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Burden and Parental Satisfaction in Informal Caregivers of People with Angelman Syndrome: A Mix-method Study

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Abstract

Background: Studies focusing on the burden of parents informally caring for offspring with chronic illnesses such as Angelman Syndrome are rare, despite the challenging task of parenting a person with such a disabling illness. The present article seeks to study the experience of being a parent and, simultaneously, an informal caregiver for a person with Angelman Syndrome, pursuing to investigate the effects of autonomy on the caregiver burden and how parental satisfaction mediates this relationship.

Methods: A mixed methods research was conducted. The quantitative cross-sectional study involved 24 parents of a person with Angelman Syndrome, 75% mothers (Mothers $M_{age} = 42.9$, $SD = 2.1$; Fathers $M_{age} = 50.00$, $SD = 4.4$). Five self-reported instruments were applied: sociodemographic questionnaire, Katz Index, Parental Satisfaction Scale and Zarit Caregiver Burden Scale. The qualitative study involved four parents (75% mothers), to whom we conducted a semi-structured interview.

Results: The results show that the greater the general satisfaction, the lower the feelings of burden. Also, an indirect effect of autonomy on caregiver burden through general satisfaction was found. The qualitative study revealed the presence of feelings such as burden, sadness, shock, fear, uncertainty, and hope, being the main complaints the lack of personal time and the multitude of responsibilities and activities. All data was collected during a global pandemic situation, inevitably affecting the results of the study.

Conclusion: Results confirm the emotional impact of being, simultaneously, a parent and a caregiver of a person diagnosed with Angelman Syndrome. Further studies with this population are needed to find out the risk and protective factors of symptoms of burden within this population.

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1. Introduction

Parenting is a challenging role. Even more so if the child suffers from an incapacitating illness, such as Angelman Syndrome (AS). AS is a neurological disorder with severe intellectual and motor implications that manifests itself from infancy, with sufferers presenting very specific physical characteristics (Buntix et al., 1995; Clayton-Smith & Laan, 2003; Williams & Frias, 1982). According to the European Medicines Agency (2019), this syndrome had a prevalence of one in 10.000 people in the European Union in 2019.

Evidence shows that caring for someone with a disability can have severe consequences on both the nuclear and extended family roles (Isa et al., 2016), with a particular focus on cases of chronic illness, involving higher dependency levels and a multiplicity of caregiving tasks (Sales, 2003). When the chronically ill person is a child, the informal caregivers are usually his/her parents. The situation requires an adaptation and reorganization of their lives to meet the child's needs, which can impact the family dynamics, changing their daily routines and relational experiences (Cardinali et al., 2019). Thus, there is a higher chance for the presence of complaints of mental and physical health and well-being by caregivers, comparatively to non-caregivers (Clyburn et al., 2000; Gérardin & Zech, 2019), with reports of a higher rate of hospitalizations, mortality (Pereira, 2015), stress and depression (Gérardin & Zech, 2019; Vasileiou et al., 2017). The concept of family burden becomes relevant in these cases, referring to the difficulties and challenges of caring for a dependent person, being that the primary caregiver experiences higher levels of burden (Sales, 2003).

To our knowledge, the scientific literature referring to the impact of informal caregiving in carers of people with AS is sparse, specifically regarding burden. Thus, the objectives of the present study are to examine the degree of burden of the caregivers and in what way it relates to the level of autonomy of the dependent, as well as to verify if parental satisfaction helps reduce the psychological impact of caregiving on the caregiver; to deepen the knowledge about the subjective experience of these caregivers of people with diagnosed AS.

1.1 Caregiving and Burden

In 1965, Harry Angelman reported the existence of three clinical cases of intellectual disability with specific physical characteristics, now known as Angelman Syndrome (Williams & Frias, 1982). This syndrome is characterized by unstable and rigid movement, convulsions, spontaneous laughter (Buntix et al., 1995; Williams & Frias, 1982), hyperactivity, sleep disorders (Williams, 2005), lack of speech or apraxia (Penner et al., 1993), among many other symptoms.

Due to the lack of stable locomotion, these individuals are dependent on walkers or other people to move around (Angel, 2018).

This illness presents many different degrees, and the severity of the symptoms depends on the type of chromosomal alteration present in each case (Keute et al., 2020). These individuals present a normal life expectancy (Angel, 2018), requiring constant care and supervision for the rest of their lives, due to lack of physical and intellectual autonomy (Maris & Trott, 2011; Trindade et al., 2017). Despite being incurable, there are many therapies that can help manage the symptoms and better the quality of life, such as speech therapy, physical therapy, and occupational therapy (Angel, 2018; Budisteanu et al., 2013).

The characteristics of people with AS and their daily needs, taken together, have an impact on parenting. It is the parents' duty to assure the child's psychological, physical, and social growth, as well as promote their autonomy and guarantee their survival (Barroso & Machado, 2010; Martins, 2013), providing a favourable environment to their healthy growth and development (Silva & Dessen, 2001).

When a diagnosis of disability is delivered, the parents' ideas, dreams, and expectations regarding their child, created during pregnancy (Martins, 2013) are destroyed, causing the family to go through a phase of deep anxiety and frustration, followed by grieving for the loss of this idealized child with feelings of denial, guilt and despair (Guerra et al., 2015; Martins, 2013; Russel, 2003). Then, through spirituality, social and/or family support resilience emerges, helping to overcome this suffering period (Guerra et al., 2015), allowing the arising of more positive feelings, with an acceptance of this new reality and child within the family, becoming a more inclusive environment (Martins, 2013; Silva & Dessen, 2001). Thus, new expectations and idealizations are created, adjusted to the child's actual capabilities (Martins, 2013; Russel, 2003). Some authors summarize in these five phases the described process of adaptation to the birth of a "different" child: shock, denial, and panic; anger and resentment; denial and demand; depression and discouragement; acceptance (Cook et al., 2008; Correia & Serrano, 2000; Nielsen, 1999).

Studies show that, despite the initial impact the diagnosis have on the family functioning and stress levels, once the family accept the child and their condition, these feelings lessen and family relationships are strengthened (Pinto et al., 2016), with most of the parents reporting a positive impact by the child (Singh et al., 2008).

Despite this adaptive process, the daily challenges and demands remain, with potential negative consequences such as burden, the negative impact on a person of household that comes from providing care to a dependent person, usually a family member with some medical condition (Chou, 2000; Pereira, 2015; Santos, 2019). Burden encompasses a set of psychological, physical, social, spiritual, and financial problems, capable of affecting emotional balance, family, conjugal and social relationships, work life and freedom of the caregiver (Chou, 2000; Papastavrou et al., 2007; Santos, 2019). Regardless of the diagnosis of the dependent person, research shows that the majority of caregivers have levels of intense burden (Brites et al., 2020; Trindade et al., 2017).

The concept of burden integrates two different types: objective burden and subjective burden (Chou, 2000; Hoffmann & Mitchell, 1998; Pereira, 2015; Sales, 2003). The first corresponds to the physical or practical problems that arise from caring (Hoffmann & Mitchell, 1998; Pereira, 2015; Sales, 2003). The second refers to the psychological factors, feelings and emotional perceptions of the caregiver in regard to caregiving (Chou, 2000; Pereira, 2015; Sales, 2003; Santos, 2019).

The lack of services and home support, and of support from family and friends, can cause physical, psychological, social, and financial problems, compromising the quality of life of the caregivers (Pereira, 2015; Santos, 2019). The presence of depressive symptoms, fatigue, stress, frustration, tension, problems of self-esteem (Martins et al., 2003), guilt, rage, discouragement (Chou, 2000), irritability, anxiety (Santos, 2019) and sleep disorders (Pereira, 2015) are very frequent in this population.

In a study conducted with caregivers of children with Dravet Syndrome, it was observed that caregiving caused substantial impact on their physical and emotional state, increasing their anxiety and depression levels, the feeling of difficulty in performing daily activities and decreasing their quality of life (Campbell et al., 2018).

There are cases where burden is seen as a positive and beneficial to the carers life (Trindade et al., 2017), resulting in feelings of satisfaction and realization in the face of improving the person's health (Fonseca, 2010). Although these results are not consensual, with some studies not finding a significant relationship between positive aspects of caregiving and caregiver burden (Manalel et al., 2022), they show that caregiving can be seen in a more positive perspective, based on solidarity and intimacy to the family member (Martins et al., 2003). This relationship of proximity and familiarity and the strong bonds that are established between caregiver and dependent, come to mitigate the feelings of burden and facilitate the adaptation to this new role as caregiver (Chou, 2000; Santos, 2019).

1.2 The present studies

When reviewing the literature, there are many studies dedicated to the burden in informal caregivers of elderly (Rangira et al., 2022; Ricarte, 2009; Santos, 2019), dementia (Brites et al., 2020; Clyburn et al., 2000; Goren et al., 2016; Papastavrou et al., 2007; Pereira, 2015) and with oncological issues (Bayen et al., 2017; Grunfeld et al., 2004; Li et al., 2022; Maronesi et al., 2014). However, studies dedicated to informal caregivers of offspring are more scarce (e.g. Canning et al., 1996) and focused on intellectual or developmental disabilities (e.g., Byrne et al., 2010; Dawson et al., 2021). Thus, many conditions have yet to be addressed, specifically the rarest conditions, such as Angelman Syndrome. The few studies on this type of disease tend to focus on genetic issues and diagnostic criteria (Buntix et al., 1995; Goldman et al., 2011; Grieco et al., 2018; Maris & Trott, 2011; Pelc et al., 2008; Penner et al., 1993; Summers et al., 1995; Teodoro et al., 2019; Thibert et al., 2013; Williams et al., 1995; Williams, 2005).

To our knowledge, few studies (Griffith et al., 2011; Thomson, 2011; Van den Borne et al., 1999) have focused on the negative psychological impact of informal caregiving in AS, leaving a relevant gap in the literature. In a study with parents of people with Angelman and Prader-Willi Syndrome, Thomson (2011) found that no specific coping strategy was associated with reduced stress. Griffith et al. (2011) found that the mothers and fathers of children with AS (and other two rare syndromes, Cornelia de Lange, and Cri du Chat) were more likely to report clinical levels of anxiety and depression symptoms than normative samples. Only one study focused on the emotional burden felt by these parents, related to the fear of losing control and powerlessness (Van den Borne et al., 1999).

Studies with caregivers of children with developmental problems show that one of the main factors that increase burden is the constant need for care, or dependence of the child (Chou, 2000; Marquis et al., 2019; Santos, 2019). Since the quality of life of caregivers is dependent on the functional capacities and the level of dependence of the child (Isa et al., 2016), we consider that, in caregivers of people with AS, autonomy will function as a predictor of burden.

In regard to parental satisfaction, no studies exclusively focused on parents of people with AS were found. However, the results of studies within other medical conditions demonstrate the importance of considering positive aspects of parenting to explore parental adjustment in rare genetic syndromes, such as AS (Fitzgerald et al., 2021). Bearing in mind the evidence that positive aspects of parenting seems to mediate the relationship between child behavior difficulties and parental stress (Blacher & Baker, 2007), we consider that it may also be negatively associated with parental burden and be a mediator between autonomy and parental burden.

In addition, the qualitative study will be a contribution towards a better understanding of the impact of AS on the family from the perspective of parent caregivers.

Study 1

2.1 Method

In this quantitative cross-sectional study, which involves a non-probabilistic purposive sample of parents of individuals with AS, four hypotheses were tested:

H1: the level of autonomy of the person with AS will predict the level of burden of his parent caregiver.

H2: parental satisfaction will be negatively associated with parental burden.

H3: parental satisfaction will mediate the association between autonomy and parental burden.

An online self-report questionnaire was created, encompassing all the research instruments, and distributed to AS associations all over Europe, having obtained answers from five European countries (Portugal, Slovenia, Croatia, Sweden, and Finland). The research was approved by the Ethics Committee of CIP-Psychology Research Centre of Universidade Autónoma de Lisboa.

2.1.1 Participants

The sample is comprised of 24 parents of persons with AS, from which 75% were women. Women had, on average, 42.9 years old ($SD= 2.1$) and men, 50 years old ($SD= 4.4$). Most of the participants held a university degree (79.2%), were married (79.2%) and were employed (83.3%). The average household of the participants was composed of 2.9 people ($SD=0.9$). The participants had, on average, 2.3 children ($SD=1$; $Min. = 1$; $Max. = 6$), one of whom was the person with Angelman. The age of this person ranged from 3 to 37 years ($M=14.7$; $SD=11.3$). When being caregivers, participants spent an average of 15.5 hours ($SD = 6.8$; $Min. = 4$; $Max. = 24$) per day caring for their child with Angelman, on average 6 days per week ($SD= 0.9$; $Min. = 3.5$; $Max. = 7$). People with Angelman's benefit, on average, from 3.71 ($SD = 1.9$, $Min. = 1$; $Max. = 8$) external assistances, such as physiotherapy, psychology, speech therapy, hippotherapy, among others.

2.1.2 Instruments

Sociodemographic and disease-related data was obtained through a brief questionnaire in order to collect personal and contextual information about the caregivers' reality. This 24-item questionnaire was comprised of two sections: the caregiver section, entailing nine direct

questions and six open questions, and the dependant section, comprised of four direct questions and three open questions.

The autonomy of the person suffering from AS was assessed through the Katz Index of Independence in Activities of Daily Living, which was previously used by Castelhana (2012) in a population of children caregivers, due to the similarities of the activities measured to the patterns of child development. This index measures independence in six different activities (bathing, dressing, toileting, transferring, continence and feeding) and each of these is evaluated according to the caregivers' observation of the dependent's capabilities, being classified as independent those who can do a certain action without supervision or assistance (Duarte et al., 2007). Higher scores on this measure means lower autonomy (being 1 the score for full autonomy).

Parental Satisfaction was evaluated through the Parent Satisfaction Scale by Halverson and Duke (1991), comprised of 30 items distributed in four dimensions: General Satisfaction (comprising all the items; item example: "Having children compensates for all sacrifices"), Parental Pleasures (10 items; item example: "I feel happy as a mother/father, in general"), Importance (8 items; item example: "Motherhood/fatherhood is the most important aspect of life") and Burdens (2 items; item example: "My kids limit my freedom") (Andrade, 2015; Martins, 2008). Responses were rated on a seven-point Likert scale ranging from 1 = always disagree to 7 = always agree (Martins, 2008). This measure shows good internal consistency in all dimensions, except for Burdens (General Satisfaction, $\alpha = .897$, Parental Pleasure, $\alpha = .901$, Importance, $\alpha = .824$, Burdens, $\alpha = .405$) (Andrade, 2015; Martins, 2008). In the present study, the Cronbach alpha was .95 for the General satisfaction, .93 for Parental pleasures, .90 for Importance and .33 for Burdens. This last dimension was removed from the analyses because of the unsatisfactory reliability value.

Caregiver Burden was measured by the Zarit Burden Interview, a 22-item scale (example item: "*Do you feel stressed by having to divide yourself between caring for your family member and your other responsibilities (work/family)?*") with responses based on a 5-point Likert scale (ranging from 1 = Never to 5 = Nearly Always) (Sequeira, 2010). This scale presents good reliability ($\alpha = .93$). In this study, Cronbach's alpha was .91.

2.1.3 Data Analysis

Data were analysed with SPSS 26.0. Descriptive statistics and Spearman correlations were used to examine study variables and their correlations. The mediation model was tested using the PROCESS macro for SPSS (model 4) (Hayes, 2015). Three simple mediation models were

examined: the independent variable was autonomy; mediators were the three dimensions of parental satisfaction (general satisfaction, parenting pleasures and parenting importance); the dependent variable was caregiver burden. Five thousand bootstrap resamples were used to estimate direct and indirect effect. Indirect effects were considered significant when the 95% confidence intervals (CIs) did not include zero. Unstandardized betas were reported.

2.3 Results

2.3.1 Descriptive Statistics and Correlations

Descriptive statistics such as means and standard deviations, as well as bivariate Spearman correlations are included in table 1. Autonomy was negatively associated with general satisfaction. General satisfaction was negatively associated with caregiver burden and positively associated with the others parental satisfaction' dimensions. Burden was negatively associated with all parental satisfaction dimensions.

Table 1. Means, Standard-deviation, and Bivariate Spearman Correlations among Study Variables (N = 24)

	M (SD)	1.	2.	3.	4.	5.
1. Caregiver's burden	63.83 (16.04)	-				
2. Parenting pleasures	83.33 (13.93)	-.663**	-			
3. Parenting importance	79.17 (17.00)	-.442*	.809**	-		
4. General satisfaction	71.33 (12.26)	-.669**	.786**	.671**	-	
5. Autonomy	5.71 (1.52)	.178	-.198	-.143	-.419*	-

Note. * $p < .05$; ** $p < .01$

2.3.2 Mediation Model

A mediation model stating that autonomy should be related to burden via general satisfaction was tested. Although there was no direct effect of autonomy on caregiver burden [$t = -0.72$, $p = .48$], a significant indirect effect was found when general satisfaction was added as a mediator [$F(2, 21) = 9.56$, $p < .001$], explaining 48% of its variance (Table 2). The remaining parental

satisfaction dimensions did not function as mediators (respectively, $F(1, 22) = 1.15, p = .30$ for parenting pleasures and $F(1, 22) = 0.54, p = .47$ for parenting importance).

Table 2. Direct, indirect, and total effects of autonomy on burden through parental satisfaction (N = 24)

	Coeff	SE	t	p	LLCI	ULCI
Aut -> GS	-3.08	1.59	-1.94	.07	-6.38	0.22
GS -> Burden	-0.95	0.22	-4.26	.000**	-1.42	-0.49
Total effect	1.64	2.23	0.74	.47	-2.98	6.26
Direct effect	-1.30	1.81	-.72	.48	-5.05	2.46
Indirect effects	Effect	Boo			Boo 95%	Boo 95%
		SE			LLCI	ULCI
Aut -> GS -> Burden	2.94	1.61			0.41	6.64
R² = .48						

Note. Significant effects are in bold. ** $p < .01$; * $p < .05$; Aut = autonomy; GS = General satisfaction; Coeff = coefficient; SE = standard error; LLCI = lower level of the 95% confidence intervals; ULCI = upper level of the 95% confidence intervals; Boo = Bootstrap results.

2.4 Discussion

The present study aimed to examine the association between the level of autonomy of the person suffering from AS and the caregiver burden, as well as verify the mediation role of parental satisfaction in this association. Specifically, it aimed to test: whether autonomy functioned as an indicator of burden (H1); whether there was an association between parental satisfaction and burden (H2); whether parental satisfaction functioned as a mediator between autonomy and parental burden (H3).

Although no significant association was found between autonomy of the AS person and caregiver burden, a significant moderate negative association was found between general satisfaction and caregiver's burden (confirming H2 but not H1). Also, an indirect effect between autonomy and caregiver burden was found, suggesting that autonomy predicts caregiver's burden through general satisfaction (confirming H3). Higher levels of autonomy are associated with higher parental satisfaction, which in turn is associated with lower levels caregiver burden.

The absence of a direct effect of autonomy on caregiver's burden was not expected, although half of the caregivers did show intense levels of burden, in accordance with the current scientific literature within other caregiving contexts (Brites et al., 2020; Trindade et al., 2017). However, this can be related to the caregivers' ability to adapt to the caregiving routine, becoming more

confident in their capabilities and diminishing their feelings of burden (Araújo et al., 2017; Gratão et al., 2013). It is also possible that, by adapting to the caregiving reality and routine, caregivers' perspective of burden is no longer affected by the dependants' lack of autonomy (Ricarte, 2009). In addition, the feeling of burden can be associated with other variables other than the autonomy of the person with AS, such as the increase in financial needs (Pereira, 2015), the lack of time for leisure, the management of all medical necessities and the detriment of family and/or social relations (Santos, 2019).

In fact, parental satisfaction was negatively associated with caregiver burden. Feelings of parental satisfaction depend on the parents' perspective of their abilities, i.e. their perceived self-efficacy (Lynch, 2002; Martins, 2008). The higher this perception, the greater the confidence in meeting the child's needs and dealing with his/her daily struggles (Lynch, 2002) and, consequently, the lower the levels of stress (Aranda, 2013; Hastings & Brown, 2002).

Finally, general satisfaction had an indirect effect on the relationship between the level of autonomy of the person with AS and caregivers' burden. There is evidence of the relationship between low levels of parental satisfaction and high levels of parental stress (Samadi et al., 2020) and of the mediating effect of the positive aspects of parenting on the relationship between children's behavioural difficulties and parental stress (Blacher & Baker, 2007). Parental satisfaction seems to function as a regulator for parenting struggles associated with the child's condition, in this case, AS.

We hypothesize that the autonomy of the person with AS can facilitate the adoption of a more positive perspective on parenting, enhancing a more positive and adaptive attitude (Albuquerque et al., 2013) and feelings of satisfaction. Often, caregivers feel pride, satisfaction, and fulfillment in caregiving, forming a greater intimacy with the dependent person (Fonseca, 2010; Santos, 2019; Trindade et al., 2017), generating positive feelings that combat the parents' feelings of burden (Chou, 2000; Santos, 2019).

Study 2

3. Method

The qualitative study involves a subgroup of parents, to which interviews were conducted to obtain a more in-depth view of the subjective experience of taking care of a dependent with AS. A total of three recorded interviews were conducted through zoom (a Portuguese couple, a Portuguese mother, and a Slovenian mother), due to the ongoing pandemic and the geographical distance, and were later transcribed for analysis.

According to Cardinali and colleagues (2019), a family member with a chronic condition leads to major changes in family life, due to the constant and daily caregiving needs, redefining relationships, daily routines, and perception of future to each family member. A semi-structured interview, developed by Cardinali and colleagues (2019), was used to better comprehend these changes, focusing in three main areas: the personal experience of caring for a sick child (*Can you tell me your story since your child was born?; How did you experienced getting the diagnosis?; How do you care for your child?*), family change (*How did the family change when the child was diagnosed?*) and the perception of social support (*Who helps and supports you? How do you feel that support?*).

The interviews were analyzed following the thematic approach by Braun and Clark, which seeks to identify patterns (themes) within the collected data, organizing them (topics) and describing them in detail (Braun & Clark, 2006). Thematic analysis was carried out independently by two researchers. A consensus was reached after a subsequent joint analysis.

First, the two researchers familiarised themselves with the procedures involved in the thematic analysis. Afterwards, the transcripts were analysed, and themes present in all the interviews were defined, in a procedure called coding. This process consists of the production of codes that succinctly describe the events reported in the data (Braun & Clark, 2006), that is, in the case of an interviewee reporting that he discussed the issue of their child's diagnosis with family members, the code would be "1. discussed with family members".

After coding, excerpts for each code were extracted from the interviews, and they were then reviewed, keeping only the most relevant points for the theme. Then, the various codes were grouped into larger themes, arriving at three key themes. Finally, a thematic map to illustrate the themes and their constituents was elaborated (Figure 1).

3.2 Results and Discussion

Three key themes were reached after the thematic analysis, namely the diagnosis process, the impact of caregiving and hope. Each theme is composed of smaller sub-themes, as seen in Figure 1.

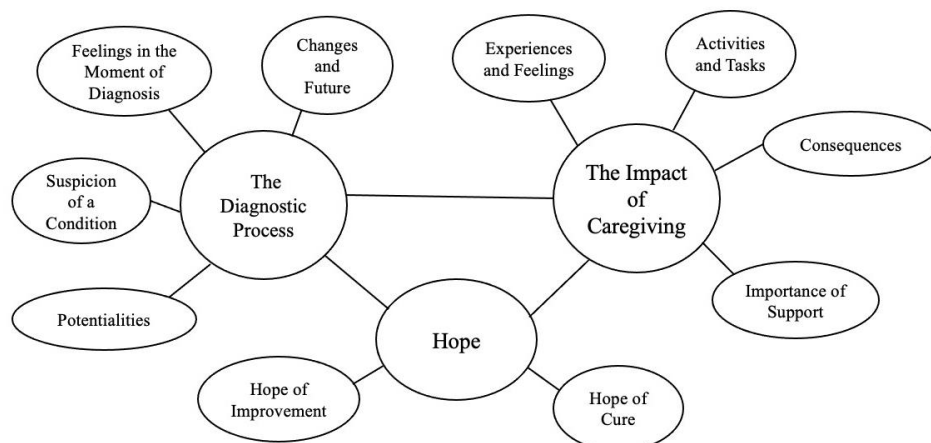


Figure 1. Final Thematic Map: Themes and sub-themes emerged from thematic analysis

Theme 1: Diagnosis process

The diagnosis process, mainly the period between the *suspicious* and the diagnosis confirmation, was described as a very difficult time for parents. A mother says:

My father-in-law, he is a doctor, he is a bit annoying, and he started to tell us that "my grandson has something, he is not being well accompanied, we should go to another pediatrician". (C, mother of a 2 y/o son)

According to Bonis (2016), parental stress levels tend to increase during the search for answers that explain the developmental challenges of their child:

The hardest thing about everything, for me was the waiting time, I was always very anxious, I would get a lot of eczema, I had a lot of flushes, my body was showing that I was really in a very stressful state. (C, mother of a 2 y/o son)

Receiving of the diagnosis had an *emotional impact on the parents*, being a moment of sadness, anxiety and loss of idealized hopes and expectations for the child, but also of relief, due to the waiting time until the confirmation of the diagnosis:

When we went to the consultation and had the confirmation, I won't say that I didn't cry, but on the other hand we cried, but also with some relief, like, we reached the end of this path to be able to confirm that A. has Angelman syndrome". (C, mother of a 2 y/o son)

This loss is a part of the grieving process, requiring a readaptation to the parents' new reality (Martins, 2013). In an initial phase of shock and denial, there is a tendency to maintain the hope that the diagnosis is wrong, and later feelings of revolt and rejection arise, that only through acceptance of this new reality and emergence of more positive feelings, is the child truly accepted and integrated into the family community, occurring the development of tolerance, empathy, and patience (Martins, 2013; Silva & Dressen, 2001):

It's better to accept the situation as it is (pause) and make the most of it? (M, mother of a 7 y/o daughter)

Fear, worry and uncertainty for the *future of the child with AS* was also very prevalent in this first theme, as well as the parents' *potentials to adapt* to the new reality and necessities of their child.

Of course, I have moments when we get very sad, when I think about the future, that's when I get sadder (...) but that's it, or when I think about the limitations that we are all going to live eventually, but above all, they are already mapped out in A.'s life. A." (C, mother of a 2 y/o son)

This uncertainty causes fear and concern to parents, feelings also verified by Delve and collaborators (2006), in which high levels of concern for the child's future was one of the central points of the parents' stress. In turn, the adjustment demonstrated by the interviewed parents, demonstrate their innate potentialities, where, according to Brites (2010), there is a progressive modeling and differentiation that seeks to integrate the most appropriate experiences to the organisms needs.

No matter how many tears we sometimes have, we dry our tears, we roll up our sleeves and every day, we don't have time to cry, because we every morning think: "what are we going to do to help A more?" (C, mother of a 2 y/o son)

Theme 2: impact of caregiving

Regarding the impact of caregiving, parents report feeling burdened by the quantity of daily responsibilities, *necessities, activities, and tasks* that caregiving for their child intakes, negatively impacting the parents lives on a professional and social level, as well as their physical and mental health.

These are things that always stress us a little... it's part of our routine but we always have to say: we can't forget this, otherwise it's serious (C, mother of a 2y/o son)

This constant need for care by children with disabilities, is one of the causes of increase in caregiver burden (Chou, 2000; Marquis et al., 2019; Santos, 2019), as well as stress levels and negative psychological, emotional, and physical *impact in caregiver* well-being (Clyburn et al., 2000; Martins, 2013; Samadi et al., 2020).

I feel that I am not a good mother, I am not a good professional, I am nothing because as I have to do everything (...) I can't do anything well (C, mother of a 2 y/o son)

Lack of time for personal and social endeavours was another issue presented by the interviewees, which can have harmful effects in physical and mental health, and productivity of caregivers (Bayen et al., 2017; Clyburn et al., 2000; Pereira, 2015).

So it's a burden in that sense, we don't have time to do also, things (pause) that we want or that we feel like. (D, father of a 2 y/o son)

The *reduced offer of specific supports* and therapies for AS and children in general, as well as a lack in scientific research and information for this syndrome, are some of the difficulties highlighted by parents. These are some of the main issues among cases of rare diseases, as well as the difficulty in obtaining a diagnosis and the management of daily care needs (Delve et al., 2006).

In terms of what Angelman syndrome is there is not much information either, not many, for example indications of almost, what kind of therapies we can try and see how the children react and then, the offer for children is not much either. (S, mother of a 5 y/o daughter)

Theme 3: hope

The last theme refers to the hope felt by parents and extended family regarding the *child's future abilities and acquisitions*, but also *hope for a cure* that comes to “fix” the child.

We are very positive (laughs) in fact, we believe that A. will really surprise us a lot. (C, mother of a 2 y/o son)

He (the ex-husband) tries to believe, you know, that sometime in the future, but the pharmaceutical industry, they will develop a drug to save K. (M, mother of a 7 y/o daughter)

This hope for the child's abilities can provide a more positive outlook on life and help the parents adapt to the uncertainties of their current situation (Truitt et al., 2011). This theme links to the previous themes, being present since diagnosis and remaining over time being associated with the evolution of the dependents' capabilities.

Given the pandemic context in which data were collected, it was also important to contextualize its effects on these families and their caregiving experiences. Reports were divergent: some parents saw this as positive, being able to dedicate most of their time to developing activities with their children:

So it could be what I wanted, which was to be a 100% mother (C, mother of a 2 y/o son)

Other parents have a less positive approach, emphasizing the lack of support and activities to help the child grow and develop basic abilities, as well as lack of personal time:

unfortunately now we have the lockdown and hum (pause) it's again you know, this structure is interrupted and she's at home with us (pause) now we're struggling, now we, it's (pause) for her and for us it's a very hard time.
(M, mother of a 7 y/o daughter)

Thus, the lack of support and therapies that help dependents progress, fell entirely on the parents, who are now full-time caregivers, burdening them even more and compromising their quality of life (Santos, 2019).

4. General Discussion

The psychological indicators associated with chronic illnesses are extended at different levels (Merlo, 2019), going beyond the person diagnosed and reaching the people closest to them, the family members who care for them. To our knowledge, this is one of the first studies that sought to exclusively study some psychological variables associated with the burden of informal caregivers of people with AS, confirming the presence of high levels of burden and highlighting the importance assigned by them to the parental dimension of their lives. The results show that being a parent and, at the same time, an informal caregiver of a person with a chronic and rare disease are not distinct and independent dimensions of these people's lives. On the contrary, the evidence seems to show that the two dimensions overlap, with parenting having a particular influence on caregiving.

The quantitative study showed that the lack of autonomy of the person with Angelman syndrome was not directly associated with the burden of their caregiver. On the contrary, the interviews evidenced the presence of feelings of burden on parents, due to the constant needs and dependence of their children to perform daily activities. In previous studies, positive associations between levels of burden and stress and greater dependence on long-term care have been found (Canning et al., 1996; Martins, 2013; Sales, 2003).

The quantitative results also showed that the greater the perceived satisfaction with the parental role, the lower the effects of burden on parents, in line with Prieto and colleagues (2022) study. They also indicated that autonomy was associated with parental satisfaction, which, in turn, was associated with a lower burden. These conclusions were corroborated by the qualitative study, in which feelings of parental efficacy in relation to caregiving were determinants of caregivers' motivation. Although there are studies that have not found a significant association between burden and positive aspects of caregiving (Mananel et al., 2022), our parents' feelings of efficacy

are closely linked to their perceived parental satisfaction, decreasing their feelings of stress, something that seems to decrease their burden (Aranda, 2013; Hastings & Brown, 2002).

This study also deepened the knowledge about the subjective experience of caring for someone with a disabling condition, understanding the complexity of feelings and the duality of experiences (negative vs. positive). It suggests that there are many variables, other than lack of autonomy, that can cause burden, which should be addressed in future studies.

5. Conclusion

The strength of this research is its contribution to knowledge about the psychological impact of informal care for people with AS, a rare and profoundly disabling disease, considering the results obtained from both quantitative and qualitative data. From a clinical Psychology point of view, it's relevant to address the psychological mechanisms associated to medical chronic conditions, both in patients and in their caregivers. Although the psychological research in medical settings has focused mainly on patients (e.g., Caputo et al., 2022; Conversano & Giuseppe, 2021; Di Giacomo et al., 2019; Shahar, 2020; Sirri et al., 2007), evidence shows the importance of studying caregivers experience related to chronic disease (Canning et al., 1996; Clyburn et al., 2000; Gérain & Zech, 2019; Pereira, 2015; Vasileiou et al., 2017), such as AS.

As for the limitations of this study, in the case of the quantitative study, the small sample size restricted the number and type of possible statistical analyses, having focused only on the mediation effects. The fact that this was a cross-sectional study also limited the possibility of establishing possible causal relationships between the variables. Finally, the lack of specificity and exhaustiveness of the Katz Index (used to assess autonomy) may have contributed to the results, specifically to the lack of a direct relationship between autonomy and burden.

Regarding the qualitative study, the reduced number of interviews may have restricted the diversity of the themes raised, failing to reach thematic saturation, since no other families were available to participate in the study. The fact that one of the interviews was conducted in a different language may also have influenced the analysis of the responses.

Finally, the fact that all the data was collected in a global pandemic situation may have affected all the participants, making the results obtained specific to the "moment", and it is impossible to determine how different they would be if the circumstances were different.

The results obtained by this study can be considered as a starting point for further studies with this population, with the need to examine the predictor variables and the possible presence of burden symptoms, as well as internal resources that may act as protective factors, in this

situation of vulnerability where people must be, simultaneously, parent and informal caregiver. Research often focuses only on the negative or the positive aspects of the caregiving process. The described results underline the need to develop comprehensive assessment protocols that consider the different dimensions and consequences of caregiving, the risk factors related with stress, but also those mechanisms that allow the person to pursue a good adaptation (Frisone et al., 2021), as a starting point for adequate and effective interventions with this population.

Ethical approval *: The research was approved by the Ethics Committee of the Research Centre where authors are affiliated (Approval 6/2020).

Informed Consent Statement: Informed consent was obtained from all subjects involved in the study.

Data Availability Statement *: The data that support the findings of this study are available from the corresponding author, [RB], upon reasonable request.

Conflict of interest statement *: The authors declare no conflict of interest.

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Author Contributions *: TF: this author developed the research model, contacted the institutions involved and collected all the data. She participated in the qualitative analysis.

RB: was the coordinator of the project where this research is integrated. She conducted the quantitative analysis and the second qualitative analysis.

TB: This author was responsible for the literature review, general discussion and conclusion.

ON: this author participated in the literature review. She was also responsible for the discussion of the qualitative study.

JH: this author participated in the literature review. He was also responsible for the discussion of the quantitative study.

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References

1. Albuquerque, S., Pereira, M., Fonseca, A., & Canavarro, M. (2013). *Deficiência e parentalidade: A influência das perceções de contribuições positivas dos pais na sobrecarga percebida e na qualidade de vida* [Disability and parenting: The influence of perceptions of positive contributions of parents on perceived burden and quality of life]. Atas do II Congresso Ibero-Americano / III Luso-Brasileiro de Psicologia da Saúde. Faro: Universidade do Algarve.
2. Andrade, N. M. (2015). *A influência do bem-estar das mães no desenvolvimento dos filhos na primeira infância* [The influence of mothers' well-being on the development of children in early childhood]. [Dissertação de Mestrado, Universidade Autónoma de Lisboa]. Repositório Camões. <https://repositorio.ual.pt/handle/11144/2445>
3. Angel. (2018). Campanha informativa sobre Síndrome de Angelman [Informative campaign regarding Angelman syndrome]. http://angel.pt/Sabia_que_completo.pdf
4. Aranda, C. (2013). *An ecological investigation of contextual factors and cognitions that impact parental responsivity for low-income mothers of preschool-age children*. [Doctorate Thesis, University of Oregon Graduate School]. Scholar's Bank. <https://scholarsbank.uoregon.edu/xmlui/handle/1794/13399>
5. Araújo, J., Cirne, G., Lima, N., Cavalcanti, F., Cacho, E., & Cacho, R. (2017). Sobrecarga de cuidadores familiares e independência funcional de pacientes pós-acidente vascular encefálico. *Revista de Ciências Médicas*, 25(107). <https://doi.org/10.24220/2318-0897v25n3a2991>
6. Barroso, R., & Machado, C. (2010). Definições, dimensões e determinantes da parentalidade. *Psychologia*, 52(1), 211-229. http://dx.doi.org/10.14195/1647-8606_52-1_10
7. Bayen, E., Laigle-Donadey, F., Prouté, M., Hoang-Xuan, K., Joël, M., & Delattre, J. (2017). The multidimensional burden of informal caregivers in primary malignant brain tumor. *Support Care Cancer*, 25, 245-253. <https://doi.org/10.1007/s00520-016-3397-6>
8. Bonis, S. (2016). Stresse and parents of children with autism: A review of literature. *Issues in Mental Health Nursing*, 37(3), 153-163. <http://dx.doi.org/10.3109/01612840.2015.1116030>
9. Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, 3(2), 77-101. <https://doi.org/10.1191/1478088706qp063oa>
10. Brites, R. (2010). *Parentalidade, auto-estima e auto-eficácia: A situação de doença crónica de um filho* [Parenting, self-esteem and self-efficacy: The chronic illness situation of a child]. [Tese de Doutoramento, Universidade do Algarve]. Sapientia: Repositório da Universidade do Algarve. <https://sapientia.ualg.pt/handle/10400.1/10621>
11. Brites, R., Brandão, T., Pereira, F., Hipólito, J., & Nunes, O. (2020). Effects of supporting patients with dementia: A study with dyads. *Perspectives on Psychiatric Care*, 56, 614- 620. <https://doi.org/10.1111/ppc.12476>
12. Budisteanu, M., Papuc, S. M., Tutulan-Cunita, A., Craiu, D., Barca, D., Iliescu, C., & Arghir, A. (2013). Angelman syndrome patient management: 5 years of clinical experience. *International Journal on Disability and Human Development*, 12(3). <https://doi.org/10.1515/ijdh-2012-0107>
13. Buntix, I. M., Hennekam, R. C., Brouwer, O. F., Stroink, H., Beuten, J., Mangelschots, K., & Fryns, J. P. (1995). Clinical profile of Angelman Syndrome at different ages. *American Journal of Medical Genetics*, 56, 176-183. <https://doi.org/10.1002/ajmg.1320560213>

14. Byrne, M., Hurley, D., Daly, L., & Cunningham, C. (2010). Health status of caregivers of children with cerebral palsy. *Child: Care, Health and Development*, 36(5), 696-702.
<https://doi.org/10.1111/j.1365-2214.2009.01047.x>
15. Campbell, J., Whittington, M., Kim, C., VanderVeen, G., Knupp, K., & Gammaitoni, A. (2018). Assessing the impact of caring for a child with Dravet syndrome: Results of a caregiver survey. *Epilepsy & Behavior*, 80, 152-156. <https://doi.org/10.1016/j.yebeh.2018.01.003>
16. Canning, R. D., Harris, E. S., & Kelleher, K. J. (1996). Factors predicting distress among caregivers to children with chronic medical conditions. *Journal of Pediatric Psychology*, 21(5), 735-749.
<http://doi.org/doi:10.1093/jpepsy/21.5.735>
17. Caputo, A., Vicario, C. M., Cazzato, V., & Martino, G. (2022). Editorial on the research topic psychological factors as determinants of medical conditions-Volume II. *Frontiers in Psychology*, 643.
<https://doi.org/10.3389/fpsyg.2022.865235>
18. Cardinali, P., Migliorini, L., & Rania, N. (2019). The caregiving experiences of fathers and mothers of children with rare diseases in Italy: Challenges and social support perceptions. *Frontiers in Psychology*, 10(1780), 1-14.
<https://doi.org/10.3389/fpsyg.2019.01780>
19. Castelhana, P. J. (2012). *A sobrecarga dos pais enquanto cuidadores principais da criança com espinha bífida [The burden of parents as primary caregivers of the child with spina bífida]*. [Dissertação de Mestrado, Escola Superior de Enfermagem de Coimbra]. Repositório Científico da ESEnfC.
<https://repositorio.esenfc.pt/rc/index.php?module=repository&target=list&clear=1>
20. Chou, K. (2000). Caregiver burden: A concept analysis. *Journal of Pediatric Nursing*, 15(6), 398-407.
<https://doi.org/10.1053/jpdn.2000.16709>
21. Clayton-Smith, J., & Laan, L. (2003). Angelman syndrome: a review of clinical and genetic aspects. *Journal of Medical Genetics*, 40, 87-95. <http://doi.org/10.1136/jmg.40.2.87>
22. Clyburn, L., Stones, M., Hadjistavropoulos, T., & Tuokko, H. (2000). Predicting caregiver burden and depression in Alzheimer's disease. *Journal of Gerontology*, 55(1), 2-13. <https://doi.org/10.1093/geronb/55.1.s2>
23. Conversano, C., & Di Giuseppe, M. (2021). Psychological factors as determinants of chronic conditions: clinical and psychodynamic advances. *Frontiers in Psychology*, 12, 635708.
<https://doi.org/10.3389/fpsyg.2021.635708>
24. Cook, R., Klein, M. & Tessier, A. (2008). *Adapting Early Childhood Curricula for Children with Special Needs* (7th Ed.). Pearson.
25. Correia, L. M., & Serrano, A.M. (Orgs), (2000). *Envolvimento Parental em Intervenção Precoce. Das Práticas Centradas na Criança, às Práticas Centradas na Família*. Porto Editora.
26. Dawson, C., Aryeetey, G., Agyemang, S., Mensah, K., Addo, R., & Nonvignon, J. (2021). Costs, burden and quality of life associated with informal caregiving for children with Lymphoma attending a tertiary hospital in Ghana. *International Journal of Care Coordination*, 23(4), 165-172.
<https://doi.org/10.1177/2053434520981357>

27. Delve, L., Samuelsson, L., Talborn, A., Fasth, A., & Hallberg, L. (2006). Stresse and well-being among parents of children with rare diseases: a prospective intervention study. *Journal of Advanced Nursing*, 53(4), 392-402. <https://doi.org/10.1111/j.1365-2648.2006.03736.x>
28. Di Giacomo, D., Ranieri, J., Nasta, L., Moscato, S., Guerra, F., & Passafiume, D. (2019). Psychological distress in Interstitial Cystitis/Bladder Pain Syndrome: A cross-sectional study on emotional patterns. *Mediterranean Journal of Clinical Psychology*, 7(2). <https://doi.org/10.6092/2282-1619/2019.7.2135>
29. Duarte, Y. A., Andrade, C. L., & Lebrão, M. L. (2007). O índice de Katz na avaliação da funcionalidade dos idosos. *Revista da Escola de Enfermagem da USP*, 41(2), 317-325. <https://doi.org/10.1590/S0080-62342007000200021>
30. European Medicines Agency. (2019). *Public summary of opinion on orphan designation Gaboxadol monohydrate for the treatment of Angelman syndrome*. https://www.ema.europa.eu/en/documents/orphan-designation/eu/3/19/2172-public-summary-opinion-orphan-designation-gaboxadol-monohydrate-treatment-angelman-syndrome_en.pdf
31. Fitzgerald, J., Wilson, C., Kelly, C., & Gallagher, L. (2021). More than a box of puzzles: Understanding the parental experience of having a child with a rare genetic condition. *European Journal of Medical Genetics*, 64(4), 1-9. <https://doi.org/10.1016/j.ejmg.2021.104164>
32. Fonseca, T. S. (2010). *Sobrecarga, depressão e generatividade em mulheres cuidadoras informais [Burden, depression and generativeness in women informal caregivers]*. [Dissertação de Mestrado, Faculdade de Psicologia]. Repositório da Universidade de Lisboa. <https://repositorio.ul.pt/handle/10451/2495>
33. Frisone, F., Sicari, F., Settineri, S., & Merlo, E. M. (2021). Clinical psychological assessment of stress: a narrative review of the last 5 years. *Clinical Neuropsychiatry*, 18(2), 91-100. <https://doi.org/10.36131/cnfioritieditore20210203>
34. Gérard, P., & Zech, E. (2019). Informal caregiver burnout? Development of a theoretical framework to understand the impact of caregiving. *Frontiers in Psychology*, 10:1748. <http://doi.org/10.3389/fpsyg.2019.01748>
35. Goldman, S. E., Bichell, T. J., Surdyka, K., & Malow, B. A. (2011). Sleep in children and adolescents with Angelman syndrome: association with parent sleep and stress. *Journal of Intellectual Disability Research*, 56(6), 600–608. <https://doi.org/10.1111/j.1365-2788.2011.01499.x>
36. Goren, A., Montgomery, W., Kahle-Wroblewski, K., Nakamura, T., & Ueda, K. (2016). Impact of caring for persons with Alzheimer's disease or dementia on caregivers' health outcomes: findings from a community-based survey in Japan. *BMC Geriatrics*, 16(122). <http://dx.doi.org/10.1186/s12877-016-0298-y>
37. Gratão, A., Talmelli, L., Figueiredo, L., Rosset, I., Freitas, C., & Rodrigues, R. (2013). Dependência funcional de idosos e a sobrecarga do cuidador. *Revista da Escola de Enfermagem da USP*, 47(1), 137-144. <https://doi.org/10.1590/S0080-62342013000100017>
38. Grieco, J. C., Bahr, R. H., Schoenberg, M. R., Conover, L., Mackie, L. N., & Weeber, E. J. (2018). Quantitative Measurement of Communication Ability in Children with Angelman Syndrome. *Journal of Applied Research in Intellectual Disabilities: JARID*, 31(1), e49–e58. <https://doi.org/10.1111/jar.12305>

39. Griffith, G. M., Hastings, R. P., Oliver, C., Howlin, P., Moss, J., Petty, J., & Tunnicliffe, P. (2011). Psychological well-being in parents of children with Angelman, Cornelia de Lange and Cri du Chat syndromes. *Journal of Intellectual Disability Research*, 55(4), 397–410.
<https://doi.org/10.1111/j.1365-2788.2011.01386.x>
40. Grunfeld, E., Coyle, D., Whelan, T., Clinch, J., Reyno, L., Earl, C., Willan, A., Viola, R., Coristine, M., Janz, T., & Glossop, R. (2004). Family caregiver burden: Results of a longitudinal study of breast cancer patients and their principal caregivers. *Canadian Medical Association Journal*, 170(12), 1795-1801.
<https://doi.org/10.1503/cmaj.1031205>
41. Guerra, C., Dias, M., Filha, M., Andrade, F., Reichert, A., & Araújo, V. (2015). Do sonho a realidade: Vivência de mães de filhos com deficiência. *Texto & Contexto Enfermagem*, 24(2), 459-466.
<http://dx.doi.org/10.1590/0104-07072015000992014>
42. Halverson, C. F., & Duke, H. P. (1991). Parent Satisfaction scale. In J. Touliatos, B. Perlmutter, & G. Holden (Eds) (2001). *Handbook of Family Measurement Techniques: Abstracts, Volume 2* (pp. 272). SAGE.
43. Hastings, R., & Brown, T. (2002). Behavior problems of children with Autism, parental self- efficacy and mental health. *American Journal on Mental Retardation*, 107(3), 222-232.
[https://doi.org/10.1352/0895-8017\(2002\)107<0222:BPOCWA>2.0.CO;2](https://doi.org/10.1352/0895-8017(2002)107<0222:BPOCWA>2.0.CO;2)
44. Hayes, A. (2015). An index and test of linear moderated mediation. *Multivariate Behavioral Research*, 50(1), 1-22. <https://doi.org/10.1080/00273171.2014.962683>
45. Hoffmann, R., & Mitchell, A. (1998). Caregiver burden: Historical development. *Nursing Forum*, 33(4), 5-11.
<https://doi.org/10.1111/j.1744-6198.1998.tb00223.x>
46. Isa, S., Ishak, I., Rahman, A., Saat, N., Din, N., Lubis, S., & Ismail, M. (2016). Health and quality of life among the caregivers of children with disabilities: A review of literature. *Asian Journal of Psychiatry*, 23.
<http://dx.doi.org/10.1016/j.ajp.2016.07.007>
47. Keute, M., Miller, M. T., Krishnan, M., Sadhwani, A., Chamberlain, S., Thibert, R. L., Tan, W., Bird, L. M., & Hipp, J. F. (2020). Angelman Syndrome genotypes manifest varying degrees of clinical severity and developmental impairment. *Molecular Psychiatry*. <https://doi.org/10.1038/s41380-020-0858-6>
48. Li, Y., Li, J., Zhang, Y., Ding, Y., & Hu, X. (2022). The effectiveness of e-Health interventions on caregiver burden, depression, and quality of life in informal caregivers of patients with cancer: A systematic review and meta-analysis of randomized controlled trials. *International journal of nursing studies*, 104179.
<https://doi.org/10.1016/j.ijnurstu.2022.104179>
49. Lynch, J. (2002). Parents' self-efficacy beliefs, parents' gender, children reader self-perceptions, reading achievement and gender. *Journal of Research in Reading*, 25(1), 54-67.
<https://doi.org/10.1111/1467-9817.00158>
50. Manalel, J. A., Sumrall, S., Davidson, H., Grewal, M., Granovetter, M. A., & Koehly, L. M. (2022). Stress, coping, and positive aspects of caregiving among caregivers of children with rare disease. *Psychology & Health*, 1-17. <https://doi.org/10.1080/08870446.2022.2057494>
51. Maris, A. F., & Trott, A. (2011). A patogênese genética e molecular da Síndrome de Angelman. *Jornal Brasileiro de Psiquiatria*, 60(4), 321-330. <http://doi.org/10.1590/S0047-20852011000400014>

52. Maronesi, L., Silva, N., Cantu, S., & Santos, A. (2014). Indicadores de estresse e sobrecarga em cuidadores formais e informais de pacientes oncológicos. *Estudos e Pesquisas em Psicologia*, 14(3), 877-892.
53. Marquis, S., Hayes M., & McGrail, K. (2019). Factors affecting the health of caregivers of children who have an intellectual/developmental disability. *Journal of Policy and Practice in the Intellectual Disabilities*, 16(3), 201-216. <https://doi.org/10.1111/jppi.12283>
54. Martins, M. (2013). *Competências parentais em pais com filhos portadores de deficiência: Um estudo descritivo*. [Masters Dissertation, Faculdade de Ciências Humanas e Sociais, Universidade do Algarve, Faro]. Sapientia Repositório da Universidade do Algarve. <https://sapientia.ualg.pt/handle/10400.1/3614>
55. Martins, S. M. (2008). *Satisfação parental e impacto familiar – Contribuição para a validação de dois instrumentos*. [Masters Dissertation, Instituto Superior de Psicologia Aplicada]. Repositório do ISPA. <http://repositorio.ispa.pt/handle/10400.12/669?mode=full>
56. Martins, T., Ribeiro, J., & Garrett, C. (2003). Estudo de validação do questionário de avaliação da sobrecarga para cuidadores informais. *Psicologia, Saúde e Doenças*, 4(1), 131-148.
57. Merlo, E. M. (2019). Opinion Article: The role of psychological features in chronic diseases, advancements and perspectives. *Mediterranean Journal of Clinical Psychology*, 7(3). <http://doi.org/10.6092/2282-1619/2019.7.2341>
58. Nielsen, L. (1999). *Necessidades Educativas Especiais na Sala de Aulas: Um Guia para Professores*. Porto Editora.
59. Papastavrou, E., Kalokerinou, A., Papacostas, S., Tsangari, T., & Sourtzi, P. (2007). Caring for a relative with dementia: Family caregiver burden. *Journal of Advanced Nursing*, 58(5), 446-457. <https://doi.org/10.1111/j.1365-2648.2007.04250.x>
60. Pelc, K., Boyd, S. G., Cheron, G., & Dan, B. (2008). Epilepsy in Angelman syndrome. *Seizure*, 17(3), 211–217. <https://doi.org/10.1016/j.seizure.2007.08.004>
61. Penner, K. A., Johnston, J., Faircloth, B. H., Irish, P., & Williams, C. A. (1993). Communication, cognition and social interaction in the Angelman Syndrome. *American Journal of Medical Genetics*, 46, 34-39. <http://doi.org/10.1002/ajmg.1320460108>
62. Pereira, F. (2015). *Necessidades dos cuidadores de doentes com demência*. [Doctorate Thesis, Universidade Autónoma de Lisboa, Lisboa] Camões Repositório Institucional da Universidade Autónoma de Lisboa. <https://repositorio.ual.pt/handle/11144/1870?locale=en>
63. Pinto, R., Torquato, I., Reichert, A., Neto, V., & Saraiva, A. (2016). Autismo infantil: Impacto do diagnóstico e repercussões nas relações. *Revista Gaúcha de Enfermagem*, 37(3). <http://dx.doi.org/10.1590/1983-1447.2016.03.61572>
64. Prieto, V., Rozmus, C., Cohen, E., & LoBiondo-Wood, G. (2022). Caregiver burden, caregiving satisfaction, and health-related quality of life among caregivers of children with medical complexity. *Pediatric Nursing*, 48(3), 111-121.
65. Rangira, D., Najeeb, H., Shune, S. E., & Namasivayam-MacDonald, A. (2022). Understanding Burden in Caregivers of Adults with Dysphagia: A Systematic Review. *American Journal of Speech-Language Pathology*, 31(1), 486-501. https://doi.org/10.1044/2021_AJSLP-21-00249

66. Ricarte, L. (2009). *Sobrecarga do cuidador informal de idosos dependentes no concelho da Ribeira Grande*. [Dissertação de Mestrado, Instituto de Ciências Biomédicas de Abel Salazar da Universidade do Porto, Porto]. Repositório Aberto da Universidade do Porto. <https://repositorio-aberto.up.pt/handle/10216/19131?locale=pt>
67. Russel, F. (2003). The expectations of parents of disabled children. *British Journal of Special Education*, 30(3), 144-149. <https://doi.org/10.1111/1467-8527.00300>
68. Sales, E. (2003). Family burden and quality of life. *Quality of Life Research*, 12(1), 33-41. <https://doi.org/10.1023/A:1023513218433>
69. Samadi, S. A., Abdollahi-Boghrabadi, G., & McConkey, R. (2020). Parental satisfaction with caregiving among parents of children with autism spectrum disorders, attention deficit and hyperactivity, intellectual disabilities and typically developing. *Early Child Development and Care*, 190(7), 1115-1122. <https://doi.org/10.1080/03004430.2018.1518903>
70. Santos, A. (2019). *Sobrecarga e depressão em cuidadores informais de idosos dependentes*. [Dissertação de Mestrado, Universidade Autónoma de Lisboa, Lisboa]. Repositório Camões. <https://repositorio.ual.pt/handle/11144/4327>
71. Sequeira, C. A. (2010). Adaptação e validação da escala de Sobrecarga do Cuidador de Zarit. *Referência*, 12, 9-16.
72. Shahar, G. (2020). Interdisciplinarity and integration: an introduction to the special issue on psychopathology in medical settings. *Journal of Clinical Psychology in Medical Settings*, 28(1), 1-5. <https://doi.org/10.1007/s10880-020-09752-2>
73. Silva, N., & Dessen, M. (2001). Deficiência mental e família: Implicações para o desenvolvimento da criança. *Psicologia: Teoria e Pesquisa*, 17(2), 133-141. <https://dx.doi.org/10.1590/S0102-37722001000200005>
74. Singh, T., India, V., & India, R. R. (2008). Impact of disability of mentally retarded persons on their parents. *Indian Journal of Psychological Medicine*, 30(2), 98-104. <https://doi.org/10.1177/0975156420080208>
75. Sirri, L., Fabbri, S., Fava, G. A., & Sonino, N. (2007). New strategies in the assessment of psychological factors affecting medical conditions. *Journal of Personality Assessment*, 89(3), 216-228. <http://dx.doi.org/10.1080/00223890701629649>
76. Summers, J. A., Alison, D. B., Lynch, P. S., & Sandler, L. (1995). Behaviour problems in Angelman Syndrome. *Journal of Intellectual Disability Research*, 39(2), 97-106. <https://doi.org/10.1111/j.1365-2788.1995.tb00477.x>
77. Teodoro, A. T., Chaves, D. Y., Crenitte, P. A., Hage, S. R., & Lamônica, D. A. (2019). Linguagem, neurodesenvolvimento e comportamento na Síndrome de Angelman: Relato de caso. *Codas*, 31(4). <http://doi.org/10.1590/2317-1782/20182018177>
78. Thibert, R. L., Larson, A. M., Hsieh, D. T., Raby, A. R., & Thiele, E. A. (2013). Neurologic Manifestations of Angelman Syndrome. *Pediatric Neurology*, 48(4), 271-279. <https://doi.org/10.1016/j.pediatrneurol.2012.09.015>
79. Thomson, A. K. (2011). *Parental and carer responses to Angelman syndrome and Prader-Willi syndrome*. [Doctorate Thesis, School of Exercise, Biomedical and Health Sciences]. Edith Cowan University Institutional Repository.

80. Trindade, I., Almeida, D., Romão, M., Rocha, S., Fernandes, S., Varela, V., & Braga, M. (2017). Caracterização do grau de sobrecarga dos cuidadores de utentes dependentes da Unidade de Saúde Familiar USF Descobertas. *Revista Portuguesa de Medicina Geral e Familiar*, 33, 178-186.
<http://dx.doi.org/10.32385/rpmgf.v33i3.12160>
81. Truitt, M., Biesecker, B., Capone, G., Bailey, T., & Erby, L. (2011). The role of hope in adaptation to uncertainty: The experience of caregivers of children with Down syndrome. *Patient Education and Counseling*, 87, 233-238. <https://doi.org/10.1016/j.pec.2011.08.015>
82. Van den Borne, H., Van Hooren, R., Van Gestel, M., Rienmeijer, P., Fryns, J., & Curfs, L. (1999). Psychosocial problems, coping strategies, and the need for information of parents of children with Prader–Willi syndrome and Angelman syndrome. *Patient Education and Counseling*, 38, 205–216.
[https://doi.org/10.1016/S0738-3991\(99\)00004-X](https://doi.org/10.1016/S0738-3991(99)00004-X)
83. Vasileiou, K., Barnett, J., Barreto, M., Vines, J., Atkinson, M., Lawson, S., & Wilson, M. (2017). Experiences of loneliness associated with being an informal caregiver: A qualitative investigation. *Frontiers in Psychology*, 8(585). <http://dx.doi.org/10.3389/fpsyg.2017.00585>
84. Willgoss, T., Cassater, D., Connor, S., Krishnan, M. L., Miller, M. T., Dias-Barbosa, C., Phillips, D., McCormack, J., Bird, L. M., Burdine, R. D., Claridge, S., & Bichell, T. J. (2020). Measuring what matters to individuals with angelman syndrome and their families: Development of a patient-centered disease concept model. *Child Psychiatry & Human Development*. <https://doi.org/10.1007/s10578-020-01051-z>
85. Williams, C. A. (2005). Neurological aspects of the Angelman Syndrome. *Brain & Development*, 27, 88-94.
<http://doi.org/10.1016/j.braindev.2003.09.014>
86. Williams, C. A., & Frias, J. L. (1982). The Angelman (“happy puppet”) Syndrome. *American Journal of Medical Genetics*, 11, 453-460. <http://doi.org/10.1002/ajmg.1320110411>
87. Williams, C. A., Angelman, H., Clayton-Smith, J., Driscoll, D. J., Hendrickson, J. E., Knoll, J. H., Magenis, R. E., Schinzel, A., Wagstaff, J., Whidden, E. M., & Zori, R. T. (1995). Angelman Syndrome: Consensus for diagnostic criteria. *American Journal of Medical Genetics*, 56, 237-238.
<https://doi.org/10.1002/ajmg.1320560224>
88. Williams, C. A., Zori, R. T., Hendrickson, J., Stalker, H., Marum, T., Whidden, E., & Driscoll, D. J. (1995). Angelman syndrome. *Current Problems in Pediatrics*, 25(7), 216-231. [https://doi.org/10.1016/S0045-9380\(06\)80036-8](https://doi.org/10.1016/S0045-9380(06)80036-8)



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