

Case report

Psychosis in a patient with Usher syndrome: a case report

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SUMMARY The present report is the case of a 26-year-old man, born with Usher syndrome. The patient had had a significant hearing impairment since birth and had developed retinitis pigmentosa. He had originally been diagnosed with a depressive disorder and treated with antidepressants, with no subsequent improvement in his mental state. Following a deterioration in his mental state he was admitted for reassessment at the Queen Elizabeth Psychiatric Hospital, Birmingham, and antidepressants were stopped. It subsequently became apparent from observations, interviews with the patient and information from the patient's carers and relatives that he had a psychotic illness. Treatment was started with the antipsychotic drug risperidone, after which he showed significant improvement. The association between Usher syndrome and psychosis is discussed.

Introduction

Usher syndrome is a combination of congenital hearing loss, either mild or severe, and retinitis pigmentosa, a progressive eye disorder usually with onset in the teens or twenties that causes night blindness, loss of peripheral vision and eventual loss of central vision [1].

It is believed that Usher syndrome is a heterogeneous group of genetic diseases and there are three recognizable phenotypes characterized by progressive retinitis pigmentosa. The Usher syndrome type 1 individuals have abnormal vestibular disturbance and profound sensorineural hearing loss. The Usher syndrome type 2 individuals have normal vestibular function and mild to severe hearing loss [2]. The rare type 3 cases show a progressive hearing and sight loss from early adult life [3].

Case history

Mr M is a 26-year-old deaf male patient living with his hearing parents. He has two brothers, both hearing. The patient is unable to communicate with other family members because of their inability to use sign language. There was no history of psychiatric illness, deafness or Usher syndrome in the family. The patient did not have a history of drug or alcohol abuse or dependence.

Over the previous 2 years he had been treated for a depressive disorder with several different antidepressants and recently had been taking citalopram 20 mg in the morning and flupenthixol 0.5 mg twice daily. From the information provided by the family, the community psychiatric nurse and social worker it became clear that his mental state was deteriorating. He

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became more isolated, spending a lot of time in his room, neglecting himself and his hygiene and walking about the house during the night. He was not eating well and losing weight. He talked to himself in sign language, laughing to himself inappropriately and on several occasions showed anger to his mother and complained of headaches. He was frequently copying passages from books.

Following this deterioration in his mental state, the family became more concerned. A planned admission to Denmark House, Queen Elizabeth Psychiatric Hospital in Birmingham, United Kingdom, was arranged for further assessment in an environment that would address his mental health and both his deafness and visual problems (loss of peripheral vision). On admission, a thorough assessment was performed on his mental state. All medication was stopped and the patient observed and interviewed on several occasions. Several times he was seen talking to himself in sign language, looking around in different directions and, while sitting in the television lounge, he would appear preoccupied and considerable effort was needed to attract his attention.

We noticed from the medical history that there had been a deterioration in the patient's ability to communicate using sign language over the past 2 years. The patient had also experienced several falls, most probably caused by his vestibular problem and compounded by the loss of his peripheral vision (a phenomenon associated with Usher syndrome type 1). The behavioural symptoms were felt to reflect a psychotic disorder rather than a mood disorder and treatment was started with the antipsychotic medication risperidone 1 mg twice daily on the first day, then 2 mg twice daily thereafter. The patient appeared to improve and was no longer talking to himself, and

accordingly risperidone was increased to 3 mg twice daily.

The outcome of the therapy seemed to confirm a diagnosis of a psychotic illness and the possibility of a depressive illness was eliminated on the basis of frequent observation and interviews during the admission period. It was concluded that the *ICD-10* diagnosis should be schizophrenia unspecified 20.9 [4].

Discussion

Database searches (Ovid: Bibliographic Records) show there are limited published observations on patients with a psychotic illness associated with Usher syndrome deaf-blindness phenomenon. Only 11 (4.6%) of all the deaf, mentally ill, hospitalized patients in a New York State hospital had a diagnosis of Usher syndrome. Of these, 3 patients showed a clinical picture of schizophrenia and 4 exhibited psychotic features associated with mental retardation [5].

The association between different forms of mental health problems and either hearing and visual impairment, or both, is reported in the literature. For example, Cooper [6] reported that although the incidence of schizophrenia in prelingually deaf people was similar to that in a hearing population, contributory factors to the etiology of paranoid illness are: the psychological and social consequences of deafness, the possible contribution of sensory deprivation phenomena, and the interference of hearing loss in attention, perception and communication. Watt [7] studied the hearing and premorbid personality of patients whose illnesses began before the age of 60 years and concluded that premorbid paranoid personality and hearing loss are not associated with paranoid states in middle age.

Eastwood et al. [8] reported an association between paranoid symptoms and moderate to severe deafness, although the small number of patients in his sample makes it difficult to draw firm conclusions.

There are few reports about the possible association between Usher syndrome and psychosis [9]. However, it is important to note that Mangotich et al. [10] reported a correlation between progressive sensory deprivation and psychotic symptoms in a patient with previously undiagnosed Usher syndrome.

Technical difficulties in the evaluation and treatment of psychiatric disturbances in a deaf-blind population are well-recognized. Seligson [11] reported difficulties in communication and advised that several methods, such as handwriting in conjugation with sign language, would improve communication. Where there is a lack of effective communication skills, careful and close observation of behavioural manifestations may be the only means of examination, as in our patient. Evaluating certain aspects of appearance and general behaviour, for example, gait and posture, dress, facial expression, manner of relating, may also be helpful although they are not always

indicative of mental illness. The author has noted that isolation and withdrawal are seen more frequently in this population, so too are hallucinatory phenomena, but phenomena such as ideas of reference and delusional beliefs are less likely to be clearly evident.

It is worth mentioning that auditory hallucinations are a common phenomena in profoundly prelingually deaf schizophrenic patients. This cannot be explained in terms of patients describing other symptoms, and illustrates that these phenomena are not restricted to those who had heard and understood language before becoming deaf [12].

Recommendations

The association between Usher syndrome and psychosis is a rare one. It is important that both psychiatrists as well as ophthalmologists should be aware of the possibilities of such an association. It is hoped that this report will encourage future research and possible further case reports similar to ours in order to document and address the mental health needs of patients who are diagnosed with Usher syndrome.

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Prevention and promotion in mental health

The WHO Meeting on Evidence for Prevention and Promotion in Mental Health: Conceptual and Measurement Issues, was convened in Geneva in November 2001 to advance the work related to prevention of mental disorders and promotion of mental health. A group of experts from all WHO Regions discussed the definitions and concepts related to promotion and prevention, shared the current state of evidence to further develop the field, and advised WHO on its role in this area.

Prevention and promotion in mental health, based on the deliberations of the WHO meeting, highlights some of the basic issues in the field of prevention and promotion in mental health with special reference to the evidence base. It also outlines the role of WHO in advancing current knowledge and disseminating information among Member States, especially among developing countries. It is hoped that this document will help promote the wider use of appropriate and effective interventions and will assist in reducing the burden of mental disorders and enhancing the mental health of populations. Policy-makers will find the document useful as it provides an overview of some of the issues often debated among researchers and policy-makers with respect to prevention and promotion in mental health. The document can be obtained from the World Health Organization, 20 Avenue Appia, 1211 Geneva 27, Switzerland. It is also available free online at: <http://whqlibdoc.who.int/publications/2002/9241562161.pdf>