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Letter to the Editor

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An Heidenhain variant of the Creutzfeldt–Jakob disease misdiagnosed as a DSM-5 conversion disorder: a case report

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To the Editor:

A 56-year-old outpatient of the ophthalmology department was referred to the liaison psychiatry team for blurred vision and transient blindness without a found somatic etiology. There was no specific personal medical history excepted an alcohol addiction.

The patient was first referred to the ophthalmology department 2 months after the symptom onset. At this time, the visual acuity, the optical coherence tomography, the retinal nerve fiber layer, and the ganglion cell layer complex (which excluded a glaucoma) were normal. But the Goldmann visual field testing, which is a subjective test, showed a transient lateral homonymous hemianopsia (LHH).

Thus, the patient was hospitalized in the neurology department. The EEG revealed a focal slowing in the bi-occipital region. The cerebral MRI (fluid-attenuated inversion recovery (FLAIR)-weighted images) evidenced nonspecific cortical hyperintensities which were not associated with the transient LHH. The Montreal Cognitive Assessment scored 19/30 which is in favor of cognitive dysfunction but was initially related to alcohol consumption. A functional etiology was then proposed by neurologists for this transient blurred vision.

Four months after the first consultation, the patient experienced a worsening of the eyesight with a total binocular blindness. Since visual acuity changed within minutes from light perception to a 20/20 score, the neurologist and ophthalmologist suggested the DSM-5 diagnosis of acute conversion disorder with visual symptoms and the patient was referred to the liaison psychiatry team. The examination evidenced a mild sadness and anxiety related to his medical condition and visual hallucinations. No anorexia, no insomnia, no suicidal thoughts, no athymhormia, no anhedonia, no other symptoms of an affective or a psychotic disorder and no symptoms of withdrawal delirium were evidenced. Fluctuating symptoms were observed, from accurate descriptions of the room to incapacity to describe the surroundings. No specific stress factor and no clear evidence for a functional disorder were identified. Of note, the patient did not meet all criteria for conversion symptoms of the Diagnostic Criteria for Psychosomatic Research (DCPR).[1]

After a multidisciplinary staff, a second cerebral MRI with diffusion-weighted acquisition was prescribed because the psychiatric examination and paraclinical abnormalities (EEG and cerebral MRI) suggested a diagnosis of medical condition rather than a diagnosis of conversion disorder. The second MRI evidenced diffusion-weighted bi-occipital and parietal hyperintensities with a low Attenuated Diffusion Coefficient and FLAIR hyperintensities in the same areas suggesting an encephalopathy.

The patient was immediately admitted to the neurology department. Ataxia and myoclonus appeared 2 days later, suggesting a Creutzfeldt–Jakob disease (CJD). A second EEG showed a worsened activity with diffuse slowing and monophasic periodic sharp wave complexes (1 Hz). The cerebrospinal fluid (CSF) 14-3-3 protein assay was weakly positive, and the CSF Neopterin level was in the usual concentration range. A probable sporadic CJD according to criteria of the Centers for Disease Control and Prevention (CDC), and, more specifically, the Heidenhain variant, was diagnosed.[2] Ten days later, the patient died, and the brain anatomopathological examination could not be performed.

In line with this case, the inadequacy of the DSM-5 classification of somatic symptom and the presentation of the DCPR have been described previously.[1] We agree that those DSM-5 criteria

have a limited clinical utility for conversion disorder, a disorder generally poorly known to practitioners.

Our case highlights that the DSM-5 conversion disorder criteria may induce overdiagnosis, especially in rare diseases, like the Creutzfeldt–Jakob disease, with uncommon clinical presentation. It also emphasizes that transient, inconsistent, or fluctuating neurological symptoms may not be functional, psychological, or psychiatric, but due to a neurological disease.

The CJD is a rare neurodegenerative disorder induced by the Scrapie Prion (PrPsc).[2] It is often associated with prodromal psychiatric symptoms such as depressive, psychotic or anxiety symptoms, sleep disturbances, and abnormal behavior. The Heidenhain variant represents 20% of the sporadic cases of Creutzfeldt–Jakob disease.[2] Its main symptoms are visual, such as disturbed perception of colors or structures, visual hallucinations, cortical blindness or visual field defects, associated with MRI abnormalities in the occipital area (occipital cortical ribbon). Its median survival time is 6 months. It has been reported that those symptoms could be transient.[2]

This case-report emphasizes that rare diseases, such as the Heidenhain variant of CJD, can have uncommon clinical presentations, like an atypical transient and inconsistent cortical blindness. Those presentations may mimic DSM-5 conversion disorder.[3] Their diagnosis requires a thorough and repeated multidisciplinary assessment, particularly with neurological clinical and para-clinical assessments, since specific abnormalities may not be identified at the beginning. For example, MRI changes, and mainly cortical ones, are frequently misinterpreted in early stages of CJD.[4]

Even if there is no curative treatment for CJD to date, its delayed diagnosis has consequences for the patient and the family, in terms of quality of life, stigmatization, and appropriate palliative care treatment.

With the DSM-5 diagnostic criteria of conversion disorder, unlike DSM-IV, the lack of stress factor before the symptom onset does not exclude anymore the diagnosis of conversion disorder, which is one of the major changes.

This change allows to avoid passing over a diagnosis of conversion disorder in case of difficulties to identify a stressful life event during a brief and unique psychiatric assessment.[3] But this leads to a possible inadequate over-diagnosis of conversion disorder, as practitioners are often deficiently trained to its diagnosis, especially in patients with rare organic diseases with psychiatric features, like the CJD. Indeed, among outpatients with unexplained neurological symptoms, 18% are diagnosed as conversion disorder.[5] Under DSM-5 definition, almost all of them could be diagnosed as conversion disorder.[1]

Conversion disorder over-diagnosis could induce a delayed diagnosis of nonpsychiatric diseases with inherent mortality and morbidity risks. We suggest using other criteria as the DCPR to assess conversion symptoms and to rethink DSM criteria take into account a stress factor preceding symptom onset.

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