

A RARE CAUSE OF KIDNEY FAILURE : CASE REPORT OF A YOUNG CHILD.

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Abstract : Action myoclonus–renal failure syndrome (AMRF) is a rare autosomal recessive disease. The first description of AMRF was in four French Canadian patients in 1986. Its exact prevalence is unknown. symptoms can appear between ages of 15 and 25, but it can appear at younger or older ages. It caused by mutations in the *SCARB2* gene. Clinical manifestations are dominated by fine tremor and proteinuria, followed by action myoclonus, renal failure and occasional generalised tonic-clonic seizures. Neurologic manifestations can appear before or after the renal manifestations. The aim of this work is to underline the clinical manifestations of this rare pathology which can lead to renal and vital prognosis if it is discovered at a very advanced stage.

MATERIALS and METHODS : Male child aged 13 years old, followed since the age of 2 years for epilepsy resistant to pharmacological treatment ; the child was initially treated with monotherapy, then by bitherapy and then by triple therapy due to the persistence of seizures ; with a good psychomotor development, he has a 3rd degree of consanguinity, and elder brother recently followed for end-stage renal failure. kidney failure was discovered accidentally during a hospitalisation for toxidermy. The clinical examination was unremarkable. The urine strip showed a cross of proteinuria and a cross of hematuria. biological test revealed blood urea at 1.19 and creatinine at 51.6. Abdominal ultrasound showed kidneys of chronic nephropathy with peritoneal effusion of low abundance. renal biopsy showed the appearance of chronic interstitial nephritis.

Keywords: Action-myoclonus renal failure syndrome, rare disease, *SCARB2* gene

INTRODUCTION :

Action myoclonus–renal failure syndrome (AMRF) is a rare autosomal recessive disease.[1] The first description of AMRF was in four French Canadian patients in 1986. Recent studies have demonstrated that this syndrome has a larger ethnic and geographical distribution, it is misdiagnosed, as cases with predominantly renal presentation and later neurological disease are under-recognised.[2]. Its exact prevalence is unknown. Just about 38 individuals with the condition have been described in the medical literature. This disease is typically begins causing symptoms between ages 15 and 25, but it can appear at younger or older ages. The age of onset and the progression of the condition differ, even among members of the same family. [1]. AMRF associates myoclonus epilepsy and renal impairment that may lead to renal failure. Sometimes it can be manifested with isolated neurological involvement.[3]

CASE REPORT :

Male child aged 13 years old. from a pregnancy followed to full term, without notable neonatal antecedents, followed since the age of 2 years for epilepsy; the child was initially treated with monotherapy, then by bitherapy and then by triple therapy due to the persistence of seizures ; the patient initially presented with palpebral and upper limb myoclonus, the evolution was marked by generalised tonicoclonic attacks without loss of consciousness, with resistance to pharmacological treatment . He has a good psychomotor development. He has a 3rd degree of consanguinity, and elder brother recently followed for end-stage renal failure. kidney failure was discovered accidentally during a hospitalisation for toxidermy. The clinical examination was unremarkable. the urine strip showed a cross of proteinuria and a cross of hematuria. albumin at 44. alkaline reserves at 22. blood urea at 1.19. creatinine at 51.6. Ferritinemia: 13.2 , Protein electrophoresis: normal. Vitamin D: 27. Vitamin B12: 1149. Vitamin B9: 275. parathormon: 275. Abdominal ultrasound showed kidneys of chronic nephropathy with peritoneal effusion of low abundance. renal biopsy showed the appearance of chronic interstitial nephritis. the genetic study was not carried out due to a lack of resources . Therapeutically, the patient benefited from peritoneal dialysis and he was referred to France for a possible kidney transplant.

Discussion

Action myoclonus–renal failure syndrome (AMRF) is a rare autosomal recessive disease.[1]. It is a rare feature of progressive myoclonic epilepsy (PME) accompanied by renal failure. It was first described in four French Canadian patients by Andermann in 1986¹, and was initially considered to be restricted to French Canadians. Nowadays it is recognized as having a large ethnic and geographic distribution including Cuban ,German, American, Australian and Portuguese patients.[4]. This disease is typically begins causing symptoms between ages 15 and 25, but it can appear at younger or older ages. The age of onset and the progression of the condition differ, even among members of the same family. [1].

The diagnosis is confirmed in individuals with biallelic (homozygous or compound heterozygous) loss-of-function pathogenic variants in SCARB2 [1]. It combines progressive myoclonic epilepsy and renal failure which can lead to end-stage renal failure. It can sometimes manifest itself as an isolated neurological impairment [3]. Neurological manifestations may appear before, simultaneously or after renal manifestations [2]. Neurological and renal manifestations progress independently. It should be noted that neurological manifestations are not the result of metabolic encephalopathy due to renal failure and are not improved by dialysis or renal transplantation [5].

Neurological symptoms begin with a slight bilateral tremor of the fingers which is noted at rest and is accentuated by a delicate movement such as writing, by the intention to move and by maintaining an attitude of horizontal extension [6].

The fine tremor is then followed by jerky movements. Over time, myoclonic tremors involve the proximal limbs; their amplitude and number increases with limb movement, and the myoclonic action can be exacerbated by anxiety, excitement, stress and fatigue [7]. Clonic-tonic-clonic seizures begin with generalised clonic jerks with consciousness preserved and continue until unconsciousness with tonic-clonic features. They are uncommon, at the rate of one per year at the beginning. [2].

Kidney involvement in this disease is poorly described, proteinuria, the first manifestation of kidney damage, is initially mild and asymptomatic. Kidney disease can progress to nephrotic syndrome and terminal kidney failure. [5]. Histological changes may include interstitial fibrosis,

atrophy, focal sclerosing glomerulonephritis, possibly accompanied by collapsing glomerulopathy (a severe variant of glomerulosclerosis), or membranous nephropathy. No storage has been reported [8].

The EEG may show normal background activity in some patients or show a slowing of the diffusion at 6.5 to 7.5 Hz. There may be relatively low, bilaterally synchronous and generalised voltage spikes and shock wave discharges, or confined to the central vertex or both occipital regions, increased by hyperventilation and intermittent photic stimulation [5]. MRI scans of the brain may be undisturbed or show slight cerebral and diffuse cerebellar atrophy [9].

Pharmacological treatment and psychosocial support is the main means of care for neurological manifestations. Response to treatment is variable and may deteriorate over time. [5]

Kidney failure requires dialysis, but the response to treatment is poor and a kidney transplant is often necessary.[5]

our clinical case is in line with the literature regarding the chronology of clinical manifestations, the resistance to antiepileptic treatment and the serious evolution towards renal insufficiency, in spite of the fact that no genetic study has been carried out due to lack of means.

CONCLUSION :

Action myoclonus–renal failure syndrome (AMRF) is a rare disease and often under-diagnosed, that combines neurological and renal manifestations, the age of onset and evolution varies from one patient to another. The risk of being diagnosed late is end-stage renal failure.

Disclosure of interest

The authors declare that they have no conflict of interest concerning this article

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