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USHER SYNDROME AND PSYCHIATRIC SYMPTOMS: A CHALLENGE IN PSYCHIATRIC MANAGEMENT

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SUMMARY

Usher syndrome, the most common case of deaf – blindness, may be associated with various psychiatric disorders. Inability of communication through spoken language in association with progressive visual impairment affects diagnostics and management in case of co-morbidity with mental disorder.

A patient with Usher syndrome and psychiatric symptoms is described and the difficulties in psychiatric assessment in her case are discussed.

A 28 years old woman with hearing impairment diagnosed at the age of 3 months and progressive pigmentary retinopathy diagnosed at the age of 19 years, has been treated for ADHD in childhood, eating disorder in adolescence and psychosis-like disorder in adult life.

Direct observation of patient behavior and the effects of pharmacotherapy were the main diagnostic procedures, since the use of sign language and handwriting was very limited. The limitations of management are discussed.

Key words: *usher syndrome - psychiatric symptoms – diagnosis - management*

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INTRODUCTION

It is not unusual for a psychiatrist to treat patients with co-morbid conditions. However, patients with sensory impairments like deafness or blindness pose a challenge in diagnosing and treating the underlying psychiatric symptoms.

Usher syndrome is an autosomal recessive genetic disorder manifested by hearing impairment, retinitis pigmentosa and variable vestibular deficit. Three clinical types of Usher syndrome have been defined (Waldeck et al. 2001, Keats 2002). Type I is characterized by profound congenital deafness, prepubertal onset of progressive retinitis pigmentosa, vestibular dysfunction and CNS abnormalities. Type II is characterized by moderate to severe congenital deafness, adolescent onset of retinitis pigmentosa, and absence of vestibular dysfunction. Type III is characterized by rapidly progressing auditory deterioration with complete hearing loss by early childhood, adolescent onset of retinitis pigmentosa, variable vestibular dysfunction and unknown CNS abnormalities. Prevalence of Usher syndrome is in range between 3 and 10 individuals per 100,000 worldwide (Waldeck et al. 2001).

Medline search based on the key words "Usher syndrome, psychiatry, psychosis, mental deterioration, hallucinations" revealed a limited number of articles, some of them with case reports. Patients presented with persecutory delusions, anger, fear, depression, anorexia, personal neglect, suicidal thoughts, bizarre behavior and deterioration in the ability to communicate using sign language (Waldeck et al. 2001, Hess-Rover et al. 1999, Jumalan & Fergusson 2003). Some authors suggested that persons with Usher syndrome may experience more psychiatric symptoms than general population (Hallgren 1959, Nuutila 1970). According to Schaefer the manifestation with psychotic symptoms might be secondary to a metabolic degeneration involving the brain more diffusely, since some cerebellar and cerebral abnormalities were also found (Schaefer et al. 1998).

Proposed explanations of relationship between Usher syndrome and psychotic illness are

similar genetic impairment, biochemical alterations and cerebral abnormalities (Carvill 2001).

Persons with Usher syndrome are unable to describe their thoughts and feelings through spoken language. Therefore, direct observation of patient behavior and detailed assessment are the most important diagnostic procedures. However, communication is only possible with handwriting and sign language in patients with partially preserved vision. In case of psychiatric symptoms, the limitation of communication may affect the diagnostic and treatment procedures.

We describe a female patient with rare and complicated co-morbidity of Usher syndrome and various psychiatric conditions in order to discuss the diagnostic and treatment possibilities in similar cases.

CASE REPORT

Ms. P.R., 28-yrs old, was born as a deaf child to normally spoken and hearing parents. Her mental and physical development in early childhood has been retarded and she started to walk at the age of 19 months. Deafness was diagnosed at the age of 3 months.

In family history two second-degree male relatives on mother's side were deaf and blind. There was no known history of psychiatric disorder in the family. No genetic evaluation has ever been performed.

At the age of eight the patient was referred to a child psychiatrist and diagnosed and treated for Attention deficit hyperactivity disorder (ADHD) for a short period. Later she did not receive any psychiatric treatment until the age of 21.

According to her mother, she learned to use one-hand sign language, had no problem with learning and later successfully graduated from secondary graphic school with modulated set-up program. Her learning abilities were excellent. She attended two years of college, but never worked. Since her early childhood she had been living with her mother, who was divorced. She never had contact with her father, but occasionally spent time with her grandmother and aunt. She was able to communicate with sign language and occasionally attended support groups for deaf people. Apart from that, she was socially isolated most of the time.

At the age of 19 she was diagnosed with Usher syndrome – retinitis pigmentosa by an ophthalmologist. Five years later her vision was reported to be "moderately affected but stable for the last 5 years, with the loss of peripheral vision".

She was treated on an outpatient basis for eating disorder at the age of 21. At that time, an alteration in her behavior was noticed. A psychotic disorder was suspected and treated with thioridazine with good response. At the age of 25 years panic - like attacks and anxiety appeared and she was treated with escitalopram, but her condition deteriorated. She became violent against her family members, with obsessive rituals. At that point she was admitted to University Psychiatric Hospital in Ljubljana. The evaluation of her mental status was very limited due to her poor sign language and written communication. Her written replies were extremely limited and finally she refused to communicate in this way. We consulted a professional interpreter of sign language, who also could not get any communication so the symptoms of disorganized thought were suspected. From the same reason psychological evaluation was not possible, although we estimated her primary intellectual potential to be within normal limits on the basis of her prior good school performance.

Diagnostic procedures like head CT- scan, EEG and blood findings showed no abnormalities. No neurological abnormalities were found. Treatment with the antipsychotic risperidone 1 mg BID was introduced with improvement in the patient's behavior. In the next 4 years she had ten hospital admissions, mostly as a consequence of poor compliance and relapse of behavioral symptoms, mostly violent outbursts, obsessive symptoms, and anorexia. We never managed to evaluate specific psychotic symptoms, however, from her behavior (like hiding under the bed, screaming, agitation) the presence of persecutory delusions or possible auditory hallucinations was suspected. Occasionally affective symptoms like depressed mood and crying was observed. She

was treated with various typical and atypical antipsychotics in low doses (haloperidol, aripiprazole, quetiapine, clozapine), in combination with antidepressants and anxiolytics. In the last year, the symptoms partly remitted and no further hospitalization was needed. She is doing well on a combination of clozapine 25mg BID and the antianxiety drug pregabalin 150mg daily. According to her mother, some obsessive rituals, opposing behavior, anorexia and phobic symptoms are occasionally present, but she can cope with everyday demands. However, her communication skills deteriorated and she abandoned her previous social contacts and spends most of her time alone or with her mother and grandmother. Throughout the treatment her mother was overprotective and expressing anxiety concerning the patient condition.

DISCUSSION

The literature review revealed a small number of articles about Usher syndrome and psychiatric disorder with the majority of them focusing on pathophysiology and etiology of the disorder (Waldeck et al. 2001, Schaefer et al. 1998, Carvill 2001). Reports about the difficulties in the evaluation, management and treatment of mental disorders in a deaf-blind population are limited (Hess-Rover et al. 1999, Jumalan & Fergusson 2003, Seligson 1983).

Authors advise careful and repeated observation of patient behavior, relevant history from family members and detailed assessment of communication such as handwriting in conjunction with sign language (Hess-Rover et al. 1999, Jumalan & Fergusson 2003). However, in case of Ms. P.R., such communication was impossible so the guidelines for diagnosis were patient behavior, history of her condition referred by her relatives, and her response to treatment. The symptoms observed in her case were similar to the ones experienced by other patients (Waldeck et al. 2001, Hess-Rover et al. 1999, Jumalan & Fergusson 2003, Wu & Chiu 2006).

We concluded that Ms. P.R. suffered from Usher syndrome type III and various psychiatric disorders. The diagnoses, based on clinical interpretation of signs and symptoms only, were ADHD in childhood, anorexia in adolescence, and obsessive – compulsive disorder, depression and organic delusional (schizophrenia – like) disorder in the adulthood. The possible presence of auditory hallucinations may contribute to this diagnosis, since they have also been found in deaf schizophrenic patients (du Feu &McKenna 1999). The deterioration of patient vision and domestic environment (over-protective mother, very limited social interactions) may contribute to the overall clinical picture. As suggested by some authors it is diagnostically important to consider the etiological, developmental, adaptive and social factors in interpreting patient condition. Our diagnostics and pharmacological treatment did not differ much from other published cases, however the best results were achieved with a combination of low-dose clozapine and a new antianxiety drug pregabalin.

CONCLUSIONS

Usher syndrome, a common ophthalmologic condition, may be associated with various psychiatric manifestations. Symptoms of organic delusional (schizophrenia-like) disorder in these patients are very rarely described. Our case is the first to be identified in Slovenia. As a consequence, psychiatrists and nursing staff may be unskilled in managing these complicated patients. In the lack of adequate communication to evaluate symptoms, a lot of speculation and “blind” introduction of pharmacotherapy, based only on patient behavior, may affect the competence and quality of treatment. The need for specialized interpreters, special diagnostic procedures, evaluation and treatment, as well as social rehabilitation pose a challenge and are vital for good clinical management.

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