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Dementia
2020, Vol. 19(5) 1474–1491
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DOI: 10.1177/1471301218801501
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"In the flesh": Narratives of family caregivers at risk of Early-onset Familial Alzheimer's Disease

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Abstract

In the growing body of literature dealing with the consequences of family caregiving amongst people with dementia, there are few studies examining the impact of Early-onset Familial Alzheimer's Disease on caregivers. This study exposes the subjective experience of a group of family caregivers who themselves possess a genetic susceptibility to develop this form of dementia. We interviewed and analyzed the accounts of 27 caregivers belonging to family lineages carrying the E280A mutation for Early-onset Alzheimer's Disease. We utilized a phenomenological method to analyze these accounts, initially tracking seven theoretical categories (Anxiety, Depression, Burden, Resilience, Self-efficacy, Social Support, and Coping Strategies) and then subsequently two additional categories which emerged (Conceptions about the Disease and Other Vital Experiences Interfering with Caregiving). The results show that caring for a loved one while simultaneously running the risk of developing the same form of Alzheimer's Disease permeates the caregivers' experience both in a negative and a positive way. The continuous exposition to emotional stress in these caregivers should be seriously considered as they may be at risk of accelerating the onset of symptoms of Alzheimer's Disease, while simultaneously, early psychological symptoms of dementia may be masked by the emotional sequelae of

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caregiving, interfering with early diagnosis. Certainly, support services for the entire family group are suggested.

Keywords
family caregivers, Early-onset Alzheimer's Disease, E280A mutation, qualitative research

Introduction

Early-onset Alzheimer's Disease is a rare condition that affects people under 65 years of age. It represents between 5% and 10% of all Alzheimer's cases, and of these, approximately 13% correspond to Familial Alzheimer's Disease, in which a hereditary genetic predisposition leads to the condition (Campion et al., 1999). Precise data about the worldwide prevalence of Early-onset Alzheimer's Disease is not available, because the numbers vary from one region to another, and there is a presumption of underreporting due to difficulties in diagnosing this condition. However, in the United Kingdom, it is estimated that there are around 18,319 people under 65 with different types of Early-onset Dementia (EOD). The prevalence is approximately 54 per 100,000 people between 30 and 64 years old, a figure that is predicted to double approximately every five years (Harvey, Skelton-Robinson, & Rossor, 2003). And in the US, EOD is estimated to affect approximately 220,000 people – including Early-onset Alzheimer's (Alzheimer's Association, 2006).

Early-onset Familial Alzheimer's Disease follows a pattern of autosomal dominant inheritance and is related to the abnormal functioning of three genes on chromosomes 21, 14, and 1 (Amyloid Precursor Protein - PPA, presenilin 1 – PS1, and presenilin 2 – PS2, respectively). While these genes account for less than 1% of all cases of Alzheimer's Disease, they are responsible for approximately 60% to 70% of Early-onset Alzheimer's cases (Harvey et al., 2003).

The risk of developing this specific form of Alzheimer's depends on the genetic status of the parents. If one of the parents is affected or has a pathogenic variant, the risk of passing on the mutation to their children is 50% (Bird, 1993; Lopera, 2012).

Early-onset Alzheimer's Disease, whether sporadic or genetic, displays the same symptoms and follows the same phases as late-onset Alzheimer's Disease, although its evolution tends to be more rapid and aggressive, with the presence of early aphasia, myoclonus and epilepsy (Calle Bernal & Lopera, 1998). In many cases the diagnosis is complicated because the initial symptomatology can be confused with other clinical pictures and some professionals may fail to associate the symptoms with dementia when they occur in young adults.

Caring for someone with Early-onset Alzheimer's Disease

In the case of the Early-onset Alzheimer's Disease, the main caregivers are usually members of the family, especially the spouse or children who, in general, share housing with the person with dementia and fully dedicate their time to caring for their loved one (Ducharme et al., 2014; Flynn & Mulcahy, 2013).

Early-onset Alzheimer's Disease often involves a long search for a diagnosis (Roach, Drummond, & Keady, 2016; Wain, Uhlmann, Heidebrink, & Roberts, 2009), a longer period of care, more adverse effects on family organization and the financial situation of the caregiver (reduced working hours or loss of job), and an important social isolation due to the loss of companionship and activities that were once enjoyed (Campion et al., 1999; Ducharme, Kergoat, Antoine, Pasquier, & Coulombe, 2013; Lockeridge & Simpson, 2013). If the caregiver is the partner of the person with dementia, disease progression causes a redefinition of their identity and the conjugal relationship (Wawrziczny, Pasquier, Ducharme, Kergoat, & Antoine, 2015). Furthermore, emotionally it is more difficult to accept the impairment of younger individuals (Tindall & Manthorpe, 1997).

It has been reported that the caregivers of younger people have a greater emotional burden and higher levels of stress than caregivers of the elderly (Freyne, Kidd, Coen, & Lawlor, 1999; Kaiser & Panegyres, 2006). Some research indicates that younger family members find it difficult to adapt to the loss of the father or mother they once knew and, at the same time, may feel embarrassed by or fearful of their behavior (Allen, Oyebode, & Allen, 2009; Gelman & Greer, 2011). Furthermore, it is often these younger individuals who end up assuming care of their family members or partners. Due to the surprise generated by these unexpected diagnoses, the caregivers often perceive themselves as not prepared to face the task (Ducharme et al., 2014).

On the other hand, some family caregivers must deal with the emotional burden of perhaps being carriers of a genetic mutation that could give rise to dementia, and furthermore, some of them will in turn become care recipients as their own dementia condition progresses.

A family history of Alzhemer's Disease

During the 1980s and 1990s, several studies found an endemic population of Early-onset Familial Alzheimer's Disease with autosomal dominant transmission in the northwest and center of Antioquia, a region in Colombia (Lopera et al., 1994). Among seven generations, around 200 cases of Alzheimer's dementia and 25 large multigenerational families with the disease were identified. This population is considered the largest population cluster of Early-onset Familial Alzheimer's Disease in the world. The responsible genetic mutation has been identified as E280A, or "paisa" mutation, because it is found exclusively among the Antioquian population, a historically geographically isolated people who self-identify as "paisa" (Lopera et al., 1997; Sepulveda-Falla, Glatzel, & Lopera, 2012). Most of those living with this condition receive care from their relatives, who they themselves sometimes carry the mutation.

The families that carry the E280A mutation are originally of rural origin (García, Lopera, Madrigal, & Ossa, 2003), but in recent years, several of these family groups have moved into urban environments, whether for work, academic pursuit, economic reasons, or to flee the violence that affected their region over the last few decades, or even to seek better medical care for their relatives. This movement essentially dispersed E280A mutation carriers to other regions of Antioquia and Colombia. In several ways, the displacement to urban areas has benefitted both people living with dementia and their families. For example, it facilitates access to more timely diagnoses, better quality medical services, and grants or subsidies from the local government. However, the dynamics of the countryside and the city are very different, and thus the landscape of caregiving inevitably changes for these families. Most of them must work outside the home and rely on a reduced support network. Living away from their families, these caregivers, sometimes carrying the mutation themselves, are often are unaware of the existence of their disease, causing their diagnoses to take them fully by surprise. The majority of those living with this condition and their relatives are treated in the Neuroscience Group of Antioquia (GNA), the organization that studies the E280A mutation.

According to a previous study with informal caregivers of people living with Alzheimer's Disease carrying the E280A mutation who utilized the GNA care services, a tendency towards Trait Anxiety was found among children of people with this type of dementia (Méndez, Giraldo, Aguirre Acevedo, & Lopera, 2006). This supported the idea that these individuals possessed a propensity to perceive otherwise benign situations as being more threatening. Nevertheless, there is a possibility that the family bonds and subsequent risk of inheriting the disease play a part in influencing this trait.

For many of the above-mentioned reasons, this paper exposes the particular experiences of family caregivers who themselves are possible carriers of the E280A genetic mutation. The title, "In the flesh", arises from a participant who talking about the history of Alzheimer's in her family, said: "*I knew it was Alzheimer's but it wasn't like it is now... now that it has touched me in the flesh*". In this paper, we present the preliminary qualitative results of a larger mixed method study about the mediators affecting the perception of anxiety, depression, and burden of family caregivers of people with Early-onset Familial Alzheimer's Disease. Within the larger project, 50 family caregivers were interviewed, but in this paper, only the testimonies of caregivers who have a consanguineous relationship with the person with dementia and who are at risk of carrying the mutation were extracted and analyzed separately.

Method

Participants

A judgment sampling was carried out, looking for maximum information density and an exhaustive understanding of the social/emotional phenomena of interest. The participants were recruited from the GNA database and among the 50 caregivers participating in the research, 23 were excluded for not fitting the following inclusion criteria: (a) Being the main caregiver of a person with Early-onset Alzheimer's Disease who carries the E280A genetic mutation; (b) Having a consanguineous tie to the person with dementia, and (c) Not receiving financial compensation for care.

The final sample consisted of 27 caregivers (21 children, 3 sisters, 1 niece, as well as 2 wives, belonging to one of the carrier families and who are cousins of their spouse with dementia). Four sisters share equally the care of their mother (one who refrained from participating in the study), and one woman who cares for two siblings at the same time was included.

The sociodemographic characteristics are detailed in Table 1.

Data obtaining and management

A semi-structured interview was designed, and the participants' responses to the standardized scales were recorded. The interviews were conducted at the participants' homes, to interfere as little as possible with their caregiving duties. The informed consent (approved by the Bioethics Committee of the Faculty of Medicine of the University of Antioquia) was read and signed before proceeding with the data collection. The interview audio information

Table 1. Sociodemographic characteristics of participants.

Sociodemographic characteristics of caregivers		Characteristics of the person with dementia	
Gender		Gender	
Female	26	Female	21
Male	1	Male	4
Age		Age	
Mean ± SD	35±12.2	Mean ± SD	55.5±5.8
Min-max.	18–60	Min-max.	43–69
Civil status		Alzheimer's stage	
Single	16	Occasional surveillance	7
Married	9	24 hours' surveillance	3
Separated/divorced	1	24 hours' assistance	9
Widow	1	Fully functional dependency	6
Schooling			
Primary	4		
High School	13		
Technical studies	5		
College career	2		
University student	3		
Occupation			
House wife	9		
Employed	8		
Independent worker	1		
Unemployed	7		
Student	2		
Kinship			
Children	21		
Sister	3		
Niece	1		
Wife (cousin)	2		
Years caring			
Mean±SD	5±4		
Min-max.	3 months–15 years		
Days a week caring			
Mean ± SD	5±2		
Min-max.	1–7		

SD: Standard deviation.

was recorded, in some cases, photographs were taken showing strategies and accommodations at home to facilitate the daily functioning of the person with dementia. The interviews were transcribed, the text was fragmented by thematic content in order to carry out codification, categorization, and construction of models.

With NVivo 11 software, auxiliary analysis was carried out as follows: (a) Coding matrix: this procedure allowed us to examine the distribution of discourse between categories, (b) Frequency of words: the most frequent words in the narrations were consulted, excluding "stop words" (less significant words for analysis), which were verified by examining the words in context, (c) Cluster analysis: we examined the discourse according to their similarity in order to find the most important links between themes and categories.

Procedure

The data were analyzed following a phenomenological method, which aims to collect and understand the meaning that participants attribute to situations according to how they name and interpret their experiences. The scheme proposed by Miles and Huberman (1994) on qualitative data analysis was utilized as follows: (a) Data reduction: each testimony was divided into textual units according to thematic criteria and each textual unit was codified into hierarchical categories. The categories were defined both deductively with a theoretical criterion (a priori according to the dimensions under analysis) and inductively (categories that emerged from the discourse), (b) Visualization: the NVivo 11 software was used to prepare maps and graphics, (c) Conclusion: the findings were contrasted with the existing literature, (d) Verification: verification was carried out during each phase of the process.

Controlling quality of data

Categories were grouped according to how they are related and how they emerged during discourse.

The coding and categorization was reviewed by 10 expert psychologists who independently analyzed 100 fragments which were chosen at random.

Subsequently, the expert opinions were analyzed through a comparative matrix and adjustments were made to both the coding and the category system. While taking into account that the constructs addressed are multifactorial and have elements in common, to avoid overlap, a conceptual map was elaborated that helped us to visualize category similarities and differences. This allowed us to carry out the codification with greater precision.

Results

Seven interconnected theoretical categories were initially tracked and two categories later emerged while analyzing the interviews, as illustrated in Figure 1.

The emerging categories were: *Conceptions of* Early-onset Familial Alzheimer's Disease and *Other vital experiences interfering with caregiving*. Regarding the former, five subthemes were highlighted: (a) misconceptions of younger onset dementia made by other people, (b) constant changes that make it difficult to adapt to the situation, (c) the process of disease diagnosis, (d) family history with the disease, and (e) Risk of developing the disease. In the second theme, personal, family, and financial problems were mentioned. These emergent categories intersect with the theoretical categories, as discussed further on in this paper.

Moreover, Figure 2 illustrates the quantity of text units encoded in each theoretical category; this reflects the topics that contain greater and lesser amounts of discursive production.

The following components of mental health were tracked in the interviews: Anxiety, Depression and Burden.

With reference to Anxiety, the following symptoms were identified: concerns about the future and health, feelings of "being at one's limit", difficulty with attention and memory, tension (headache and neck pain, inability to relax), irritability, insomnia, increased intake of food, and motor restlessness. On the other hand, caregivers expressed that the activities of

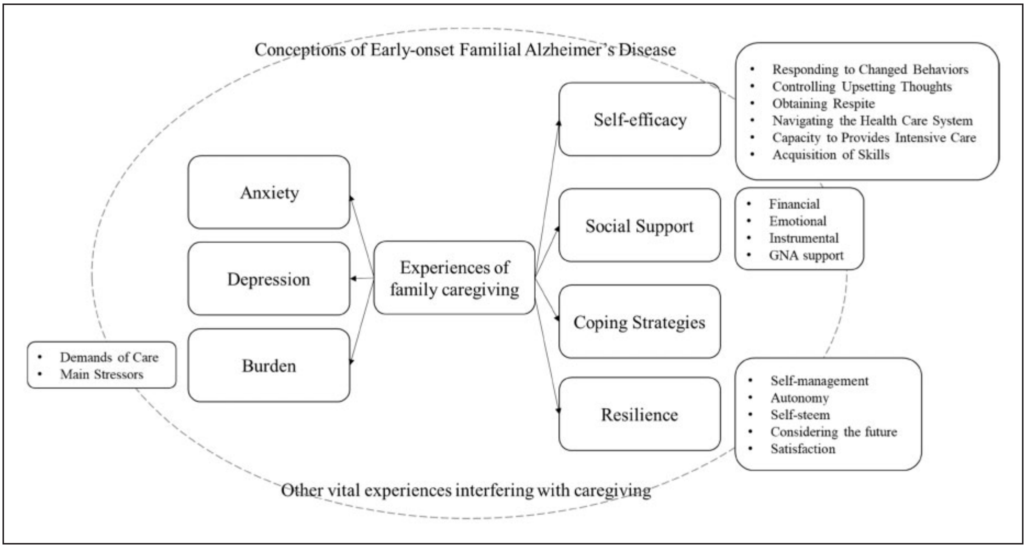


Figure 1. Concept map for categories and themes.

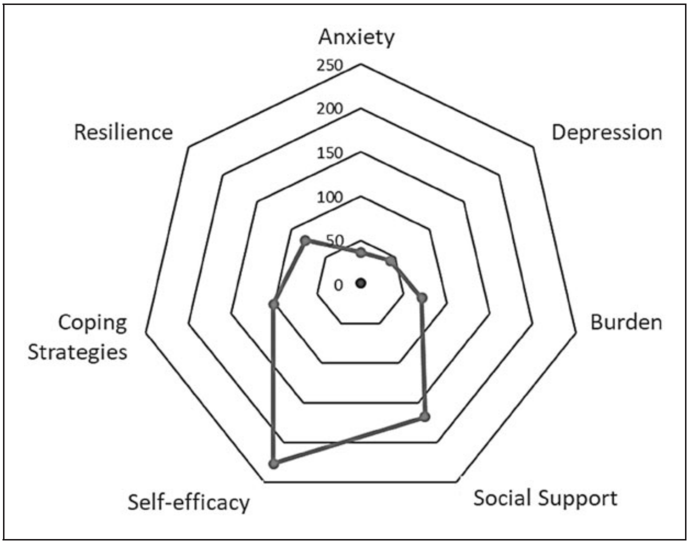


Figure 2. Radial graph with the results of the coding matrix. Nvivo 11 software.

their labor do not generate as much anxiety as other life experiences, such as family quarrels or economic difficulties. One of the caregivers was diagnosed with an anxiety disorder and is currently medicated.

Regarding Depression, four caregivers define themselves as being depressed, and two of them sought professional help and are taking medication. Several caregivers report feeling down at times, having no appetite or often being overly sensitive and crying for "any reason".

In connection with Burden, two themes were identified: *Demands of care* and *Main stressors*. Some caregivers spoke of the progressive deterioration of their relative and the growing demand for care, making it difficult to adapt completely to the situation. Just when they manage to adjust to a recent change in their loved ones functional status, and feeling that they have mastered the necessary skills for care, the disease progresses yet again and new skills must be acquired.

> The disease is progressing... I've adapted to her being in bed, bathing, dressing her, everything in the bed... Now, we're dealing with respiratory problems, we've moved on to the (gastric) tube so we had to start handling the tube. It is a disease that is progressing, every day brings something harder. (M., daughter, 36 years old)

Some caregivers distinguish that the stage where their relative requires constant surveillance and 24 hours' support is the most complex from an emotional perspective due to the changed behavior of the person with dementia, which can include mood swings, aggression, delusions, crying, ambulation, escape attempts, among others. While the stage of total dependence is often complex from a logistics and instrumental point of view: gastric tubes, avoiding bedsores, mechanical ventilation, etc., it may be more "easy to handle" emotionally.

Caregivers recognize that an important source of anxiety, depression or stress, is the sense of "confinement", feeling lonely and having few opportunities to vary ones' activities.

> Later what really stressed me out was the confinement. I felt my waist hurt, my whole body was in pain, not from caregiving but from the confinement, because it's not only being 'locked up', but people forget everything, no relative says: "Let's go to visit J. one day, or this afternoon, tonight", it is like you are totally abandoned. (J., daughter, 28 years old)

Self-efficacy

Self-efficacy in *Responding to changed behaviors*, and *Controlling upsetting thoughts* is related to how one decides to be a caregiver. Those who feel that they chose to be a caregiver tend to manifest less difficulty in handling their loved ones' behaviors, their concerns about the future and the personal sacrifices they make. For those who view their caregiving role as an imposed situation or as a result of there being "no other option", it is more difficult to cope with emotional burden of caregiving. In addition, participants more favorably viewed their ability to respond to their relatives' difficult behaviors if they "understood the disease".

Participants' self-efficacy in *Obtaining respite* is often affected by their difficulty in delegating care to someone else, often related to the lack of availability and dissatisfaction with the social support they are able to access. Caregivers who have been caring for an extended period of time learn to set aside time for themselves, they see the necessity in even simple activities such as taking a walk in the park.

> Having seen my aunt who just died in January, her caregiver spent 11 years taking care of her. She raised her daughter, buried her mother and she was there at home, confined, confined. So... at first it was like that for me [not being able to take a break from caregiving], but after seeing so

> many reflections of my life in that family I made sure that it not going to be like that anymore. (M.F., daughter, 24 years old)

Meanwhile, self-efficacy in *Navigating the health care system* for their relative is one of the areas that displays greatest improvement over time. And while some caregivers feel that they have wasted time managing social aid for their relative and finally give up, most of them know and resort to legal mechanisms for guaranteeing health services for their loved one.

Regarding the *Capacity to provides intensive care* (during late-stage dementia), by developing both a code language to communicate with their loved ones and forming a routine of stable activities, caregivers can better understand their relatives needs and can maintain their well-being.

> I already know when she's uncomfortable, if she has any pain, when she's hungry. It's like when you have babies, you say: 'she is cold, she is hungry'. I know everything about her. (G., daughter, 37 years old)

The theme, *Acquisition of skills*, is linked to the gradual acquisition of caregiving skills and the knowledge obtained from caring for previous family members with the disease.

> Most of all, I learned through caring for him [uncle], not so much through the [GNA] talks. In the talks... well, they guide you, but you really learn in the moment when you are experiencing and doing things. I learned a lot from my uncle and that is the reason why, thank God, I understand my mom so well and I know how I will take care of her. (A.L., daughter, 46 years old)

> [since] I grew up here and here they have taken care of sick relatives, I have seen ... I know. I don't have to be taught about how to take care of my aunt. (S., niece, 18 years old)

Social Support

We distinguish five themes pertaining to *Social Support*: financial, emotional, instrumental, and GNA support, as well as satisfaction with the support received. Financial help is the most common, although insufficient. Caregivers recognize that some family members "help out" with money, food, diapers, etc., but they don't want to take care of their relative with dementia even for a few moments. Finances are often a source of family disputes, often occurring due to unfulfilled commitments.

Emotional support usually comes from the caregivers' partner and the children for those who have them; otherwise, this type of support is frequently perceived as lacking. However, some caregivers with other family members who have gone through or are currently going through a similar situation do indeed provide guidance and a listening ear. It is difficult to find emotional support from people outside of the family, like neighbors or friends, due to ignorance about the condition and its implications.

> Since people see the problem from the outside, they never experience what I experience, so I feel like people never understand the problem. (N., daughter, 25 years old)

We define "instrumental support" as directly assisting in tasks such as bathing, dressing, or preparing/giving food to the relative. The caretaker perceives this as being an unshared burden because "nobody else fully assumes this role". For most participants, the drive to ask for help decreases when they do not receive an answer they expected or are not satisfied with the help they receive, until finally they give up on asking others for help with these tasks. However, participants generally express high level of satisfaction with the help provided by the GNA, via technical aids, financial support, and professional advice, as well as through workshops of cognitive stimulation and arts for the relative with Alzheimer's Disease. A caregiver who did not know about the history of the disease in his family explains the importance of receiving assistance from this group of professionals:

> I say that if it was not for him [GNA nurse], we would not have known how to take care of her. Because he was the one who explained everything to me, all the stages that she was going to pass through, he always gave me a warning. Then when she began with the aggressive behaviors, he [nurse] had already warned me so I knew how to handle the situation. Otherwise I do not know how we would have done it. (M.B., daughter, 32 years old)

Some caregivers avoid contact with the GNA when they still have difficulty coming to terms with the diagnosis or because they fear the possibility of finding out that they themselves are beginning to develop Alzheimer's Disease symptoms.

> There when we went [to GNA], they explained everything to us, they gave us a talk. When you go for medical exams, they explain many things to you. ... But, we did not want to inquire more about the Alzheimer's, no... As things get to us, with the love we have for her, we will see how to deal with it. (E., daughter, 33 years old)

Coping Strategies

The general *Coping Strategies* mentioned by the participants were: Waiting, search for social support, dealing the burden in private, cognitive avoidance, denial, patience/tolerance, dialogue/family agreement, religion/spirituality, positive reevaluation, resignification/acceptance, seeking professional support, problem solving, and humor. The difference between *positive reevaluation* and *resignification/acceptance* is that the latter does not involve looking for positive interpretations or searching for meaning, but acceptance of the situation as it is and assuming the consequences.

Regarding the possibility of inheriting the mutation, we identified three main ways our participants dealt with this: denial, over-intellectualization in attempts to stay one step ahead, and cognitive avoidance.

> I was called to participate [in a GNA project] ... but I am afraid that they will tell me: "you have it". (L., sister, 60 years old)

> Even though you don't want to get sick, you say: 'what if I get sick? what if another relative gets sick?' Then you have to be ... I don't know, one step ahead. (J., daughter, 28 years old)

> If I think about it, I get sad and I cannot live well every day, because it seems to me that you have to enjoy each day and if you don't know when you're going to die, live happily. But then, I do think long term, and if I have it, my daughter can have it. (Y., daughter, 31 years old)

Resilience

Self-management and *Autonomy* are resources that emerge from the need to resolve situations on their own. *Being a model* for others, also appears as a theme related to the social acknowledgment received for the work they do (by neighbors, family, and friends). The aspects associated with resilience that are most affected by caregiving are as follows: Self-esteem: some participants have noticed a decrease in their sense of self-worth and personal value, that sometimes is reflected in a lack of interest in personal grooming; Considering the future: as it is uncertain as to how long they will perform their role as caregiver, their initiative and ability to make plans for the future are affected; Satisfaction: participants find other aspects of their life are limited so they often feel dissatisfied not with their caregiving duties, but rather with other areas such as family, personal, or professional relationships and one's career.

According to cluster analysis, coping strategies such as religion, the search for social support, patience/tolerance, positive reevaluation, resignification/acceptance, dialogue/family agreement, problem solving, and humor seem to be related to resilience.

We also see that sense of personal growth is revealed by several of our participants.

> My vision of the world is particular, and I don't feel more than people, but I do feel with a greater objectivity, I feel I am too understanding, I even feel sometimes that I understand people more than professionals who work with them (...) because I had to face many things, and had to learn to understand my sisters. I say: the day that I live with someone, I will be very happy, because I've learned to yield, to grant, to understand. (D., daughter, 40 years old)

Representative meanings

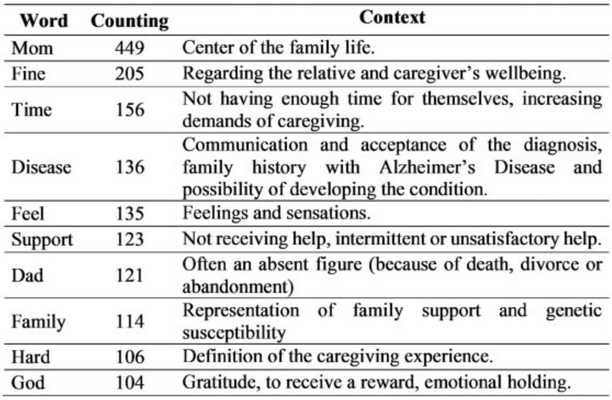
In order to delve into relevant themes, we looked for the most frequently occurring words in the interviews (Table 2).

Most caregivers are in charge of caring for their mothers and this seems to reflect the cultural value attributed to this maternal figure. In contrast, the father figure is often described as absent, uncooperative, or disengaged from his economic responsibilities. And while in some cases, the father is highlighted as participating in the care and supportive duties, his work is often viewed with dissatisfaction.

> When there is a mother and her children, there is a family. (R.A., sister, 53 years old)

> It's good [the help that the father provides] although how do I explain it? I mean, he has not learned, for example, how to change the diaper, the food ... he does try to help me but things don't work out as I expect. (M.B., daughter, 36 years old)

Table 2. Results of Word Frequency query using Nvivo 11 software.



Word	Counting	Context
Mom	449	Center of the family life.
Fine	205	Regarding the relative and caregiver's wellbeing.
Time	156	Not having enough time for themselves, increasing demands of caregiving.
Disease	136	Communication and acceptance of the diagnosis, family history with Alzheimer's Disease and possibility of developing the condition.
Feel	135	Feelings and sensations.
Support	123	Not receiving help, intermittent or unsatisfactory help.
Dad	121	Often an absent figure (because of death, divorce or abandonment)
Family	114	Representation of family support and genetic susceptibility
Hard	106	Definition of the caregiving experience.
God	104	Gratitude, to receive a reward, emotional holding.

Note: The more a specific word appears in the textual data, the bigger it appears in the word cloud.

The word "fine" often appears in context of whether their relative is "being well cared for". In interviews, the status of well-being of the person with dementia is judged on whether he or she smiles, is clean, and has the basic needs covered; or for those people with dementia who are in bed, the prevention or control of health complications. A perception of welfare in the relative gives tranquility and emotional well-being to caregivers.

The mentioning of "time" is often related to the limited time available to devote to other members of the family (husband, children), or even to themselves. Time is also mentioned in relation to the gradual increase in demands for care as a consequence of functional deterioration.

With the presence of Alzheimer's Disease, the first difficulties to arise for participants is coming to terms with the diagnosis, along with the fear of also carrying the genetic mutation. Although there is a history of familial Alzheimer's, family members do not feel prepared for the diagnosis.

> Obviously, it did come as a surprise (...) in the family we have many relatives already sick, my mom has nephews and nieces with this disease, but given my mom's age, and how she still seemed sane... because, actually, you don't believe that things like that will happen to you. (A.L., daughter, 46 years old)

The word "feel" often appears associated with the following words: lonely, guilty (for losing patience with their relative), tired, exhausted (some of them clarify that it is not physical but emotional fatigue), busy, down, frustrated, sad; as well as fear, pain, fat, skinny, or sick. However, feel has also been associated with being satisfied and happy, in fact, a sense of satisfaction appears to act as a stress buffer.

> I do feel very happy to be here, taking care of her. I don't know, sometimes it's strange how I assimilate everything (...) that doesn't mean that I don't feel sad, that I don't feel bad about seeing her like that, obviously I do, but I try to be well, especially for her. (M.B., daughter, 36 years old)

Fear of inheriting the condition has also been noted in interviews, regarding ones' ability to continue taking care of their loved ones if they themselves became ill. They also fear for someone else in the family developing Alzheimer's Disease and no longer being able to manage the situation, and finally, they fear transmitting the mutation to their children. As they get closer to the age of onset, there seems to be more concern and perhaps over interpretation of symptoms.

> So, I didn't say anything to my children but I have been starting to think: Could it be that my memory is already 'fading'? I don't know. I feel so sad about my memory. (L., sister, 60 years old)

Words "support" and "family" are related, as caretakers usually expect more help from family than friends. Caregivers express the sadness in having a big family and yet still feeling lonely. Some interpret these issues as stemming from the inherent difficulty in processing the emotions associated with the condition and their role as the caregiver.

> My little brother doesn't live with us, but I think he doesn't come home sometimes in order to not to face it (the situation). (Y., daughter, 31 years old)

Many of the caregivers who take care of their mothers' express that the illness implies the loss of the support that she used to give them.

> I would like to have my mom healthy, I have two very young children who need their grandmother (...) and also, as a daughter I would like to have [her] healthy, to receive advice from her. (A.L., daughter, 46 years old)

> When I had some difficulties I always went to her, to tell her, to ask her what I should do, what she thought, what advice she gave me, and she guided me in all those things. (A.G., daughter, 33 years old)

In several cases, participants report that increased solidarity within the family has been one of the positive consequences of the Alzheimer's Disease and that this helps to cope with the burden.

On the other hand, the caregiving experience is mostly seen as being "hard". Participants point out the difficulties in combining their various responsibilities, leaving their own projects, the experience of seeing multiple family members getting sick and dying of the same condition, as well as anticipating and overcoming successive periods of mourning with each death.

And finally, "God" emerges as an important source of emotional holding. They express gratitude for what they have, and many hope to receive material or spiritual reward for their "good labor". Religiosity also helps to make sense of their experience; sayings such as "everything happens for a reason", "if God gave us this it is for a reason", or "God knows why he did these things" are common expressed beliefs.

Conclusions and discussion

There are a few studies that address the social and emotional consequences of Early-onset Alzheimer's Disease in family caregivers, and even fewer dealing with those who poses a long history of dementia in the family. Even if there are families living with a hereditary form of Alzheimer's Disease around the world, we know very little about how their caregivers deal with their tremendous burden.

This work is one of the first studies that exposes the caregivers' experience within extended families carriers of a genetic mutation causing Early-onset Familial Alzheimer's Disease. We describe the subjective experience of care in its participants and also further analyze the ways in which this experience manifests itself physically (through fatigue, pain, loss or gaining of weight), mentally (through thoughts, ideas, preconceptions), and emotionally (Alcantud Marín, 2012; Ayala Carabajo, 2008; Moreno, 2014).

The results of this study shows that caring for a loved one while running the risk of later developing the same condition, permeates the caregivers' experience both in a negative and a positive way.

For some caregivers, witnessing the impairment of their relative is a constant reminder of a possible future with Alzheimer's Disease, and it represents an additional emotional burden. For the others, the history of family care has been vital in understanding the disease and acquiring skills of care. When there is a long tradition of care in the family, due to the presence of several generations of affected members or simply due to the extended duration of caregiving, the construction and transmission of knowledge as to how to care of his relative often takes place (de la Cuesta, 2005). Furthermore, this knowledge can eventually help buffer the tremendous burden placed on all those involved (Deví & Ruiz Almazán, 2002).

In agreement with previous research conducted by de la Cuesta (2004, 2005), Gelman (2014), and Roff et al. (2004), it was found that despite the stress caused by the disease, a deep affective relationship between the person with dementia and their caregiver can generate feelings of satisfaction and mastery, aspects that are associated with resilience (Becoña, 2006), decreasing the trend towards depression and helping to minimize the feeling of being overwhelmed (Au et al., 2010; Ávila-Toscano, García-Cuadrado, & Gaitán-Ruiz, 2010). Among interviewed caregivers, the greatest source of anxiety did not come from providing care, but from the risk of carrying the genetic mutation.

Implications for practice

It is common for people with Alzheimer's Disease to have symptoms affecting their mood. Some of the most frequently expressed emotions are anxiety, depression, tension, irritability, apathy, sadness, loss of energy, and social isolation.

Regarding depression, it has not yet been possible to determine whether it is a risk factor for developing dementia or if rather it is an early symptom, as there is evidence pointing towards both hypotheses (Deví Bastida, Puig Pomés, Jofre Font, & Fetscher Eickhoff, 2016). However, in subjects with Early-onset Familial Alzheimer's Disease, it has been found that depression can be a preclinical symptom and not necessarily a reactive depression to the perceived loss of memory (Kasuga et al., 2009; Ringman et al., 2004). This aspect should be seriously considered in relation to caregivers who belong to families that carry a genetic mutation associated with Alzheimer's Disease. First, these caregivers may be at risk of accelerating the onset of symptoms when exposed to situations of emotional stress (Bengtsson, 2013; Blair et al., 2013; Mah, Szabuniewicz, & Fiocco, 2016) and second, the onset of the symptoms of Alzheimer's Disease could create overlap. Specifically, memory loss, attentional problems, irritability, and other psychological symptoms mentioned above can be confused with the emotional consequences of care and may not be identified in time as early symptoms of dementia.

In large familial lineages where Alzheimer's Disease is caused by a genetic mutation, the continued stress generated by caring for multiple relatives with dementia significantly interferes with the quality of life of the caregiver and the family group. Caregiving indefinitely postpones their personal projects and their life plans are forced to take the backseat, since in many occasions the same caregiver "inherits" the care of multiple family members. We believe that supporting services must be made available to family caregivers after the death of their relative, as they usually require additional support to overcome successive period of mourning due to the loss of multiple family members.

In light of these results, it is important to design support programs that address the entire family group as a dynamic and changing system, not only taking the caregiver into account. These support programs should seek to: (a) help build networks of solidarity and family cohesion, to maintain the balance of the family system, and even to restore its functioning, (b) to guide the development of strategies for family adaptation with the purpose of the caregiver receiving and perceiving additional support from their relatives. Because, in the words of some caregivers, other family members do not assume any caretaking duties in order to avoid both the emotions associated with the diagnosis and managing their relative's deterioration.

Limitations and future research

This study included family caregivers living in urban areas; however, there is still a large population affected by the E280A mutation concentrated in rural areas, and hence it would be important to extend the study to these regions to contrast the results between both populations.

It would also be beneficial to conduct focus groups with psychosocial and healthcare professionals in Antioquia (Colombia), to investigate their level of knowledge about Early-onset Dementia and especially Early-onset Alzheimer's Disease, in order to establish actions that increase timely diagnoses, appropriate treatments for people with younger onset dementia and simultaneous care for the family, in coordination with the services carried out by the GNA.

Declaration of Conflicting Interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The author(s) disclosed receipt of the following financial support for the research, authorship, and/or publication of this article: This research was possible thanks to the Santander International Scholarship for doctoral studies.

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