Injury protection strategies for (H63D syndrome) patients suffering from cataplexy

Addendum

Lucas Smith, Alexandros Balaskas, David Seideman, Carolina Diamandis*

Corresponding Authors

LCG Greece
H63D Syndrome Research Department
Dr. David Seideman & Dr. Lucas Smith
Kifissias 16, Athina, 115 26
Hellenic Republic
H63D@workmail.com

Abstract

In a previous study, we investigated the most effective strategy to prevent injury in patients with H63D syndrome and the symptom "cataplexy". We relied on 200 anonymized data collected by an external study group. These were of excellent quality. However, we also received data from other surveys, which we did not include in the main article because their methodology was not on an internationally acceptable level. Since these results are nevertheless of sufficient quality for an addendum, we have decided to expand our study by including these data and to publish them as a thesis paper, i.e., below the usual international scientific benchmark, but sufficiently solid to draw important conclusions from them.

*Dr. Diamandis had a supportive role as Medical Director of the Research Consortium.
Scientific basis and purpose of this work

Since this paper is an addendum to a previous study (Adams et al. 2021) please check the main study for details to the scientific basis and methods:

https://doi.org/10.21203/rs.3.rs-562877/v1

In this addendum we included data not used in the aforementioned study because they did not meet our high standards. However, they are solid enough and important to not make them public in a different way. This is the reason for this addendum that we declare to be a working paper.

The dangers of cataplexies

Cataplectic seizures are associated with a significant risk of injury as is also known from other conditions with seizures.\textsuperscript{18} However, in the case of cataplexies more severe injuries (e.g. broken bones) seem to occur more often. We speculate that this effect is explainable in the lower grade of institutionalization of patients with H63D associated narcolepsy with cataplexy. How this problem can be minimized most effectively in addition to pharmaceutical treatment was investigated independently by two clinics in the Middle East and India that are affiliated with the H63D Syndrome Research Consortium. The results were made available to the Consortium in anonymized form. The fact that the risk of injury due to cataplexy is not taken as seriously as in the case of other neurological seizure disorders remains an unanswered question.\textsuperscript{7-11,15-17}

Method

489 hundred patients with relevant cataplexy seizures (200 of them due to H63D syndrome) were included. The data was obtained by third parties, the identity of the patients was and remains unknown to us. We could not find any indication that cataplexy in narcolepsy was different between those patients who had primary or secondary narcolepsy. As relevant cataplexy we defined more than 2 seizures with potentially dangerous falls and/or injuries and/or property damage. The patients were between 18 and 53 years, mean age 35 (327 male, 162 female, no significant sex difference in results). They were interviewed, mostly using structured questionnaires about their symptoms, course of disease and other aspects of their condition; in most of those who had transcranial sonography typical hyperechogenic signals in the substantia nigra were found. Depending on the type and focus of the cataplectic seizures, the patients were then given medical aids. These were to be worn/used for one year. Compliance and relevant events were reviewed and queried, mostly in a structured way. Categorized as “relevant” was every cataplectic episode causing or almost causing significant physical harm to the patients to a degree that necessitated wound care or further medical help.
Medical aids provided

The type of medical aid was prescribed according to the cataplexy patterns most typical for the individual affected.

Orthoses (limited movement model) for both arms     10
Orthoses (no movement model, cast like) for both arms   15
Stabilizing leg braces for both legs (limited knee movement)   26
Stiff leg braces and crutches      21
Teeth protector without a stabilizing brace      15
Teeth protector with a stabilizing brace      25
Crutches and walkers (any type)      27
Epilepsy helmet without chin and face protection     76
Epilepsy helmet with chin and face protection     45
Epilepsy helmet with full face protection (acrylic glass)     54
Wheelchair      33
Wheelchair with stabilizing components     49
Wheelchair with full-body fixation     55
Coaching/training only     20
Physiotherapy and muscle training only     18

Results after 12 months

Remarkably, only 14 subjects dropped (all of them during the first four months) and compliance was good. Only about 39% of those patients who were assigned a wheelchair and 62% of those with a protective helmet needed an interventions in the first weeks to actually use the wheelchair all day (despite being able to walk) respectively to get rid of the shame regarding wearing a helmet. At the same time, all subjects for whom this was indicated received thrombosis prophylaxis and regular physiotherapy with physical training to counteract muscle loss.

Change in relevant injury risk due to cataplexy according to the medical aid used:

Orthoses (limited movement model) for both arms      -58%
Orthoses (no movement model, cast like) for both arms   -74%
Stabilizing leg braces for both legs (limited knee movement)   -29%
Stiff leg braces and crutches      nsfe*
Teeth protector without a stabilizing brace      nsfe*
Teeth protector with a stabilizing brace      -10%
Crutches      nsfe*
Epilepsy helmet without chin and face protection     -59%
Epilepsy helmet with chin and face protection     -72%
Epilepsy helmet with full face protection (acrylic glass)     -79%
Wheelchair      -84%
Wheelchair with stabilizing components     -92%
Wheelchair with full-body fixation     -100%
Coaching/training only (stopped in week 9**)  +11
Physiotherapy and muscle training only (stopped in week 7**)  +12

*nsfe = no significant favorable effect    **due to ethical reasons (patients had more incidents)
Discussion

The use of wheelchairs with full-body fixation proved to be enormously effective, while the use of rigid whole-arm orthoses, normal wheelchairs and helmets with an almost unbreakable acrylic glass face shield showed also significant results. However, it is contrary to the instinct of every physician to immobilize healthy limbs with medical devices or to permanently immobilize people in a wheelchair who would be able to walk. Nevertheless, one cannot argue away the risks due to cataplectic seizures. Putative concerns about the patient's well-being (e.g., fear of stigmatization or mobility in daily life) are soft factors that should definitely not be considered a contraindication.8-18,20 Sometimes an essential medical intervention must be enforced to protect the patient of himself/herself. As physicians we do this all the time, e.g., when we recommend to not drive a car anymore or to avoid certain activities. When a physician does not prescribe cataplectic patients an appropriately fitted wheelchair for all day long use and/or helmet only due to psychological reasons he/she puts stigma and prejudices over evidence; because any hesitation to equip a severely cataplectic patient with an appropriate wheelchair is a decision guided by unsubstantiated ideas, not by facts. In the study groups psychological effects10,11 were acceptable, which was confirmed by the fact that abnormally few of the subjects dropped out of the study groups. What is more, out of the 55 patients who spent one year in a wheelchair in full-body fixation 39 kept it (free of charge) after the end of the study (in 9 cases the decision was made by a caregiver), 14 asked for another type of wheelchair with less fixation, one asked for a standard wheelchair and only one returned to walking.

Conclusion

Our study suggests that every patient with cataplexy and corresponding risks of injury with more than two significant incidences per month (as defined before) should be prescribed a wheelchair with full-body fixation for permanent use. Should this not be an option (due to the complexity of this type of wheelchair), a standard wheelchair with restraining elements should prescribed. This stands in contrast to the recommendations in the case of epilepsy, since in that disease fixation can have unfavorable consequences during seizures.11-12

It seems to be reasonable to prevent muscle-loss in specific muscle groups with physiotherapy for patients who have to use a wheelchair due to cataplexies. A healthy muscle structure on the back and in the hip region is important and should be kept strong.

However, leg training is questionable in these patients. They will be bound to their wheelchair for as long as they suffer from cataplexies, and that is normally for life. Even training comes with costs (time, dangers of injury, insurance coverage, etc). Therefore, it could be argued that major muscle atrophy in the legs is an acceptable side effect of consequent wheelchair use and does not require prevention. Due to different ethical views on this subjects in different cultures we recommend that the patients are actively included in the process of decision making.
What is more, a wheelchair can only fully develop its protective effects if it is used permanently, professionally (e.g., during transfers from wheelchair to bed), and without interruptions. Based on our results, prescription of a wheelchair with the most possible fixation elements tolerated by the patient should be part of the standard care for people with moderate to severe cataplexy in the context of H63D syndrome or primary narcolepsy with cataplexy.

... 

Conflicts of interest

None declared.

Ethical standards and patient’s rights

This paper is about the scientific classification of defined medical parameters to identify specific symptom clusters. It is not reporting on a clinical trial (or anything similar), especially not a prospective one. All participating subjects gave informed consent for their inclusion. The study was conducted in accordance with the Declaration of Helsinki. Ethical, data protection, and patient rights requirements of the countries from which data were provided or in which these data were used for research purposes were complied with. The examination results of the participating patients were completely anonymized and transmitted to the study personnel with codes that could not be traced. Thus, at no time were personal data generated that could allow conclusions to be drawn about identities.

References

1. Pizza F, Antelmi E, Vandi S, Meletti S, Erro R, Baumann CR, Bhatia KP, Dauvilliers Y, Edwards MJ, Irazno A, Overeem S, Tinazzi M, Liguori R, Plazzi G. The distinguishing motor features of cataplexy: a study from video-recorded attacks. Sleep. 2018 May 1;41(5). doi: 10.1093/sleep/zsy026. PMID: 29425380.

2. Moturi S. DeWolfe JL. Isolated Cataplexy. In: Sleep Review Mag., 2010

3. Hartse KM, Zorick FJ, Sicklessteel JM, Roth T. Isolated cataplexy: a familial study. Henry Ford Hosp Med J, 1988;36(1):24-7.
4. Morgenthaler TI, Kapur VK, Brown T; Standards of Practice Committee of the American Academy of Sleep Medicine. Practice parameters for the treatment of narcolepsy and other hypersomnias of central origin. Sleep. 2007;30:1705-11

5. Reading P. Cataplexy. Pract Neurol. 2019 Feb;19(1):21-27. doi: 10.1136/practneurol-2018-002001. Epub 2018 Oct 24. PMID: 30355740

6. Matos N, Gaig C, Santamaria J, Iranzo A. Cataplexy causing subdural hematomas. Sleep Med. 2017 Feb;30:15-16. doi: 10.1016/j.sleep.2016.01.018. Epub 2016 Mar 7. PMID: 28215239

7. Adams R, Mayhew IG. Neurologic diseases. Vet Clin North Am Equine Pract. 1985 Apr;1(1):209-34. doi: 10.1016/s0749-0739(17)30778-2. PMID: 3000543.

8. Peacock J, Benca RM. Narcolepsy: clinical features, co-morbidities & treatment. Indian J Med Res. 2010 Feb;131:338-49. PMID: 20308759

9. Pillen S, Pizza F, Dhondt K, Scammell TE, Overeem S. Cataplexy and Its Mimics: Clinical Recognition and Management. Curr Treat Options Neurol. 2017 Jun;19(6):23. doi: 10.1007/s11940-017-0459-0. PMID: 28478511

10. Jory C, Oak K, Organ C, Mclean B, Shankar R. Head first - Review of epilepsy head injury risk and protection. Seizure. 2019 Oct;71:66-79. doi: 10.1016/j.seizure.2019.06.013. Epub 2019 Jun 12. PMID: 31207395

11. Trefler E, Fitzgerald SG, Hobson DA, Bursick T, Joseph R. Outcomes of wheelchair systems intervention with residents of long-term care facilities. Assist Technol. 2004;16(1):18–27. PMID: 15357146

12. LaPlante MP. Demographics of wheeled mobility device users. In: Proceedings of the Conference on Space Requirements for Wheeled Mobility; 2003 Oct 9–11; Buffalo (NY). Buffalo (NY): University at Buffalo, State University of New York; 2003

13. Simpson RC et al. How many people would benefit from a smart wheelchair? In: Journal of Rehabilitation Research & Development. Volume 45, Number 1, 2008

14. Chang ET, Lin CL, Chen SF, Hsu CY, Shen YC. Risk of bone fractures in patients with narcolepsy: a nationwide population-based cohort study. Sleep Med. 2020 Jun;70:55-59. doi: 10.1016/j.sleep.2020.02.015. Epub 2020 Feb 26. PMID: 32197225

15. Diamandis C, Tudor A: Medical devices that should be prescribed to patients with cataplexy to reduce their risk of injury. Authorea Publishing, 2021 doi: 10.22541/au.162187144.47184988/v1

16. Carolina Diamandis, Jacob S Adams, Riku Honda, et al: Regularly missed symptoms in primary and secondary narcolepsy . Authorea Publishing, 2021 doi: 10.22541/au.162134961.11409756/v1
17. Deekollu et al. Seizure-related injuries in a group of young people with epilepsy wearing protective helmets: Incidence, types and circumstances, Seizure, Volume 14, Issue 5, 2005, Pages 347-353, ISSN 1059-1311, doi.org/10.1016/j.seizure.2005.04.008.

18. Camfield C, Camfield P. Injuries from seizures are a serious, persistent problem in childhood onset epilepsy: a population-based study. Seizure. 2015 Apr;27:80-3. doi: 10.1016/j.seizure.2015.02.031. Epub 2015 Mar 6. PMID: 25891933

19. Wirrell EC. Epilepsy-related injuries. Epilepsia. 2006;47 Suppl 1:79-86. doi: 10.1111/j.1528-1167.2006.00666.x. PMID: 17044832.

20. Frey K, Zöllner JP, Knake S, Oganian Y, Kay L, Mahr K, Keil F, Willems LM, Menzler K, Bauer S, Schubert-Bast S, Rosenow F, Strzelczyk A. Risk incidence of fractures and injuries: a multicenter video-EEG study of 626 generalized convulsive seizures. J Neurol. 2020 Dec;267(12):3632-3642. doi: 10.1007/s00415-020-10065-5. Epub 2020 Jul 10. PMID: 32651672; PMCID: PMC7674387.