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Schizophrenia like psychosis in Alport syndrome

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ABSTRACT A 28 year old male with Alport syndrome presented with preoccupied behaviour, hearing voices, sleep disturbance and abusive behaviour for past 5 years. He was diagnosed as a case of Schizophrenia like psychosis and followed up. This case is presented to focus on the clinical similarities between the presentation of this disorder and schizophrenia, suggesting a possible similar genomic location or a genetic mechanism of transmission for both disorders.

KEYWORDS : Alport syndrome, psychosis, schizophrenia

INTRODUCTION:

Alport syndrome is an inherited disorder characterised by hematuria and mild proteinuria (less than 1-2g/24hours), thinning and splitting of glomerular basement membrane followed by chronic glomerulosclerosis leading to renal failure in association with sensorineural deafness, lenticonus of anterior lens capsule, dot and fleck retinopathy and rarely, mental retardation or leiomyomatosis[1]. Most patients have defects in type IV collagen chainsfound in the basement membrane due to mutations in the collagen genes. Alport syndrome is also called hereditary nephritis. Alport syndrome was first identified in a British family by University of Edinburgh Medical School graduate CecilA. Alport in 1927.This genetic disorder affects 1 in 50,000 children. Four forms of Alport syndrome are now recognised. Classic Alport syndrome is the most common form, which is an X-linked disorder. Other forms of Alport syndrome are an X-linked form associated with diffuse leiomyomatosis, an autosomal recessive form and an autosomal dominant form. The diagnosis of classic Alport syndrome is based on X-linked inheritance of hematuria, sensorineural deafness and lenticonus. Primary treatment is control of systemic hypertension and use of ACE inhibitors to slow renal progression. Dialysis is needed and renal transplantation is usually successful inspite of the development of anti GBM antibodies against the collagen deficient in their native kidneys.

CASE REPORT:

The routine blood biochemistry panel done during a surgical workup for crush injury-right hand in a 28 year old man revealed, serum creatinine of 8.4mg/dL, microscopic hematuria, albuminuria and associated hypertension. He was diagnosed with chronic kidney disease stage 5. He was started on a salt, fluid and protein restricted diet, supplementation of iron& calcium along with antihypertensive medications and peritoneal dialysis by the nephrologist. Analysis of the pedigree chart revealed a possible X-linked inheritance. His mother succumbed to chronic renal failure at 36 years of age. Ophthalmic examination revealed the presence of lenticonus in right eye. Audiogram detected bilateral sensorineural hearing loss. He was placed on maintenance hemodialysis twice a week and waitlisted for cadaver kidney transplant.

He was referred from nephrology department to the psychiatry outpatient department, with the complaints of being preoccupied, hearing voices, sleep disturbance, talking to self, anger outbursts and abusive behaviour on and off for past 5 years and worsening for the past 6 months. It was of insidious onset and progressive in nature.

There was no previous history of psychiatric illness, no history of any substance use and no significant. Family history revealed alcohol related complications leading to death of his father.

His birth and developmental milestones were normal. He had average scholastic skills and completed 12th standard with a score of 50 percent. He worked as a machine operator in a private company with normal pre-morbid functioning.

On mental status examination he was co-operative for the interview and was dressed appropriately with sustained gaze contact. He was oriented to time, place, person, and was found to be occasionally preoccupied. His talk was relevant and coherent with sad, restricted affect. He had auditory and visual hallucinations. He described the voice of Lord Vinayaka speaking to him in soothing words frequently. He also claimed that he could converse with the Lord. He also could describe vivid images of the Lord Vinayaka in front of him which was suggestive of visual hallucination. There was no evidence ofany formalthought disturbance.

His attention was arousable with intact memory, average intelligence and impaired abstraction. His judgement and insight was preserved. On PANSS this patient scored 74 on day 1, 50 on day 15 and 38 after 1 month. We made a diagnosis of schizophrenia like psychosis (non-organic psychosis) and started him on Haloperidol 1.5mg twice daily, Trihexyphenidyl 2mg once daily and Alprazolam0.5mg at night. Despite repeated requests our patient refused to come for further assessment after 1 month. Nevertheless his caregivers confirmed to us that he was maintaining well and on regular medication.

DISCUSSION :

To our knowledge, this is the first case report in India describing the occurrence of schizophrenia like psychosis in a patient with Alport syndrome. We present this case in order to document its rarity and to stimulate further research work in this area.

Alport syndrome is a rare cause of renal failure and the manifestations of psychiatric symptoms is also a rarity. Jais et al.(2000) studied the natural history in 195 families with X-linked Alport syndrome, over a period of 4 years, and discussed about genotype and phenotype correlations in affected males[2].

A review of literature done on 58 cases of Alport syndrome by Gubler et al. (1981) over a period of 20 years, has mentioned about the presence of microscopic hematuria in all cases[3].

A study done by Thomas N. Wise(1977), mentions two male patients with hereditary chronic nephritis (Alport syndrome) presenting with acute psychiatric reactions while on maintenance hemodialysis[4].

Shields et al. (1990) have studied the relationship of Alport syndrome and psychosis among the members of a family. Alport syndrome has been mapped to the long arm of the X-chromosome. Some studies also support a genomic locus on the X-chromosome in at least some cases of manic- depressive disorder and schizophrenia[5].

The advanced kidney disease was not present when psychosis was diagnosed in our patient. Therefore the occurrence of psychotic symptoms does not appear to be a consequence of uremic toxicity to the central nervous system. Both Alport syndrome and schizophrenia have clinical similarities. Clinical onset in adolescence, with peak illness at about 40 years of age, severe course in males are similarities

that are not aetiologically related. Nevertheless, one can safely speculate that the two illnesses are likely to have a common genomic location or genetic mechanism of transmission. We strongly recommend further studies in this area to explore for candidate genes and linkage to the pseudoautosomal regions (long arm) of the X-chromosome.

CONCLUSION:

The psychiatric manifestations in Alport syndrome is a pointer towards transmission of at least some cases of schizophrenia through the X-chromosomal genes. Future research can clarify our knowledge in this exciting area and possibly lead to the identification of a specific gene for schizophrenia.

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