



Movement Disorders Rounds: Expert commentary

Billiards-related dystonia: A new task-specific dystonia

In this beautifully illustrated paper, Smilowska and colleagues describe a very unusual patient with a task-specific dystonia related to billiard playing. Focal task-specific dystonia (FTSD) is one of the strangest and most fascinating disorders in all of neurology. By definition, in this condition dystonic movements are triggered in one part of the body (usually the arm or hand) selectively by a specific activity. In all other actions and tasks, the hand performs its required activities without any observable impairment. First described hundreds of years ago, the unique phenomenology of FTSD attracted the attention of prominent nineteenth century neurologists including Sully, Hammond, Duschenne, Oppenheim and Gowers. Early authors described all manners of FTSD patients, including musicians, telegraph operators, clerks, watchmakers, and jewelers, among others [1]. More recently, sporting activities such as golfing, archery, darts, pitching and now billiards have been added to the list of activities that can trigger FTSD.

When seen in the form demonstrated in this case, the differential diagnosis of FTSD is virtually non-existent—it is dystonia, and nothing else. The term “geigenneuroses” was coined by early authors to refer to FTSD, and most original descriptions emphasized the organic nature of the condition, noting the possibility of spread of dystonia to other tasks performed by the affected limb and even the spread of dystonia to other body parts [1]. Unfortunately, skepticism about FTSD in the first half of the twentieth century lent the term geigenneuroses a psychogenic connotation, and little of value was written about the condition. David Marsden's seminal observation in the 1970's linking various forms of focal dystonia (blepharospasm, torticollis, spasmodic dysphonia) [2], and the realization that isolated writer's cramp occurred in some patients with DYT-1 while other affected patients in the same family manifested generalized dystonia, FTSD was recognized as an organic disorder [3]. We now recognize that patients such as the one illustrated in this case are virtually never functional—most patients have devoted thousands of hours to hone their craft, and stand to benefit little from loss of their skill. Further, the presence of an imagined sensory trick in this billiard player and the purity of the phenotypic pattern of muscle involvement secure an organic etiology.

Rather than a curiosity or nuisance disorder, history reveals the impact of FTSD on patients. Telegrapher's cramp was such a significant issue in early twentieth century England (likely due to the poor design of the telegraph machine) that special legislation was passed to help support affected individuals. The impact of FTSD on patient's lives was first brought to wide attention by the American pianist Gary Graffman, who wrote publicly about his struggles with FTSD affecting his right hand at the piano. At the same time, his colleague and friend, the wonderful American pianist Leon Fleisher was suffering from the same disorder. Partnering with the Dystonia Medical Research Foundation in a public awareness campaign in 2004, Fleisher shared the story of his

struggle with FTSD and his triumphant return to play selected repertoire at the piano with the aid of botulinum toxin injections performed at the National Institutes of Health. Fleisher's story of struggle and heartbreak at the loss of his career as a two-handed pianist for 35 years resonated deeply with musician and non-musician patient affected by dystonia, and helped deepen the appreciation of the impact of the disorder on patients. In the current case, the authors demonstrate beautifully that physiology (EMG) can help define muscles that are overactive during performance of a specific task. Identifying these muscle groups, and avoiding injecting compensatory muscle groups, is critical to successful treatment with botulinum toxin. Early attempts to treat musicians' dystonia with toxin yielded disappointing results. More recent work has been very promising, the result of application (as in this patient) or careful localization techniques.

In the last decade and a half, there has been a remarkable growth in research into the etiology and treatment of FTSD. It is still unclear whether or not FTSD is best viewed as a cousin of other focal dystonias, (like blepharospasm, cervical dystonia and spasmodic dysphonia), or as a distinct disorder [4]. FTSD shares many similarities with the focal dystonia, including the presence of sensory tricks, null point, and the possibility of spread of dystonia to other tasks in the affected limb or other body areas. However, several features of FTSD are distinct, as beautifully illustrated in the detailed description of billiard's dystonia in the present case report. FTSD affecting musicians (and likely also athletes) has a marked male predominance, 4:1 M:F, whereas most forms of focal dystonia affect women slightly more often than men. Further, the response of FTSD to treatment with botulinum toxin is often more sustained than in focal dystonia such as blepharospasm and cervical dystonia, where the effects of toxin often dissipate in 12–14 weeks [5–7].

Many questions remain unanswered regarding the mechanism of FTSD. The prevalence of the disorder, 1/200 in professional musicians, is far more common than initially realized and much more common than focal dystonia in the general population. It is unknown whether similar numbers apply to sporting dystonia. Involvement of the hand performing the more complex task (right hand in pianists, left hand in violinists), and the protective influence of early life instrumental instruction in reducing the risk of the disorder, suggests that there are prominent environmental influences on the development of FTSD [8]. Aberrant plasticity, disruption of overlearned motor routines, and genetic predispositions to the disorder have been proposed, however work continues.

This case description illustrates a fascinating example of FTSD, providing another window into this unusual disorder of motor control.

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