‘You have to sit and explain it all, and explain yourself’. Mothers experiences of caring for an adult with a rare genetic intellectual disability syndrome.


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Abstract

Background: The experiences of mothers caring for an adult offspring with a rare genetic syndrome has not been previously explored in research.

Methods: Eight mothers of adults with Angelman, Cornelia de Lange, or Cri du Chat syndrome were interviewed. Interpretative Phenomenological Analysis (IPA) was used to interpret the interviews.

Results: Four themes emerged from the analysis: (i) Social inclusion and stigma, (ii) Uneven medical and social care service provision, (iii) The inertia of social care services, and (iv) Mothers as advocates.

Conclusions: These findings suggest that barriers to social inclusion affect parents as well as the person with intellectual disabilities, and that accessing appropriate social care services is a lengthy and complex process for mothers. These data may help inform care service providers about how best to support young adults with rare genetic syndromes and their carers.

Key Words: Angelman syndrome, Cri du Chat syndrome, Cornelia de Lange syndrome, qualitative research, maternal, support services.
Introduction

Research on parents of individuals with intellectual disabilities has largely focused on the childhood years. Quantitative research has suggested that the increased demands due to a child’s personal, medical, and educational needs may place parents at increased risk for stress and depression when compared to parents of typically developing children (Olsson & Hwang, 2001). When parents are asked directly about their experiences of raising a child with an intellectual disability in qualitative research, they identify a number of problematic issues, such as shock and distress around the time of diagnosis of their child (Kearney & Griffin, 2001). As their child grew up, parents have described having many worries and fears about their development, and, at times, feelings of hopelessness when trying to help their child. As well as this, many parents reported that coping with negative attitudes of others (including professionals) towards their child added an additional burden (Kearney & Griffin, 2001).

Of course, not all experiences are negative and many positive dimensions have also been identified in qualitative studies. Gray (2004) found that parents of children with autism employ many different coping strategies during their child’s upbringing, and generally felt that they were successful in their coping, despite the challenges. Additionally, parents report feeling great joy and pride in their child’s achievements, and having a sense of hope for the future, particularly if their child surpassed initial negative prognoses (Hastings & Taunt, 2002; Scorgie & Sobsey, 2000). In other research, parents were keen to stress that they felt the experience of raising a child with an intellectual disability had made them better people; some reported it had made them stronger and more tolerant (Kearney & Griffin, 2001). The considerable positive impact of having a child with intellectual disabilities can occur concurrently with any
negative impact (Glidden & Johnson, 1999; Hastings & Taunt, 2002). Experiencing both positive and negative feelings at the same time has been described as a ‘tension’ for parents, whose joy in their child is also a part of the pain they experience (Kearney & Griffin, 2001).

Generally, less is known about parental experiences in the mid and late stages of their parental career, although many adults with intellectual disabilities continue to live in the family home well into their middle age (McConkey, 2005; Todd, Shearn, Beyer, & Felce, 1993). In the UK, it is estimated that around 50-60% of adults with intellectual disabilities are cared for by their parents (McGrother, Hauck, Bhaumik, Thorp & Taub, 1999), and around 60% of people with intellectual disabilities in the United States and 59.8% in Australia live with family caregivers (Braddock, Emerson, Felce, & Stancliffe, 2001) Caregiving for an adult is reported to be both rewarding and stressful for parents, and parental experiences of caring for their adult offspring with intellectual disabilities is an area of increasing interest. When interviewed, parents reported a felt difference between their lives and those of parents of typically developing adult offspring, and felt they were living in ‘frozen animation’ as many caregiving tasks had changed little since their offspring’s childhood (Shearn & Todd, 1997). Their parenting role had been extended indefinitely; some parents found this limiting and reported wanting a life beyond parenting, but the practicalities of taking care of their offspring somewhat prevented this (Todd & Jones, 2005). Additional worries about what would happen to their offspring after their death were also a source of concern (Todd & Shearn, 1996).

Like qualitative research on parents with children with intellectual disabilities, these more negative experiences of parents caring for adults were described along with many positive rewards, such as finding the extended parenting role very
rewarding, (Todd & Shern, 1995). Research findings also confirm the importance of considering change over time. Longitudinal research on parents of children with autism has found that parental coping strategies change between childhood and adulthood. Parents of adults with autism reported more emotion-focused coping and less reliance on family or formal services, which may reflect assimilation to having offspring with a disability, and an improvement in their offspring’s behaviour in adulthood (Gray, 2006).

Although there have been research studies focused on the experiences of parents who care for adults with intellectual disabilities, developments in the field generally relating to genetic syndromes have been rarely considered. There is a growing interest in differences related to genetic aetiology in the wider literature surrounding intellectual disabilities and the family (Hodapp, 1997). Although parental experiences of caring for children with rare syndromes has started to be explored in small numbers of both quantitative (Abbeduto et al., 2004; Sarimski, 1997) and qualitative studies (Strehle & Middlemiss, 2007), research about the experiences of parents of adults with rare syndromes is scarce and we could find no such studies published in peer review journals. Therefore, the purpose of the current study is to explore the experiences and perceptions of caring for an adult with one of three rare syndromes associated with intellectual disability: Angelman syndrome (AS), Cornelia de Lange syndrome (CdLS), and Cri du Chat syndrome (CdCS). The three syndromes are each associated with intellectual disabilities and have unique behavioural phenotypes and facial characteristics.

CdLS is estimated to affect 1:40,000 live births (Beck 1976: Beck & Fenger, 1985). The majority of individuals with CdLS have profound (45.6%) or severe (30.43%) intellectual disabilities (Berney, Ireland & Burn, 1999). Health problems are
a dominant feature of CdLS, commonly including gastro-intestinal disorders.
(Jackson, Kline, Barr & Koch, 1993). Common behavioural features include: anxiety,
oversensitivity, sensory self-stimulation, self-injurious behaviour, and compulsivity
(Basile, Villa, Selicorni, & Moltini, 2007).

Prevalence for AS is estimated at around 1:10,000 to 1:40,000 live births
(Buckley, Dinno, & Webber, 1998; Clayton-Smith, 1993). The severity of intellectual
disability ranges from moderate to profound, with severe intellectual disability being
the most common (Clarke & Marsten, 2000). Around 80% of individuals with AS
have epileptic seizures (Clayton-Smith & Laan, 2003). Frequent smiling and laughing,
hyperactivity, and sleep disorder are common behavioural features of the syndrome
(Clayton-Smith & Laan, 2003; Horsler & Oliver, 2006).

CdCS is so named because of a characteristic ‘cat-like’ cry, apparent
immediately after birth. The prevalence of CdC is estimated at 1:50,000 live births
(Niebuhr, 1978). The degree of intellectual disability ranges from profound to
moderate (Cornish & Bramble, 2002; Sarimski, 2003). Self injurious behaviour and
hyperactivity are associated with the syndrome (Cornish, Bramble, & Munir, 1998;
Cornish & Bramble, 2002).

By interviewing mothers who care for adult offspring with CdLS, AS, and
CdCS syndromes, we sought to describe mothers’ experiences. Due to the scarcity of
research in this area, this study is exploratory in nature.

Method

Methodological approach

As the current research focuses on the previously unexplored population of
mothers of adults with rare syndromes, Interpretative Phenomenological Analysis
(IPA: Smith, Jarman, & Osborn, 1999) has been used, as it is well suited to
investigating novel areas of personal experience. Unlike Grounded Theory, which examines social processes and attempts to construct an explanatory framework for the phenomenon under investigation (Willig, 2001); IPA focuses on individual experiences and psychological processes. IPA aims to reveal, not inhibit, the diversity and richness of individuals’ experiences within similar situations and the interviewee is regarded as an expert in their own experiences. It is a ‘bottom-up’ approach, and the data are coded without trying to fit them into a pre-existing theme. Therefore, some identified themes may be dissimilar to the questions that were asked of the participants (Braun & Clarke, 2006).

A key process in IPA is the dynamic interpretation of the interviews, as the researcher is active in conducting the research and in interpreting the participant’s responses. It is recognised that researcher interpretative activity is inherent in the method of IPA, and that the researchers’ own preconceptions will necessarily influence this process (Smith et al., 1999). In the current study, the primary researcher was a research student with interests in parental adjustment to having a child or adolescent with CdCS, AS, or CdLS, and who had previous experience of interviewing parents of children with intellectual disabilities over the telephone for a quantitative study focused on these same syndromes.

Typically, IPA studies use fairly small samples of five to ten participants. This facilitates the analysis as it allows the researchers to recall individual accounts, and thus helps reduce the loss of any subtle nuances between them (Smith, Michie, Stephenson & Quarral, 2002). Previously, IPA has been used to analyze samples from single case studies to studies using 30 participants (Broki & Wearden, 2006).

Participants
Eight mothers of adults with rare syndromes were interviewed. The ages of mothers ranged from 51 years to 72 years (M= 55.87 years, SD= 6.75), and their offspring ranged from 24 years to 44 years of age (M= 29.33 years, SD= 6.18). All participants were biological mothers, and all were married and living with their husbands, and had no other children with an intellectual disability (apart from one mother who had two daughters with AS). Three mothers had sons with a rare syndrome, and five had daughters. Two mothers had offspring with CdLS, three had offspring with AS (one of these mothers had two daughters with AS), and three had offspring with CdC syndrome. All names and places in the present paper have been changed or deleted to protect the identity of the participants. See Table 1 for details on individual mothers and their offspring.

----Insert Table 1 about here----

Procedure

The mothers originally responded to a mail shot campaign recruiting participants for research about challenging behaviour in children and adolescents with CdLS, AS, and CdCS. However, because their offspring did not meet the criteria for the study (because they were over the age of 19), they were invited to take part in the current study. A cover letter, information leaflet, and consent form were posted to the mothers. Of the ten mothers contacted in this manner, seven consented to take part in the study. One mother was also recruited from a syndrome support group meeting, in which information packs about the study were handed out. The only inclusion criterion was that they were the primary carer of an adult (aged over 19) with, CdLS, AS, or CdC, and there was no upper-age limit for participating in this research.

Once the mothers had returned the consent form with their written consent, a researcher contacted them to arrange a suitable interview time and to answer any
questions. Interviews were conducted over the telephone rather than face-to-face to facilitate participation, as the participants lived in various geographical locations across the UK. Interviewing via telephone has been affirmed as a useful method for conducting qualitative research (Sturge & Hanarahan, 2004), and has been used successfully in IPA research as a method of interviewing parents of individuals with intellectual disability (Reilly, Huws, Hastings, & Vaughan, 2008).

In addition to written consent, verbal consent to record the interview was gained just before the interview commenced, and all interviews were recorded on a digital recorder. They lasted from 54 to 96 minutes (Mean = 76.12 minutes) and were one-time interviews. A semi-structured interview was developed for the purpose of this study and it included subsidiary questions and prompts. The interview started with general demographic questions to help the mothers become familiar with the researcher and to feel more at ease. The remainder of the interview was designed to explore parental experiences of caring for adult offspring with a rare syndrome, questions were asked about their offspring’s transition from childhood to adulthood, social care services received, medical services, encounters with professionals, and issues surrounding the rare genetic syndrome status of their offspring. These questions were flexible to allow pursuit of any topics that arose during the interviews which had not been identified by the researcher. For example, issues around social stigma and inclusion emerged because of this flexibility, although it was not specifically asked about during the interviews (see Results). Mothers did not see a copy of the semi-structured interview, but did receive a general outline of the topics to be explored in the information sheet provided.

Data analysis
Firstly the recorded interviews were fully transcribed by the researcher. The process of transcription helps the researcher become familiar with the interviews, and some regard it as a key phase of data analysis (Bird, 2005). The researcher then read through the transcriptions line by line, noting points of interest or significance on the transcript in the left hand margin. The readings were repeated and emerging themes were noted on the right hand margin. This was repeated until the researcher was satisfied that all possible themes had been identified. These themes and supporting quotes from the interviews were compiled in a separate document, and were then grouped and connections between them were noted. This process was repeated for all eight transcripts. The themes from all the interviews were then compiled and compared, producing a list of ‘master’ themes which best represent the interviews, along with emerging sub-themes. To ensure validity of these themes, this process involved a co-author reading each transcript and developing emergent and master themes via discussion, ensuring that the themes were grounded in material from the transcripts. As is usual practice during IPA, the list of master themes changed throughout the analysis and write-up, until the final four themes emerged (Smith et al., 1999).

Results

The four master themes which emerged were: (a) Social inclusion and stigma (b) Uneven medical and social care service provision, (c) The inertia of the social care system, and (d) Mothers as advocates.

When providing extracts from the interviews the following conventions are used:
… Short pause
[text] Explanatory information provided by author
(…) Words omitted to shorten quote
Theme 1. Social inclusion and stigma

Not all mothers expressed concerns about social inclusion, as they were not specifically asked about this during the interviews. The three mothers who did talk about these issues spoke spontaneously on the subject at some length, an indication of how closely these issues are intertwined with their experiences of caring for their adult offspring.

Although all three rare syndromes are associated with specific craniofacial features which are recognisable if one is familiar with the syndrome, because public awareness is so low some young adults are not immediately perceived as having an intellectual disability. Mothers often used Down syndrome (of which public awareness is relatively high) as a comparison group when explaining the difficulties surrounding public perceptions of their offspring.

Natalie: I find with Sarah because she, to look at, unlike Down syndrome (…) there is nothing to tell you its Cri du Chat, it’s only her mannerisms that give her away. When they see her messing around they just think she’s naughty, they don’t realise.

Disapproving public attitudes make Joanna feel conflicted about her daughter’s physical appearance; she wants Julia to look nice, and this is not to try to hide her intellectual disability, but simply “to make the best of what she’s got”. However, by doing so Joanna feels she may be exacerbating the problem of negative reactions from members of the public, and finds these situations distressing and difficult to handle.

Joanna: She is normally fantastic, normally very well behaved. But there are odd occasions where she does misbehave or is rather silly… and people will have a go at me, and I think- because she doesn’t really look disabled – so they
think I’m not doing my job properly (...) so it does become quite difficult
sometimes, when people don’t realise.

The non-visibility of an intellectual disability can lead to members of the public
deciding by inappropriate standards and questioning the capability of mothers. The
attitudes of others towards their offspring were very important to mothers, and if they
had difficult encounters the event was recounted in considerable detail; an indication
of the impact the insensitivity of others can have. Natalie described a situation that
arose while she was out shopping.

Natalie: And they were giving it “She should be locked up you should have
drowned her at birth” and all this. So you can imagine my tongue ran away
(...) and I sobbed and from that day on, even now, I will not go up to town on a
Saturday for that reason and that’s been a good seven or eight years. It’s the
ignorance of people. I mean they were two elderly ladies old enough to know
better.

On a similar note, mothers talked about instances when members of the public
would stare at their offspring, but on the whole felt that attitudes were shifting
towards people with intellectual disabilities and that public awareness and
understanding was increasing.

Megan: I think that people are becoming more aware of these rare syndromes
as time goes on, I think people are more tolerant now.

Although tolerance may have increased, there are still difficulties with social
integration. Everyday social occasions were made more difficult for Sarah because of
other people’s well-intentioned attitudes, as people often speak to her mother rather
than to Sarah herself.
Natalie; Well it’s a case of “Does she take sugar? Does she want a biscuit?”

“Well I don’t know ask her, I’m not Sarah, you ask her.” She knows in a minute when you are talking stupid to her.

A major concern for some mothers was their offspring’s lack of a peer group. Due to the lack of available opportunities; one mother was involved in setting up a local network of activities for people with intellectual disabilities in order to encourage friendships. It was apparent to her that many people in her local area with intellectual disabilities were socially isolated; potentially leading to depression and health issues.

Olivia: I don’t think you can dismiss the fact that people do like to be around similar people, who laugh at the same things, you know you don’t want to be with people who are totally alien to you. And there is a great comfort isn’t there? In being with people who you have similarities with.

Theme 2. Uneven medical and social care service provision

Mothers reported widely different experiences of medical and social care provision for their offspring, although they were largely negative. Within medical services for example, half the mothers reported that they had experienced a form of prejudice against their offspring. One participant believed that the reason her son’s health care was inadequate was due to a ‘Medical system that doesn’t allow for disability’ (Olivia), whereas Natalie believed that within the medical system ‘Our kids are treated as second-class citizens’. Olivia felt the diagnostic process for her son was impeded by faults in the medical system. Over a period of five years, her son, John’s weight dropped to around 38 kilograms, and Olivia went to an enormous effort to get the medical consultant to investigate her son’s health thoroughly.
Olivia: So with the letter from the social worker going to the consultant, with the letter from the college going to the consultant, with my own doctor’s letter going to the consultant, and with the learning disability nurse coming along with us to the appointment - they started to look a bit closer.

After this effort, the consultant diagnosed John with hyperthyroidism. The length of time it took to get this diagnosis was felt to be a subtle form of prejudice that was inherent in the medical system, as the consultant did not take John’s health seriously. A further barrier to accessing healthcare was some staff’s lack of experience with people with intellectual disabilities. At times, mothers found themselves teaching healthcare staff about intellectual disabilities, which was both frustrating and time consuming.

Sophie: Well if they [medical staff] are involved in disabled people they are good (...) But if you go to other places that don’t deal with disabled people it can be different, ’cos then you have to sit and explain it all and explain yourself.

At times, this was made more difficult because of the rareness of their offsprings’ syndrome, as some medical professionals have little knowledge about rarer syndromes and as a result may not be able to give specific medical advice.

Natalie: They won’t admit to you they don’t know much about Cri du Chat. (...) And they are asking me questions about Cri du Chat, and I’m saying “Are you not supposed to be telling me? You’re the professionals, I’m just a Mum”.

Additional medical complications surrounding intellectual disability are not always addressed sufficiently. Sophie’s daughter, Charlotte was diagnosed with polycystic ovaries, but if given any medication containing artificial colours her
challenging behaviour is likely to increase for around six months. The medical
consultant’s advice on this matter seems wholly inadequate:

Sophie: Well they just said to me ‘Go to your chemist and ask them what is in
it [the medication] and find out - and then if it’s too much trouble for yourself
to handle it, because of her behaviour, then leave it out’. That’s the answer.

Some mothers felt that medical staff’s lack of experience and expertise
contributed to a lengthy diagnostic and/or treatment process for their offspring, and
that this was unacceptable practice.

Widely different experiences of social care provision were also reported by
mothers. Appropriate day centre or living placements were scarce or non-existent, so
most of the mothers reported having to take what was available in their local area and
‘making the best of it’ (Katie). The most pervasive problem with day, respite, and
residential care services was the frequent turnover of care staff, which created
multiple problems for both mothers and their offspring. Mothers believed that high
staff turnover led to challenging behaviours and a reluctance on the part of their
offspring to attend day and respite placements.

Joanna: The trouble is though the turnover of staff is so…regular (...) she gets
to know them and then they leave. Then it’s like starting over again, from
scratch because she finds it very, very difficult. Anything new, it takes her a
long time to get used to.

Social care services were often regarded as unreliable and mothers did not trust
them to deliver the agreed service.

Katie: They listen to what I say and put it down on paper, and we have action
plans and then it doesn’t materialise and then we have another hiccups and the
action plans fall by the wayside.
These situations occurred frequently and led to many mothers being uncertain about the quality of support their offspring received from social care services. Because they did not trust the day or residential care providers, many mothers felt they had to get deeply involved with them to ensure their offspring’s well-being.

Sophie’s daughter Charlotte has been in the same supported living placement for six years, during which time it was run by three different companies. The staff turnover was so high that often they were not trained, and sometimes were not even competent.

Sophie: They [staff employed by the first care provider organisation] didn’t know how to cook, they didn’t know how to wash her clothes. They dress her in summer clothes on a winter’s day or the reverse way round, there was so many problems, they stole her money the first week she was there. They kept having to change the staff, I gave complaint after complaint after complaint until they got rid of them. And then they left and they got another company in and they were there for two years and I fought tooth and nail with them- because of the care.

In addition to dealing with constant problems with day or residential care services, two mothers reported that their offspring had experienced physical abuse from social care staff. Ross came home from the day centre with bruises and he and other service users were the subject of a thorough police investigation, with two staff members being removed from their jobs as a result. Natalie’s daughter, Sarah, had been physically abused as a teenager on two occasions by care staff working at respite services. Sarah suddenly became reluctant to go to respite, and after some careful questioning by Natalie, indicated that a member of staff had been physically pushing her hand up behind her back.
Natalie: She actually showed me what they did to her. So we took it further but because Sarah, with her speech, we were basically, we were told it was her word against this other persons, so basically you haven’t got a leg to stand on have you?

Although this should have been the basis of an investigation by police and social care services, Natalie was unable to take it any further due to Sarah’s difficulties with communication. This experience left both Megan and Natalie with deep reservations about the standard of care provided by social care services.

In contrast, three mothers reported receiving good day or residential services at times, although only one mother was totally satisfied with her offspring’s supported living placement. Helen’s daughter, Lisa had been living in the same supported living home for eight years; the grounds of the home were attractive and well kept, and there was a low turnover of staff. If there were ever any issues (for example when Lisa got hit by another service user) both Lisa’s parents and social services were immediately informed, which Helen found reassuring.

Helen: She always looks well, she looks happy, she wants to go back. You know she loves coming home, and she just slots in as though she has never been away, but you take her back and they say the same.

Mothers’ most commonly reported area of satisfaction was that day centres enabled their offspring to experience a varied and full life.

Natalie: I wouldn’t mind going [to the day centre] myself. And she has a brilliant time she does horse riding, swimming, bowling, yachting, canoeing, she does all sorts, every day she’s out. It’s just up Sarah’s street, she loves it.

Likewise, three mothers felt they had received good medical care for their offspring over the years.
Grace: We’ve never had any problems when we have turned up with him anywhere, and I know some people do, but locally here, we seem to be ok really.

There appears to be an uneven quality in health and social care service provision received by these mothers, with the majority of mothers reporting a poor standard of care. However it is important to note that a minority of mothers did feel their offspring were receiving a good standard of care from social care services. This was often seen as an atypical situation, and parents frequently described themselves as ‘lucky’ to receive such provision. Likewise, Todd and Jones (2003) found that parents felt that to receive a high standard of social care was an unusual event, and also attributed this to luck.

Theme 3. The inertia of the social care system.

Social care services were regarded as a large, complex system within which mothers had to work hard to make themselves heard. Social care services seldom responded to requests unless repeatedly chased by mothers. Mothers rarely talked about individuals within social care services as being difficult or causing problems, it was the organisation of a faceless ‘system’ that they perceived as being problematic and inert. Social care services were often referred to in the third person (e.g. ‘them’ ‘they’) and this reflected a felt separateness. There was also little parental report of a collaborative effort between themselves and social care services to support their offspring. Thus, in addition to negative or positive experiences with social care services, the system itself was perceived as rigid.

Natalie: Social services is a waste of time. The excuse you get as well that they are short-staffed, it’s this it’s that it’s the other. They’ve got a lot on, and
you just basically think to yourself “I think I’m wasting my time here” and I think that’s what they hope you’re going to do, that you’re going to give up in the end, and you do, inevitably. You get cheesed off with waiting for them to call you and you calling them.

Although a role of social workers is to help parents access the services they need, getting and retaining a supportive social worker was difficult. The majority of mothers had had many different social workers over the years, and two mothers at the time of interview did not have one at all, and were told there were none available.

*Megan; You just get used to one, you fill out all the paper work “That’s fine” they say “We’ll go away we’ll do this, that, and the other”...Silence. So then we try and find out what’s going on and they say “Oh sorry that social worker’s left and there’s nobody in post” So you get those sorts of problems.*

Even from the top of the staff hierarchy at social care organisations mothers experienced problems. Joanna found that the managers of the local intellectual disability team had little experience with people with intellectual disabilities, and as a result had many misconceptions.

*Joanna: A few of the top people of the social services and [name of] County Council have never come across or never dealt with disabled people. And they are put in these positions without realising what they are going to do and they expect half of them [people with intellectual disabilities] to be able to work, and they can’t work. (...) They don’t want to know, and I’m thinking ‘You’ve got all these top jobs.’ What are they doing?’*

Problems with the hierarchy were also reported by Olivia. She found that during meetings with social care services, no single person takes control of making a
decision and ensures it is followed through; this slows down the decision-making process. A lack of an expeditor is seen as a fundamental difficulty when communicating with social services.

Olivia: *That is the whole problem with statutory services, they have no word for expedite. They just don’t understand about moving things on, and that is the whole problem with everything, (...) whatever I’ve come to, nobody has had the responsibility of making it happen, they all make judgements, but nobody is actually making sure it happens.*

As well as mothers reporting problems with the organisational structure of social care services, trying to get basic day-to-day questions answered by social care services was problematic. Despite some mothers having received services from their local authority for over a decade, they still receive an indifferent response when they try to communicate with people working for local social services.

Joanna: *Whenever I ring up I always speak to somebody different, you never speak to the same person ... but you go and ring somebody up now and ask them a question they will say “Does she have to have help? Does she need help?” Well would I be ringing if I didn’t? I mean I wouldn’t bother to ring would I?*

All mothers interviewed found communicating with social services problematic, and were aware of other issues besides the well-being of their offspring which influence care workers’ decisions. Distrust of social care services was implicit throughout many of the interviews.

Olivia: *The issue is they don’t have one agenda do they? (...) They might have my son’s interest at heart, but they also have the budget at heart and keeping*
their jobs at heart, and a lot of other things are influencing their decision as to how good my son’s care is.

Theme 4. Mothers as advocates

Mothers often referred to attempts to access appropriate social care for their offspring as ‘fights’ and a sense of ‘us’ versus ‘them’ emerged from the interviews. Mothers were forced into a role of advocate for their offspring, because if they did not advocate, they felt their offspring would receive sub-standard social care.

*Katie: The services don’t come to you, and you don’t get the changes unless you are out there fighting, which isn’t the right thing to be doing really, you don’t want to be fighting for everything you get, but sometimes it feels like that.*

Throughout the interviews mothers described having to ‘fight’ or ‘battle’ with social care or medical services to receive anything beyond minimum provision. Placements that were first offered by social services were largely found to be unsuitable – for example it was suggested that a residential home for the elderly would provide respite care for Joanna’s 30-year old daughter. This was totally unacceptable to Joanna, so she then had to ‘fight’ this in order to get suitable respite care. Social care services would only respond to mothers’ requests if they repeatedly and assertively ‘fought’ for an appropriate care service.

*Katie: You get your basics but you know if you’re wanting anything that you think is what should be acceptable for her [my daughter], then you have to have a battle about it, you have to have a battle.*

Mothers needed to be persistent in their communications with social services, and could not rely on anyone else (including social workers) to get acceptable services for their offspring. Thus, mothers were forced into a role of being advocates
and ‘fighters’. This seemed to be a large part of their identities as mothers, as the role of advocate was at a level of involvement not usually experienced by parents of typically developing offspring.

_Helen_: You have to fight all the way. You have enough of a fight often just with day to day living. And if you let things get on top of you it can be really difficult.

Mothers often felt as though they were not listened to and so were forced to go to a higher authority to get suitable services. In all, seven out of the eight mothers had reported doing this. Four mothers had contacted their local Member of Parliament (MP) about getting services for their offspring. This was done after mothers had tried and failed to get access to appropriate services for their offspring via standard routes.

_Olivia_: The only time things started to change was when I said to the MP, and said look, can you help me? And once ... the awful thing is that it takes that, for people to be doing anything.

Another parent threatened legal action against their local social services, another went to a local legal tribunal, and another went to the High Court with around 50 other families to protest against her local council trying to close all respite and day services within her local area. Some mothers felt conflicted about their role as an advocate, and felt that they were a nuisance when asking for acceptable services for their offspring. However, they also felt they had little choice but to fight on behalf of their offspring.

_Megan_: You do feel guilty at sometimes having to shout a bit loud, whereas normally I would sit back and let it go, but you have to be the spokesperson, don’t you?

_Interviewer_: So what would happen if you did sit back and not say anything?
Megan: Probably it would go all to pot really.

The frustration and negative psychological impact of dealing with the network of social services was apparent throughout most of the interviews.

Natalie: We never had any help, it was always a fight all the time to get anything done. Over the years you get so tired of getting your knickers in a twist all the time. When you ask nicely, nothing gets done. It was the Mums at the school used to say “Natalie you have to get nasty because that makes them listen”. And that proved to be correct. But it does get tiresome.

Some mothers reported that the stress of advocating for their offspring affected their health, some attributing high blood pressure or nervous breakdowns to the additional burden of constantly making sure their offspring was cared for appropriately by social care services. At the time of the interview, Sophie had been waiting for eight months for a suitable supported living placement for Ellie, her younger daughter.

Sophie: I can’t manage myself I know, I can’t carry on. I want to give it up [the care] because of my three breakdowns and I want my life back. I’ve had 30 years of it and it was easier when they were children...you think it isn’t, but it was.

Clearly, involvement with social services added a major dimension into these mothers’ lives throughout their offspring’s adult years, a dimension which was largely negative and frustrating. Some mothers found that their role as ‘fighters’ was made easier by becoming involved with the social care system in other ways, such as being a parent representative on local intellectual disability advisory boards or working for intellectual disability charities. These proactive strategies involved a lot
of commitment but mothers felt they were more likely to be listened to by social care services as a result.

*Grace:* I’ve been for years a campaigner of some form or another, (...) when you say what your name is they do know, you know, who it is. And I think you have got to be very vocal and just keep on, just keep on making yourself heard all the time.

Some mothers felt that a reputation of determination and persistence helped them be listened to by social care services; but it took years of hard work to get to this point.

*Olivia:* Am I listened to by services? Yes I am now, I am now, but that is only because half my life is spent...and they know that we are helping them. It’s not because they love me. But I would say the only reasons I am listened to is because they know that if they are not listening to me, somebody else will be.

**Discussion**

These mother’s accounts of caring for an adult with a rare syndrome highlight a number of important dimensions. They describe issues surrounding negative public reactions, and of the difficulties surrounding the social inclusion of their offspring. In relation to social care services, mothers reported largely negative experiences similar to those reported in previous research (Shearn & Todd, 1997). They described the difficulties encountered when trying to get acceptable social care for their offspring in day, respite, and residential services, having to become an advocate for their offspring, and the frustration and stress experienced in this role. Issues surrounding the rareness of their offspring’s diagnosis did not emerge as a strong theme, and possible reasons for this will be discussed further.
Social inclusion and stigma

Some mothers had experienced negative public reactions towards their offspring, which was directed towards them, as mothers. This increased mother’s self-consciousness in public places and some mothers felt angry, awkward, and frustrated as a result of such difficult encounters. Previous research suggests that this is a shared experience with mothers of both children and adults with other kinds of intellectual disabilities. When going to public places, some mothers of children with intellectual and developmental disabilities experience disproval and a lack of understanding from members of the public (Gray, 2002; Ryan, 2005). Although some parents claim dealing with negative public reactions becomes easier as their child gets older (Ryan, 2008), parents of adults with intellectual disabilities still report feeling upset and frustrated in these situations (Todd & Shearn, 1997).

Encounters were more problematic because their offspring’s disability was not immediately obvious. This is not unique to mothers of adults with rare syndromes, and has been reported in earlier research. A visible intellectual disability may moderate other people’s reactions because their expectations of that person’s behavior changes, therefore they may be less likely to judge that individual negatively (Todd & Shearn, 1997). Parents may benefit from support in how best to inform others about their offspring’s rare syndrome, or how to explain their offspring’s behavior in difficult public situations.

Some mothers were unsure about how best to help their offspring develop a social circle, and their offspring forming friendships was regarded as very important by two of the mothers. It is recognized that many people with intellectual disabilities are socially isolated, yet the friendship needs of this population have often been ignored by policy makers (Emerson & McVilly, 2004). There is scope for social care
Caring for an adult with a rare syndrome 26

services to provide opportunities for encouraging friendships among those with intellectual disabilities.

*Uneven social care and medical services*

Although social and medical services are intended to bring positive benefits for service users and their families, the current research suggests that any benefits are often coupled with effort and frustration when trying to access these services. Mothers want good services for their offspring, and not simply the bare minimum. It is here the conflict arises between mothers and social care providers. Mothers described their role as advocates as one of having to ‘fight’ social care services rather than having a collaborative partnership, and this is consistent with earlier research (Shearn & Todd, 1997; Todd & Jones, 2003).

On a more positive note, two mothers (Grace and Helen) reported that overall, they were satisfied with the support they had received from both medical and social care services. Like other mothers, both acknowledged that negotiating the social service system was difficult and required much involvement, and there was no noticeable difference between their dedication and knowledge of social services to that of the other mothers interviewed. So why did these mothers report satisfaction with social care services whereas others did not? One possible explanation, which was alluded to by several mothers, is that the quality of local social care services is dependent on the standards of the local authority, and is therefore inconsistent throughout different areas in the UK. This difference is social care provision is evident even among the eight mothers interviewed.

Parental experiences of medical services for their adult offspring with intellectual disability have not before been directly explored in qualitative research, although health discrepancies between people with and without intellectual disabilities
are well documented (Krahn, Hammond, & Turner, 2006). Health service provision for people with intellectual disabilities is known to be inconsistent (Lennox & Kerr, 1997). This was reflected in the current research, with some mothers reporting satisfaction with the medical care their offspring had received, whereas the majority of mothers reported a felt prejudice towards their offspring and reported difficulties in accessing appropriate medical care. The inexperience of some medical care staff and a failure to recognise complications arising from intellectual disability were issues highlighted by mothers. A need for specialist training and guidelines for medical staff is needed to assist them in best to addressing these issues.

**Rareness of the syndromes**

The experiences which emerged from the analysis are unlikely to be unique to mothers of adults with rare syndromes, as their accounts have much in common with mothers of adults with other types of intellectual disabilities (Shearn & Todd, 1997). Seven out of eight mothers felt the rareness of their young adult’s syndrome did not affect their access to social care services. Mothers found that their offspring were categorised as having an intellectual disability by social care services, with little recognition of their rare syndrome, yet this was not necessarily regarded as a problem by mothers.

*Grace: All they [social care services] are really interested in is that this person has a learning disability (...) I mean it doesn’t bother us in the least if he is lumped in with other people with learning disabilities, because he has a learning disability, and therefore, you know, we just live with that.*

The rareness of the syndrome did however, create additional difficulties for mothers in two areas; (i) lack of knowledge of the syndrome among medical professionals, which hindered access to medical care (this has also been referred to in the theme
‘Medical and social care service provision’) and (ii) the lack of information about likely developmental outcomes during adulthood, such as life-span, specific health risks, or behaviour changes. Some mothers felt they were ‘Living in the dark’ (Sophie) with little knowledge of what the future was for their offspring, and this was an additional source of concern for some mothers.

Mothers felt that they had the same opportunities to access social care services as other parents, and the rareness of their offspring’s intellectual disability made little difference in their relationship with social care services. However this may reflect the lack of awareness of the importance of syndrome-specific issues by many professionals, rather than the diagnostic status being of little importance in itself.

Methodological limitations

There are some methodological limitations of the current study which need to be considered. The small sample size of this exploratory study makes it difficult to construct generalizations from the findings and how representative these mother’s experiences are is unknown. Additionally, all mothers are members of the relevant rare syndrome support group, and had responded to a research recruitment leaflet. It is possible that their experiences differ to those parents who are not involved in support groups or who are unlikely to respond to research. The interpretative nature of the research was influenced by the researcher’s perspectives. This was counter-balanced with efforts to ensure that the interpretation was fully grounded in the data, by discussions about the analysis and subsequent themes with the second and third authors.

Although relatively small in scale, this study supports earlier research on the difficulties mothers experience in relation to services (Shearn & Todd, 1997). There is an ongoing need to take into account how parents regard the quality of services to
help policy makers bridge the gap between the standards of parents and those of services. Furthermore, acknowledging the day-to-day struggles that some mothers encounter with social care and medical services is important for policy makers, as additional stress may compromise some parents’ ability to continue caring for their offspring at home. An appreciation of the additional burden that social care services can bring to parents is needed in order to facilitate a mutually beneficial collaboration between parents and service providers.
References


Table 1. Characteristics of mother’s and their offspring, current living status and services received.

<table>
<thead>
<tr>
<th>Mother (age)</th>
<th>Young adult (age)</th>
<th>Diagnosis</th>
<th>Current living status and services received</th>
</tr>
</thead>
<tbody>
<tr>
<td>Natalie (55)</td>
<td>Sarah (25)</td>
<td>Cri du Chat syndrome</td>
<td>Lives in family home; goes to day centre on weekdays. Does not use respite services.</td>
</tr>
<tr>
<td>Grace (72)</td>
<td>Chris (44)</td>
<td>Cri du Chat syndrome</td>
<td>Lives in family home; goes to day centre weekdays and uses respite services.</td>
</tr>
<tr>
<td>Olivia (52)</td>
<td>John (24)</td>
<td>Cri du Chat syndrome</td>
<td>In supported living full time</td>
</tr>
<tr>
<td>Megan (52)</td>
<td>Ross (27)</td>
<td>Cornelia de Lange syndrome</td>
<td>Lives in family home; goes to day centre weekdays and uses respite services.</td>
</tr>
<tr>
<td>Joanna (55)</td>
<td>Julia (30)</td>
<td>Cornelia de Lange syndrome</td>
<td>Lives in family home; goes to day centre weekdays and uses respite services.</td>
</tr>
<tr>
<td>Katie (56)</td>
<td>Holly (32)</td>
<td>Angelman syndrome</td>
<td>In supported living full time</td>
</tr>
<tr>
<td>Helen (54)</td>
<td>Lisa (27)</td>
<td>Angelman syndrome</td>
<td>In supported living full time</td>
</tr>
<tr>
<td>Sophie (51)</td>
<td>Charlotte (30)</td>
<td>Angelman syndrome</td>
<td>In supported living full time</td>
</tr>
<tr>
<td>Ellie (24)</td>
<td></td>
<td>Angelman syndrome</td>
<td>Lives in family home; goes to day centre weekdays and uses respite services.</td>
</tr>
</tbody>
</table>