BAOJ Psychology
An Open Access Journal

BIO ACCENT

BAOJ Psychol
Case Report
Volume 3

A case report of usher's syndrome with autistic spectrum disorder: Challenges in diagnosis and management

Roufael R[1]*; Yassein K[1]; Kuntal V[2]; Joseph S[2]

[1]Health Education and Improvement Wales (HEIW), UK.

[2]Cwm Taff Morgannwg University Health Board, UK.

***Corresponding Author: Roufael R**

Health Education and Improvement Wales (HEIW), United Kingdom.

Email: blessed_rose_1979@hotmail.com

Received: Nov 14, 2022

Accepted: Jan 03, 2023

Published: Jan 05, 2023

Keywords:
Usher's Syndrome;
Autistic Spectrum Disorder;
Vision;
Hearing;
Communication;
Ritualistic;
Idiosyncratic Speech;
Theory of the mind.

Abstract

Usher's syndrome is a rare genetic disease that affects both hearing and vision. It is defined by the association of sensorineural hearing loss and visual impairment due to retinitis pigmentosa. There is paucity in research describing patients with Usher's syndrome and comorbid Autistic Spectrum Disorders (ASD). Assessment and management of mental disorders, in association with Usher's Syndrome, can be particularly challenging due to dual sensory. The authors describe a case of a patient with Usher's Syndrome referred to mental health services with behavioral difficulties. The patient required extensive support with her Activities of Daily Living (ADLs), however did not engage with staff support leading to high risk of self-neglect. She was also requesting to leave the supported accommodation to live independently. The assessors depended in their assessment mainly on collateral history and behavioral observation. The patient showed multiple features suggestive of ASD; including difficulties in maintaining a two way conversation, inflexible adherence to a pattern of ritualistic behaviors, stereotyped repetitive motor movements and idiosyncratic speech, in addition to hyper-reactivity to different sensory stimuli. These difficulties appear to have stemmed since her childhood, as her family used to struggle with providing her care because of her same behavioral patterns. The assessors made recommendations based on their findings, including referral for specialist diagnostic assessment for ASD and conducting mental capacity assessment for the patient's ability to decide on her future accommodation. The authors also discuss the possible underlying etiologies for such presentation, including delayed development of Theory of Mind (ToM) because of the limited social experience caused by early visual loss and hearing impairment.

Citation: Roufael R, Yassein K, Kuntal V, Joseph S. A Case Report of Usher's Syndrome with Autistic Spectrum Disorder: Challenges in Diagnosis and Management. BAOJ Psycho. 2023; 3(1): 1001.

Introduction

Usher's syndrome is is a rare genetic disease that affects hearing and vision. It is defined by the association of sensorineural hearing loss and visual impairment due to retinitis pigmentosa [1]. The prevalence of Usher's syndrome ranges from 3.2 to 6.2 per 100,000 [2]. It may be associated with various psychiatric disorders [3]. Inability of communication through spoken language in association with progressive visual impairment affects diagnostics and management of comorbid mental disorders [4]. Herein, the authors report a case of Usher's syndrome with autistic symptoms and highlight the challenges faced with assessment and management of this uncommon presentation.

Case report

Miss X, a female patient in her fifties, was referred to mental health services with behavioral difficulties. She has a diagnosis of Usher's syndrome. She was living in a supported accommodation, requiring extensive support with all her Activities of Daily Living (ADLs), however she was resisting support leading to high risk of self-neglect and poor physical health. She was also requesting to move on to live independently in the community, which raised considerable concerns about her ability to understand her care needs.

Collateral history

Miss X used to live with her mother in her childhood. She attended main stream education. There is limited information about her developmental history. She was registered blind in her early twenties. Her mother used to provide her with extensive support with her ADLs, however Miss X used to be resistive to interventions. In her adulthood, Miss X struggled to live independently in the community, so she then moved in to a supported accommodation with a supported care plan in place.

Miss X showed the same pattern of behavior towards the supporting staff; requiring support however resisting it. Staff described that she had inflexible adherence to a pattern of ritualistic behaviors. She would struggle to engage with any task outside her rigid routine. Her acceptance of support was very specific, for example, accepting food, but declining personal care. She was very specific about time frames for different tasks.

She showed patterns suggesting difficulties with executive functioning. Miss X would voice her need to do a particular task, however did not actually execute the task unless the exact steps were explained to her. For example, she required prompting to drink, even when she was able to identify feeling thirsty.

Miss X seemed to present with some sensory difficulties. She appeared to be hyper-reactive to the sensory input of having water on her skin, leading to reluctance to engage in tasks related to hygiene.

Miss X vision was pin-hole vision and she required two hearing aids in order to hear clearly. She was not on any psychotropics. She developed recurrent infections secondary to self-neglect.

Mental state examination

The assessors depended mainly on behavioral observation, due to Miss X limited engagement. Miss X showed signs of self-neglect. She went through a number of rituals including taking tablets in a particular order with a particular number of sips from her drink for each tablet. She showed a pattern of stereotyped repetitive motor movements, for example, she would tap her cup against her lip particular number of times before drinking. She showed idiosyncratic use of language, spelling some words out instead of saying them. She could not maintain a two way conversation. She had a number of repetitive stock answers that she used when she struggled with maintaining a conversation; those answers were out of context. Her mood was subjectively and objectively anxious, with reactive affect. There was no evidence of depressive symptoms. There was no evidence of delusional thoughts, hallucinatory experiences or any other psychotic phenomena. She was oriented to time, place and person. Miss X had no insight into her difficulties. She expressed her wish to live independently in the community and could not understand professionals concerns and views regarding her care needs, which raised the question about her understanding and capacity to plan for her future accommodation.

Working diagnosis

Miss X showed features suggestive of ASD; including difficulties in maintaining a two way conversation, inflexible adherence to a pattern of ritualistic behaviors, stereotyped repetitive motor movements and use of repetitive stock words, in addition to her hyper-reactivity to different sensory stimuli. These difficulties appear to have stemmed since her childhood, as her mother used to struggle with providing her care because of the same behavioral patterns that her carers are finding now.

Management

The assessors made recommendations based on the findings. The first recommendation was to include input from the autism specialist service for specialist diagnostic assessment for ASD. It was highlighted that there might be some practical difficulties with conducting the standard assessments, because of Miss X difficulties with maintaining conversation and engaging in any activities outside her rigid routine. The second recommendation was to conduct mental capacity assessment to look into Miss X capacity to decide on the future accommodation that would meet her care needs, taking into consideration her difficulties in understanding other people's concerns which is caused by the autistic traits.

Discussion

There are 3 subtypes of Usher's syndrome that have been identified [5]. People with Usher's are congenitally deaf, and start to lose vision early in life, with difficulties in balance. Individuals with Usher's II also experience hearing loss, are not profoundly deaf, with no noticeable balance problems. Individuals with Usher's III are not congenitally deaf, but gradually lose their sense of hearing and vision and some of them experience balance difficulties [6,7]. Different mental and behavioral disorders have been reported among adults with Usher's Syndrome including; psychosis [8,16], mental retardation [17], anorexia nervosa [18], Attention Deficit Hyperactivity Disorder (ADHD) [8], anxiety, social isolation and depression [19,20].

Dammeyer, J. found that a quarter of children with Usher's Syndrome had comorbid mental and behavioral disorders. The types of disorders observed were not of uniform presentations and included mental retardation, schizophrenia, and conduct disorder. Also, two out of the 26 children in the review showed atypical autism. Some different mechanisms that could influence the development of mental and behavioral disorders were stated, including; genes associated both with Usher's syndrome and mental disorders, reaction to stress due to progressive loss

of vision, and the dynamic interplay of hearing, vision, and language impairment [21].

As for association between Usher's Syndrome and autism, literature shows that patients with Usher's Syndrome have limited access to information caused by visual loss, reducing the degree of their social experience, thereby affecting the development of their Theory of the Mind (ToM) [22], which is similar to patients with ASD. Other studies also showed that ToM understanding, differed significantly between groups of deaf children when compared to hearing controls. Limited early access to family conversations about thoughts and feelings in hearing families, was thought to contribute to the delayed development of ToM in hearing impaired children [23]. It is to be noted that children with dual sensory impairments can present with symptoms of communicative impairments, similar to autism, however such symptoms can sometimes disappear, when visual or oral communication has adequately been developed [24], which is different to patients with Usher's Syndrome as their sensory impairments are progressive irreversible.

Assessment of mental and behavioral disorders, especially autism, in patients with Usher's Syndrome can be particularly challenging and can depend mainly on collateral history and behavioral observation. Management of these associated mental disorders would include optimizing medical treatment if indicated, optimizing their sensory aids, as well as support with functioning. Intervention by means of communication and language rehabilitation can be important in preventing mental and behavioral disorders and psychosocial difficulties, including the risk of social isolation and depression in this population [21].

Conflicts of interest: There are no conflicts of interest.

Financial support and sponsorship: Nil.

References

1. Petit C. Usher Syndrome: From Genetics to Pathogenesis. Annu Rev Genomics Hum Genet. 2001; 2: 271–297.
2. Koenekoop RK, Arriaga MA, Trzupek KM, Lentz JJ. Usher Syndrome Type I. 1993.
3. Bailly D, Dechoulydelenclave MB, Lauwerier L. Hearing impairment and psychopathological disorders in children and adolescents. Review of the recent literature. Encephale. 2003; 29: 329-337.
4. Rijavec N, Grubic VN. Usher syndrome and psychiatric symptoms: a challenge in psychiatric management. Psychiatria Danubina. 2009; 21: 68-71.
5. Keats BJ, Savas S. Genetic heterogeneity in Usher syndrome. Am J Med Genet A. 2004; 130A: 13-16.
6. Yan D, Liu XZ. Genetics and pathological mechanisms of Usher syndrome. J Hum Genet. 2010; 55: 327-335.
7. Williams DS. Usher syndrome: animal models, retinal function of Usher proteins, and prospects for gene therapy. Vision Res. 2008; 48: 433-441.
8. Rijavec N, Grubic VN. Usher syndrome and psychiatric symptoms: a challenge in psychiatric management. Psychiatr Danub. 2009; 21: 68-71.
9. Mangotich M, Misiaszek J. Atypical psychosis in Usher's syndrome. Psychosomatics. 1983; 24: 674-675.
10. Hess-Röver J, Crichton J, Byrne K, Holland AJ. Diagnosis and treatment of a severe psychotic illness in a man with dual severe sensory impairments caused by the presence of Usher syndrome. J Intellect Disabil Res. 1999; 43: 428-434.
11. Jumaian A, Fergusson K. Psychosis in a patient with Usher syndrome: a case report. East Mediterr Health J. 2003; 9: 215-218.
12. McDonald C, Kenna P, Larkin T. Retinitis pigmentosa and schizophrenia. Eur Psychiatry. 1998; 13: 423-426.
13. Viala A, Nicot T, Levy F, Vacheron MN. A case of Usher's syndrome associated with psychotic symptoms: diagnosis and follow-up in a psychiatric unit. Encephale. 2009; 35: 286-291.
14. Koizumi J, Ofuku K, Sakuma K, Shiraishi H, Iio M, et al. CNS changes in Usher's syndrome with mental disorder: CT, MRI and PET findings. J Neurol Neurosurg Psychiatry. 1988; 51: 987-990.
15. Wu CY, Chiu CC. Usher syndrome with psychotic symptoms: two cases in the same family. Psychiatry Clin Neurosci. 2006; 60: 626-628.
16. Rais T. Acute Psychosis with Paranoid Features in a Young Patient with Usher's Syndrome: A Case Report. J Tenet Hum. 2006; 18: 57-88.
17. Sharp CW, Muir WJ, Blackwood DH, Walker M, Gosden C, et al. Schizophrenia and mental retardation associated in a pedigree with retinitis pigmentosa and sensorineural deafness. Am J Med Genet. 1994; 54: 354-360.
18. Chapman V, Valmana A, Lacey JH. A case report of Usher's syndrome and anorexia nervosa. Int J Eat Disord 1998; 23: 223-226.
19. Miner ID. Psychosocial implications of Usher syndrome, Type 1, throughout the life cycle. J Visual Impair Blin 1995; 89: 287-297.
20. Damen GWJA, Krabbe PFM, Kilsby M, Mylanus EAM. The Usher lifestyle survey: maintaining independence: a multi-centre study. Int J Rehabil Res. 2005; 28: 309-328.
21. Dammeyer J. Children with Usher syndrome: mental and behavioral disorders. Behav Brain Funct. 2012; 8: 16.
22. Henricson C, Frölander HE, Möller C, Lyxell B. Theory of Mind and Cognitive Function in Adults with Alström or Usher Syndrome. J Vis Impair Blind. 2016; 110: 349-366.
23. Peterson CC (2016) Empathy and Theory of Mind in Deaf and Hearing Children. J Deaf Stud Deaf Educ. 2016; 21: 141-7.
24. Hoevenaars-van Boom den MA, Antonissen AC, Knoors H, Vervloed MP. Differentiating characteristics of deaf blindness and autism in people with congenital deafblindness and profound intellectual disability. J Intellect Disabil Res. 2009; 53: 548-558.