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Case report

THOMSEN'S DISEASE AND PSYCHIATRIC MISDIAGNOSIS: A CASE REPORT

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INRODUCTION

Thomsen's congenital myotonia is a rare disease with a frequency of 1/23,000. The transmission mode may be autosomal dominant, as for Thomsen myotonia, or autosomal recessive, as for Becker myotonia (Sun et al. 2001).

As a genetic disease of the neuromuscular chlorine channels, it affects the striated muscle fibres (Lossin et al. 2008).

The disease is believed to manifest in the first decades of life, but it can begin in the third or fourth decades.

The main symptom of the disease is a delay in contracted muscle relaxation generating muscle stiffness, cramps and muscle hypertrophy.

Myotonia can affect all muscles after contraction with a higher frequency in the lower limbs and hands. Great phenotypic variability exists for an identical mutation with different clinical presentations, thus hampering diagnosis (Colding-Jorgensen 2005).

Paradoxically, myotonia usually subsides on exertion (“warm-up” effect).

Muscle stiffness is therefore reduced by repeated exercises or movements, although the effect only lasts for the duration of the activity.

The aetiology is a mutation in the CLCN1 gene encoding chlorine-dependent voltage channels (CIC-1). The defective channels generate inappropriate hyperexcitability of skeletal muscle cells causing repeated depolarization and myotonia (Platt et al. 2009).

Electomyography (EMG) is used to confirm myotonia and the cold test coupled with effort can guide towards a genetic molecular diagnosis (Phillips et al. 2018).

Life expectancy is normal and prognosis favourable although functional impairment can diminish the quality of life (Gutmann et al. 1991).

This study illustrated the difficulty in diagnosing Thomsen's disease and the too-rapid interpretation of symptoms as a psychiatric pathology through the description of the case of a patient in 2021 who was referred for psychiatry consultation at the stress clinic at UCL Namur Godinne University Hospital Centre. Patient follow-up lasted one year and was based on data collected during 12 consultations.

CASE REPORT

In 2021, a 45-year-old patient was referred for a psychiatric consultation to improve stress management. She was divorced, her parents were deceased, and she had a brother and a son. The patient described having little contact with her family and having little information about her family's health history.

Two years earlier, due to relationship difficulties, she experienced a severe documented depressive episode that required three weeks of hospitalisation in a psychiatric ward. Afterwards, she was treated for two years with 10 mg of escitalopram and 2.5 mg of lorazepam.

She continued psychological and psychiatric follow-up care and was diagnosed with depression in partial remission. Her treatment plan was to take a more specific approach to stress management.

During the first consultation, the patient's mood had improved since the depressive episode of 2019, but she stated that she still had difficulty falling asleep and believed she needed lorazepam to be able to relax in the evening. She had moderate concerns about having a more serious health problem that doctors had not yet detected.

The patient reported that before her depressive episode, she practised sport intensively. She also stated that fatigue and diffuse muscle pain prevented her from taking up sport again.

She mentioned that she attended a physical medicine consultation, which concluded a likely diagnosis of fibromyalgia and treatment of her depression.

Follow-up care focused on stress management and the psychotherapies of somatization mechanism psychotherapies. During consultations, the patient mainly spoke of cramps and myalgia.

A relaxation session using the Jacobson technique (alternating contraction/relaxation) was prepared and performed. The session proved impossible as each contraction generated cramps and pain; the session had to be stopped.

Subsequently, the patient described eyelid spasms that occurred several times a day.

Given the symptomatology presented, another physical medicine consultation with coordination was organised with an EMG. The patient was also referred for a neurology consultation.

After a genetic consultation a year later, Thomsen's myotonia was diagnosed.

In a follow-up visit in 2022, the patient felt reassured to have an explanation for her difficulties and regained a correct quality of functioning.

DISCUSSION

The clinical case presented in this paper illustrated the difficulty of diagnosing rare diseases, especially when the family medical history is limited.

The case also demonstrated that although the diagnosis of depression was not erroneous, the notion of psychiatric history could bias the somatic assessments with the consideration of "non-specific" symptoms, such as muscle pain in this case, as somatization phenomena caused by chronic stress. The initial physical medicine consultation also focused on optimising her depression treatment.

In most of the diagnostic criteria for mental illnesses included in the DSM V, the criterion "the disorder is not attributable to the physiological effects of a substance or another medical condition" is of paramount importance in psychiatric follow-up care.

In this case, without the relaxation exercises with the direct observation of the muscular contracture phenomena, the diagnostic assessment would probably have been further delayed.

CONCLUSION

Diagnosing rare diseases is a difficult issue; thorough rigorous and repeated examination of personal and family history is preponderant. Diagnosis often takes time and requires careful observation of the evolution of symptoms when the family history is non-contributory.

In the particular case of psychiatric follow-up care, non-specific symptoms should be considered particularly, which are too quickly interpreted as somatization phenomena.

The contextual analysis of non-specific symptomatic manifestations must be conducted, even if a psychiatric diagnosis has been given. This emphasize the need of multidisciplinary approach.

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