ABSTRACT
Taking into account the crescent interest in the field of dystonia genetics, we have organized a didactic and fast algorithm to diagnose the main forms of hereditary dystonias. The key branch of this algorithm is pointed to dystonia classification in primary, plus, or paroxysmal. The specific characteristics of each syndrome will reveal the diagnosis.

Key words: dystonia, Parkinsonism, algorithm.

RESUMO
Levando em consideração o interesse crescente no campo da genética das distonias, organizou-se um algoritmo rápido e didático com a finalidade de auxiliar no diagnóstico das principais formas de distonia hereditária. O ramo principal desse algoritmo é a própria classificação da distonia: primária, paroxística, ou plus. As características específicas de cada síndrome podem sugerir o diagnóstico.

Palavras-Chave: distonia, parkinsonismo, algoritmo.
autosomal recessive mode of transmission, DYT1 and DYT6 have an incomplete penetrance (30 and 60%, respectively), which can be easily confounded and misinterpreted as sporadic, or be due to an autosomal recessive inheritance.\(^4\,^7\)

### DYSTONIA PLUS

If it is dystonia plus, the issue is about following the symptom: myoclonus or Parkinsonism? Is there levodopa response?

#### Myoclonus

In myoclonus dystonia, myoclonus usually is prominent and dystonia is represented as torticollis or writer cramp. Occasionally, dystonia can be the only disorder manifestation. Symptoms respond to alcohol. There are genetic heterogeneity and two known causative loci (DYT11 and 15).\(^8\) Gene was described only in DYT11 (sarcoglycan epsilon – SGCE).\(^9\)

#### Parkinsonism

If Parkinsonism is the other following symptom in dystonia plus, the questions to be answered are:
- Is there levodopa response?
- What is the inheritance?
- Was the onset sudden?

#### Is there levodopa response?

If dystonia respond to levodopa, dopa-responsive dystonia (DRD) is likely. There are two main genes responsible for DRD: GCH1 and TH. GCH 1 (GTP cyclohydrolase 1) is transmitted as an autosomal dominant way, with complete remission of symptoms after low doses of levodopa administration and there is a typical diurnal fluctuation (DYT5a).\(^10\) Tyrosine hydroxylase (TH) is transmitted in an autosomal recessive fashion and is also called recessive Segawa syndrome or recessive DRD (DYT5b). There is also marked improvement with levodopa, diurnal fluctuation, and differently from dominant DRD, frequent motor and speech delay.\(^11\)

#### Inheritance is X-linked or recessive?

In X-linked dystonia, only men will be affected in pedigree. Lugbag (Filipino dialect for twisted) occurs predominantly in families from the Philippines. Besides Parkinsonism, chorea and ballism have been described.\(^12\) If dystonia Parkinsonism is recessive and unresponsive to levodopa, DYT16 should be considered. Some DYT16 patients do not have Parkinsonism and present a generalized dystonia form, with prominent speech involvement.\(^13\)

#### Was the onset sudden?

DYT12 has a typical rapid evolution of bulbar and limb dystonia in a cranium-caudal spread with Parkinsonian features frequently without tremor and succeeding to psychological and physical triggers.\(^14\)
**PAROXYSMAL DYSTONIA**

Paroxysmal dyskinesias are intermittent movement disorders manifested by dystonia, chorea, and ballismus. If paroxysmal, one should check if it is kinesigenic or nonkinesigenic.

**Kinesigenic**

Paroxysmal kinesigenic dystonia is characterized by brief episodes of involuntary movements induced by sudden voluntary ones. Attacks last several seconds in a daily basis (usually less than one minute), with no loss of consciousness, and with good response to antiepileptic drugs. If kinesigenic, the questions to be answered are:

- Is there epilepsy?
- Is there spasticity or episodic ataxia?
- Are there other conditions, such as cognitive impairment or hemolytic anemia?

**Epilepsy**

Several patients have epilepsy as comorbidity (usually infantile benign seizures). There is genetic heterogeneity, and testing is possible for only DYT10.

**Spasticity or ataxia**

If kinesigenic and associated with spasticity, or episodic ataxia, DYT9 is probable, there is no genetic testing for this condition and in some patients there is acetazolamide response. It is clinically similar to DYT8: in both episodes, it can be triggered by alcohol, stress, and fatigue, but only in DYT9 exercise can precipitate the episodes.

**Other conditions**

If paroxysmal exercise dyskinesia induced is associated with epilepsy (absence seizures in majority) and/or ataxia, mild cognitive impairment, hemolytic anemia, reticulocytosis, and hypoglycorrachia, DYT18 is possible, it can be tested and there is a chance of response to ketogenic diet.

**Nonkinesigenic**

Finally, if nonkinesigenic and when precipitated by alcohol, stress, and caffeine with good response to benzodiazepines, DYT8 is likely and can be tested. If there are no obvious triggers, DYT20 is possible, however it unfortunately cannot be tested.

**References**