authors and encouraging interaction among the networks of authors publishing in the journal[8], promoting scientific ties with researchers from Spanish-speaking countries[5], as well as international Spanish researcher collaborations and interdisciplinary connection of clinical research and basic scientific and epidemiologic activity[9]. Furthermore, the policy of prioritising the English language in a publication can lead to unintended and perhaps undesired effects, such as reducing the importance of the publication as a dissemination vehicle for the community which it serves. In this sense, different aspects of great importance have been rightly pointed out from the area. These should be taken into account by all clinically-oriented publications, particularly in a situation characterised by the increasing number of journals indexed in the desired (and at the same time controversial) SCI-Expanded: namely, that these publications are generally a vehicle of expression for the interests of a scientific society and serve a large community of readers beyond the scientists who publish in them and the role they occupy in the qualifying rankings of publications[10].

G. González-Alcaide [a,*], S. Mosbah-Natanson [b] and Y. Gingras [b]
[a] *Instituto de Historia de la Medicina y de la Ciencia López Piñero, Departamento de Historia de la Ciencia y Documentación, Universitat de València-Consejo Superior de Investigaciones Científicas, Valencia, Spain*
[a] *Centre Interuniversitaire de Recherche sur la Science et la Technology, Université du Québec à Montréal, Montréal, Québec, Canada*

* Author for correspondence.
E-mail: gregorio.gonzalez@uv.es (G. González).

References

1. Matías-Guiu J. Las citaciones y las publicaciones médicas en español. Rev Neurol. 1996;24:904-6.
2. Matías-Guiu J. Revista de Neurología. El reto de la difusión de las neurociencias en español. Rev Neurol. 2000;30:35-40.
3. Martínez Vila E, Martín R. ¿Por que publicar en Neurología y en español? Neurología. 2005;20:1-3.
4. Aleixandre Benavent R, Valderrama Zurián JC, Alonso Arroyo A, Miguel Dasit A, González de Dios J, De Granda Orive JI. Español frente a inglés como idioma de publicación y factor de impacto de Neurología. Neurología. 2007;22:19-26.
5. González de Dios J, Sempere AP, Aleixandre Benavent R. Las publicaciones biomédicas en España a debate (I): estado de las revistas neurológicas. Rev Neurol. 2007;44:32-42.
6. Bracho-Riquelme RL, Pescador Salas N, Reyes-Romero M. Repercusión bibliométrica de adoptar el inglés como idioma único de publicación. Rev Invest Clin. 1997;49:369-72.
7. Matías-Guiu J, García Ramos R. El factor de impacto y las decisiones editoriales. Neurología. 2008;23:342-8.
8. Matías-Guiu J, García Ramos R. Autores y autoría en las publicaciones médicas. Neurología. 2009;24:1-6.
9. Bermejo Pareja F. La investigación neurológica en España, presente y futuro. Neurología. 2004;19 Suppl 1:64-72.
10. Alfonso F. Revistas biomédicas españolas: relevancia académica, impacto científico o factor de impacto. ¿Qué es lo que importa? Rev Neurol. 2009;48:113-6.

Adult onset of leukoencephalopathy with vanishing white matter

Leucoencefalopatía con sustancia blanca evanescente de inicio en edad adulta

Dear Editor:

We read with great interest the letter published in the previous issue of your journal by Pato Pato et al.[1], which presented a case having characteristics similar to ours. The publication of that case leads us to comment on the possible existence of an underdiagnosis of this disease, which may be more prevalent in the general population and go unnoticed by the entire medical population.

Male, 20 years old, healthy, with no remarkable medical history, son of non-consanguineous parents. In 1994, after a minor traffic accident without head trauma and in which the remaining occupants of the vehicle were unharmed, he presented an episode of psychomotor agitation. Computed tomography (CT) showed bilateral white matter hypodensity, which was initially interpreted as cerebral oedema, requiring mechanical ventilation and admission to the intensive care unit. After extubation nine days later, the patient presented episodes of agitation with echolalia and palilalia, alternating these with periods during which he was able to maintain simple conversations. He also suffered occasional epileptic crises and right spastic hemiparesis. Brain magnetic resonance imaging (MRI) showed almost complete bilateral affectation of white matter.

The studies performed to rule out common leukoencephalopathy aetiologies were normal: creatine kinase, lactate, protein and immunoglobulins, cerebrospinal fluid analysis, including IgG index, autoimmunity, serology (syphilis, HIV, *Salmonella*, herpes virus), basal cortisol, ACTH, amino acids in urine, very–long-chain fatty acids, analysis of fibroblast culture, urinary arylsulfatase A, phytanic acid, copper and ceruloplasmin, and muscle and sural nerve biopsy.

The patient experienced gradual improvement that became consolidated in the following months; he was able to walk unaided and the epileptic crises disappeared.

Throughout the years of follow-up, he presented progressive deterioration in both motor and cognitive functions, spontaneously or coinciding with fever episodes. Hospitalisation was required in some of them.

The genetic study identified a G338A homozygous mutation in the *eIFGB5* gene. Both parents were carriers of this mutation.

Currently, after 14 years, he is totally dependent in his daily life: he presents marked cognitive impairment, severe

spasticity and dystonic postures. The recent MRI (fig. 1) shows an important progression of leukoencephalopathy with images of associated cystic degeneration.

Initially, leukoencephalopathy with vanishing white matter was called CACH (childhood ataxia with diffuse central nervous system hypomyelination) or MCD (myelinopathia centralis diffusa). The disorder was defined as such by Van der Knaap et al.[2] in 1997 using radiological and diagnostic criteria, although several descriptions had already been published in the literature. In 1998, Van der Knaap described milder forms and late-onset variants of the same disease, leading to the exclusion of the age at onset from the diagnostic criteria[3]. Since then, 263 cases have been reported and the number of mutations has risen to 121[4].

Currently, we know that ovary-leukodystrophy and vanishing white matter disease form part of the spectrum of the same disease, so they are referred to as eukaryotic initiation factor (eIF2B)-related disorder.

Approximately 75% of patients have an eIF2B5 gene mutation[5], of which the most frequent is G338A, as was the case in our patient and the one previously presented in this publication. This is mainly associated with adult onset forms, although cases of the classic form have also been reported. There is a genotype-phenotype correlation, as some mutations are associated with milder forms of the disease and others with severe variations[4].

Despite having no specific treatment, early diagnosis is essential to prevent infections, fever and minor trauma, which are aggravating factors, as well as to carry out suitable genetic counselling.

It should be noted that this may be an underdiagnosed entity and that there are presymptomatic patients (as was evidenced in our patient's CT) in whom any banal situation can trigger the disease. So, it seems important to bear in mind that, despite being a "rare" disease, the geographical proximity of both cases is unusual.

M.S. Piñeiro*, M. López and J. Sánchez-Herrero

Servicio de Neurología, Hospital Universitario Xeral-Cíes de Vigo, Vigo, Spain

*Author for correspondence.
E-mail: Socops@hotmail.com (M.S. Piñeiro).

References

1. Pato Pato A, Lorenzo González JR, Cimas Hernando I, Rodríguez-Constenla I. Leucoencefalopatía con sustancia blanca evanescente: caso clínico de inicio en adulto. Neurología. 2009;24:504-6.
2. Van der Knaap MS, Barth PG, Gabreëls FJ, Franzoni E, Begeer JH, Stroink H, et al. A new leukoencephalopathy with vanishing white matter. Neurology. 1997;48:845-55.
3. Van der Knaap MS, Kamphorst W, Barth PG, Kraaijeveld CL, Gut E, Valk J. Phenotypic variation in leukoencephalopathy with vanishing white matter. Neurology. 1998;51:540-7.
4. Maletkovic J, Schiffmann R, Gorospe JR, Gordon ES, Mintz M, Hoffman EP, et al. Genetic and clinical heterogeneity in eIF2B-related disorder. J Child Neurol. 2008;23:205-15.
5. Van der Knaap MS, Pronk JC, Scheper GC. Vanishing white matter disease. Lancet Neurol. 2006;5:413-23.

Figure 1 T1 sequence of magnetic resonance imaging: extensive bilateral hypodensity of the white matter with associated areas of cystic degeneration.