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Diagnostic and Therapeutic Challenges due to Psychosis and Catatonia in Non-syndromic Retinitis Pigmentosa: A Case Report

To the editor,

Retinitis Pigmentosa (RP) is a group of heterogeneous, inherited retinal disorders characterized by the degeneration of the photoreceptor cells and, in extreme cases, the retinal pigment epithelium. The patients initially experience night blindness and eventually progressive constriction of the visual field, leading to central vision loss or even complete blindness.[1] The prevalence of RP is 1:3000 to 1:7000.[2] RP is classified into three types: syndromic (involving special senses of the nervous system such as hearing), non-syndromic (not affecting other organs or tissues), and systemic (affecting multiple organs). Similar to the other variants, the mode of inheritance of non-syndromic RP include autosomal dominant (15%–25%), autosomal recessive (5%–20 %), X-linked (5%–15%), unknown: simplex (40%–50%), and digenic (very rare).[3] Few reports exist regarding the intriguing relationship between syndromic RP (ushers syndrome) and psychiatric illnesses, the most common being schizophrenia with manifestations of delusions and hallucinations.[4] However, whether psychiatric disturbance can co-occur in non-syndromic RP and whether it can have other varied psychopathology has not been studied earlier.

We describe a rare case of a young female with non-syndromic RP presenting with psychotic features, including catatonic symptoms and auditory hallucinations, with a difficult-to-treat episode requiring electroconvulsive therapy (ECT).

Case Presentation

A 20-year-old female with no past or family history of psychiatric illness presented with an acute onset behavioral disturbance lasting for one month, characterized by irrelevant speech, wandering behavior, hallucinatory behavior (claiming to hear voices, without perceiving their images), agitation, and disturbed sleep and appetite. Her medical history revealed progressive deterioration of vision since childhood. There was no family history of visual impairment. Her enduring visual impairment led to difficulties in assessing her present state perceptual abnormalities. Her blood biochemistry (complete blood counts, renal and liver function tests) and electrocardiogram returned normal. A diagnosis of acute and transient psychotic disorder, based on the *International Classification of Diseases, Tenth Revision*, was made. She scored 39 in the Brief Psychiatric Rating Scale (BPRS).

Ophthalmology opinion was sought and a comprehensive evaluation was done for the visual impairment. The fundi examination revealed retinal pigment clumping, waxy disc pallor, and arteriolar attenuation. Subsequently, the possibility of a syndromic RP was evaluated. The absence of hearing impairment (noted in the clinical examination) ruled out usher syndrome. The lack of ophthalmoplegia, ataxia, dysphagia, and cardiac conduction deficits eliminated the possibility of Kearns–Sayre syndrome. Absence of steatorrhoea and peripheral neuropathy and normal peripheral blood smear ruled out abetalipoproteinemia. A normal sexual and psychological development ruled out Bardet–Biedl syndrome.

She was started on T. Risperidone 2 mg/d and the dose was gradually titrated to 6 mg/day. Risperidone was chosen primarily because, as an atypical antipsychotic, it was safer than the typical antipsychotics (especially, chlorpromazine and thioridazine), which have an increased propensity to cause ocular side effects such as retinopathy.[5] She was also on T. clonazepam 3mg/day for sedation. Her psychotic symptoms improved significantly within two weeks, as reflected by the BPRS score of 28. While tapering down the dose of clonazepam from 3mg/day to 2 mg/day, she developed catatonic symptoms of excitement, verbigeration, negativism, withdrawal, and perseveration. She scored 11 on Bush Francis Catatonia Rating Scale (BFCRS). With lorazepam (up to 8 mg/day), she showed minimal improvement in catatonic symptoms, and her oral intake was compromised. ECT was administered, and the catatonia resolved after two sessions (BFCRS = 0, BPRS = 25). After discharge, lorazepam was tapered and stopped over four weeks. Psychosocial interventions such as supportive counseling and educating on personal safety and the provision for disability benefits were done. The patient was on anti-psychotic prophylaxis for six months, was maintaining well (BPRS = 20), and did not have further relapses.

Discussion

This report highlights that apart from the syndromic variety of RP, non-syndromic RP also can have psychiatric complications. Additionally, psychosis in non-syndromic RP can present with catatonic symptoms. Psychoses in syndromic RP had mostly presented with hallucinations and delusions, and catatonic symptoms appear to be a rare phenomenon.[2,6] As noted in our patient, a thorough medical history and clinical examination are necessary to eliminate the variants of syndromic RP and to arrive at a diagnosis of non-syndromic RP.

The etiopathogenesis of psychotic symptoms in RP remains unclear. Studies were able to formulate certain postulates regarding the origin of psychotic symptoms in syndromic RP: an independent genetic predisposition towards developing both ushers syndrome and schizophrenia together,[4] a diffuse involvement of the central nervous system in RP leading to polymorphic clinical presentations involving perceptual and behavioral abnormalities,[7] or the psychotic states in RP being a stress-related response consequent to progressive sensory impairment.[8] Our report will add to the literature that psychosis in non-syndromic RP can present with brief-lasting psychotic states and catatonia and may require intensive treatment strategies such as ECT.

Conclusion

The report will inform the clinician that enduring sensory impairment can lead to poor communication, with consequent diagnostic difficulties. Hence, a thorough medical history and comprehensive mental status examination may be required to overcome such hardships. Future studies should explore the genetic underpinnings of the association of non-syndromic RP and psychosis.

Informed consent was taken for patient management and write-up of the report.

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Duration of Illness at the Time of Disability Certification for Mental Illness and Neurological Disorders

The Government of India has issued guidelines for assessment and certification of disability according to Rights of Persons with Disabilities Act 2016.[1] Disability due to mental illness is measured and quantified by Indian Disability Evaluation and Assessment Scale (IDEAS). While the assessment for certification of neurological disability "is usually done six months after the onset of disease," no minimum duration is specified for mental illness.[1] The Rehabilitation Committee of Indian Psychiatric Society, which drafted IDEAS, had recommended that the duration of illness should be a minimum of two years at the time of certification, and the number of months disabled in the last two years is scored.[2] In the gazetted version of IDEAS, the minimum duration of illness is not specified and the domain "the number of months disabled in the last two years" was replaced with the duration of illness (a score of 1 for duration < 2 years and 4 for duration > 10 years).[1] As per law, disability due to mental illness can be certified even during the first consultation for mental illness.

According to the National Mental Health Survey 2015–2016, the median duration of time taken from the onset of symptoms of mental illness to seeking professional care varies from 2.5 months (for depressive disorder) to 11 months (for bipolar affective disorder).[3] The mean duration of illness before seeking consultation at a tertiary center was around 5.5 years.[4] There is a lack of literature about