

## Validation of a guideline for the diagnosis of cervical dystonia

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*Introduction:* Cervical dystonia (CD) is the most frequent form of focal dystonia. Due to the lack of a diagnostic test, CD diagnosis is based on clinical examination, and it is therefore open to bias [1, 2].

*Objective:* This study aims to provide practical guidance for clinicians in confirming or refuting suspected CD.

*Method:* Participants were video-recorded according to a standardized protocol assessing 6 main clinical features possibly contributing to CD diagnosis: presence of repetitive, patterned head/neck movements/postures inducing head/neck deviation from neutral position (item 1); sensory trick (item 2); and red flags related to conditions mimicking dystonia that should be absent in dystonia (items 3 to 6). To estimate sensitivity and specificity, the gold standard was CD diagnosis reviewed at each site by independent senior neurologists.

*Results:* The validation sample included 43 idiopathic CD patients and 41 control subjects (12 normal subjects, 6 patients with isolated head tremor, 4 with chorea, 6 with tics, 4 with head ptosis due to myasthenia or amyotrophic lateral sclerosis, 7 with orthopedic/rheumatologic neck diseases, and 2 with ocular torticollis). The best combination of sensitivity and specificity was observed considering all the items omitting the item related to capability to voluntarily suppress spasms. Indeed, the final algorithm yielded a sensitivity of 96.1% and a specificity of 81%.

*Conclusions:* An accurate diagnosis of CD can be achieved if, in addition to the core motor features, we also consider some clinical features related to dystonia mimics that should be absent in dystonia. The diagnostic algorithm without the item “ability to voluntarily suppress spasms” was sensitive and specific enough to be proposed as a guideline for presumptive diagnosis of CD, though it needs to be further expanded and validated in a larger sample.

### References:

- [1] Albanese A, Bhatia K, Bressman SB, et al. Phenomenology and classification of dystonia: A consensus update. 2013 *Movement Disorders* 28:863–873.
- [2] Romano M, Bagnato S, Altavista MC, et al. Diagnostic and therapeutic recommendations in adult dystonia: a joint document by the Italian Society of Neurology, the Italian Academy for the Study of Parkinson’s Disease and Movement Disorders, and the Italian Network on Botulinum Toxin. 2022 *Neurol Sci*.