grief and pain on receiving a diagnosis, and a sense of trauma attached to these memories. More than one parent referenced having experienced a 'breakdown' following diagnosis, indicating a direct and significant impact on mental health. For some parents their child had been ill following birth and removed for specialist care, whilst for others their child had been relatively well until diagnosis later in childhood. Parents described experiencing worry, for some about bonding early on, and for many about how diagnosis and medical regimes would impact on relationships with the child who had been diagnosed and other children in the family too. Research has linked maternal resolution of a child's diagnosis with secure attachment outcomes (Barnett et al., 2006). Crucially these findings suggest that with time family adjustment and attachment styles can improve where they have become difficult following diagnosis. In addition, Howe (2006) indicates an interplay between parental state of mind regarding a child's disability and attachment style. Research thus indicates benefit in supporting parents in this early period to help sustain well-being and maintain positive attachment relationships.

In line with other research (Motyer et al., 2020), parents also indicated that difficulties of this time may be mediated somewhat by access to information. It is clear that this varied across parents with one indicating that excessive information prior to diagnosis had been upsetting and unhelpful. This indicates a need for clinicians to be led by parents. Nonetheless, this supports that provision of early support could be considered preventative in addressing parental worries and distress and thereby protecting parent-child relationships within the family.

Parents indicated further stress and frustration experienced in structural issues encountered in accessing support for their child, with many having to exercise agency in seeking answers and information. Some parents recounted issues accessing referrals on account of internal politics, with clinician interest in rare cases an obstacle in transferring their child's care to a specialist team. Another parent had to seek out a specialist on recommendation from another parent, not from her assigned medical lead. These are clear instances in which structures delay care provision and exacerbate the stress felt by parents trying to navigate their way through. Some parents indicate that this continues beyond diagnosis, with one recounting the difference finding a dietician with specialist knowledge made to the care of their child. Granted these conditions may be rare, but with internet access successfully guiding parents to relevant knowledge and support, it seems the medical world is lagging behind.

The subtheme of questioning knowledge moved beyond the impact of diagnosis to offer insights into factors that may be helpful in supporting parental well-being. In seeking out other sources of knowledge, the parents interviewed found charities offering access to information and communities of parents also caring for children with FA/DBA. As explored in other literature, this experience allowed parents to find understanding, information, and hope in these connections (Hartman et al., 1992). This indicates the role of peer support in the maintenance of emotional well-being. Indeed, this is further supported in the social losses and gains parents spoke of, indicating how isolative this early period could feel. Whilst social support has been indicated as beneficial to the well-being of families (Armstrong et al., 2005), it is perhaps timely to question why parents must source these connections independently and how services could be working to make and promote better links.

Moreover, some parents indicated levels of social judgement encountered which may worsen feelings of isolation or exclusion. From judgement regarding family planning choices, to well-meant but painful comments from other parents, distress may be encountered in an ongoing way beyond diagnosis. Indeed, parents spoke of

the complexities of sharing this news socially, on account of these responses. This may be exacerbated by the likelihood that hearers will not be familiar with the diagnosis and may disqualify concerns based on common conceptions of anaemia.

One parent shared the difficulties encountered in seeking support for a child with physical disabilities and complex needs. Noting a nursery who refused to care for their child, as well as NHS appointments which could not be offered virtually, there are clear structural access issues faced by those with disabilities thereby exacerbating stress. This may indicate differing levels of need and difficulty for families according to a wide range of intersecting factors. Indeed, other parents touched on geographical distance to specialist services as mediating factors in accessing support. Reminded of Crenshaw's (1989) work on intersectionality, it is crucial that research begin to consider these factors so that provision may actually and accurately meet need. It is also interesting to reflect on how COVID-19 will likely have improved virtual access for many denied it until now. Perhaps the bigger issue here is not creativity of thought but will. When required, the will of those in power was enough to make a change that has benefited many families who can now access services online. This should signal a clear need to rethink and update the status quo in how healthcare is presently delivered, particularly where access appears unequal.

These findings further support the value of the systemic literature base. Accounts from parents touch on the reciprocity of relationships and health as well, but highlight another perspective in how powerful the connection to positive supports can be. These findings also touch on issues of access and equality as conceptualised by Social Ecological Theory (Bronfenbrenner, 1979). The findings suggest that access to support is largely unequal with complex and multifaceted mediating factors. It is perhaps timely to consider the utility of such theoretical frameworks not as a mere abstraction, but as a tool to support in conceptualising issues of access and justice, and thereby as a tool to facilitate better service provision and design.

Family: The above sections have attempted to link the results with research question one regarding the psychological impact of FA/DBA. In these sections the focus, although socially mediated, has been on the individual or the parent. The third theme outlined related more to family and elements of psychological impact that were more relational in nature.

Strain on the parental relationship was experienced to some degree across all parents interviewed. Whilst the wider research on childhood illness supports this picture of relational strain (Silva-Rodrigues et al., 2016), here in this research is a sample of parents who experienced this strain and survived the impact. In the spirit of the systemic frame, perhaps this is a better area of focus in terms of understanding what has gone well. Parents touched on this, indicating that feeling united in their efforts to access and provide the best care was helpful, as well as renewing and remembering former shared interests. Helgeson et al. (2018) have reported that a shared appraisal of illness and feeling in collaboration in managing illness is linked with adaptive adjustment. This offers room to consider how parents may be supported to draw on the strengths of their relationship in this difficult time.

Whilst the results indicated a clear tendency for parents to be on different pages regarding diagnosis initially, leading to feelings of isolation, there was also an idea around both partners coping differently or having different coping needs. Whilst research has indicated the assumptive nature of this, particularly when the sample is disproportionately female (Christie & Khatun, 2012), I wish not to make gendered claims of difference here, but to indicate that humans may generally have different styles of coping. Many parents in the sample reported having different coping methods and needs from their partner, which sometimes led to tension within the relationship. With family members perceived to be on different pages, how might support be offered to trace out the spaces of unity considered helpful?

There is perhaps though a gendered issue to reflect on briefly regarding the role division that arose for many parents, with mums typically the partner to leave or reduce work hours and thereby take on the greater bulk of childcare needs. Whilst the results chapter outlined the structural ways in which this happened for parents, with maternity leave producing a knowledge gap, or views of motherhood preventing access to social care support, there is much to be considered structurally in how families are supported. Parents and wider research have linked work with fulfilment and well-being (Pratt & Ashforth, 2003), but many parents feel there is no choice but to leave or reduce work hours due to medical demands. Work is thereby lost as a source of wellbeing, potentially increasing financial strain and exacerbating stress (Gannoni & Shute, 2010). Research conducted during COVID-19 has highlighted that the burden of childcare remains predominantly with mothers (Sevilla & Smith, 2020). This signals a need for wider consideration not only of how parents are being supported in the transition to parenthood, but to interrogate how the structures in place may continue to uphold these divisions and impact on well-being.

There was also pressure and concern from both parents and individuals about relationships with siblings. There were concerns about resentment as well as painful reflections on how relationships may have changed, with other siblings perhaps receiving less attention at times or feeling less important, as is supported by wider literature (Velleman et al., 2016). At the same time research has reported increased family closeness in contexts of childhood chronic illness (Wennick & Huus, 2012). Whilst this research did not reach siblings directly, other research on sibling experience indicates risk of emotional distress, worry and uncertainty about the future (Velleman et al., 2016). Whilst the impact on siblings is not straightforward and requires further research, what is clear is that there is potential for negative impact on the well-being of siblings, parents, and individuals in these relational shifts. It is crucial that research consider how practice might support families as a whole system in navigating this impact and maintaining relationships, to develop their strengths and manage difficulties.

Perhaps linked with this, there was a clear trend in learned communication patterns of not talking about FA/DBA. For many this was protective in nature to prevent worry or upset, but resultantly talk of death or prognosis comes to be experienced as taboo with patterns of silence reciprocated across the family (Hutson & Atler, 2007). This may indicate some therapeutic value in creating a space where families can find ways of talking together. This perhaps explains why self-selection was not a successful recruitment method.

Findings focused on family touch more explicitly on relational issues and lend further weight to how relationships are impacted upon and in turn impact on the health condition (Stiell et al., 2007). Parents note initial relational strain, as well as concerns regarding sibling and child relationships within the family. It is within this context that findings indicate these protective communication patterns of not talking that arise within family systems. It is not unfeasible that these patterns go some way to maintaining a double bind as well, as individuals are unable to address the tension between agentic and passive positions as this would require talking about the illness. These findings would lend support to the merits in providing the opportunity for families to access spaces in which to talk and think about reciprocal interactions between health and the family.

4.1.2 What is the current demand for psychological support and how suitable has provision been deemed?

In addressing this research question there are two elements to consider, firstly demand and secondly provision; these will be addressed separately. This section has not been separated by family member on account of less variation in responses. Whilst reference may be made to other sections, the focus here is on the final theme presented from the data of Mental Healthcare as Necessary but Inadequate.

Demand: It was clear across the sample that there was in fact a demand for psychological, counselling or psychotherapeutic support. Six of nine parents interviewed, and four of six individuals had sought some form of therapeutic support

at various points since diagnosis. That is 66% of the sample had *sought* out support. Emphasis is placed here on having sought out support to qualify and situate this number within its context in which, as will be discussed, support typically had to be actively asked for. Demand therefore cannot be measured by access numbers alone. It is not possible to know how many individuals might accept support were it to be offered routinely and proactively. This is crucial to hold in mind.

Analysis of the results would suggest that this issue is largely one of unequal access as outlined in subtheme two, *Unequal Access*. This is not a straightforward issue to unpack, with several strands addressed here, and possibly many more experienced in actuality. There was reference to knowledge with Parent 7 indicating that 'luckily I know what to ask for' as contrasted with Individual 5, who noted later realising that their experience was 'massive', and that no support had been offered. This is suggestive of an agency required to access support, and thereby highlights lack of knowledge of available provision as a potential barrier. Knowledge alone does not appear to open the way to support though, with Parent 10 recounting the repeated visits to a GP who recommended, however well intentioned, tea, antidepressants, and eventually self-referral to a mental health service. This required repeated visits and clarifications by a parent regarding her desire to talk to someone about the situation and how they were feeling. Whilst this is just one account, it is representative of the experience of many parents in this sample who had to push to receive any therapeutic support. In the context of the policy frameworks outlined (*1.6.3 Policy landscape),* where there is ongoing focus on achieving a parity of esteem between mental and physical healthcare, this stands as evidence of the gaps yet to be closed. That said, many of this group of individuals must also exercise agency in addressing physical healthcare too, a situation exacerbated by limited knowledge. Perhaps this picture speaks more to groups, such as those with rare conditions, who face obstacles in access to high quality care and therein may also be considered excluded from the general standards held as part of best practice care (Kole & Faurisson, 2010). While knowledge remains limited, these individuals

may be further excluded in the lack of any standardised approach to their care.

It must be noted that there were families included who had not encountered obstacles in accessing support. These accounts perhaps shine a light on the opposite side of the same coin, and thereby further add to the idea of unequal access. One family shared having been approached by and informed of the psychological support available at their treatment hospital. Another family indicated that support would have been difficult to arrange on account of their being far away from a treatment hospital. This may indicate how resources and specialist knowledge can vary according to location, service, and funding. Access is not just unequal; it is therein political. There are documented critiques across the literature regarding the disparities due to the 'post code lottery' nature of NHS provision (Russell et al., 2013). This appears to be the experience of these families both in terms of physical health and mental health. Again, in a policy context that is geared toward ensuring equal standards of care, work is yet needed.

Privilege was also raised as a mediating factor, with more than one family sharing that they had been fortunate enough to be able to pay for private psychological support. This offers a rationale against homogenising these families as one and the same. To do so is to cover over all of the intersecting variables that may significantly impact on the experience of living with FA or DBA, for the worse or the better. Research needs to be able to consider these differences to ensure the provision of support is anti-exclusionary and is able to meet the needs of any family.

A final obstacle noted amid the results which may impact on demand and access for psychological support concerned stigma. Parent 9 noted the risk as a parent of being perceived as not coping, the worry about what it would mean to be seen as seeking help. Other research has indicated that fear around negative perception may well prevent parents and families from seeking mental health support (Bruland et al., 2017). To contextualise this finding, it is worth noting the work in recent years to improve perinatal mental health access. Delivered in a tiered structure there is at

least an ideal aim of offering a minimum universal level of support for all new parents (Wave Trust, 2014). Whilst initiatives like this may help to normalise seeking support, it does seem again that there may be gaps with particular groups of parents not yet reached by provision.

Indeed, this gap is supported in the wider literature which indicates a greater body of research and presence of psychologists in services specific to other health conditions such as sickle cell disease or cystic fibrosis (Anie & Green, 2015; Goldbeck et al., 2014). It is unclear what has led to the presence of psychology in these areas and the relative absence of similar clinicians inputting to the support of families with FA/DBA. A tentative hypothesis here makes a connection between the lack of literature on FA/DBA as contrasted with the relative abundance of psychological literature for the two conditions mentioned here. The reason for this discrepancy has been articulated more in the *Introduction Chapter (2.3 One Condition or Many?)*, but it is worth noting the real-life practical impact of this unequal research base as leading to unequal levels of support.

Provision: Whilst this last point touches on provision, the emphasis here is on how provision has been deemed as outlined under the *One Size does Not Fit All* subtheme. This theme was so titled because views and experiences of provision differed.

Whilst this was so, there was repeated reference to the ways in which mental health and physical health are separated in regard to the provision of care. Whilst obstacles in accessing psychological support may illustrate psychology's peripheral nature in medical health care, individuals expressed a clear desire to have mental health and well-being asked about during their physical healthcare appointments, indicating a desire that clinicians better understand the wider experience of living with FA/DBA. This is in keeping with research that suggests medical consultations typically focus on physical aspects only (Coventry et al., 2011), and indicates progress still needed regarding the integrated care agenda.

This separation may be upheld by the mental health arena too. In discussing the psychological support received, Individual 6 reflected that this support overlooked the context of their health condition and how this might have been impacting on their depression. Moreover, this perhaps touches on a desire for specialised knowledge by those providing psychological support, echoed by other individuals represented in this sample. Interestingly, Parent 10 reflected that this knowledge need not be specific to DBA/FA but reasoned that many of the concerns or worries may be shared by parents of children with other complex health conditions. This may indicate that the real concern is not how important it is for supporting clinicians to understand the medical intricacies of the health condition, but to understand the emotional experience and the difficulties that may arise.

For some then, provision was deemed as lacking relevance, on account of lack of knowledge. For others the therapist modality was experienced as inflexible and limited. Individual 4 notes how despite feeling low in mood, a modality led by psychometric scales did not indicate low mood and therein was dismissed. Parent 10 recounted a modality which prescribed the use of cognitive strategies such as worry time, which were experienced as unhelpful. Whilst these strategies may have a place, without sufficient attention to the reality of concerns regarding FA/DBA, these strategies risk indicating that one is capable of thinking away real distress in their focus on the individual (Smail, 2005). It has elsewhere been noted that ethical mental health support should contextualise distress (Patel, 2020) which appears to have been missing for some. This critique is not aimed at any modality in particular, for all modalities have the potential to overlook context. It is offered instead to reiterate the importance of support that seeks to place the person or people in their context of living complex, multi-layered lives and with a diagnosis of FA/DBA.

In terms of provision that had been deemed most helpful, parents cited the ability to have an ongoing relationship with a therapist or counsellor, which was most often achieved in having sought private support. This indicates another perceived limit on

provision regarding how psychological support is routinely structured in the UK with discrete periods of time limited support. Carey et al., (2013) have written extensively of the merits in service user led services. This term is used to denote the ability of service users to lead on their care via opportunities to self-book appointments, and to collaborate in deciding on the number of sessions they receive. As envisioned by the authors, these services do not formally end periods of care but allow individuals to return as and when they see fit. This research has indicated that this is not in fact any more demanding on resources. Such initiatives may improve the experience of care for families.

Finally, both parents and individuals expressed frustration that mental health support had been reactive in delivery, sourced and provided when it became a need, rather than delivered proactively. This touches on a wider issue documented across mental health care around a need to focus on prevention (Kumari, 2020). One is left grappling again with a sense that the families represented here, and perhaps by other families living with rare conditions, remain outside of developments in general mental health provision. It seems some effort is required to fully acknowledge and address these discrepancies.

4.2 Critical Review

4.2.1 Reflexivity
Although widely accepted as a requirement of qualitative research, the concept of reflexivity is described in multiple and conflicting ways across the literature. To be explicit, the iteration of reflexivity employed in this research is at odds with those accounts which claim to use reflexivity to achieve objectivity as rooted in positivist epistemology (Lynch, 2000). The reflexivity attempted here is in keeping with the idea of reflexivity as critical thinking, in which one aims to articulate how the research and the knowledge it produces may have been shaped by various contexts and processes (Lazard & McAvoy, 2017). Reflexivity in this form is engaged with

throughout the research process to allow consideration and transparency regarding how the researcher's own views have given rise to and influenced the focus of research and the interpretations offered (Willig, 2013). To help in this, Wilkinson's (1998) account of reflexivity based on three domains (personal, epistemological, and disciplinary) is drawn upon.

4.2.2 Personal Reflexivity

An effort was made to spend time in this domain from the earliest conceptualisation of the research, and throughout the process. In this way I have sought to understand and clarify how my personal experiences, beliefs and attitudes have shaped the research. Berger (2015) notes a vast array of things that may be worth consideration, including gender, age, political and ideological stance. At the outset of the research I reflected on my personal experience as a family member of someone living with a lifelong and life-limiting condition. It is based on this that I was drawn to give voice to a group of families whose experience was missing from research. I have also had the fortune of being supervised by someone with a personal connection to the conditions considered here. This will have informed the research design and output with layers of insight and reflection. It also facilitated a shared reflective process of how both our experiences may be shaping interaction with the data. In this way the feedback and drafting process has been embedded with a double layer of personal reflexivity. No research is entirely apolitical. Indeed, this effort is in line with my own political outlook and interest in critical health literature, which has given rise to opinions about how services may be improved. These views will undoubtedly have shaped the research and analytic process involved (Braun et al., 2006).

4.2.3 Epistemological Reflexivity

These personal views are of course reflected in my positioning the research from a critical realist lens. The initial idea for the research stems directly from a tension between this lens and a positivist scientific one regarding how illness is

conceptualised. The researcher's own experience has in part led to an understanding of illness as a social experience, more than symptoms alone. In this way the research has also been framed using a systemic lens to explore these ways of thinking. The very idea of this research as necessary arises out of this position and perhaps in contention with mainstream ways of thinking about health and illness. I have not sought to filter out this positioning but have further used this stance to remain open to hearing the multiple perspectives that may arise in thinking about illness and well-being, some of which may pertain to contrasting ways of thinking (Healey et al., 2000). This stance will have influenced the research questions used as well as the mode of analysis chosen. The latter will be discussed with regards to quality.

4.2.4 Disciplinary Reflexivity

This domain encourages consideration of how topics or methods come to be considered more or less important in disciplines (Marecek, 2012). Historically psychology and health psychology have been allied with medicine and thereby their research base has been heavily influenced by positivism with a reliance on research methods affiliated with this stance (Stam, 2000). It has been argued that health research often privileges third-person accounts and data considered to be empirical over first-person accounts (Carel & Kidd, 2014). Personal testimony then may be viewed as less important within health research, thereby impacting practice. Moreover, health psychology has been slow to consider the political nature of healthcare provision and resource allocation (Stam, 2000). This has allowed me to understand my position regarding the scope and remit of psychology as a discipline, as one which can support in bringing service-users into health research and questioning the established system.

4.2.5 Quality of Research

Linked closely with the epistemological position of the research, quality was endeavoured for in line with the standards set out by Clarke and Braun's (2020)

reflexive thematic analysis, which points out that bids to manage bias and achieve objectivity are to misunderstand the philosophical underpinnings of this approach. For these reasons more traditional measures of quality such as coding frameworks or second coders were not employed. Rather researcher subjectivity was utilised as a measure of quality control (Terry et al., 2017). The results and analysis presented relied on deep immersion with the data achieved by one researcher. Themes were not pre-conceptualised but sculpted and shaped by the researcher. The results presented then have not been reduced by a need for agreement with another researcher, a technique often employed to offer objectivity, but which may actually obscure the meaning of the data for the sake of consensus. Instead, the researcher has remained reflexive and transparent about the processes and decisions made in conducting the research, as well as her own socio-political positioning and views which will likely have influenced the resulting analysis. Supervision has been used to remain mindful of these views and their impact too.

Nonetheless, to satisfy more traditional concerns regarding quality, the researcher has given thought to the four criteria set out by Lincoln & Guba (1985) and considers these to have been met without any tension upon the epistemological and methodological frameworks applied. Regarding credibility, there is a clear alignment throughout the research in terms of the epistemology and methodology employed. Dependability is assured in that another researcher could clearly follow the steps applied here, although, in line with the epistemology it would not be unlikely if their analysis differed. With regards to confirmability, the researcher has made a detailed use of description and quotes to share the thought processes behind analysis. Finally, the context in which the research was documented has been clearly set out, thus it is hoped the findings presented are transferable in that they resonate with other parents. Moreover, it is hoped that the analysis presented will make clear that it would be difficult to transfer all findings neatly, as those very findings indicate a range of important contexts by which the experience of families may vary. It is these variations to which further research must pay attention.

4.2.6 Strengths

As far as the researcher is aware, this is the first piece of research within the UK which considers the experience of families living with FA and DBA from a psychological perspective. The research sought to understand the psychological impact on families of living with either condition. It enabled individuals to reflect on and share their experience of receiving and living life following diagnosis, and to add their voice to an otherwise medically oriented research landscape. Whilst medical research continues to grow, this is the first research which considers the lived experience of the individuals who are recipients of the medical science.

The sample of family members involved included individuals diagnosed as infants and children as well as those who received a diagnosis later in adulthood. It also included parents whose children ranged in age at diagnosis from three months to 13 years old. In this way the sample offers a good range of variation as a beginning in mapping the experience of families.

The researcher considers the broad focus on psychological impact a strength in having facilitated a wide consideration of the emotional well-being and mental health of family members. This frame has allowed consideration of the impact on families as well as their sources of strength and resilience in mitigating this impact. Whilst this breadth may be open to critique it is considered fitting for research that seeks to begin an ongoing exploration of familial experience. More applied research is welcomed as a development, which may wish to be specific in the domain of impact it explores.

4.2.7 Limitations

Nonetheless there were limitations of the sample of family members represented here. The majority of the parent sample was comprised of mums with just two dads represented, and all of the individuals were female. In addition, no siblings were

reached in the recruitment processes meaning that reference to siblings is minimal here and based only on the perceptions of those relationships made by other family members. Whilst still useful, this could be strengthened by having the direct voice of siblings included.

Furthermore, participants were all above the age of 18 years. Whilst the ethical rationale for this has been indicated, it does limit the findings here as ones which must be contextualised as the views of adults reflecting retrospectively on their earlier years. Whilst this is useful insight, it is difficult to know if the reflections by adults on what might have been helpful as children would actually be acceptable to and accepted by children. This may therein limit the utility of findings in their application directly to children. It is hoped that having begun a conversation, more research can follow to empower children living with either condition to speak to their own needs in service design and provision.

There may have been several factors at play in shaping the sample represented here. It is possible that recruitment via charities, which by their positioning are a source of help, has led to gaps in the pool of families reached, missing out those who may not be actively seeking help or engaged in online communities. It may be that siblings are not active on these support forums. It may also link to possible gender differences in help seeking (Nam et al., 2010). Whilst these limitations might mean there are gaps in the understanding presented here, it remains useful to reflect on these potential gaps, and how research might be improved to reach those missed. These are all considerations for future research.

It should be noted as well that all the parents represented here were parents of individuals diagnosed as children. The results then may not be representative of the experience of parents whose children were diagnosed in adulthood. To the researcher's knowledge, all those parents represented were also in relationships with their child's father, overlooking single parent families or other variations on the family unit. Support needs may look very different for these families.

Finally, there are perhaps limitations in having a combined focus on FA and DBA. Whilst similar in many ways, both conditions are distinct, and these differences may

be reduced in the results presented here. At the same time, however, this does perhaps speak to a growing body of literature which encourages research that focuses more on the experience of living with rare conditions more generally (von der Lippe et al., 2017).

4.3 Implications of the Research

4.3.1 Clinical Implications

This research indicates a desire among families affected by FA or DBA for access to psychological supports or support around wellbeing more generally. Having framed this desire within the wider literature on chronic illness supports the likelihood that this finding will hold true for UK families beyond those represented in the sample here. Whilst this is so, the current research also indicates that families may face multiple obstacles in accessing therapeutic support. In addition, when accessed, the support is not always experienced as helpful or relevant; the support received has been reported to overlook the impact of FA/DBA producing a sense of separation between physical health and mental health. It is also considered to be reactive in focus.

This demand indicates a duty to consider how the findings from this research, and the necessary future research, may be applied practically to improve and meet demand for support. This will be given consideration with regard to services, policy, research, and clinical training.

4.3.2 Implications for Services

Families identified several barriers experienced in accessing psychological support, and their varied responses indicate that there is presently no clear pathway by which families living with FA/DBA might seek support if required. Responses also raise questions regarding whether mainstream mental health services are meeting the needs of individuals who seek support without any clear understanding of their

health condition, and in some cases with care delivered via modalities and in ways which actively did not facilitate space for this. Understandably then families raised the idea of physical health and mental health being separate in the care they receive. Given that an individual may be receiving care from multiple health services at any one time there is a further sense of fragmentation to care provision. This is not a necessary but unfortunate consequence of having multiple health needs, rather it is an unfortunate consequence of how services are currently structured. Individuals expressed a desire for medics to ask about mental health, and for mental health to include physical health understandings, as indicated by the agenda for integrated care. These ideas are not new or novel; they already exist in other health areas with integrated health and psychological support; they are possible. It is hoped that clinicians and commissioners might be responsive in using the experiences of FA/DBA families to work toward achieving more integrated care provision, and to carve out clear pathways of support which may be clearly outlined to families at the point of access.

On this note, services could be structured so as to facilitate proactive support that is geared toward prevention as endorsed by the World Health Organisation (WHO, 2002). The accounts presented by the families here indicate that periods of stress and distress may occur in relation to FA/DBA right across the lifespan and in conjunction with other life events (Rolland, 1984). Whilst it is accepted that mental health falls under the remit of the human right to health care (WHO, 1946), services must begin to respond to ensure FA/DBA families are receiving relevant support. It is not enough to claim that individuals can access support via general primary care methods if this support lacks the relevance or focus required to be helpful. Services must be guided by the experience and voice of service users in exploring and responding to unmet needs. All parents for instance indicated that the initial period following diagnosis was a really difficult time. A response to such findings might be to offer every family who receives a diagnosis a set number of initial supportive sessions. To provide a universal offer of support in this way might be to remove

some of the stigma or concern that would prevent parents from seeking support (Eaton et al., 2016).

Research has also indicated the importance of offering choice to service users (Mayor, 2016). Given also the impact outlined across the family system it would seem fitting to offer individuals the choice of family sessions or one-to-one sessions. Moreover, whilst further research may be needed, creativity and innovation are encouraged in responding to the views of families expressed here. In particular, families touched on the importance of ongoing relationships in health care. Particularly in the context of complex and chronic health conditions, and the bid for integrated care, one is drawn back to the work of Carey (2013) in advocating for mental health provision delivered in a similar way to physical health care, where individuals may book as and when required. This is not to assume that all individuals or families will need or desire support, but this provision recognises and normalises that support is available if required from time to time in the same way as from one's GP.

Finally, the peer support celebrated by the families represented here, highlights the potential for services to foster stronger links with charitable and supporting organisations who are often the source of this peer support. In this way better links are established whereby professionals are better able to signpost to various supports, removing much of this burden from families currently left to seek out connection on their own. These relationships would also serve to formally recognise the specialist knowledge and work of such organisations.

Ultimately these suggestions are the researcher's based on conversations with the family members represented. Whilst this is a useful step, further development will be to include families in service development and design as experts by experience.

4.3.3 Policy implications

Much of the above requires change, and change requires facilitation. A paper from the Psychological Professions Network (PPN, 2019) considers how psychology can support in implementing the NHS Long Term Plan (NHS, 2019). In this they note the lack of representation of psychologists in leadership roles across the NHS, with no requirement - even for mental health providers - to have representation from psychology at Board level. NHS Improvement (2019) recognises the contribution psychologists could make toward leadership and achieving system change. Given the agenda of achieving more integrated care (Naylor et al., 2016), it seems timely that psychologists are included at a leadership level. This will ensure that those trained to think psychologically about health can share and develop this thinking in conjunction with the specialist knowledge of those who are medically trained. Psychologists are also trained to appreciate a role and function for qualitative research and are therefore well placed to create policy that stems from the voice of those it is designed to protect and serve. In this way change at a policy level could be achieved which would formalise a standard of care for FA/DBA families.

4.3.4 Implications for training

The findings from this research may also make contributions to clinical training. At present the content of health psychology teaching may vary across training centres, with health-related placements an optional requirement. The PPN (2020) recommend that the content of clinical training is reviewed to ensure sufficient skills and knowledge are acquired for working in areas of physical health. This research prompts some further development of this point and encourages a critical consideration of what is included in this content. Responses from families included here have indicated that the psychological support received has overlooked FA/DBA or has prescribed certain strategies which have not felt helpful in isolation. It is imperative that clinical psychology training facilitates engagement with critical health discourses to support in developing clinicians who can work alongside families to develop meaningful ways of thinking about the complex experience of living with chronic illness. This will also foster clinicians who are able to support in

contextualising this distress by understanding the complex interplay between health and social determinants of distress (Allen et al., 2020). In this way clinicians will be skilled enough to move away from a focus that rests largely on health behaviours and behaviour change (Sainsbury et al., 2018), to offer interventions and modalities flexibly based on individual or family need.

4.3.5 Implications for Research

Developing clinicians in this way is likely to have an impact on research practices too. Whilst the *Introduction* chapter set out a research landscape that is unequal in focus regarding how much attention different conditions might receive, the results indicate how this plays out in practice with no specialised psychological support available to those families represented here who live with FA/DBA. Training clinical psychologists to critically engage with this landscape is likely to have benefits in beginning to address some of these gaps specifically regarding research on FA/DBA, and more generally to increase the psychological research base for physical health conditions. It is hoped that this would begin to change the clinical picture too by improving the research base from which interventions and supports are drawn.

More specifically, ensuring that training is critically focused is likely to mean that research teaching is epistemologically broad. This may help to move away from the 'methodolatry' that has been noted to arise in health research (Chamberlain, 2000). Sophisticated research in health and health psychology benefits from both quantitative and qualitative methods. Whilst the results here indicate a need for more research that gives voice to the experience of individuals living with FA/DBA, to focus on this at the expense of medically focused research would of course be problematic. Seidlien & Salloch (2019) note that the risk in health-related research is that these two foci, one on medicine, one on patient experience, do not often map onto or relate to each other well. As proposed by the authors, there is an ethical imperative that health research must progress to encompass both, to produce medical research that is guided by patient experience. Certainly, the results from this

research indicate that there would be much benefit to such research in mapping experience and need onto support provision.

4.4 Conclusion

The current research set out to initiate an understanding of how individuals and families may be psychologically impacted on in living with the life-long, life-limiting conditions that are Fanconi Anaemia and Diamond Blackfan Anaemia. It also sought to consider if there was demand for psychological support and, where accessed, how support has been regarded.

The accounts of the parents and individuals represented here indicate a complex picture regarding psychological impact experienced across the family and across the lifespan. At the individual level, both parents and individuals describe an impact on mental health and wellbeing. For individuals this encompassed the stress of uncertainty, awareness of mortality, and indeed at times impacted on the development of one's sense of self as 'ill' or otherwise. For parents this impact ranged from feelings of grief following diagnosis, to stress, pressure and isolation in adjusting to life with their child's diagnosis as well as managing the uncertainty beyond this. Both also reflected on social elements of impact, namely lack of understanding and identification, as well as sometimes encountering explicitly negative judgements and interactions.

Impact was felt beyond the individual level, most notably in concerns regarding relationships within the family. This included parental relationship strain, as well as concerns regarding the relationship with siblings in the family. It was noted that families often use protective communication patterns of not talking about the conditions to prevent causing upset and distress. The direct voice of siblings was missing in this research, which is indicated as a direction for future endeavours.

In the context of these findings, it is understandable that many individuals and

parents had sought or tried to seek out psychological support. Support overall was critiqued for being largely separate from medical care, overlooking the impact of FA/DBA, and being reactive in focus. Most notably access was unequal, with several potential structural issues at play.

These findings offer implications for future research and practice. Crucially they indicate a desire and need for support from many families. This is not limited to psychological support with indication that peer support is much welcomed. There is a clear need here to consider the provision of care for families living with FA/DBA, as well as benefit in collaborating with these families to improve provision and service design.

The contribution of this research is to highlight that there are support needs regarding well-being across these families, and that provision is not deemed as adequate by many in its current form. The voice and experience of these families has been missing in the research base until now. Research must now rise to the challenge of uncovering and addressing the gaps and inequalities that have led to the current status quo; it must seek to establish clear and adequate pathways and a standard of care for the well-being of all families living with FA/DBA.

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APPENDIX A - Invite Letter

University of East London

PARTICIPANT INVITATION LETTER

You are being invited to participate in a research study. Before you agree it is important that you understand what your participation would involve. Please take time to read the following information carefully.

Who am I?

I am a postgraduate student in the School of Psychology at the University of East London and am studying for a Doctorate in Clinical Psychology. As part of my studies I am conducting the research you are being invited to participate in.

What is the research?

I am conducting research to understand what it is like for individuals and families to live with Fanconi Anemia and Diamond-Blackfan Anemia.

Research Title: **Exploring the Impact of Living with Fanconi Anemia & Diamond-Blackfan Anemia: A Systemic Consideration.**

Research Questions:
1. What is the psychological impact on an individual and their family of living with Fanconi Anemia or Diamond Blackfan Anemia.

2. What is the current demand for psychological support and how suitable has any provision been?

My research has been approved by the School of Psychology Research Ethics Committee. This means that my research follows the standard of research ethics set by the British Psychological Society.

Why have you been asked to participate?

You have been invited to participate in my research as someone living with either condition. I am hoping by participating that your experience can help develop knowledge in this area particularly around supporting families.

I emphasise that I am not looking for expert answers, but your lived experience. You will not be judged or personally analysed in any way and you will be treated with respect. You are free to decide whether or not to participate and should not feel coerced.

What will your participation involve?

If you agree to participate you will be asked to share your experience as someone who lives with either condition. I am interested in hearing more about the impact of this condition on your life and family, and to hear your ideas about the support you have received or could receive.

I will ask to chat with you for up to 1 hour during which time I will ask some questions about how it was to receive your diagnosis and to live with your condition, and how this may have changed over time.

Our chat will be relaxed and informal, as I would like you to feel as comfortable as possible in chatting with me. Our chat will be recorded so that I can type up what I have learned from you for my research.

I expect that we will meet for up to 1 hour. Given the current restrictions on contact, we will be able to meet online via Microsoft Teams.

I will also invite you to ask immediate family members to join focus groups of other family members, eg a focus group of parents, or siblings. This is optional on your part. This will allow a space to hear their perspective on living with either condition as a family.

I will not be able to pay you for participating in my research, but your participation would be very valuable in helping to develop knowledge and understanding of my research topic.

Your taking part will be safe and confidential. Your privacy and safety will always be respected. You will not be identified by the data collected, on any written material resulting from the data collected, or in any write-up of the

127

research
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You do not have to answer all questions asked and can stop your participation at any time or request a break if you would like one. It is not expected that you will be harmed in any way by taking part.

What will happen to the information that you provide?

I will securely store your personal contact details and the record of our discussion by password protecting them. I will then be able to look at our discussion alongside those of other people to see where there might be common thoughts, ideas and experiences.

Any details that could identify you (e.g. Your name) will be anonymised by the substitution of numbers allocated to each person involved.

Only myself, my supervisor, and examiners will see this anonymised data. If any of the research is published then this information becomes more publicly available, but your information will not be identifiable from this information. All anonymised data will be deleted after 5 years of completing the research.

All identifiable data will be destroyed on completion of my study write-up. I will remind you at the end of our interview, that you can of course withdraw your data until 3 weeks after we meet, as then I will need to begin writing about this.

What if you want to withdraw?
You are free to withdraw from the research study at any time without explanation, disadvantage or consequence. Separately, you may also request to withdraw your data even after you have participated, provided that this request is made 3 weeks of the data being collected (after which point the data analysis will begin, and withdrawal will not be possible).

If you would like further information about my research or have any questions or concerns, please do not hesitate to contact me.

Stacey Barkley, u1826607@uel.ac.uk

If you have any questions or concerns about how the research has been conducted please contact the research supervisor Paula Corredor Lopez,

Programme Director (Clinical), Professional Doctorate in Clinical Psychology, School of Psychology, University of East London, Water Lane, London E15 4LZ,

Email: Paula Corredor-Lopez - P.Corredor-lopez@uel.ac.uk

 or

Chair of the School of Psychology Research Ethics Sub-committee: Dr Tim Lomas, School of Psychology, University of East London, Water Lane, London E15 4LZ.

(Email: t.lomas@uel.ac.uk)

APPENDIX B – Consent Form

UNIVERSITY OF EAST LONDON

Consent to participate in a research study

Title: Exploring the Impact of Living with Fanconi Anaemia & Diamond-Blackfan Anaemia: A Systemic Consideration.

I have read the information sheet relating to the above research study and have been given a copy to keep. The nature and purposes of the research have been explained to me, and I have had the opportunity to discuss the details and ask questions about this information. I understand what is being proposed and the procedures in which I will be involved have been explained to me.

I understand that my involvement in this study, and particular data from this research, will remain strictly confidential. Only the researcher(s) involved in the study will have access to identifying data. It has been explained to me what will happen once the research study has been completed.

I hereby freely and fully consent to participate in the study which has been fully explained to me. Having given this consent I understand that I have the right to withdraw from the study at any time without disadvantage to myself and without being obliged to give any reason. I also understand that should I withdraw, the researcher reserves the right to use my anonymous data after analysis of the data has begun.

Participant's Name (BLOCK CAPITALS)

……………………………………………………………………………………

Participant's Signature

……………………………………………………………………………………

Researcher's Name (BLOCK CAPITALS)

……………………………………………………………………………………..

Researcher's Signature

……………………………………………………………………………………

Date: ……………………….……

APPENDIX C – Interview Schedule

<u>**INDIVIDUALS**</u>
PRE-DIAGNOSIS

1.How old were you when you were diagnosed with FA/DBA?

2. How did you learn about your condition [FA/DBA]?

3. What did this mean for you at the time/ Did you have any particular concerns or worries?

4. Were you able to talk about these? (Consider who with/why/how)

Y - How was it to talk about these?

N - How do you think this impacted on you? (What were the barriers?)

- **POST-DIAGNOSIS**

6. How do you feel [FA/DBA] impacted on your school life or education?

7. What has [condition] meant for you as a family?/How has [condition] affected you as a family? Prompts: Relationships

-Relationships with parents

-Sibs

-Friends

8. How has the impact of your health condition changed as you have gotten older?

-What does it mean for you day-to-day?
9. How do you think living with [FA/DBA] affects how you think about the future?
10. I wonder if and how you feel your condition has affected your mental health?

11. Have you ever sought support from a professional or someone else to think about any of this?

Y – Were you satisfied with this? Was there space to think about how [FA/DBA] might be contributing?

N – Do you feel you could or **would*** have benefited from a space to talk about this?

- Would you have accepted this if it had been offered as part of health check-ups?
*Were there any barriers to accessing support?

12. Have you got any ideas of types of support not currently available that you and your family or others living with [FA/DBA] might benefit from?

PARENTS

- **PRE-DIAGNOSIS**

1. How was the process of getting a diagnosis for your child?
-What was the length of journey pre-diagnosis? - Any other diagnoses?

-obstacles/helpful elements

2. Could you describe what happened for you personally on learning of your child's diagnosis?

3. Did this impact your parenting/your interactions with your child with FA/DBA at all?

-Sources of support?
 -What helped?
-Obstacles?
-Adjustments to day-to-day life?

4. Are there additional supports you could envision being helpful for parents at this early stage of finding out about their child's diagnosis?

- **POST-DIAGNOSIS**

5. How has your child's condition affected family life?

-Relationships within? Other children. Roles.

6. How has this changed as your child has gotten older?

-adolescence / peers

-adulthood

-independence

7. How do you think raising a child diagnosed with [FA/DBA] affects how you think about the future?

8. I wonder if and how you feel your mental health has been affected as someone who cares for a child with [FA/DBA]? (How?)

9. Have you sought professional support to talk about your mental health or the impact of living with this health condition as a family?

Y- Has this been satisfactory? (Space to think about impact of caring role? Impact of child's health?)

N- Do you feel you could or would have benefited from a space to talk about this? - Would you have accepted this if it was offered regularly? Any barriers?

10. Have you got any ideas of types of support not currently available that you and your family or others living with this condition might benefit from? -What's lacking re support?

SIBLINGS

- **PRE-DIAGNOSIS**

1. How do you remember learning about your sibling's health condition [insert]?

2. What did this mean for you at the time? Did you have any particular concerns or worries?
3. Were you able to talk about these? (Consider who with/why/how)

Y - How was it to talk about these?

N - How do you think this impacted on you? How did you manage these?

- **POST-DIAGNOSIS**

5. 7. How do you think that [condition] has affected you as a family?
Prompts:

-Relationships with parents

-Sibs

-Friends

- What does it mean for you day-to-day?

6. How do you think having with a sibling who lives with [FA/DBA] affects how you think about the future?

7. (Optional as based on above responses) - I wonder if and how you feel your sibling's condition has affected your own wellbeing? (How?)

8. Have you ever sought support from a professional or someone else to think about this?

Y – Were you satisfied with this? (Consider if space to discuss sib's health and family situation).

N – Do you feel you could or **would*** have benefited from a space to talk about this?

 - Would you have accepted this if it had been offered? ***Were there any barriers to accessing support?***

9. Is there anywhere or anyone that you feel understands how it is to be a sibling of a child with this condition?

9. Have you got any ideas on types of support not currently available that you and your family or others living with [FA/DBA] might benefit from?

APPENDIX D – Conventions Used in Transcribing

(Banister, Burman, Parker, Taylor, & Tindall, 1994, pp.64)

For pragmatic reasons of adhering to a word count whilst also aiming to accurately represent the data and resultant analysis, minor changes were applied to the text selected for inclusion. Repetitive and filler words (such as, 'sort of', 'umm', 'you know') were removed to improve readability and clarity.

Conventions used to convey any changes are as follows:

… to indicate the omission of words or sections of text
[text] addition of content to provide clarity, or to remove specific text to ensure anonymity whilst maintaining clarity.

APPENDIX E– Ethical Approval & Amendment

School of Psychology Research Ethics Committee

NOTICE OF ETHICS REVIEW DECISION **For research involving human participants** BSc/MSc/MA/Professional Doctorates in Clinical, Counselling and Educational Psychology

REVIEWER: Ava Kanyeredzi

SUPERVISOR: Paula Corredor Lopez

STUDENT: Stacey Barkley

Course: Doctorate in Clinical Psychology

Title of proposed study: Exploring the Impact of Living with Fanconi Anemia & Diamond-Blackfan Anemia: A Systemic Consideration

DECISION OPTIONS:

1. **APPROVED:** Ethics approval for the above named research study has been granted from the date of approval (see end of this notice) to the date it is submitted for assessment/examination.

2. **APPROVED, BUT MINOR AMENDMENTS ARE REQUIRED BEFORE THE RESEARCH COMMENCES** (see Minor Amendments box below)**:** In this circumstance, re-submission of an ethics application is not required but the student must confirm with their supervisor that all minor amendments have been made before the research commences. Students are to do this by filling in the confirmation box below when all amendments have been attended to and emailing a copy of this decision notice to her/his supervisor for their records. The supervisor will then forward the student's confirmation to the School for its records.

3. **NOT APPROVED, MAJOR AMENDMENTS AND RE-SUBMISSION REQUIRED** (see Major Amendments box below)**:** In this circumstance, a revised ethics application must be submitted and approved before any research takes place. The revised application will be reviewed by the same reviewer. If

in doubt, students should ask their supervisor for support in revising their ethics application.

DECISION ON THE ABOVE-NAMED PROPOSED RESEARCH STUDY
(Please indicate the decision according to one of the 3 options above)

2. Approved – Very minor amendments

Minor amendments required *(for reviewer)*:

Q.4.4 Please clarify that the interviews will be conducted on teams and not an audio recorder Unless this is a number solely for the purposes of research, please remove your telephone number from the Invitation Letters.

Major amendments required *(for reviewer)*:

Confirmation of making the above minor amendments *(for students)*:

I have noted and made all the required minor amendments, as stated above, before starting my research and collecting data. Student's name *(Typed name to act as signature)*:   Stacey Barkley Student number: u1826607 Date: 21/10/20 *(Please submit a copy of this decision letter to your supervisor with this box completed, if minor amendments to your ethics application are required)*

ASSESSMENT OF RISK TO RESEACHER *(for reviewer)*

Has an adequate risk assessment been offered in the application form?

YES / NO

Please request resubmission with an adequate risk assessment

If the proposed research could expose the <u>researcher</u> to any of kind of emotional, physical or health and safety hazard? Please rate the degree of risk:

☐ HIGH

Please do not approve a high risk application and refer to the Chair of Ethics. Travel to countries/provinces/areas deemed to be high risk should not be permitted and an application not approved on this basis. If unsure please refer to the Chair of Ethics.

☐ MEDIUM (Please approve but with appropriate recommendations)

☒ LOW

Reviewer comments in relation to researcher risk (if any).

Reviewer *(Typed name to act as signature)*:   Ava Kanyeredzi

Date: 20/10/2020
This reviewer has assessed the ethics application for the named research study on behalf of the School of Psychology Research Ethics Committee

<u>RESEARCHER PLEASE NOTE:</u>

For the researcher and participants involved in the above named study to be covered by UEL's Insurance, prior ethics approval from the School of Psychology (acting on behalf of the UEL Research Ethics Committee), and confirmation from students where minor amendments were required, must be obtained before any research takes place.

For a copy of UELs Personal Accident & Travel Insurance Policy, please see the Ethics Folder in the Psychology Noticeboard

University of East London Psychology

REQUEST FOR TITLE CHANGE TO AN ETHICS APPLICATION

FOR BSc, MSc/MA & TAUGHT PROFESSIONAL DOCTORATE STUDENTS

Please complete this form if you are requesting approval for proposed title change to an ethics application that has been approved by the School of Psychology.

By applying for a change of title request you confirm that in doing so the process by which you have collected your data/conducted your research has not changed or deviated from your original ethics approval. If either of these have changed then you are required to complete an Ethics Amendments Form.

HOW TO COMPLETE & SUBMIT THE REQUEST

1. Complete the request form electronically and accurately.

2. Type your name in the 'student's signature' section (page 2).

3. Using your UEL email address, email the completed request form along with

associated documents to: Psychology.Ethics@uel.ac.uk

4. Your request form will be returned to you via your UEL email address with reviewer's response box completed. This will normally be within five days. Keep a copy of the approval to submit with your project/dissertation/thesis.

REQUIRED DOCUMENTS

1. A copy of the approval of your initial ethics application.

Name of applicant: Stacey Barkley

Programme of study: DClinPsy - ProfDoc

Name of supervisor: Dr Paula Corredor-Lopez

Briefly outline the nature of your proposed title change in the boxes below

Proposed amendment	Rationale
Old Title: Exploring the Impact of Living with Fanconi Anemia & Diamond-Blackfan Anemia: A Systemic Consideration.	Former title contains American spelling: anemia rather than anaemia.
New Title: Exploring the Impact of Living with Fanconi Anaemia & Diamond-	

Blackfan Anaemia: A Systemic Consideration.	

Please tick	YES	NO
Is your supervisor aware of your proposed amendment(s) and agree to them?	x	
Does your change of title impact the process of how you collected your data/conducted your research?		x

Student's signature (please type your name):   Stacey Barkley

Date: 17/03/2021

TO BE COMPLETED BY REVIEWER		
Title changes approved	YES	
Comments		

Reviewer: Glen Rooney

Date: 18/03/2021

APPENDIX F – Sample Coded Transcript

Interviewer: Ok. And I wonder if you remember or know much about how the process was for your parents to get that diagnosis?

Participant: Erm yeah, so from what I understand it was actually quite easy erm mainly because I was under the care of [Name] Hospital and then there was actually a specialist, well, someone who'd heard of my illness before and so was able to diagnosed well quite quickly and I was able to get the right treatment from there.

> Specialist/knowledge/familiar with condition, made it easier to be diagnosed.

Interviewer: Great, OK. And I wonder do you remember how you learned about your condition?

Participant: Erm, yeah, but I just think I've grown up kind of knowing that there's something kind of, you know, it's just an illness that I have, that I've just got to eh, I've just got, not to keep going, but it's just part of me.

> Grown up aware of – 'illness' Part of person.

Interviewer: Yeah, yeah, fair.

Participant: Erm it's never really been a big deal up until, uh, well, quite recently. Ah, I've always just kind of thought I'm just a normal person, just kind of living my life getting on with things.

> Change in impact
> Idea of 'normal'

Interviewer: And we'll maybe come on to talk about recently a little bit later. I wonder if you remember the first time you were aware of having DBA?

Participant: Um No, not really. I mean I've had constant checkups, and constant hospital appointments, irregular blood transfusions and medication every day, so it's kind of one of those things like I know I've got two arms, it's just like I knew I've got DBA. And I knew that other kids didn't have it, erm but I was always in the mentality that I wasn't going to let it kind of rule my life. It's not, it's a part of me,

> Constant medical input

> Awareness of difference, other kids didn't have.

> Mentality re condition

144

but it isn't me. That was my mentality.

Interviewer: Yes, I see, ok. You said there you were aware that other kids didn't have this. Can you say a little bit about that or or how you think you came to realize that?

Participant: Erm well, I have brothers and sisters, so I've always been aware that they don't have erm the medical appointments I do. But I've just I've always taken um, like I understand kind of the numbers of it all and what's actually happening,
happening em physically with my body and what the symptoms of it were. And so I've always just kind of been aware and also keen to understand what is happening to me as well.

| Awareness based on medical treatment |

| Interest in process/seeking to understand. |

Interviewer: So when you were little [Name], you started to notice the medical treatments other people didn't have. Did you have any particular concerns or worries that you remember?

Participant: Erm no, not really, no. I just, I think my parents were always quite erm, they were always quiet like, oh, this is just something we have
to do. It's not a problem, it's not an issue. I've always kind of grown up not, like this is gonna sound, well yeah, when I've - I've gone to ah, DBA
meetings before, I've gone to a couple to follow up until a couple and um, I didn't find it that
helpful. There's a lot of people were of the mentality that like it was a really negative thing and it's really restrictive. And obviously with DBA, there's lots of different, like not strands of it but it affects
people really differently. Whereas I've always kind of been like this is just something that I
have. So I'm really lucky in the sense that my parents were like that and let me do what I wanted to do instead of kind of holding me back.

| Parental perspective re DBA |

| Different perspectives, negative/restrictive |

| Parental perspective – impact not held back |

Interviewer: Yeah, maybe that answers the question. I was going to ask what do you think made the difference for you? So from some
people in those groups where some people

saw it as something quite negative.

Participant: It was just my parents, um... And I also, um. I think I've got kind of like the the mentality of um kind of really pushing myself as well and wanting, like kind of a personality of like wanting to be perfect and an and I wanted like I didn't want that to define how I was going to be. And I always hated it when people were like, oh, I'm really tired but I can't be as tired as you because of your illness. I'm kind of, uh, a lot of the time because it wasn't really spoken about I kind of pushed it to the back of my mind and I had this illness.

Not wanting to be defined by diagnosis

Interviewer: OK, that that makes sense and that that mentality you you're speaking about where do you think that comes from for you?

Participant: I actually don't know. So um, kind of last, so the middle of last year I actually had a mental breakdown. Erm I've kind of unpacked a lot of, a lot of that in therapy, but not really got any resolution, so I'm gonna be having more therapy soon. But I've just always been like it, so I've always been stressed, I've always, so, if there was homework that I forgot I had, I would like breakdown and cry and just wanna be like that perfect person and I'm such a perfectionist. And yeah, I don't know. I've just always been like. And my mum I've also spoke to my mum and she said that yeah, you never you, you just worried about everything.

Mental health – 'breakdown'

Accessed therapy

Perfectionism

Interviewer: Do you think that links with having had DBA or having had the experiences you've had when you were little?

Participant: Yeah, it's really hard, it's really hard to know erm because I have always been like it, and I just wouldn't know because I've always lived with DBA erm and I've always had this drive and I wouldn't know if I didn't have

Difficult to separate DBA

DBA if I would have that drive to king of succeed and push beyond my limitations.

Interviewer: Yeah.

Participant: Probably not push beyond my limitations because I wouldn't be striving to be like a like a in inverted commas, normal person. I would just be trying to be, well just be myself.

Push to be 'normal' – idea of not being normal?

Interviewer: Yeah, yeah.

Participant: I push more, I push through because, I push through because I don't want to be seen as someone that's weak and that someone who has those limitations.

Negative associations – weak/limitations

APPENDIX G – Thematic Map

INDIVIDUALS

KNOWLEDGE

ILLNESS CONCEPT

Medical Uncertainty

Knowledge Acquisition

Relationship to Condition

Social Evaluation

PARENTS

KNOWLEDGE

SOCIAL RESPONSES

Medical Uncertainty

Questioning Knowledge

Losses & Gains

Social Judgement

JOINT

FAMILY DYNAMICS

MENTAL HEALTH CARE NEC. BUT INADEQUATE

Parental Relationship Strain

Protective Communication Patterns

Concerns re Siblings

Impact Across Family

Unequal Access

One Size Does Not Fit All