Case study

Alacrymia revealing triple A syndrome: About a case report

Abstract :

Introduction :allgrove's syndrome is a rare hereditary disease associating alacrymia, achalasia and Addison's disease. Neurological signs are observed in a third of the cases. We report the observation of a 10-year-old child with congenital alacrymia in the context of Allgrove syndrome.

Patients and methods: We report the case of a 10-year-old patient with triple A syndrome,.

Results: The ophthalmological examination revealed a superficial punctate keratitis with an altered Schirmer's test. The somatic examination found a perioral melanoderma, with amyotrophy of the leg and hollow foot. The explorations performed showed an Addison's syndrome in both children associated with achalasia.

Discussion:Congenital alacrymia is the most frequently reported sign, it concerns the basic lacrimal secretion and often also the reflex secretion. Adrenocortical insufficiency is manifested during the first decade of life by severe hypoglycemic crises. Achalasia is revealed between 6 months and 15 years of age by vomiting and dysphagia.

Conclusion: In front of any alacrymia of the child or the young adult must evoke a syndrome of Allgrove. A careful search for adrenal insufficiency, achalasia and associated neurological signs is necessary and allows to watch out for complications that could put at risk the vital prognosis.

key words: alacrimia, triple A syndrome, pediatric population,

INTRODUCTION:

Allgrove syndrome or triple syndrome A is a rare hereditary multisystemic disease associating alacrymia, achalasia and adrenocortical insufficiency not responding to adrenocorticotropic hormone.Neurological signs are observed in one third of cases. This polymalformative syndrome is transmitted in an autosomal recessive fashion [1] .

The most probable pathogenic hypothesis of Allgrove syndrome is the degeneration of cholinergic neurons of the autonomic nervous system.The therapeutic management is symptomatic.

The objective is to recall and describe the clinical presentation and to know how to evoke the syndrome in time in order to avoid the sometimes fatal complications

CLINICAL CASE :

We report the case of a 10 years old patient, the third of four siblings, from a consanguineous marriage, who consulted for congenital alacrymia. His antecedents were phonological delay, with the notion of intermittent dysphagia. His prenatal and perinatal history was without particularity. Her 2 month old brother died following vomiting (acute dehydration) with a notion of alacrymia.

The ophthalmological examination found a corrected visual acuity of 10/10 in both eyes, absence of the palpebral part of the lacrimal gland bilaterally, the slit lamp examination showed conjunctival hyperemia, presence of superficial punctate keratitis(figure 1)

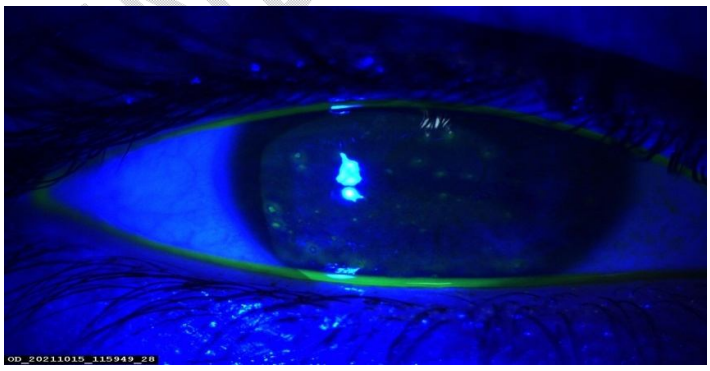


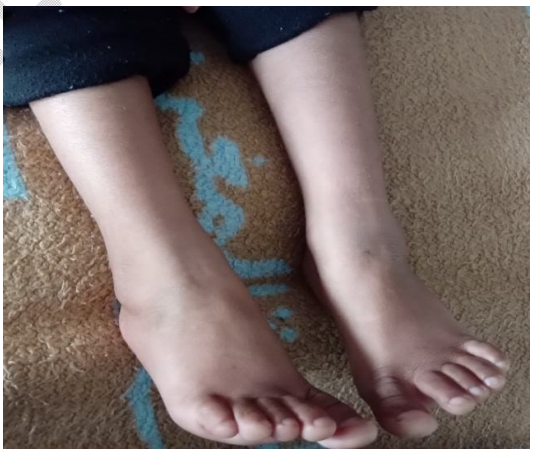
Figure 1 : superficial punctate keratitis

The Schirmer's test is altered; showed a wetting of 1mm and 2mm respectively in the right and left eyes(figure 2). Fundus examination was normal in both eyes.

Figure 2:altered shirmer test, wetting of 1mm in the right eye and 2mm in the left eye

The clinical examination revealed a delay in staturo-weight (-3DS), melanoderma especially in the perioral region(figure 3), with amyotrophy of the leg ,hands and hollow foot(figure 4).

Figure 3: Perioral melanoderma



Figures 4 : amyotrophy of the lower extremities

A biological check-up was requested revealing an iron-deficient microcytic hypochromic anemia, a collapsed 8H cortisol level, an elevated ACTH level. Adrenal CT scan showed bilateral adrenal atrophy.

In view of the presence of alacrymia as shown by the Schirmer's test as well as the presence of dysphagia and adrenal insufficiency in the biological work-up, the diagnosis of triple A syndrome was retained.

The patient was hospitalized in the pediatric ward for further treatment; a barium esophagastroduodenal transit(figure 5) showed a bird's beak aspect of the esophageal junction, and esophageal manometry revealed an aperistalsis of the body of the esophagus.esogastroduodenal fibroscopy showed a narrowing of the lower esophagus, which was easily crossed without reflux or signs of esophagitis**.**

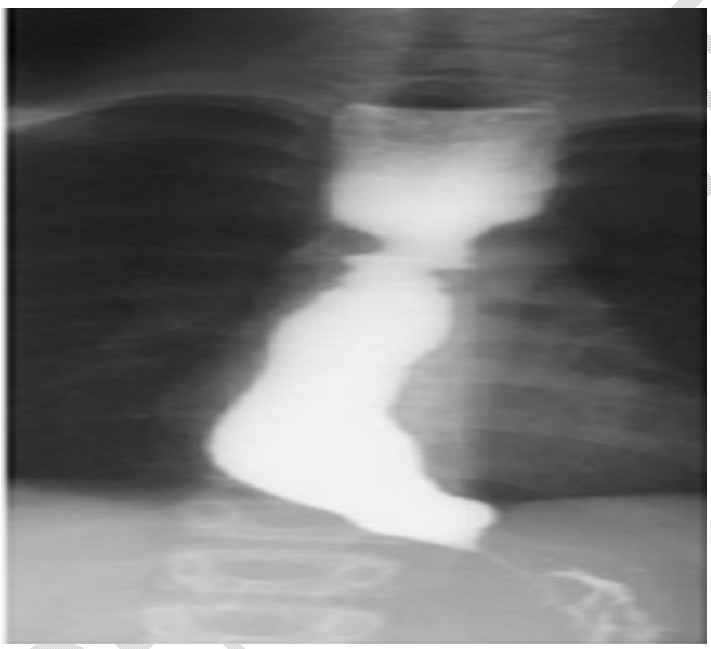


Figure 5: Esogastroduodenal transit with regular narrowing of the lower esophagus

The patient underwent endoscopic dilatation: Heller cardiomyotomy with simple postoperative procedures. Frequent ocular lubrication with topical tear substitutes was administered three times a day. With a substitution treatment based on hydrocortisone in one morning dose or in two doses, associated with fludrocortisone.

Discussion :

Allgrove syndrome is a rare condition whose prevalence may be underestimated. It should be considered in any patient presenting with any of the triad of abnormalities.

Alacrymia is the first and most consistent sign of this syndrome [2] Other features, such as cardiac achalasia or insensitivity to ACTH may present over a variable period of

time [3].

Triple A syndrome is the consequence of a genetic mutation (substitution, deletion, inversion....) of the AAAS gene (Achalasia - Addisonianism - Alacrima Syndrome). This AAAS gene codes for a protein, the exact role of this protein in Triple A syndrome is not yet known. However, researchers believe that it may have a regulatory role on the receptors of the hormone produced by the adrenal glands, cortisol, and a degenerative role on the nervous system [4].

The main ophthalmological manifestations in triple A syndrome include alacrymia with atrophy of the lacrimal glands and absence of lacrimation, keratoconjunctivitis, pupillary abnormalities including slow pupillary movement, accommodation disorders, amblyopia [5].

Other symptoms may be present; microcephaly related to the recurrence of hypoglycemic crises, mental retardation, hyperpigmentation, hyperkeratosis, cracking of the palms of the hands and soles of the feet, recurrent bronchopneumopathies due to dysphagia and achalasia.

The treatment of Allgrove syndrome is symptomatic, based on substitution by artificial tears in case of alacrymia, by glucocorticoids associated with fludrocortisone in case of mineralocorticoid insufficiency. Achalasia is treated by esophageal dilatation or Heller cardiomyotomy

CONCLUSION

Triple A syndrome is a neuroendocrine disease with a significant diagnostic erraticity. A great heterogeneity of clinical pictures intervenes, making the approach difficult within this disease. In front of any alacrymia of the child or the young adult must evoke a syndrome of Allgrove. A careful research of an adrenal insufficiency of achalasia and associated neurological signs is essential and makes it possible to look out for complications being able to put at stake the vital prognosis.

CONSENT

As per international standard or university standard, patient(s) written consent has been collected and preserved by the author(s)

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s

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