Is Skater's Cramp a Task-Specific Dystonia?

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Introduction

Known in The Netherlands by the moniker *zwabbervoet*, and in Japan as *bura bura*, skater's cramp has ended the careers of many Olympic speed skaters and promising talents over the years. It was first observed to be a lessening of control and a jerking of the skate before it lands on the ice after a completed skating stroke (1). The onset appeared sudden and insidious, where skaters went from unaffected performance to having their careers end suddenly and prematurely in a matter of months. Despite this extreme and unlikely turn of events, the real cause of the disorder has remained a mystery. This thesis attempted to help solve this mystery.

The first indication of what skater's cramp might be, came in 2014. A group of neurologists, writing in a popular physiotherapy journal suggested a possible diagnosis: task-specific dystonia (TSD)(2). Prior suggestions had included flawed skating technique, arterial occlusion, sport specific compartment syndrome, and peripheral neurogenic damage; however, no quantitative evidence was ever provided and treatments based on these theories appeared unsuccessful (1). In the wake of these unsatisfactory explanations, prof. Marina Tijssen and other neurologists suggested TSD, based on their expert clinical assessment and from reading accounts from the media of famous speeds skaters falling prey to the condition such as Olympic great Gerard Kemkers (3,4).

TSD is part of a larger group of disorders known simply as dystonia. Dystonia is a group of movement disorders described as "sustained and intermittent muscle contractions causing abnormal and often repetitive movements, postures or both."(5). Although still not fully understood, many lines of evidence indicate dystonia is caused by abnormal functioning of the brain (6). Human movement is partly driven by a complex interaction between the motor cortex, cerebellum, thalamus and many individual sub-structures of the basal ganglia (7). These interconnected structures engage in a highly precise balance of excitement and inhibition governed by the regulation of neurotransmitters (8). In dystonia this precise balance is disturbed (6). Researchers have found myriad evidence of this in over-active (dis-inhibited) brain regions including the basal ganglia (9); as well as cortical, and subcortical structures like the primary sensorimotor-cortex, premotor areas, cerebellum and thalamus (6). These dis-inhibitions result in abnormal over-activation in the spinal cord (10,11) and co-contraction of muscles (12) resulting in the "sustained and intermittent contractions" (5) witnessed in dystonia.

Dystonia that only occurs while executing a particular practiced skill is referred to as *task specific* dystonia i.e. TSD. The most prevalent and known form of TSD is writer's cramp(13), but it also occurs in any skills requiring high dexterity like watchmakers and hairdressers(14,15), and occurs most commonly where motor control challenges are highest, like in musicians and sportsman (16,17). Symptoms usually present as patterned jerking of a limb while trying to perform the skilled task, and develop suddenly as a kind

of "attack" on a skilled movement (18). Clinical features previously associated with other TSDs include onset in middle age (19–21), a sudden (weeks) onset, low remission rate (20), and an imbalanced sex ratio (1:4 m/f) (22,23). As with dystonia more broadly it is related to abnormal brain function, but the exact reason for this disorder - the pathophysiology - is not fully understood. One hypothesis holds that it is related to the corruption of complex motor engrams or motor programs in the brain that develop when you practice a skill over many years (24). The flaws in the motor program are thought to dysregulate the correct execution of expert movements related to the skill (25). Fascinatingly, sudden external changes in the performance of a skilled movement due to injury or a change in equipment are implicated in an increased risk of corrupting these motor programs and developing TSD (18). While additional factors related to TSD like genetics (26), cortical/sub-cotical dis-inhibition (27,28) and stress (29) have also been found in dystonia more generally (6), the corruption of skill-specific motor engrams appears to be unique to TSD.

The reason why neurologists first hypothesized that skater's cramp was a TSD, had to do with the identification of key corresponding features in impacted skaters. Skater's cramp appeared to occur in highly accomplished skaters, suggesting it affected highly optimized motor programs, similar to TSD. Skater's cramp occurred suddenly without prior indications of a problem after many years of healthy skating. Impacted skaters general health and other skills appeared to remain unaffected. These factors suggested a corrupted motor program with an outside trigger. Also, cursory visual observations of skater's cramp resembled a TSD: a twitching jerk of the skater's foot often in a lateral direction at the moment the skate was placed on the ice. That skater's cramp appeared from initial video recordings to be an active jerk (not resultant of instability) was also key, because TSD also presents as an over-activity and not a loss of coordination or a weakness as in many other movement problems. This distinct presentation, it's task-specificity and sudden insidious onset in expert speed skaters lead researchers to the hypothesis that skater's cramp was a TSD.

This PhD aimed to build upon these initial observations with a more quantitative approach. This meant looking at other forms of TSD in sports, such as in golf, darts, snooker, running, and rowing to see what evidence researchers collected that helped them in their identification of the condition. Initial research revealed there were many different features that could be measured through quantitative means that helped to suggest that a movement disorder was a task-specific dystonia. The primary modes of investigation we chose were 1) kinematics 2) muscle activity 3) inter-muscular coherence 4) and psychometrics. Using these methods this thesis tested affected skaters for signs of TSD as a way of investigating our major thesis question: *is skater's cramp a TSD*? We will now briefly describe how these quantifiable features relate to TSD.

Kinematics

Studies of movement using accelerometry (kinematics) have provided features that aid in the observation of TSD. Kinematically, TSD presents as a highly consistent, and repetitive jerking (5,30) that is unrelated to the execution of the skill (maladaptive). The consistency and repetitiveness of jerking in TSD is thought to be caused by the corruption in motor-engrams that code for highly practiced tasks in the brain (31). These engrams are thought to be built from a series of consecutive smaller motor-instructions (chunks) that become fused together into a single long string that scales to different speeds and intensities(32). In TSD damage is limited to only certain sections of the string, therefore symptoms present repetitively only at a particular moment in the movement and across many intensities (33). Employing kinematics is a way to test for moment-specificity, consistency and stability at different intensities, thereby implicating the central nervous system, and making other explanations (bio-mechanical or neuropathic) less likely, as they would present more randomly over multiple strokes and intensities.

Muscle Activity

Measuring over-active muscles with electromyography (EMG) sensors aids in the observation of TSD. In TSD impacted muscles have been shown to produce surplus activity unrelated to the practiced task, often in combination with co-contraction, where agonist- antagonist muscle pairs co-activate maladaptively. This muscular-overflow appears unaffected by changes in intensity (16,34). It also occurs only at a specific moment in the movement that corresponds with other dystonic symptoms like repetitive jerking. This highly stereotypical set of features is suggestive of TSD, as it is unlikely to present in peripheral injuries like neuropathy, where muscle weakness is more probable. Consistent task- and moment- specific over-activity and co-contraction have been noted to suggest TSD in musicians (35), and in various lower limb TSDs in sports specifically (36), and is thought to arise due to a dis-inhibition of cortical and subcortical networks in the brain and subsequently down the motor-pathways of the cortico-spinal tract (37).

Inter-Muscular Coherence

In addition to over-activity, EMG can further be used to look for possible features of TSD through the analysis of inter-muscular coherence (IMC). IMC is a linear correlation-like coefficient determined by the consistency of phase differences between two oscillating signals from two muscles, ranging from 0 to 1 (no coherence to perfect coherence). Previous studies have found higher coherence between frequency 3-7 (theta) and 15-25 (beta) in impacted muscles involved in dystonia generally (38). In TSD specifically there is scanter evidence for higher IMC, however in two studies of writer's cramp, higher coherence was noted in the theta band (39,40). Similar to muscular over-activation and co-contraction, higher IMC is also thought to be driven by maladaptive dis-inhibition of cortical and subcortical motor pathways. Studies in TSD have specifically proposed subcortical structures like the basal-ganglia and pedunculopontine nucleus as key drivers

(40). To the extent that maladaptive IMC is driven neurologically, finding this feature in affected individuals is supportive of dystonia (40,41).

Psychometrics

Another way of observing possible features of TSD is through personality research. Those with TSD may share a distinctive personality profile typified by higher sensitivity to negative affect (leading to higher anxiety) as expressed by higher scores in "neuroticism" (42) a factor of the well validated NEO Five-Factor Inventory (NEO-FFI) (43). It is thought the reason for higher neuroticism in TSD is a shared neuro-circuitry of dis-inhibition between motor (44,45) and limbic networks regulating anxiety sensitivity(46). Specifically it is thought the frontal cortex fails to adaptively inhibit limbic loops that also drive motor circuits relying on similar brain regions (42,47).

Importantly, at the current state of art, there are no measurements that can unequivocally diagnose TSD reliably (it's diagnosed clinically by an expert neurologist). For this reason any positive findings for the kinematic, muscular, coherence and psychometric features used to test for TSD in this thesis cannot be confirmatory, but rather exist as individual pieces of a larger and incomplete mosaic of the pathophysiology of TSD.

Objectives

This thesis had two objectives: 1) To quantitatively investigate the hypothesis that skater's cramp was a task-specific dystonia, 2) and do a preliminary investigation of possible ways to treat it.

Outline

In **chapters 2** the movements of 5 affected participants were monitored while speed skating in a case-control study using IMUs to test if the movement patterns and clinical features of skater's cramp matched with TSD. In **chapter 3** we performed an in-skating investigation of whether electromyography and kinematic features in skater's cramp were suggestive of TSD in a larger cohort of 14 affected skaters comparing them to 14 age and ability matched controls. **Chapter 4** tested if inter-muscular coherence in the impacted leg of affected skaters was higher while they skated and in a stationary resisted task compared to their non-impacted leg and a control group, suggesting a centrally driven problem such as a TSD. **Chapter 5** investigated whether aspects of personality found to relate to TSD such as higher neuroticism were also higher in a population with skater's cramp. Finally, in all **chapters 2-5** we also marked clinical features that have previously been associated with TSD in other studies. In **chapter 6** we reviewed all interventions in TSD in sports. Despite the majority of this thesis being dedicated to the investigation of whether skater's cramp is a TSD, it is important to acknowledge a need for immediate medical treatment of those currently affected. For this reason doctors have already begun to treat skater's cramp based on their expert opinion that it is a TSD. To aid in this process we systematically reviewed all interventions previously attempted in TSD in sports.

References

1. Meijer L. Een schaatser met een zwabbervoet. Fysiopraxis. 2013 Aug;(Year 22 Number 8):22–5.
2. Marina de Koning-Tijssen. Over de schaatser met de zwabbervoet. FysioPraxis. 2014 FysioPraxis | maart;pag. 22-25.
3. Geest M van der. Het mysterie van de zwabbervoet en de golf-bibbers en waarom het juist geoefende sporters treft [Internet]. de Volkskrant. 2022 [cited 2023 Jan 25]. Available from: https://www.volkskrant.nl/wetenschap/het-mysterie-van-de-zwabbervoet-en-de-golf-bibbers-en-waarom-het-juist-geoefende-sporters-treft~b34369b4/
4. Redactie. Zwabbervoet velt Boeve [Internet]. destentor.nl. 2011 [cited 2023 Jan 25]. Available from: https://www.destentor.nl/overig/zwabbervoet-velt-boeve~a9c818d9/
5. Albanese A, Bhatia K, Bressman SB, DeLong MR, Fahn S, Fung VSC, et al. Phenomenology and classification of dystonia: A consensus update. Mov Disord. 2013;28(7):863–73.
6. Madeo G, Pisani A. Pathophysiology of Dystonia. In 2017. p. 929–50.
7. Rothwell JC. Overview of neurophysiology of movement control. Clin Neurol Neurosurg. 2012 Jun 1;114(5):432–5.
8. DeLong MR, Wichmann T. Circuits and Circuit Disorders of the Basal Ganglia. Arch Neurol. 2007 Jan 1;64(1):20–4.
9. Quartarone A, Hallett M. Emerging concepts in the physiological basis of dystonia. Mov Disord. 2013;28(7):958–67.
10. Berardelli A, Rothwell JC, Hallett M, Thompson PD, Manfredi M, Marsden CD. The pathophysiology of primary dystonia. Brain. 1998 Jul 1;121(7):1195–212.
11. Hallett M. Neurophysiology of dystonia: The role of inhibition. Neurobiol Dis. 2011 May 1;42(2):177–84.
12. Cohen LG, Hallett M. Hand cramps: clinical features and electromyographic patterns in a focal dystonia. Neurology. 1988 Jul;38(7):1005–12.
13. Goldman JG. Writer's cramp. Toxicon Off J Int Soc Toxinology. 2015 Dec 1;107(Pt A):98–104.
14. Horisawa S, Takeda N, Taira T. Watchmaker's Dystonia. Mov Disord Clin Pract. 2016 Feb;3(1):102–3.
15. Giorelli M, Zimatore GB. Hairdresser's Dystonia: An Unusual Occupational Dystonia. Tremor Hyperkinetic Mov N Y N. 2013;3.
16. Lenka A, Jankovic J. Sports-Related Dystonia. Tremor Hyperkinetic Mov N Y N. 2021;11:54.
17. Altenmueller E, Ioannou CI, Raab M, Lobinger B. Apollo's Curse: Causes and Cures of Motor Failures in Musicians: A Proposal for a New Classification. In: Levin, MF, editor. PROGRESS IN MOTOR CONTROL: SKILL LEARNING, PERFORMANCE, HEALTH, AND INJURY, VOL 826. AMTI; ANT Neuro; DelSys; Jintronix; Noraxon USA; No Digital; Magstim; Phoenix Technol; Polhemus; Rogue Res; Vicon; 2014. p. 161–78. (Advances in Experimental Medicine and Biology; vol. 826).
18. Altenmüller E, Jabusch HC. Focal dystonia in musicians: phenomenology, pathophysiology and triggering factors. Eur J Neurol. 2010;17(s1):31–6.
19. Ramdhani RA, Frucht SJ. Adult-onset Idiopathic Focal Lower Extremity Dystonia: A Rare Task-Specific Dystonia. Tremor Hyperkinetic Mov N Y N. 2013;3.
20. Katz M, Byl NN, San Luciano M, Ostrem JL. Focal task-specific lower extremity dystonia associated with intense repetitive exercise: a case series. Parkinsonism Relat Disord. 2013 Nov;19(11):1033–8.
21. Schneider SA, Edwards MJ, Grill SE, Goldstein S, Kanchana S, Quinn NP, et al. Adult-onset primary lower limb dystonia. Mov Disord Off J Mov Disord Soc. 2006 Jun;21(6):767–71.
22. Moura RC, de Carvalho Aguiar PM, Bortz G, Ferraz HB. Clinical and Epidemiological Correlates of Task-Specific Dystonia in a Large Cohort of Brazilian Music Players. Front Neurol. 2017;8:73.
23. Altenmüller E, Baur V, Hofmann A, Lim VK, Jabusch HC. Musician's cramp as manifestation of maladaptive brain plasticity: arguments from instrumental differences. Ann N Y Acad Sci. 2012 Apr;1252:259–65.
24. Sadnicka A, Rosset-Llobet J. Chapter 21 - A motor control model of task-specific dystonia and its rehabilitation. In: Ramat S, Shaikh AG, editors. Progress in Brain Research [Internet]. Elsevier; 2019 [cited 2022 Aug 16]. p. 269–83. (Mathematical Modelling in Motor Neuroscience: State of the Art and Translation to the Clinic. Gaze Orienting Mechanisms and Disease; vol. 249). Available from: https://www.sciencedirect.com/science/article/pii/S0079612319300871

25. Rosset-Llobet J, Fàbregas-Molas. Rehabilitation and Plasticity of Task-Specific Focal Hand Dystonia.
26. Hebert E, Borngräber F, Schmidt A, Rakovic A, Brænne I, Weissbach A, et al. Functional Characterization of Rare RAB12 Variants and Their Role in Musician's and Other Dystonias. Genes. 2017 Oct 18;8(10).
27. Bäumer T, Schmidt A, Heldmann M, Landwehr M, Simmer A, Tönniges D, et al. Abnormal interhemispheric inhibition in musician's dystonia - Trait or state? Parkinsonism Relat Disord. 2016 Apr;25:33–8.
28. Beck S, Richardson SP, Shamim EA, Dang N, Schubert M, Hallett M. Short Intracortical and Surround Inhibition Are Selectively Reduced during Movement Initiation in Focal Hand Dystonia. J Neurosci. 2008 Oct 8;28(41):10363–9.
29. Ioannou CI, Furuya S, Altenmüller E. The impact of stress on motor performance in skilled musicians suffering from focal dystonia: Physiological and psychological characteristics. Neuropsychologia. 2016 May;85:226–36.
30. Adler CH, Temkit M, Crews D, McDaniel T, Tucker J, Hentz JG, et al. The Yips: Methods to Identify Golfers with a Dystonic Etiology/Golfer's Cramp. Med Sci Sports Exerc. 2018 Nov;50(11):2226–30.
31. Sadnicka A, Kassavetis P, Pareés I, Meppelink AM, Butler K, Edwards M. Task-specific dystonia: pathophysiology and management. J Neurol Neurosurg Psychiatry. 2016 Sep;87(9):968–74.
32. Ramkumar P, Acuna DE, Berniker M, Grafton ST, Turner RS, Kording KP. Chunking as the result of an efficiency computation trade-off. Nat Commun. 2016 Jul 11;7(1):12176.
33. Sadnicka A, Kornysheva K, Rothwell JC, Edwards MJ. A unifying motor control framework for task-specific dystonia. Nat Rev Neurol. 2018 Feb;14(2):116–24.
34. Smith AM, Malo SA, Laskowski ER, Sabick M, Cooney WP 3rd, Finnie SB, et al. A multidisciplinary study of the "yips" phenomenon in golf: An exploratory analysis. Sports Med Auckl NZ. 2000 Dec;30(6):423–37.
35. Altenmueller E, Jabusch HC. Focal Dystonia in Musicians Phenomenology, Pathophysiology, Triggering Factors, and Treatment. Med Probl Perform Art. 2010 Mar;25(1):3–9.
36. Cutsforth-Gregory JK, Ahlskog JE, McKeon A, Burnett MS, Matsumoto JY, Hassan A, et al. Repetitive exercise dystonia: A difficult to treat hazard of runner and non-runner athletes. Parkinsonism Relat Disord. 2016;27:74–80.
37. Ahmad OF, Ghosh P, Stanley C, Karp B, Hallett M, Lungu C, et al. Electromyographic and Joint Kinematic Patterns in Runner's Dystonia. Toxins. 2018 Apr;10(4):166.
38. Grosse P, Edwards M, Tijssen M a. J, Schrag A, Lees AJ, Bhatia KP, et al. Patterns of EMG-EMG coherence in limb dystonia. Mov Disord Off J Mov Disord Soc. 2004 Jul;19(7):758–69.
39. Butz M, Timmermann L, Gross J, Pollok B, Dirks M, Hefter H, et al. Oscillatory coupling in writing and writer's cramp. J Physiol-Paris. 2006 Jan 1;99(1):14–20.
40. Choudhury S, Singh R, Chatterjee P, Trivedi S, Shubham S, Baker MR, et al. Abnormal Blink Reflex and Intermuscular Coherence in Writer's Cramp. Front Neurol. 2018;9:517.
41. Furuya S, Hanakawa T. The curse of motor expertise: Use-dependent focal dystonia as a manifestation of maladaptive changes in body representation. Neurosci Res. 2016 Mar 1;104:112–9.
42. Enders L, Spector JT, Altenmüller E, Schmidt A, Klein C, Jabusch HC. Musician's dystonia and comorbid anxiety: Two sides of one coin? Mov Disord. 2011;26(3):539–42.
43. Costa P, Mccrae R. Neo PI-R professional manual. Psychol Assess Resour. 1992 Jan 1;396.
44. Ridding MC, Sheean G, Rothwell JC, Inzelberg R, Kujirai T. Changes in the balance between motor cortical excitation and inhibition in focal, task specific dystonia. J Neurol Neurosurg Psychiatry. 1995 Nov;59(5):493–8.
45. Watanabe T, Yoshioka K, Matsushita K, Ishihara S. Modulation of sensorimotor cortical oscillations in athletes with yips. Sci Rep. 2021 May 14;11(1):10376.
46. Wassermann EM, Greenberg BD, Nguyen MB, Murphy DL. Motor cortex excitability correlates with an anxiety-related personality trait. Biol Psychiatry. 2001 Sep 1;50(5):377–82.
47. Lencer R, Steinlechner S, Stahlberg J, Rehling H, Orth M, Baeumer T, et al. Primary focal dystonia: evidence for distinct neuropsychiatric and personality profiles. J Neurol Neurosurg Psychiatry. 2009 Oct;80(10):1176–9.