Clinical and evolutionary aspects of Allgrove Syndrome :Algerian experience

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INTRODUCTION

Allgrove syndrome is a rare autosomal recessive disorder involving alacrymia,achalasia, Addison's disease (3A) and neurological disorders (4A), it results from mutations in the AAAS gene located on chromosome 12q13 which codes for a protein known as ALADIN (ALacryma Achalasia aDrenal Insufficiency Neurologic disorder).
Alacrymia is diagnosed by Schirmer's test, achalasia by esophageal manometry while adrenal insufficiency is confirmed by the determination of cortisol and ACTH.

AIM

To describe the clinical and evolutionary aspects of patients who presented Allgrove syndrome in our department.

MÉTHOD

It is a retrospective, descriptive study, spanning the period 2010 to 2020; were eligible for the study all patients who were hospitalized for Allgrove syndrome.
Several parameters were evaluated, age, sex, diagnostic elements, dose of treatment and evolution.

RÉSULTS

Age at diagnosis: 5 years
diagnostic deadline: 3 years.
Sexe ratio: 3 girls for 1 boy.
Consanguinity in 1 patient (syndromeAllgrove family).
Age of the first symptoms: 2 years.
Alacrymia confirmed by the test of Schirmer
Achalasia confirmed by endoscopy esogastro-duodenal and TOGD.
All the patients were treated by glucocorticoids, tears artificial.
On average, 5 dilation sessions were performed [2-11].
Evolution : We have an average follow-up of 4 years
(6 months - 8 years).
Improvement in adrenal insufficiency was achieved in all patients.
Successful dilations were obtained in 2 cases, the other 2 underwent a successful Heller cardiomyotomy.

parameters	Case1 (4 A)	Case 2 (2A)	Case 3 (3A)	Case 4 (3A)
Hypoglycemia	+		+	
Dehydration		+		
Vomiting				+
Melanoderma	+	+	+	+
Dysphagia	+	+	+	+
Alacrymia	+	+	+	+
Neurological disorders	+			
Cortisol	Low	Low	Low	Low
ACTH	High	High	High	High

COMMENTS

These 4 patients illustrate the heterogeneity of triple A syndrome in all terms. Alacrymia is the most constant symptom, it has been found in all of our patients, but which unfortunately has been trivialized by parents and has gone unnoticed by doctors, the same for melanoderma. In our series, one patient had a heavy, strongly suggestive family history (the brother presented with alacrymia and achalasia and sister presented with alacrymia), which unfortunately were completely ignored, resulting in delayed diagnosis. Adrenal insufficiency was certainly improved in all our patients, but the diagnosis was made only at the stage of decompensation with the need for management at the level of the intensive care unit for one patient.

CONCLUSION

Allgrove syndrome is a rare disease, early diagnosis and treatment improves the prognosis. The education of parents and patients must take place at each consultation, the only guarantee of a good progression of the disease.

REFERENCES

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