presented MND syndrome[2]. Recently, Nanetti et al.[3] described another 66 year-old woman with SCA2 affected with progressive weakness and fasciculation[3]. Our patient was younger than the previous reports and presented MND disease shortly after SCA2 diagnosis.

In 2006, the 43-kDa TAR DNA binding protein (TDP-43) was identified as the major disease protein in ALS and frontotemporal lobar degeneration with ubiquinated inclusions[4]. Recently, Elden et al.[5] pointed out to *ATXN2* gene as a relatively common suitability gene to ALS. They demonstrated that *ATXN2* is a potent modifier of TDP-43 toxicity in animal and cellular models. In addition, 6 patients with ALS were evaluated and disclosed different *ATXN2* localization in spinal cord[5].

This report highlights that unusual phenotypes such as an ataxia-parkinsonism-motor neuron disease syndrome may be found in SCA 2 individuals. This raises several questions such as whether or not patients investigating MND with or without known family members with cerebellar ataxia should be routinely screened for *ATXN2*. Future studies with larger series are welcome to address these questions.

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APRESENTAÇÃO CLÍNICA DE SCA2 COMO UMA SÍNDROME ATAXIA-PARKINSONISMO-DOENÇA DO NEURÔNIO MOTOR

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Facial grimacing as a clue for the diagnosis of GM1 type 3 gangliosidosis

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GM1 Gangliosidosis is an autosomal recessive lysosomal storage disease caused by the deficiency of beta-galactosidase. Only few cases have been reported in the literature, owing to the rarity of the condition but also possibly due to its underrecognition in clinical practice[1,2]. Reports of GM1 gangliosidosis type 3 patients and recent literature review shows that oromandibular dystonia producing the aprearance of grimacing is a common feature of this disorder[2]. Herein we describe a patient in wich proeminent facial grimacing served as a clue to the diagnosis of GM1 gangliosidosis type 3. The patients legal guardian gave consent to publish this case.

A 20 year-old female patient had a normal development until the age of 3 years, when the parents noted speech impairment wich worsened to the point of inteligibility in the following years. With 5 years cognitive deterioration in other areas was noted and the child was never able to attend school. Gait abnormality also developed and by the age 11 she was unable to walk or stand. On examination the patient had short stature and moderate thoracic kyphosis. Tongue and orofacial dystonia where present, giving the appearance of grimacing (Figure). There was also dystonia of the feet, dystonic posturing of the hands while at rest and increased tonus in the legs. No bradykynesia or dysmetria were noted. Strenght was normal with brisk reflexes and flexor plantar responses. There was no corneal clouding. Slit-lamp examination and fundoscopy were normal. Bone radiographies revealed kyphoscoliosis and femoral dysplasia. Routine brain MRI showed T2 hypointensity of the globus pallidus and hydrocephalus caused by a incidental ependimoma of the fourth ventricule. Routine blood and CSF examination were unremarkable. An abdominal ultrasound showed no abnormalities. Beta-glicuronidase, galacto-6-sulphatase and hexosaminidase A (testing for mucopolysaccharidosis IV, VII and Tay-Sachs disease, respectively) were normal. Leukocyte beta-galactosidade activity measured in serum was 5.7 nmol/h/

Table. Causes of facial dystonia.

Neurodegenerative causes	Secondary
Progressive supranuclear palsy	Drug induced (e.g. Neuroleptics, levodopa)
Multiple system atrophy	Peripherally-induced (e.g. after local trauma)
Corticobasal degeneration	Vascular (e.g. thalamic hemorrhage)
Wilson disease	Paraneoplastic (e.g. anti-Ri, anti-NMDA)
Neuroacanthocytosis	Autoimmune (e.g. Sjoegren syndrome, APL)
Neuroferritinopathy	Psychogenic (e.g. fixed dystonia of the lower lip)
PKAN	
Lesch-Nyhan disease	

mg (normal range 78-280), confirming the diagnosis of GM1 Gangliosidosis.

Type 3 GM1 gangliosidosis is characterized by onset around the second decade of life with slowly progressive extrapiramidal signs, such as dystonia and parkinsonism[1]. There is also a high prevalence of gait disturbance and dysarthria. Other symptoms are short stature, bone abnormalities, cognitive impairment, ataxia and cardiac disorders[3]. Orofacial dystonia is a common feature of type 3 GM1 gangliosidosis, with a prevalence of 87.5% according to a recent report[2].

Facial dystonia with proeminent involvement of oromandibular muscles is a frequent manifestation of neuroleptic induced movement disorders[4,5]. However, there is also a number of dystonia syndromes in wich proeminet orofacial involvement occur, and their presence should alert the clinician to their possibility (Table).

We suggest that in patients with early-onset dystonia, the occurance of facial grimacing should lead to the consideration of type 3 GM1 gangliosidosis, particularly when associated with speech and cognitive impairment, gait disturbances and bone abnormalities.

Figure. Facial grimacing and tongue dystonia.

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FACIAL GRIMACING COMO PISTA PARA O DIAGNÓSTICO DE GANGLIOSIDOSE GM1 TIPO 3

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Huntington's disease presenting as posterior cortical atrophy

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Neuroimaging and neuropathological studies on Huntington's disease (HD) have historically focused on striatal atrophy[1]. In posterior cortical atrophy (PCA), there is a progressive impairment of high-level visual functions and parietal damage[2]. The conundrum of PCA is that while the clinical presentation is relatively homogeneous, the nosological status remains something of a puzzle. We report a case of HD presenting as PCA.