Case Report

Primary Growth Hormone Deficiency And Usher Syndrome: A Case Report

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Abstract:

Introduction: The Usher syndrome (USH) is an autosomal-recessive disorder refers to The combinded bilateral sensorineural hearing loss, retinitis pigmentosa (RP), and in some cases vestibular dysfunction. There are three clinical types of Usher syndrome: type 1, type 2, and type 3. Type 3 is characterised by progressive hearing loss and variable age of onset of retinal degenerationand he or she will usually require hearing aids by mid- to late adulthood. Night blindness usually begins sometime during puberty.

Case Presentation: The present case reports is a 13 years-old male with type 3 of usher syndrome syndrome who developed a previously undescribed growth hormone de- ficiency.

Conclusion: We sugesst usher syndrome type 3 could be a primery GH deficiency disorders.potential link between usher syndrome and GH deficiency is still unclear and needs further studies.

Keywords: Usher syndrome,GH deficiency, Retinitis pigmentosa

Introduction:

Usher syndrome is an autosomal-recessive disorder that causes bilateral sensorineural hearing loss, retinitis pigmentosa (RP), and occasionally vestibular dysfunction(1). von Graefe was first to report the association of retinitis pigmentosa and congenital hearing loss(2). Three subtypes are recognized by the International Usher Syndrome Consortium: Type 1 is characterised by profound congenital deafness, retinal degeneration beginning in childhood, and progressive vestibular dysfunction; Type 2 is characterised by moderate to severe hearing impairment, later onset of retinal degeneration, and normal vestibular function; Type 3 is characterised by progressive hearing loss and variable age of onset of retinal degeneration(2).It is considerd to be the most frequent cause of deaf-blindness in adult(2).

In this paper, we report on an Iranian boy with usher syndrome who developed a previously undescribed growth hormone de-ficiency.

Case Presentation:

The present case reports is a 13 years-old male, white , elementary student who is born of normaly spoken and hearing parents and they are consanguineous (iranian first cousin) . one presented with congenital sensorineural hearing loss. That had been diagnosis progressive when he was 1 years old. In family history two second – degree female relative s on father s side were deaf .

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there was no known history of Grows hormone deficiency disorder in the family. No genetic evaluation has . ever been performed. According to her mother she learned to use one –hand sign language . had no problem whith learning and later successfully graduated from basic school with modulate set-up program.her learning ability were excellent . Vestibule function and gait is normal.at the age of 5 he was diagnosed with usher

patient afflicated with loss of sight and night vision in -5 years old and detect of peripheral vision and referred to ophtalmologist that diagnosed retinit pigmentosa in aquatorial regien and usher's syndrome.and in ophtalmological exam patient visial acuity was9/10 bilaterally that was refined to 10/10 by glass. Right eye plano – 1/00 ×20 and left eye plano – 0/75 × 170.patient IOP and slit lapt examination was normal.In funduscopy RPE atrophy was visible and not good feveal reflex but not prominent foveal atrophy and bone spicule was visible . this patient was diagnosed as mild case of retinit Pigmentosa cararact Not detected. Conjuctival and sclera and cornea was normal.

He had hearing aid that confirmed hearing loss in ENT consult. In primetry generalized field constriction was detected in both of eyes. In examination no cell and flare detected in anteriorchamber and anterior viterous. Audiology examination and paraclinic data usher syndrome was confirmed.

Discussion:

Usher syndrome 1, 2, and 3 can be distinguished by differences in audiovestibular features and there are many variations in the clinical symptomes in usher syndrome patiants . there for the classification of usher type 1 ,2 and 3has been complicated(1). Also usher syndrome type 3 can mimic other type of usher syndrome (3, 4).

We reviewed the literature for usher syndrome and the association GHD. There are no cases reported but there have been few reports of psychotic and another disorder in relationship with this disorder.

in a series of papar of Praharaj SK Mania episode in a 30-year-old male with Usher syndrome type II was analyzed. Various neuropsychiatric disorders have been reported to occur in those with Usher syndrome, including schizophrenia-like disorder, atypical psychosis, recurrent depressive illness, neurotic disorder, and mental retardation; however, bipolar disorder is not common in those with Usher syndrome(5). Totally Some patients of any types of usher syn develop a psychotic illness, the etiology of which is still debated (2). another report article about association usher syndrome and psychotic disorders had similar result. They report a 57-year-old man usher syndrome with sever psychosis that required several hospitalisations, for acute states with disruptive behaviour, aggressiveness against his mother, persecutory delusion and auditory hallucinations, self-talking, major anxiety, and depressive affects, without dissociation(2).

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Another disorder association with usher syn reported is Secondary vasoproliferative retinal tumor associated with Usher syndrome type 1. Secondary vasoproliferative retinal tumor that generally affect healthy persons between their fourth and sixth decades and may lead to severe vision loss as a result of intraretinal hemorrhages and exudates. They describe a case of unilateral vasoproliferative retinal tumor associated with retinitis pigmentosa in a patient with Usher syndrome type 1(2). lynch SG describe an individual with type 1 ushers syndrome and multiple sclerosis-like illness(6). Mri scan showed vermain atrophy on T1-weighted images and multiple white matter images although MRI demonstrate increase signal intensity on weighted images are reported in some individuals whith usher syndrome .they describe oligoclonal bands were present in the spinal fluid. The possibility of linkage between the two disease is raised(6).

Anothers reports talking about the relationship between Usher syndrome and psycosis with capgras syndrome. They illustrate a case that clearly demonstrate psychotic syndrome in ushers syndrome type 3 and several theoriesaround this association have been proposed these theories of

Association include a genetic link between the gens responsible for schizophrenia and the genes for usher syndrome(7). Our case suggests that usher syndrome should be taken into account when there is of growth hormone deficiency of unknown cause. we sugesst usher syndrome type 3 could be a primery GH deficiency disorders.potential link between usher syndrome and GH deficency is stiill unclear and needs further studies.

Conflict of Interests:

The authors declare that there is no conflict of interests regarding the publication of this paper.

Acknowledgment:

We are thankful to our colleagues who provided expertise that greatly assisted the research.

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