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Case report

Alacrymia revealing allgrove disease (SD Triple-A)

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Abstract

Allgrove's disease (Triple-A syndrome) is characterized by a symptomatic triad composed of alacrymia, cardiac achalasia and adrenal insufficiency. To this clinical picture are added peripheral neuropathic lesions. These are very diverse among patients but all induce a disabling long-term attack. The diagnosis is then clinical. We report the case of a child from consanguineous marriage, in which the ophthalmological examination revealed a severe dryness with keratitis invading the visual axis. Radiological assessment: Oesogastroduodenal transit (TOGD) revealed a mega esophagus. A treatment based on artificial tears and then scleral lenses is instituted immediately, as well as a laparoscopic surgical treatment. The knowledge of this pathology will allow an awareness of the severity of this disease in addition to suggesting its management.

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Introduction

Allgrove's disease (Triple-A syndrome) is characterized by a symptomatic triad composed of alacrymia, cardiac achalasia and adrenal insufficiency, neurological signs are observed in one third of cases. This polymalformative syndrome is transmitted according to the autosomal recessive mode; studies have demonstrated that Allgrove Syndrome gene "AAAS gene" at chromosome 12q13 is bound between markers D12S1629 and D12S312. This gene codes for a protein called ALADIN (alacrimia, achalasia, adrenal insufficiency, neurologic disorder) [1,2].

Patient and observation

We report the case of a 10 years old child from a first-degree consanguineous marriage with a brother who died 2 years ago with encephalitis and a brother who was followed for adrenal insufficiency. He was followed since the age of 8 years for adrenal insufficiency for which he was put under hydrocortisone. The patient consulted in ophthalmology for a reduction of bilateral visual acuity quantified at 2/10 in the right eye and at 4/10 at the left eye not improvable, with sensation of sand in the eyes, the interrogation reveals the absence of tears even during crying, he also reported chronic dysphagia with vomiting all evolving in a context of deterioration of the general state without fever. Ophthalmologic examination found diffuse superficial punctate keratitis (Figure 1) with a decrease of the BUT and an altered Schirmer test < 5mm. The endoscopic assessment with hypopharyngoscopy was normal. A supplement by Oesogastroduodenal transit objectified a motility disorder esophageal with lack of relaxation and stagnation of the contrast product above the lower esophageal sphincter (Figure 2). The patient was treated with artificial tears with frequent and regular instillation, emphasizing the need for continuous instillation and the esophageal achalasia was surgically treated laparoscopically: Heller's cardiomyotomy associated with an anti-reflux gesture with simple postoperative course. The four-month follow-up was marked by the persistence of KPS despite regular treatment with stabilization of visual acuity without improvement. The patient was then equipped with a scleral lens with a slight improvement in visual acuity at 6/10 in the right eye and 8/10 in the left eye.

Discussion

Allgrove syndrome or triple A syndrome is a hereditary syndrome whose incidence is still unknown and difficult to determine because of the clinical variant of the disease and infant mortality due to adrenal insufficiency crises, it is a The rare gene that remains rare, of autosomal recessive inheritance. The gene responsible for the disease, located on chromosome 12, would code the protein ALADIN (for alacrima-achalasia-adrenal insufficiency neurologic disorder), which belongs to the family of regulatory proteins. The exact role of this protein in Triple A syndrome is not yet known. But researchers believe it may have a regulating role on hormone receptors produced by the adrenal glands, cortisol, and a degenerative role on the nervous system [3,4]. Congenital alacrimia is the earliest cardinal sign, present in 90% of cases, objectified by SCHIRMER test. It appears secondary to progressive degenerative damage to the cholinergic vegetative innervation of the secretory lacrimal system, Alacrima generally presents from early infancy, while symptoms of achalasia may appear in individuals as young adulthood [1,5]. Other ophthalmologic signs have been described such as corneal hypoaesthesia, optic atrophy, anisocoria and disorders of pupillary motility by involvement of the cranial nerves [6]. In the absence of adequate treatment of complications can occur, such as ulcerations, superficial punctate keratitis, which can lead to blindness due to many

central ulcerations and opacification of the cornea. Achalasia is observed in 75% of cases, and adrenocortical insufficiency discovered during the first decade of life, especially by severe hypoglycaemic attacks with often isolated glucocorticoid insufficiency and ACTH negative test. Adrenal insufficiency can be discovered in adulthood, during the 2nd decade, or may never appear [7,8]. This deficiency may be life-threatening and should be carefully investigated for other clinical signs that may be associated with mental retardation, Parkinsonism, epilepsy, or orthostatic hypotension. The study of the mutation of the AAAS gene on chromosome 12q13 confirms the diagnosis, the treatment consists of a replacement treatment with artificial tears, with glucocorticoid, mineralocorticoid and DHEA replacement therapy and muscle relaxants if recent achalasia or cons indication to invasive gestures, endoscopic treatments: repeated pneumatic dilation, injection of botulinum toxin or surgical treatment [8].

Conclusion

A rare genetic disorder, Allgrove's disease is life-threatening for the child through adrenal, digestive and neurological involvement. It seems important to emphasize the role of the ophthalmologist in the multidisciplinary care of this entity. Indeed, if they are unrecognized or neglected by the severity of the general picture, complications of the ocular surface related to the associated alacrymia can lead to blindness and generate a lifetime disability.

Competing interests

The authors declare no competing interests.

Authors' contributions

All authors have participated to this study. All have read and agreed to the final manuscript.

Figures

Figure 1: diffuse superficial punctate keratitis

Figure 2: oesogastroduodenal transit (stagnation of the contrast product above the lower esophageal sphincter)

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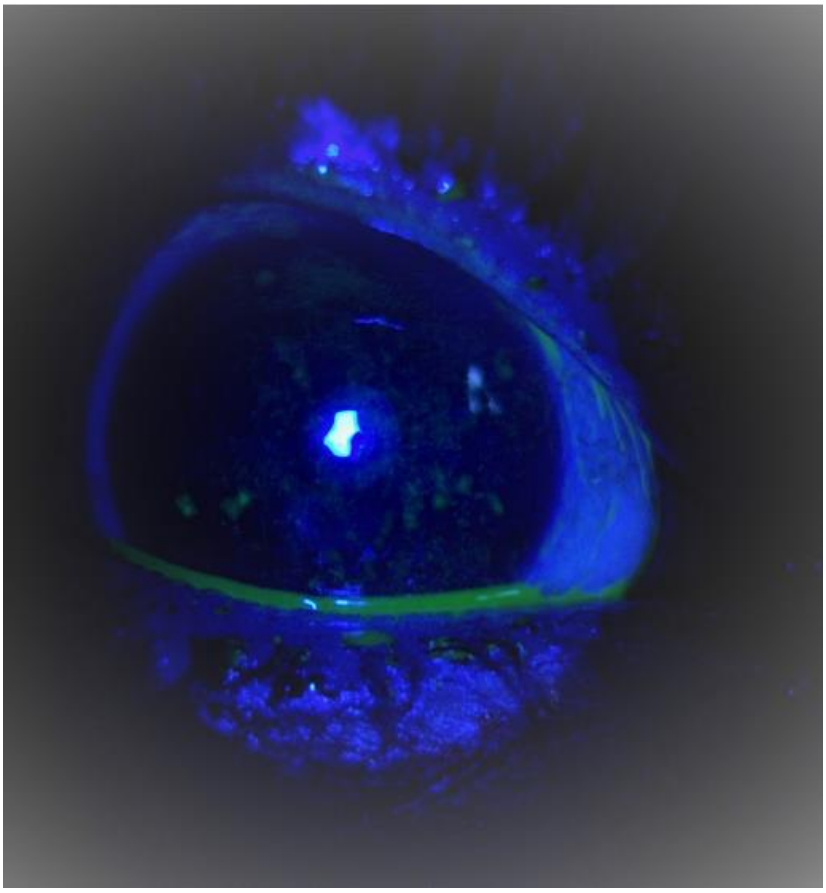

Figure 1: diffuse superficial punctate keratitis

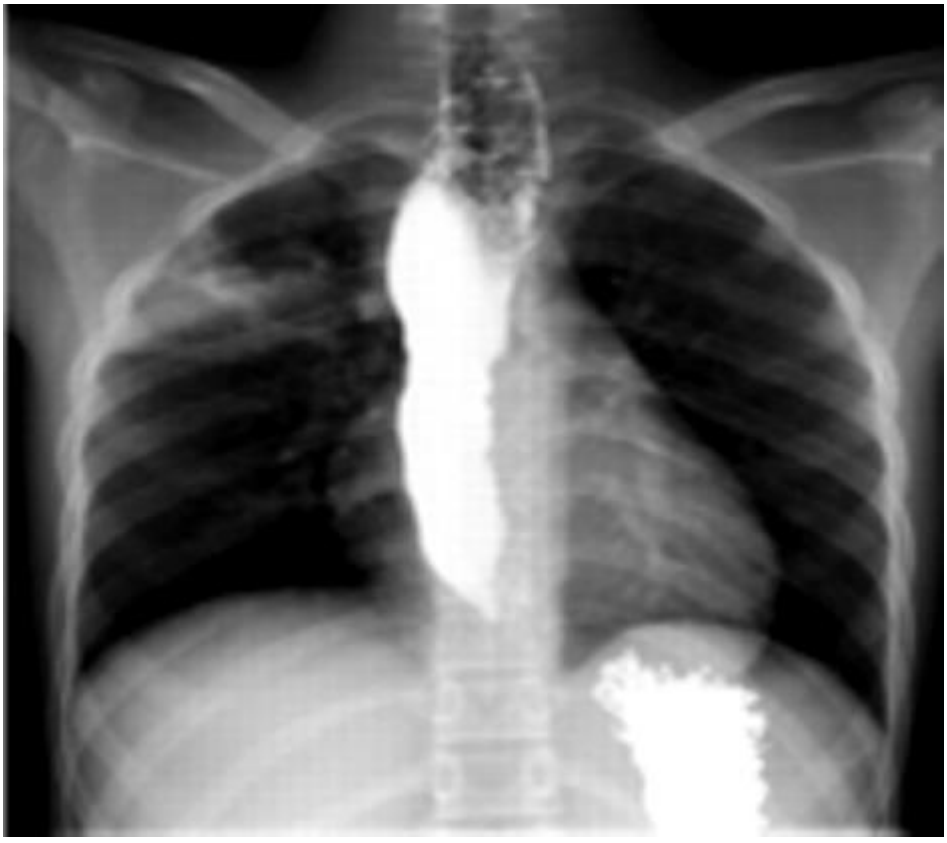

Figure 2: oesogastroduodenal transit (stagnation of the contrast product above the lower esophageal sphincter)