Learning from errors

Onset of neuropsychiatric symptoms after psychological trauma may result in erroneous diagnostic bias

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Summary

We report a case of neuroacanthocytosis, which was misdiagnosed as conversion disorder. Because the onset was after a psychologically stressful physical trauma, the patient's symptoms were interpreted in terms of psychological regression. The case clearly points out the defects of descriptive psychiatric diagnosis, especially in the area of somatoformal disorders. It also reminds the clinicians to keep in mind rare neurological disorders as possible causes for psychiatric symptoms.

BACKGROUND

In this case report, we describe a patient with neuroacanthocytosis (NA), who was misdiagnosed as suffering from conversion. The symptoms broke out after a psychologically traumatic event, and this led clinicians astray. Although the illness is not very common, we think this case is educative because

1. It shows that inferences about causality are often made in psychiatry solely by temporal sequence of events, and,
2. It highlights the problems with descriptive psychiatric diagnosis, especially somatoformal and dissociative disorders, which lack clear, positive criteria.

CASE PRESENTATION

The turning point of our patient's health was when he lost his right-hand fingers II–IV in an accident at the age of 25. His first symptoms were cold soreness and stiffness of the hand. At the age of 26, he started having dysarthria, and he contacted a otorhinolaryngologist who did not find a cause for the complaint. He remitted the patient to a psychiatrist who assumed that it was caused by a post-traumatic conversion.

When he was 27 years old, neuropsychological tests failed to detect any prominent findings. In the same summer the patient went to a neurologist, owing to other conditions he had to endure during that time, such as increased salivation, dysarthria, motor restlessness and curling of lips. The patient attributed the symptoms to the accident happened. No neurological diagnosis was established.

Soon after this, the patient drove out of the highway with his car. After this accident he was confused, disoriented and aggressive and had a generalised seizure. However, his EEG was not clearly irritative. Brain MRI- and CT-scans revealed non-specific findings in the right temporal pole. Because a brain tumour was suspected as a cause of the symptoms, a stereotactic biopsy was taken. There were to some extent degenerative axons, but no tumour. The only diagnosis was epilepsy suspecta.

The patient was referred to a psychiatric clinic at the age of 28. He had moved back to his parents, considering the risk and insecurity involved in living alone while experiencing these new epileptic seizures. Speech and eating had slowly deteriorated, and changes in personality occurred. He mutilated his head on the wall, especially when the accident was discussed. The neurologists suspected Gilles de la Tourette syndrome, even though the symptoms had emerged late. But still the final diagnosis remained unestablished. The main focus in psychiatric treatment was social rehabilitation, because the patient had started to isolate.

Until the age of 29, the main diagnosis was conversion symptoms related to the psychological trauma as a consequence of the accident. The psychiatric staff assumed that the patient had regressed to an earlier developmental level after the accident. They thought that this idea was supported by the fact that the patient had moved back to his parents and did not have a girlfriend. His psychiatrist, however, still suspected a neurological illness and consulted Turku PET-centre.

INVESTIGATIONS

Because the patient's symptoms were puzzling, a FDG-PET investigation was performed. The imaging revealed an unexpected finding, namely, 'mute' caudate and putamen nuclei bilaterally (figure 1).

Glucose metabolism was significantly reduced ($>-2SD$) in the caudate and putamen bilaterally when compared with eight control subjects. Because of clear movement symptoms, the patient also underwent a [18F]dopa PET-scan.

The caudate and putamen Ki^{occ} values, obtained with tissue (occipital cortex) reference were over 3 SD smaller compared to seven healthy volunteers (figure 2). A brain MRI was also performed (figure 3). The images were shown to a experienced neurologist who happened to be a specialist in NA (JR), and he raised the suspicion of NA. The diagnosis was confirmed by a significant number (20%) of acanthocytes in peripheral blood smear. Laboratory investigations

Figure 1 An example of the patients 18F-fluorodeoxyglucose (FDG) PET scan at the level of the basal ganglia. Note the absence of uptake in the caudate nucleus and putamen.

revealed no change in liver enzymes. Serum creatine kinase was elevated (1620 IU), but this test was taken after the PET investigations when NA was already suspected as a diagnosis.

DISCUSSION

Unclear neuropsychiatric symptoms that emerged after a psychologically traumatic event led the clinicians to suspect a post-traumatic conversion with motor and sensory symptoms. The patient's attributes of his life being ruined because of the accident, unsuccessful social rehabilitation,

peculiar behaviour and no neurological diagnosis gave way to speculations of psychological regression in this case.

NA is characterised by orofacial dyskinesias, chorea, tics, peripheral neuropathy, seizures and behavioral problems.¹ Typical findings are atrophy, neuronal loss and gliosis in the striatum, especially in the caudate. Globus pallidus, thalamus and substantia nigra are less affected. Structural brain imaging shows atrophy of the caudate and putamen, cerebral atrophy and ventricular enlargement.²

PET imaging has shown reduced blood flow and glucose metabolism affecting the striatum and frontal cortex.²

Figure 2 An example of 18F-fluorodopa (FDOPA) PET scan showing reduced uptake in the basal ganglia with putaminal predominance.

Involvement of the dopaminergic system in NA has been demonstrated with PET showing impaired putaminal [18F]dopa uptake and reduced striatal [11C]raclopride uptake ratios.³

Behavioural changes are seen in approximately one-third of the cases: irritability, aggression, obsessive-compulsiveness and disinhibition. However, involuntary movements are the most common initial presentation and behavioural problems are reported in about 13% at onset. In our patient the most probable diagnosis is autosomal recessive chorea-acanthocytosis or, possibly X linked McLeod syndrome, both of which could be nowadays confirmed by genetic testing.⁴ Our patient is a possible case of chorea-acanthocytosis in the absence of formal testing.

Unfortunately at the time of investigation of our patient, these genetic tests were not available.

The diagnosis of neuroacanthocytosis may be difficult if the physician is not aware of NA and its various symptoms and variable phenotype. This case, however, clearly points out the defects of some descriptive psychiatric diagnoses like dissociative disorders and somatoformal disorders, which are mainly based on the exclusion of a known physical reason for the symptoms. Although many psychiatric illnesses are triggered by stressful life events, some rare neurological disorders can lead the clinicians astray and give way to loose speculations about the aetiology of the symptoms.

Figure 3 An example of brain MRI transaxial slice at the level of the basal ganglia with no lesions which could account for the defects in FDG and FDOPA scans.

Learning points

- ▶ Inferences about causality are often made in psychiatry solely by temporal sequence of events, and this may be problematic.
- ▶ Descriptive psychiatric diagnosis, especially somatoformal and dissociative disorders, which lack clear positive criteria, may bear the highest risk to be confused with physical illness.
- ▶ In cases where the patient presents with symptoms that are atypical for dissociative or somatoformal disorders, the possibility of a rare neurological illness should be kept in mind.

Competing interests None.

Patient consent Obtained.

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