



# Neuroimaging in idiopathic adult-onset focal dystonia

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## Abstract

We aimed to study the attitude of Italian neurologists in the use of conventional MRI in patients with idiopathic adult-onset focal dystonia. Patients were included in the Italian Dystonia Registry by experts working in different Italian centers. MRI was available for 1045 of the 1471 (71%) patients included in the analysis. Using logistic regression analysis, we found that MRI was more likely to be performed in patients with cervical dystonia, spasmodic dysphonia, or non-task-specific upper limb dystonia, whereas it was less likely to be performed in patients with blepharospasm or task-specific upper limb dystonia. We did not find differences in the number of MRIs performed between neurological centers in Northern, Central, and Southern Italy. We conclude that although the diagnosis of idiopathic adult-onset dystonia is mainly based on clinical grounds, many movement disorder experts rely on MRI to confirm a diagnosis of idiopathic dystonia. We suggest that neuroimaging should be used in patients with adult-onset focal dystonia to rule out secondary forms.

**Keywords** Blepharospasm · Cervical dystonia · Laryngeal dystonia · Arm dystonia · Magnetic resonance imaging

## Introduction

Idiopathic focal dystonias are clinical entities characterized by the presence of involuntary movements and postures in focal segments of the body [1]. Blepharospasm (BSP), cervical dystonia (CD), spasmodic dysphonia (SD), and upper limb dystonia (ULD) are the most frequent idiopathic adult-onset focal dystonias (AOFDs) [2]. Diagnosis of AOFD forms is usually based on history and clinical examination. In AOFD, conventional neuroimaging studies are usually normal [3]. Although some experts maintain that MRI studies are not necessary in most AOFDs [4], other experts suggest that a normal MRI or CT is useful to rule out symptomatic dystonia forms [5–7].

The Italian Dystonia Registry (IDR) is a multicenter data storage system that collects the diagnostic and clinical data of patients with different types of dystonia from several specialized neurological centers throughout Italy [8–11]. The IDR questionnaire also includes a question on whether patients had undergone an MRI examination, and, if so, whether it identified lesions in the basal ganglia, thalamus, or other structures possibly involved in symptomatic dystonia [12–14]. In order to see whether Italian movement disorder specialists consider neuroimaging necessary in idiopathic AOFD patients, we analyzed whether these patients underwent MRI examination. We also evaluated whether the use of MRI in diagnosing AOFD differed between centers located in

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Northern, Central, and Southern Italy. This information may be useful to understand the approach Italian neurologists take when diagnosing AOFD.

## Methods

The Italian Dystonia Registry was started on January 2016, and methods of data collection for the IDR are described elsewhere [8]. In the analysis, we included patients with a diagnosis of idiopathic AOFD, namely BSP, CD, ULD, and SD. Patients with a diagnosis of acquired dystonia, dystonia due to known structural causes, or with a confirmed genetic mutation were excluded from the study. We included all patients present in the IDR on November 30, 2019. We analyzed MRI examinations performed in the overall IDR population and in the different dystonia patient subgroups. Using a logistic regression model, we evaluated the clinical and demographic variables significantly associated with MRI for the patients enrolled in the IDR (dependent variable). Independent variables included type of dystonia, duration of symptoms, age, gender, and location of the center (Northern, Central, or Southern Italy). The study was approved by the ethics committees throughout Italy [8].

## Results

The IDR included data from 1471 patients with idiopathic AOFD (508 males and 1008 females; mean age:  $63.6 \pm 14.0$  years; mean age of onset:  $55.8 \pm 10.4$  years; mean disease duration:  $8.6 \pm 9.2$  years). Of the 1471 dystonia patients enrolled, 572 came from Northern Italy, 538 from Central Italy, and 360 from Southern Italy. Clinical and demographic data of AOFD patients classified according to dystonia phenotype at onset are shown in Table 1.

MRI had been performed in 1045 of the 1471 patients included in the analysis (71%), and on MRI, none of these

patients showed lesions in brain areas possibly causing dystonia. The percentage of MRI performed in the various forms of dystonias is reported in Fig. 1. By stratifying dystonia patients according to the three geographical areas, we found that MRI had been performed in 422 patients (73.7%) in Northern Italy, 396 patients (73.6%) in Central Italy, and 227 patients (63%) in Southern Italy.

Logistic regression analysis disclosed that MRI was more likely to be performed in CD, SD, and non-task-specific ULD patients than in BSP or task-specific ULD patients. The analysis also disclosed that the location of the centers was not associated with the number of MRI procedures (Table 2).

## Discussion

IDR data showed that conventional MRI studies were performed in approximately 70% of Italian patients who had a diagnosis of idiopathic AOFD. CD, SD, and non-task-specific ULD patients were more likely to undergo MRI than patients with BSP or task-specific ULD. No significant differences were present in the number of patients who underwent MRI in the different geographical areas of Italy.

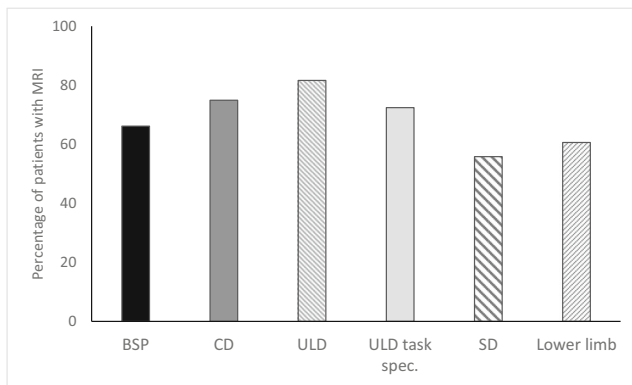
It is not clearly established whether MRI should always be performed in AOFD patients. Since dystonia due to structural lesions in the brain may be clinically indistinguishable from idiopathic dystonia [12–15], Mounier et al. [6] and European Federation of Neurological Societies (EFNS) guidelines [7] suggest that a normal MRI study is usually a prerequisite in order to determine whether dystonia is idiopathic. Conversely, Fung et al. [4] indicate that MRI is not necessary in most AOFDs and should probably be restricted to patients with early-onset dystonia or dystonia combined with other movement disorders. In our study, we found that about two-thirds of AOFD patients included in the IDR had undergone an MRI study. Our results suggest that although AOFD diagnosis is usually based on patient history and a complete clinical examination, Italian movement disorder experts perform MRI

**Table 1** Clinical and demographic data of idiopathic adult-onset dystonia patients included in the Italian Dystonia Registry classified according to the site of dystonia onset

Dystonia phenotype	No. of patients	Gender M/F	Age (Y)	Age of onset (Y)	Symptom duration (Y)
BSP	514	159/355	$69.4 \pm 10.6$	$60.8 \pm 8.1$	$8.2 \pm 8.6$
CD	662	234/128	$60.7 \pm 14.2$	$54.4 \pm 12.7$	$5.7 \pm 13.1$
SD	86	19/87	$67.9 \pm 12.5$	$52.9 \pm 18.1$	$15.6 \pm 12.1$
ULD	147	66/81	$56.2 \pm 16.8$	$49.8 \pm 16.3$	$7.8 \pm 16.1$
Task-specific ULD	29	9/20	$51.5 \pm 17.4$	$35.4 \pm 18.3$	$17.9 \pm 17.2$
LLD	33	16/17	$52.8 \pm 18.3$	$33.0 \pm 24.3$	$15.5 \pm 16.4$

Data are expressed as mean  $\pm$  standard deviation

Y years, BSP blepharospasm, CD cervical dystonia, SD spasmodic dysphonia, ULD upper limb dystonia, LLD lower limb dystonia



**Fig. 1** Percentage of patients who had MRI according to the type of dystonia

studies in order to diagnose idiopathic dystonia and exclude symptomatic forms of dystonia associated with specific lesions in the brain.

Considering the various types of dystonia, logistic regression analysis disclosed that non-task-specific ULD, SD, and CD patients were more likely to undergo an MRI study than patients with BSP or task-specific ULD. The finding that patients with non-task-specific ULD were more likely to undergo MRI is consistent with the observation of Strader et al. [13] that the upper limbs were the most commonly affected body site in symptomatic dystonia due to brain lesions. The high rate of MRI in SD patients may be due to the observation that since SD is a rare condition, clinicians are often uncertain about the diagnosis [16]. MRI was also frequently performed in CD patients, possibly due to the desire to rule out brain or cervical spine lesions [12].

Conversely, MRI was performed less frequently in BSP and task-specific ULD patients. Possible reasons MRI was performed less frequently in these two forms of dystonia may be due to the fact that physicians consider it unlikely that structural lesions cause simultaneous involvement of bilateral

structures, as in BSP, or that structural lesions, only rarely described in the brainstem, cause BSP [15]. In addition, structural lesions very rarely cause task-specific dystonia [3, 5]. The observation that MRI studies were performed with a similar frequency across the various geographical areas of Italy suggests that the number of MRIs performed is not conditioned by differences in regional healthcare policy, but more likely reflects the desire of Italian movement disorder experts to use conventional MRI to support a diagnosis made only on clinical grounds.

The strength of this study is that the analysis of MRIs performed included a large group of AOFD patients. A limitation is that patients enrolled in the IDR were followed in specialized centers, and therefore, the findings of this study cannot be generalized to the entire population.

In conclusion, a large percentage of movement disorder experts in Italy rely not only on clinical grounds but also on conventional MRI, to diagnose AOFD.

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**Table 2** Logistic regression analysis model showing demographic and clinical features associated with the dependent variable “MRI availability” (Yes = 1, No = 0)

Variable	Odds ratio	95% CI	<i>p</i> value
Gender (female)	1.02	0.86–1.22	0.76
Age	0.99	0.98–1.00	0.21
Disease duration	0.99	0.99–1.00	0.38
BSP	1.04	0.75–1.43	0.81
CD	1.44	1.02–2.04	0.03
SD	0.52	0.32–0.85	0.01
Task-specific dystonia	1.60	0.89–2.89	0.11
Non-task-specific ULD	1.95	1.17–3.25	0.009
Tremor	0.84	0.53–1.32	0.45
Geographical region	1.00	0.88–1.15	0.90

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## Compliance with ethical standards

**Conflict of interest** The authors declare that they have no conflict of interest.

**Ethical approval** Ethical approval has been obtained in all centers participating to the Italian Dystonia Registry.

**Informed consent** All participating patients have given provided informed consent.

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