

Evolution of the concept of dystonia

Evolução do conceito de distonia

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ABSTRACT

The term *dystonia* was proposed by Hermann Oppenheim in 1911, but reports of cervical dystonia are finding since the Roman Empire. The differentiation of the dystonia between a neurological and a psychiatric disease turned a lot. Sometimes was proposed, further, the exclusion of the dystonia as a different phenomenon among the movement disorders. The hypothesis of emotional etiology of the dystonias increase in the decade of 1960 and the conclusion of the etiology of dystonia only happened in the decades of 1970 and 1980 with a series of works of Charles David Marsden. These researches defined dystonia as a movement disorder caused by lesion in the basal ganglia. In February of 1984, a committee of members of Scientific Advisory Board of the Dystonia Medical Research Foundation developed the definition for dystonia accepts until 2013 when an international committee developed the new concept.

Keywords: dystonia, cervical dystonia, history, movement disorders.

RESUMO

O termo *distonia* foi proposto por Hermann Oppenheim em 1911, porém, quadros de distonia cervical são encontrados desde a antiguidade. A categorização da distonia entre uma doença neurológica ou psiquiátrica mudou muitas vezes sendo proposta, inclusive, a retirada da distonia como um fenômeno distinto entre os distúrbios do movimento. A etiologia emocional das distonias teve grande força na década de 1960 e o fim da discussão etiológica da distonia somente ocorreu nas décadas de 1970 e 1980 com uma série de trabalhos de Charles David Marsden colocando a distonia como um distúrbio do movimento por lesão nos gânglios da base. Em fevereiro de 1984, um comitê formado de membros da *Scientific Advisory Board of the Dystonia Medical Research Foundation* desenvolveu a definição para distonia aceita até 2013 quando um comitê internacional desenvolveu a definição aceita atualmente.

Palavras-chave: distonia, distonia cervical, história, distúrbios dos movimentos.

Dystonia, comes from modern Latin, from *dys-* + Greek *-tonos*¹. It is defined as: a state of disordered tonicity, especially of muscle tissue. The word tone itself had musical connotations. It derives from 13th century old French – *ton*, of the voice. Latin *tonus*, meant a stretching, quality of sound, tone, or accent; this derived from the Greek *tonos* similarly translated as stretching, tension, raising of voice, pitch. In modern usage, dystonic is applied to abnormal tension resulting in abnormal postures present in many disorders².

The disease of Roman emperor Tiberius Claudius Drusus Nero Germanicus, born in 10 BC, had been a source of speculation. In childhood, Claudius began to manifest a disorder marked by gait disturbance, tremulous involuntary movements of head and limbs. Abnormal movements of head and hands were worsened by postural and action maneuvers. There is evidence that movement fluctuated in severity. This case was probably the first description of a dystonic patient³. In 16th century, the important physician

and dramaturge François Rabelais described, in his book *Pantagruel*, a giant with movement disorders of the neck. He used the term, from Latin, *torti colli*⁴. In 1901, Destarac used the term *torticollis spasmodique* to describe the neck and pelvic dystonic movements in a 17-year-old girl. Destarac emphasized that these spasmodic movements occurred during activity and that they were relieved by rest and by various maneuvers⁵. In 1893, Sir William Richard Gowers used the term *tetanoid chorea* to describe the features of dystonia in a youth, which was later recognized to have been a case of Wilson disease⁶.

Despite these earlier descriptions, Marcus Walter Schwalbe is credited with the actual discovery of the dystonia. In his doctoral thesis from 1908, he first clearly recognized the condition as distinct from previously recognized movement disorders. He also recognized the hereditary nature of the disease, which he observed in a Jewish family. He considered the disorder to be partly psychiatric,

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however, calling it *tonic cramps syndrome with hysterical symptoms*^{5,6}.

The term *dystonia* only was coined by Oppenheim (Figure) in his 1911 paper describing 4 unrelated children, all were Jewish from Galicia and Russia¹. Initially, Oppenheim could not decide if he were seeing patients with hysteria or idiopathic bilateral athetosis. But then he realized that neither of these diagnoses was appropriate and that this was a new condition. Features that these children had in common and what impressed him were (1) muscle spasms affecting limbs and trunk that resulted in twisted postures; (2) worsening on walking, sometimes with markedly flexed spine that reminded Oppenheim of a dromedary; (3) the movements could be rapid and also rhythmic; (4) the symptoms progressed, leading eventually to sustained fixed postural deformities; (5) muscle tone could be hypotonic at one occasion and with tonic (sustained) muscle spasms at another, usually but not exclusively elicited on volitional movements; and (6) the absence of weakness, atrophy, sensory loss, impaired perception, sphincter disturbances, significant psychological abnormalities, or alteration of electrical excitability. Oppenheim suggested *dystonia musculorum deformans* as name^{1,6}. Ernest Herz elucidated this description of hypotonia and muscle spasms in dystonic patients in 1944. He demonstrated, by electromyography, simultaneous contraction of agonist and antagonist muscles in patients with dystonia⁷.

The genetic predisposition and the psychiatric genesis of dystonic symptoms generated controversies during the following years. Beyond Oppenheim, the Poland Edward Flatau and Wladyslaw Sterling described the condition in 2 Jewish boys, emphasizing its hereditary nature and calling it *progressive torsion spasms*^{5,6}. Arguments of neurological nosology shifted dystonia's categorization several times

and even threatened to eliminate dystonia as a phenomenon distinct from other movement disorders. Dystonia as a nosographic entity was vehemently argued at the Tenth International Neurological Meeting in Paris in 1929 by leading speakers including van Bogaert, Froment, Marinesco, Barré, Meige, and Lhermitte. Thereafter, though neurological investigators largely dropped their interest in dystonia for over a decade, psychiatrists, especially under the strong psychoanalytic influence of the period, avidly offered Freudian hypotheses to explain the contorted and disfiguring postures of focal and generalized dystonia⁸. In the 1960's psychiatrists claimed that the disorder results from castration anxiety, the stiff neck representing an erect phallus, or a symbolic turning away from the world⁹.

In 1924, Antonio Austregesilo, the founder of the first service of Neurology in Brazil (Rio de Janeiro – 1912), described a first case of posttraumatic dystonia¹⁰. This and other relates contributed for organic origin of dystonic movements. However, the end of discussion about psychiatric etiology of dystonia occurred only with the papers from Charles David Marsden (Figure), in 1970's and 1980's. Marsden was the most important researcher of movement disorders in the 20th century. He participated of an ad hoc committee of Scientific Advisory Board of the Dystonia Medical Research Foundation that developed the concept of dystonia in 1984, “a syndrome consisting of sustained muscle contractions, frequently causing twisting and repetitive movements, or abnormal postures”, accepts until 2013^{11,12}.

The definition from 1984 was seminal, but several shortcomings had recognized: (1) the expression “sustained muscle contractions” refers to one specific manifestation of dystonia, and implies exclusion of less sustained manifestations; (2) the quality of “abnormal postures” was not specified; (3) certain characteristic qualities of dystonia, such as the patterned and stereotypical nature of movements within an individual, the role played by movement initiation, and overflow activation of extraneous muscles, are not adequately represented. In view of these limitations of the 1984 definition, an international Consensus Committee, in 2013, proposes the following revised definition:

Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both. Dystonic movements are typically patterned, twisting, and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation¹².

In the next years, we expect a new discussion about the concept of dystonia. Maybe, the increase genetics discovery will be able to influence the next consensus dystonia definition.

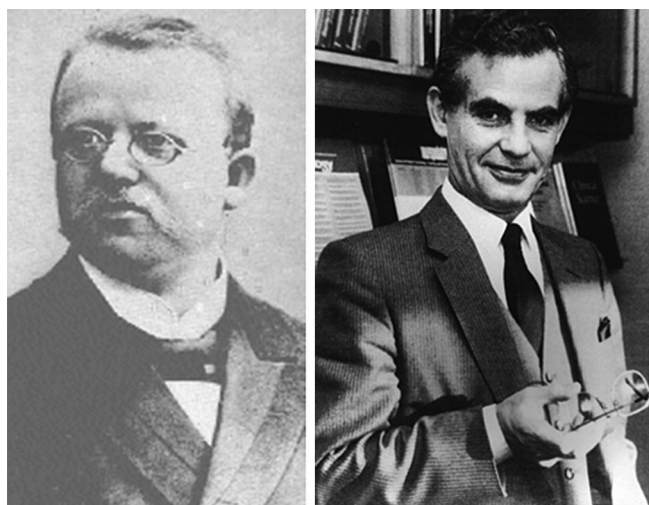


Figure. Hermann Oppenheim and C David Marsden: important researches in the history of dystonia.

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