

RESEARCH

Open Access



Communicating the Spinal Muscular Atrophy diagnosis to children and the principle of autonomy

Isabella Araujo Mota Fernandes^{1,2*}, Renata Oliveira Almeida Menezes³ and Guilhermina Rego¹

Abstract

Introduction: The trinomial relationship between physicians/children/guardians is essential in the process of communicating a disease and its prognosis.

Objective: Analyzing the exercise of autonomy by this trinomial relationship in communicating the diagnosis of spinal muscular atrophy (SMA).

Methodology: Caregivers of SMA patients answered a questionnaire containing a structured interview and the Event Impact Scale – Revised.

Results: The sample comprised 50 volunteers, 94% of whom were female caregivers. Psychological trauma was predominantly reported when caregivers communicated the diagnosis to children. 22% have a high risk of post-traumatic stress, relating the feeling of unpreparedness in communicating this to the child.

Conclusions: It was identified that the failure in communication is the main factor for negative repercussions on the autonomy of children and their guardians, with self-reported psychological trauma, besides the high risk for post-traumatic stress syndrome.

Keywords: Bioethics, Communication, Physician–Patient Relations, Spinal muscular atrophy

Introduction

The patient-physician relationship is documented by Hippocrates since the beginning of medicine, and it is surrounded by characteristics intrinsically related to the principles of bioethics, autonomy, beneficence, non-maleficence, and justice; emphasizing the care and well-being of the patient, through a wide knowledge of their current and previous history, avoiding behaviors that bring them harm [1]. The essence of this connection has importance and impact until the present day, especially

when it comes to progressive, degenerative, and disabling diseases.

Spinal muscular atrophy (SMA) is a rare autosomal recessive and neurodegenerative genetic disease, with the age of onset in early childhood, generally. Its pathophysiology results from the absence of the SMN1 gene, on chromosome 5, leading to the deficiency or absence of the SMN protein, responsible for the survival of motoneurons [2]. SMA is characterized by a progressive and variable loss of strength, predominantly appendicular and proximal, limiting the ability to walk and perform manual activities. Furthermore, a particular deficit in the axial and bulbar muscles was sometimes found, which may lead to the inability to speak, eat orally, and breathe, without compromising cognition [2, 3]. SMA has five clinical subtypes, with an earlier onset of clinical signs

*Correspondence: isabella_mota@yahoo.com.br

² Lauro Wanderley University Hospital, Federal University of Paraíba, Cidade Universitária Campus I, 58051-900 João Pessoa, Paraíba, Brasil
Full list of author information is available at the end of the article



and maximum motor function acquired pre-pharmacological intervention being associated with more severity [2, 3].

Communicating the diagnosis of a serious illness, such as SMA, requires an understanding of the patient in terms of cultural background, social context, autonomy in decision-making and resilience abilities [4, 5]. Aspects such as availability, care, compassion, empathy, communication skills, and personal characteristics, such as patience, kindness, calm, good humor, and security increase the bond with the patient and are essential for the strengthening and continuity of this relationship [6]. Interpersonal continuity of care generates a more individualized medicine and more satisfied patients [7].

When the diagnosis involves a child, there is the emergence of a doctor-child-guardian trinomial relationship [8]. In addition to the aforementioned aspects, trust and familiarity are essential for child involvement [8]. Cognitive maturity does not always match the chronological one and, therefore, the ability to consent must be ascertained in agreement with the possibility of abstraction and understanding of information [9]. According to a theory of the mature minor, if the capacity and moral maturity of children or adolescents is demonstrated, their autonomy and individuality must be respected [10], and, as a result, their decision-making regarding general aspects of their lives [11].

The literature on models for communicating the diagnosis of neurodegenerative diseases to children is limited [12–14]. The transmission of this information about SMA is especially challenging for doctors, as children face progressive functional losses, and parents, besides the responsibility of transmitting information about the diagnosis to their children, have their parental model and the relationship with the child and with others affected.

Because of this, we aim to analyze, from the main point of view of bioethical autonomy, the communication of the SMA diagnosis to children and/or adolescents, looking at the perception of family caregivers. The relevance of expanding knowledge on this topic is to enable the development of communication techniques to reduce the negative impact generated by poor communication for those involved.

Methodology

The ethics and research committee of the HULW-UFPB, Brazil, has approved this research under the number 5.176.679. The invitation to take part in the research and completion of the form took place between September 2020 and January 2022. This is a cross-sectional study with simple random sampling. Caregivers of patients with SMA, present at the time of this diagnosis, were invited, through patient associations throughout the

country, to participate. The contacts of the volunteers were forwarded to the main researcher and, after signing the free and informed consent form, data collection began.

Communication took place via teleconsultation. At first, a structured interview was prepared and forward to families/caregivers included in the study. The interview was prepared after reviewing the literature related to the topic [12–, 13–18], in addition to the Event Impact Scale – Revised to assess post-traumatic stress related to the moment of diagnosis. The Event Impact Scale – Revised assessment score ranges from 0 to 88, with 0 to 23 representing no risk, 24 to 32 low risk, 33 to 36 a probable diagnosis, and 37–88 high risk of post-traumatic stress syndrome [19]. This structured interview is available in the [supplementary material](#).

Data were categorized and tabulated in a digital spreadsheet for further descriptive and inferential statistical analysis. The R software, version 4.1.1, was used and a significance of 5% was considered. Descriptive analysis was represented by measurements of absolute and relative frequency, besides measures of central tendencies, such as mean and standard deviation. For inferential analysis, initially the Kolmogorov–Smirnov test was performed to prove the normality of the data, later the Fisher's Exact test and Pearson's correlation test were performed [20].

Results

The sample is composed of 50 family members of patients with SMA, 94% (47) comprised female caregivers, 86% (43/50) were mothers, and 8% (4/50) aunts or others. Among men, 6% (3/50) were fathers. Their average age was of 38.60 (± 11.64) years old, with variable family income, without predominance of social classes, throughout the Brazilian territory (Table 1). When asked about the time between symptom onset and diagnostic completion, 54% of respondents reported a time greater than one year between the two events. Among the patient subtypes, 18% presented SMA type 1, 48% SMA type 2, and 34% SMA type 3.

When volunteers were asked about the investigation of the disease until its diagnosis; with the possible answers being yes, no, or partially; the results were that 42% (12) of them felt included in the health services they sought, 78% (39) believed that the environment was adequate, and 68% (34) understood the diagnosis (Table 2).

The physician communicated the diagnosis to parents in 100% (50/50) of cases, but in only 22% (11/50) children/adolescents were present. When asked who informed the children/adolescents about the diagnosis, 76% (38/50) were parents without the presence of the doctor, and, of these, 81.6% (31/38) of parents reported

Table 1 Sample characterization of family caregivers of patients diagnosed with SMA

Variables	Caregiver	
	n	%
Sex		
Male	3	6,0%
Female	47	94,0%
Color		
Yellow	2	4,0%
White	24	48,0%
Black	4	8,0%
Brown	19	38,0%
Other	1	2,0%
Education		
Illiterate	1	2,0%
Complete primary education	2	4,0%
Incomplete primary education	5	10,0%
Complete high school	17	34,0%
Incomplete high school	2	4,0%
Higher education	11	22,0%
Postgraduate studies	12	24,0%
Region from Brazil		
Center-West	2	4,0%
Northeast	28	56,0%
North	0	0,0%
Southeast	15	30,0%
South	5	10,0%
Family income		
< 1 minimum wage	14	28,0%
> = 6 minimum wages	6	12,0%
between 1–2 minimum wages	15	30,0%
between 3–5 minimum wages	15	30,0%

not having received or partially received medical support related to how to communicate the diagnosis to the child (Table 2).

Among the sources of information for a better understanding of the diagnosis, 48% (24) reported medical advice, 28% from patients associations, 8% from other health professionals, 6% from social networks, 6% from scientific sources (6%) and 4% (2) other sources (Table 2).

In Table 2, we have associated the self-perception of parents and guardians regarding psychological trauma related to the moment of diagnosis with environmental and emotional factors.

Fifty percent of the volunteers had some risk of post-traumatic stress, including somatic consequences, found in 22% (11) of this sample (Table 3). The Event Impact Scale – Revised showed statistical significance

($p = 0.021$) when correlated with how prepared the parents felt to talk to the children about the diagnosis on a scale from 1 to 5, but it did not correlate with their perception about the level of distress of the children when talking about the diagnosis (Table 4).

Discussion

Parents were responsible for the communication of the diagnosis of SMA to their children in 76% of cases. Although the doctor informed the parents of the name of the diagnosis in 100% of cases, only 48% of caregivers reported that the main source of information for clarifying and orienting the diagnosis was the attending physician. Sixty-two percent (31/50) of parents did not receive or partially received professional support on how to inform their children about the diagnosis. This finding seems to be related to the increase in psychological trauma in parents responsible for communicating the diagnosis to their children compared to when doctors inform the diagnosis directly to patients ($p = 0.043$).

In this way, poor communication has a double negative impact: on parents, for not having received the information they thought was necessary, having to seek answers from other sources; and on children, who received information from emotionally shaken parents with no technical ability to approach the topic. Parents have reported insecurity in disclosing the diagnosis of genetic diseases because of the difficulty in expressing themselves, dealing with emotions, and determining the ideal moment, besides doubts regarding the level of understanding of their children, possible interferences in their self-esteem, discriminatory events, and the use of this diagnosis as an excuse to face their difficulties [21].

Firth (1983) reported that few parents initially understood or had a superficial understanding of the diagnosis of Duchenne dystrophy, a rare, genetic, and neurodegenerative disease [12], diverging from the findings of the current research in which only 6% reported not having understood the diagnosis. This understanding was not a sufficient requirement to feel prepared to disclose the information to the affected child. The approach centered on the patient and their families allows us to understand what the information needs are at any given moment, what their values, preferences, psychosocial and existential concerns are. In addition, emotional support, empathy and the opportunity for discussion are factors that strengthen support for the family during the communication process [22].

Thus, poor communication of the diagnosis of this neurodegenerative disease compromises the autonomy of the physician/child/family trinomial due to the inability to make well-informed decisions. The communication between physicians and children can shift between a

Table 2 Association between psychological trauma and emotional and environmental aspects at the moment of diagnosis reported by family caregivers of SMA patients

Variables		Has the investigation trajectory until the moment of diagnosis left any psychological trauma?			p-value
		NO n/n%	YES n/n%	MAYBE n/n%	
Caregiver present during the trajectory to the Spinal Muscular Atrophy diagnosis	Yes	18 36.0%	24 48.0%	8 16.0%	–
	No	0 0.0%	0 0.0%	0 0.0%	
Feeling of inclusion in the health services sought regarding the environment and health professionals	No	2 15.4%	10 76.9%	1 7.7%	0.135
	Partially	7 43.8%	7 43.8%	2 12.5%	
	Yes	9 42.9%	7 33.3%	5 23.8%	
The diagnostic environment was suitable	No	0 0.0%	2 100.0%	0 0.0%	0.289
	Partially	2 22.2%	6 66.7%	1 11.1%	
	Yes	16 41.0%	16 41.0%	7 17.9%	
Understood the diagnosis	No	1 33.3%	2 66.7%	0 0.0%	0.622
	Partially	4 30.8%	8 61.5%	1 7.7%	
	Yes	13 38.2%	14 41.2%	7 20.6%	
Caregivers know about the diagnosis	No	0 0.0%	0 0.0%	0 0.0%	–
	Yes	18 36.0%	24 48.0%	8 16.0%	
Person responsible for telling the diagnosis to the patient	Doctor	7 63.6%	3 27.3%	1 9.1%	0.043*
	Other	0 0.0%	0 0.0%	1 100.0%	
	Father/ Mother	11 28.9%	21 55.3%	6 15.8%	
Medical support to help caregivers communicate the diagnosis to the child	No	7 35.0%	9 45.0%	4 20.0%	0.476
	Partially	4 36.4%	4 36.4%	3 27.3%	
	Yes	7 36.8%	11 57.9%	1 5.3%	

Table 2 (continued)

Variables		Has the investigation trajectory until the moment of diagnosis left any psychological trauma?			p-value
		NO	YES	MAYBE	
		n/%	n/%	n/%	
Main sources of guidance regarding the disease of the patient	Patients association	2 14.3%	10 71.4%	2 14.3%	0.344
	Scientific sources	1 33.3%	2 66.7%	0 0.0%	
	Doctor	11 45.8%	9 37.5%	4 16.7%	
	Other Health Professional	2 50.0%	2 50.0%	0 0.0%	
	Other	0 0.0%	1 50.0%	1 50.0%	
	Social Network	2 66.7%	0 0.0%	1 33.3%	

Fisher's exact test; significance $p < 0,05^*$

Table 3 Data referring to post-traumatic stress at the diagnosis of SMA in family caregivers

Variables	Family Caregivers	
	Average	SD
Total Stress	23,22	18,510
Post-traumatic stress classification	N	%
None	25	50.0%
Low	12	24.0%
Probable	2	4.0%
High	11	22.0%

Table 4 Correlation between levels of post-traumatic stress and emotional aspects at the time of diagnosis reported by family caregivers of SMA patients

Variables	Test statistics	p-value
Total post-traumatic stress		
The child has shown feelings of distress for talking about the diagnosis	-0.258	0.070
How prepared did you feel to talk to your child about the diagnosis?	-0.325*	0.021*

Pearson's Correlation; significance $p < 0,05^*$

deliberative model, in which there is a constant dialogue, with the participation of the children and exchange of knowledge in an appropriate and individualized language for each part of the trinomial, with the use of the informed consent form from the children and their legal guardians when necessary; and a paternalistic model, in which the child is a vulnerable and submissive being, who accepts the behavior of the physician/legal guardian who holds all technical and scientific knowledge. These formats differ in terms of the perception of the moral structure of the children and its impact on decision-making, which must be individualized [23].

The autonomy of children and adolescents in health care and research must be evaluated according to their maturity, personality, independence, and problem-solving ability in their daily lives and this is not always directly related to age. The professional is responsible for providing information and technical support for decision-making between parents and children. Thus, the perception and opinion of children and adolescents are relevant in the context of health decisions [11].

The autonomy of the child-family is directly proportional to the communication of the diagnosis. The quantity and quality of information and the sensations surrounding the moment are intrinsically related to the decision-making process [24–26]. Parents of children with oncological diseases that were asked to participate

in clinical studies after the moment of diagnosis reported, afterward, not having understood what randomization was nor the difference between treatment and research. They make an ill-considered decision because they are emotionally distressed, with no understanding of the objectives of the study or the possible benefits [27].

There is little discussion in the literature about transmitting the diagnosis of neurodegenerative diseases in children [12–14]. Firth (1983) highlighted failures in communicating the diagnosis to parents of children with Duchenne diagnosis [12], Metcalfe (2008) described a gap in communicating the diagnosis of genetic diseases to the affected children, leaving parents with this difficult mission [13], and Goodwin (2015) revealed that 70% of parents of children with Down syndrome have not discussed the diagnosis with their children [14].

Caregivers, mainly mothers, were responsible for disclosing the results of genetic tests related to abnormalities in the X chromosome to affected daughters without support from specialists in most cases [15–, 16–18]. They have reported difficulties in transmitting the diagnosis to their children [16], lack of genetic counseling, vague information, and the need to search for information in different sources [24], such as search engines, followed by support groups, with less than 5% of the information obtained being from the physician [25].

In the current investigation, less than 50% of family members have received genetic counseling, which is similar to Duchenne/Becker muscular dystrophy screening studies [13–, 14–18]. Järvinen (2000) and Fraser (2018) stated that the majority of genetically tested young women did not seek genetic counseling upon reaching adulthood, declaring that the result did not impact their lives. Genetic counseling provides families with knowledge, skill, confidence, and resilience in adaptive coping related to illnesses [26]. When the patient is a child or adolescent, counseling adjusts family communication patterns, supports, and educates relatives about genetic disorders in a flexible way and adapted to needs, age, maturity, education level, and psychosocial characteristics [26].

The present study has demonstrated that the less prepared the parents felt during the communication of the diagnosis, the greater the probability of post-traumatic stress syndrome ($p=0.021$), which was observed in 50% of the family members, with 22% at high risk. People with a greater understanding of their genetic diagnosis are more likely to talk more openly about it [26]. Post-traumatic stress syndrome, related to unpreparedness in transmitting the SMA diagnosis to the child, may have psychiatric consequences, in addition to triggering an inflammatory cascade, affecting the immune system and

physical health, such as metabolic syndrome and cardiovascular diseases [28].

Although there may be a predisposition to SPTD, environmental factors can be triggers [29], in this case, the moment of diagnosis of a serious illness in the child, without assistance in communicating the diagnosis, associated with anguish, fears, and uncertainties related to neurodegenerative diseases.

Techniques for transmitting difficult diagnoses have been part of the medical course curriculum [30–32], but reports of inadequate communication have been described in the last three decades [12–, 13–18]. The lack of emotional support, focus on technique, and insufficient time for a better physician–patient relationship have generated dissatisfaction with the quality and nature of the information [32]. Eenennaam et al. (2020) emphasized the importance of the interdisciplinary team in establishing prognostic communication guides, as well as the autonomy of the patient of having a family member present and the right not to know [33].

Respect for autonomy must be present in the physician/child/guardian trinomial, and physicians must inform the family of the diagnosis [34] and participate in the moment of informing the children, either indirectly, as a support, or directly, if the child feels welcomed, confident and familiar with the medical professional. Relatives without technical and emotional preparation may carry traumatic experiences for years, affecting their physical and mental health.

Thus, it is recommended that parents seek support from a specialist to obtain information and clear up their doubts to start the communication process early, using simple and clear language, calmly and honestly, without ambiguities, so that the child understands the informed message. They must also be open to listen to their feelings and questions. Emphasis must be given to the abilities of the children and the differences that make each one of them unique, in addition to demonstrating that there are other people with the same diagnosis. This approach can be gradual, with the support of books, figures, and films to illustrate it playfully [21].

Communication barriers on topics such as the end of life and palliative care cause insecurity in the medical environment, for fear of a negative impact on the lives of patients [35, 36]. However, care and quality of life can be improved by honesty, empathy, hope without lying or deceiving, and the individualizing of each case [36], in addition to the understanding of prognosis and therapeutic limitations [35]. Multifamily discussion groups are beneficial in the process of communicating with families who carry hereditary genetic disorders [37]. The support of a multidisciplinary team, composed of psychologists, social workers, occupational therapists, physiotherapists,

and speech therapists, among others, favors the establishment of service goals, expands the support network, the sense of security, and the clarification of the disease [38, 39].

Living with an illness that gradually limits strength and consequently functionality for simple tasks, progressing to vital functions, and maintaining a normal level of consciousness from childhood, is a prison in itself and coping with this requires an empathic physician/child/family relationship, honest and barrier-free communication initiated at the time of the diagnosis.

Conclusions

This study has demonstrated that the communication of the diagnosis of SMA to affected children or adolescents was predominantly conducted by parents who did not feel prepared to talk about the topic, without professional support, with a predominant search for information from a source unrelated to the attending physician. They were consequently more predisposed to post-traumatic stress syndrome and stated that this experience left psychological trauma in their lives.

In this way, the failure in the dialogue between the physician/child/guardian trinomial, during the diagnosis of SMA, has a negative impact on the autonomy of those involved, since this bioethical principle is directly related to the clarification of information so that there is freedom and consent during the decision-making process regarding the disease.

Limitations

This study has a few limitations. The answers are related to memories about the moment of diagnosis of a serious and disabling illness, which may have contributed to a predominance of negative experiences and self-reported psychological trauma. There was a predominance of female volunteers, which is to be expected, as culturally, mothers are more participative in the medical consultations of children, but there may be some bias related to sensitivity in capturing information.

Supplementary Information

The online version contains supplementary material available at <https://doi.org/10.1186/s12887-022-03552-3>.

Additional file 1. Structured interview.

Acknowledgements

We are grateful for the associations related to spinal muscular atrophy, such as DOENMUS, DONEM and AAME that supported this research, as well as all family members and patients diagnosed with spinal muscular atrophy who

contributed with their experiences during the moment of disclosure of this diagnosis.

Authors' contributions

I.A.M.F., R.O.A.M. and G.R. idealized the research, performed the literature review and methodology. I.A.M.F. performed data collection analysis and wrote the main manuscript text. R.O.A.M. and G.R. reviewed the manuscript. The author(s) read and approved the final manuscript.

Funding

No funding.

Availability of data and materials

The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate

This research was conducted in agreement with the Declaration of Helsinki and was approved by the ethics and research committee of Hospital Universitário Lauro Wanderley in Brazil under number 5,176,679. The informed consent was obtained from all subjects and/or their legal guardian(s) for minor and uneducated participants.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests as defined by BMC, or other interests that might be perceived to influence the results and/or discussion reported in this paper.

Author details

¹Faculty of Medicine, University of Porto, Alameda Prof. Hernâni Monteiro, 4200-319 Porto, Portugal. ²Lauro Wanderley University Hospital, Federal University of Paraíba, Cidade Universitária Campus I, 58051-900 João Pessoa, Paraíba, Brasil. ³Federal University of Rio Grande Do Norte, Caicó, 59300-000, Rio Grande do Norte, Brazil.

Received: 10 February 2022 Accepted: 8 August 2022

Published online: 17 August 2022

References

- Breitenfeld T, Jurassic MJ, Breitenfeld D. Hippocrates: the forefather of neurology. *Neurol Sci*. 2014;35(9):1349–52.
- Dubowitz V. Ramblings in the history of spinal muscular atrophy. *Neuromuscul Disord*. 2009;19(1):69–73.
- Burghes AH, Beattie CE. Spinal muscular atrophy: why do low levels of survival motor neuron protein make motor neurons sick? *Nat Rev Neurosci*. 2009;10(8):597–609. <https://doi.org/10.1038/nrn2670> (Epub 2009 Jul 8. PMID: 19584893; PMCID: PMC2853768).
- Fritz Z, Schlindwein A, Slowther AM. Patient engagement or information overload: patient and physician views on sharing the medical record in the acute setting. *Clin Med*. 2019;19(5):386–91.
- Simond AK. Ethics and decision making in end stage lung disease. *Thorax*. 2003;58(3):272–7.
- Yaffe MJ, Hovey RB, Rodriguez C. Use of patients' unsolicited correspondence to a family doctor to describe and understand valued components of a doctor-patient relationship: A Hermeneutics approach. *BMC Fam Pract*. 2019;20(1):136.
- Saultz JW, Albedaiwi W. Interpersonal continuity of care and patient satisfaction: a critical review. *Ann Fam Med*. 2004;2:445–51.
- Luchtenberg M, Maeckelberghe ELM, Locock L, et al. Understanding the child-doctor relationship in research participation: a qualitative study. *BMC Pediatr*. 2020;20:353. <https://doi.org/10.1186/s12887-020-02243-1>.
- Menezes ROA. Paciente terminal e o direito ao livre desenvolvimento da personalidade. *Curitiba: Juruá*, 2017, 47–57.

10. Jacob MS. El menor maduro *Bol Pediatr.* 2005;45:156–60.
11. Marsh V, Mwangome N, Jao I, Wright K, Molyneux S, Davies A. Who should decide about children's and adolescents' participation in health research? The views of children and adults in rural Kenya. *BMC Med Ethics.* 2019;20(1):41. <https://doi.org/10.1186/s12910-019-0375-9>.
12. Firth MA. Diagnosis of Duchenne muscular dystrophy: experiences of parents of sufferers. *Br Med J (Clin Res Ed).* 1983;286(6366):700–1.
13. Metcalfe A, Coad J, Plumridge G, et al. Family communication between children and their parents about inherited genetic conditions: a meta-synthesis of the research. *Eur J Hum Genet.* 2008;16:1193–200.
14. Goodwin J, Schoch K, Shashi V, et al. A tale worth telling: the impact of the diagnosis experience on disclosure of genetic disorders. *J Intellect Disabil Res.* 2015;59(5):474–86.
15. Goldman A, Metcalfe A, MacLeod R. The Process of Disclosure: Mothers' Experiences of Communicating X-Linked Carrier Risk Information to At-Risk Daughters. *J Genet Couns.* 2018;27(5):1265–74.
16. Järvinen O, Lehesjoki A, Lindlöf M, Uutela A, Kääriäinen H. Carrier testing of children for two X-linked diseases: a retrospective evaluation of experience and satisfaction of subjects and their mothers. *Genet Test.* 1999;3:347–55.
17. Järvinen O, Lehesjoki AE, Lindlöf M, Uutela A, Kääriäinen H. Carrier testing for two X-linked diseases: a retrospective study of comprehension of the test results and social and psychological significance of the testing. *Pediatrics.* 2000;106:1460–5.
18. Parsons EP, Clarke AJ, Hood K, Lycett E, Bradley DM. Newborn screening for Duchenne muscular dystrophy: a psychosocial study. *Arch Dis Child Fetal Neonatal Ed.* 2002;86(2):F91–5.
19. Caiuby, Andrea Vannini Santesso et al. Adaptação transcultural da versão brasileira da Escala do Impacto do Evento - Revisada (IES-R). *Cadernos de Saúde Pública* [online]. 2012, v. 28, n. 3 [Acessado 25 Julho 2021], pp. 597–603. Disponível em: <<https://doi.org/10.1590/S0102-311X2012000300019>>. Epub 12 Mar 2012. ISSN 1678–4464. <https://doi.org/10.1590/S0102-311X2012000300019>.
20. Daniel WW. *Biostatistics: A Foundation for Analysis in the Health Sciences.* Ed 9. New York: John Wiley e Sons; 2009.
21. Dennis A, Howell S, Cordeiro L, Tartaglia N. "How should I tell my child?" Disclosing the diagnosis of sex chromosome aneuploidies. *J Genet Couns.* 2015;24(1):88–103.
22. Aoun SM, O'Brien MR, Breen LJ, O'Connor M. "The shock of diagnosis": Qualitative accounts from people with Motor Neurone Disease reflecting the need for more person-centred care. *J Neurol Sci.* 2018;15(387):80–4. <https://doi.org/10.1016/j.jns.2018.01.026> (Epub 2018 Feb 3 PMID: 29571877).
23. Díaz-Pérez A, Navarro Quiroz E, AparicioMarengo DE. Moral structuring of children during the process of obtaining informed consent in clinical and research settings. *BMC Med Ethics.* 2020;21:122. <https://doi.org/10.1186/s12910-020-00540-z>.
24. Fraser HG, Redmond RZ, Scotcher DF. Experiences of Women Who Have Had Carrier Testing for Duchenne Muscular Dystrophy and Becker Muscular Dystrophy During Adolescence. *J Genet Couns.* 2018;27(6):1349–59.
25. Ablon J. Parents' responses to their child's diagnosis of neurofibromatosis 1. *Am J Med Genet.* 2000;93(2):136–42.
26. McConkie-Rosell A, Heise E, Spiridiglozzi GA. Genetic risk communication: Experiences of adolescent girls and young women with families with Fragile X Syndrome. *J Genet Couns.* 2009;18:313–25.
27. Alahmad G. Informed Consent in Pediatric Oncology: A Systematic Review of Qualitative Literature. *Cancer Control.* 2018;25(1):1073274818773720. <https://doi.org/10.1177/1073274818773720>.
28. Hori H, Kim Y. Inflammation and post-traumatic stress disorder. *Psychiatry Clin Neurosci.* 2019;73(4):143–53. <https://doi.org/10.1111/pcn.12820> (Epub 2019 Feb 21 PMID: 30653780).
29. Auxéméry Y. L'état de stress post-traumatique comme conséquence de l'interaction entre une susceptibilité génétique individuelle, un événement traumatogène et un contexte social [Posttraumatic stress disorder (PTSD) as a consequence of the interaction between an individual genetic susceptibility, a traumatogenic event and a social context]. *Encephale.* 2012;38(5):373–80. French. <https://doi.org/10.1016/j.encep.2011.12.003>. Epub 2012 Jan 24. PMID: 23062450
30. Sweeny K, Shepperd JA, Han PK. The goals of communicating bad news in health care: do physicians and patients agree? *Health Expect.* 2013;16(3):230–8.
31. Kiluk JV, Dessureault S, Quinn G. Teaching medical students how to break bad news with standardized patients. *J Cancer Educ.* 2012;27(2):277–80.
32. Anestis E, Eccles F, Fletcher I, French M, Simpson J. Giving and receiving a diagnosis of a progressive neurological condition: A scoping review of doctors' and patients' perspectives. *Patient Educ Couns.* 2020;103(9):1709–23.
33. van Eenennaam RM, Kruihof WJ, van Es, MA, et al. Discussing personalized prognosis in amyotrophic lateral sclerosis: development of a communication guide. *BMC Neurol.* 2020;20:446.
34. Código de Ética Médica: Resolução CFM nº 2.217, de 27 de setembro de 2018, modificada pelas Resoluções CFM nº 2.222/2018 e 2.226/2019 / Conselho Federal de Medicina – Brasília: Conselho Federal de Medicina, 2019.
35. You JJ, Downar J, Fowler RA, et al. Barriers to goals of care discussions with seriously ill hospitalized patients and their families: a multicenter survey of clinicians. *JAMA Intern Med.* 2015;175:549–56.
36. Brighton LJ, Bristowe K. Communication in palliative care: talking about the end of life, before the end of life. *Postgrad Med J.* 2016;92(1090):466–70.
37. Socio-Psychological Research in Genomics (SPRinG) Collaboration, Eisler I, Ellison M, Flinter F, Grey J, Hutchison S, Jackson C, Longworth L, MacLeod R, McAllister M, Metcalfe A, Murrells T, Patch C, Pritchard S, Robert G, Rowland E, Ulph F. Developing an intervention to facilitate family communication about inherited genetic conditions, and training genetic counsellors in its delivery. *Eur J Hum Genet.* 2016;24(6):794–802. <https://doi.org/10.1038/ejhg.2015.215> (Epub 2015 Oct 7. PMID: 26443265; PMCID: PMC4820094).
38. Bendixen RM, Houtrow A. Parental Reflections on the Diagnostic Process for Duchenne Muscular Dystrophy: A Qualitative Study. *J Pediatr Health Care.* 2017;31(3):285–92. <https://doi.org/10.1016/j.pedhc.2016.09.002> (Epub 2016 Oct 12. PMID: 27743907; PMCID: PMC5389929).
39. Anestis E, Eccles FJR, Fletcher I, Simpson J. Neurologists' current practice and perspectives on communicating the diagnosis of a motor neurodegenerative condition: a UK survey. *BMC Neurol.* 2021;21(1):34. <https://doi.org/10.1186/s12883-021-02062-6>. PMID:33482753;PMCID:PMC7821644.

Publisher's Note

Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

Ready to submit your research? Choose BMC and benefit from:

- fast, convenient online submission
- thorough peer review by experienced researchers in your field
- rapid publication on acceptance
- support for research data, including large and complex data types
- gold Open Access which fosters wider collaboration and increased citations
- maximum visibility for your research: over 100M website views per year

At BMC, research is always in progress.

Learn more biomedcentral.com/submissions

