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Unusual Finding in Gillespie Syndrome

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Conflicts of Interest: Nil.

Abstract

Aniridia is a rare condition whose presence should alert clinicians to the possibility of other abnormalities. We report the case of a 4 months old girl, the clinical examination showed a bilateral partial aniridia with scalloped pupil border and iris strands extending into the anterior lens surface, and diffuse retinal hypo pigmentation in the funduscopic examination. Thorough investigation showed an interatrial communication and cholelithiasis without recognizable predisposing factors or disease. The genetic study showed a normal karyo type, without mutation in PAX6 gene. This report is relevant not only for the documentation of one more case of Gillespie syndrome, but also because of the description of a previously unreported association with cholelithiasis. As there are no other systemic alterations that could justify the presence of cholelithiasis, it could represent either an incidental association or a true finding not observed before.

Introduction

Aniridia is a rare condition whose presence should alert clinicians to the possibility of other abnormalities. One of the differential diagnoses that should be considered is Gillespie syndrome, in which aniridia is associated with cerebellar ataxia and mental retardation [1,2]. The goal of this paper is to report the case of an infant who presented with findings of the Gillespie syndrome, in addition to heart defect and asymptomatic cholelithiasis.

Case Report

We report the case of a 04 months old girl, born after uneventful pregnancy from healthy nonconsanguineous parents. She was admitted for nystagmus, photophobia and bilateral pupillary are flexical my driasis.

An examination under anesthesia was then performed. The cornea was clear. The anterior chamber showed normal depth. A bilateral partial aniridia with the pupil border of the iris containing a scalloped (festooned) edge and iris strands extending into the anterior lens surface were present (fig.1). Lenses were transparent within normal position. Intraocular pressure was 08 mmHg in both eyes. Gonioscopy was normal. Funduscopic examination revealed a diffuse retinal hypopigmentation. The electroretinogram and the visual evoked potential were normal. Thorough investigation showed an interatrial communication and cholelithiasis without recognizable predisposing factors or disease (fig 2). Simple monitoring was advocated. The pediatric neurological examination was normal for age.

Magnetic Resonance Images (MRI) of the brain showed a bilateral temporo-insular cortical and sub cortical atrophy. A genetic study was performed and showed a normal karyotype. No mutation in PAX6 gene was detected.

Follow-up sonographic studies showed a spontaneous resolution of cholelithiasis.

At the age of 13 months, the patient presented a motor delay and hypotonia : the child did not yet held the sitting position, she held just her head and showed no equilibrium reaction. Brain MRI remained unchanged. The diagnosis of Gillespie syndrome was suggested.

Discussion

Gillespie syndrome is usually diagnosed in the first year of life by the presence of fixed dilated pupils in a hypotonic infant. It associate aniridia to cerebellar ataxia and mental retardation [1. At the time of initial presentation, neurological involvement and radiological abnormalities can be absent or delayed. Ophthalmologic evaluation reveals eye findings that are characteristic. They usually consist of bilateral partial aniridia with the pupil border of the iris containing a scalloped (festooned) edge, and iris strands extending onto the anterior lens surface at regular intervals. It can be accompanied with additional ocular findings such as foveal, patchy iris and/or optic nerve hypoplasia, retinal hypopigmentation, and/or pigmentary macular changes leading to reduced visual acuity. Typically, the cornea and lens are clear [2]. Other associated signs include hypotonia, cerebellar hypoplasia, ptosis, partial fusion of cervical vertebrae [2,3] and heart abnormalities such as a heart murmur, and/or pulmonary artery stenosis [4,5]. There is no case associated with cholelithiasis reported in literature. This is an additional finding in our case.

The loss-of-function mutation in PAX6 gene causes the high proportion of cases of aniridia. There could be unidentified mechanisms for disrupting PAX6 function in infant with undetected mutation at the PAX6 locus [6].

Conclusion

This report is relevant not only for the documentation of one more case of Gillespie syndrome, but also because of the description of a previously unreported association with cholelithiasis. As there are no other systemic alterations that could justify the presence of cholelithiasis, it could represent either an incidental association or a true finding not observed before.

Competing interests

No competing interests.

References

[1].Luquetti, D. V., Oliveira-Sobrinho, R. P., Gil-da-Silva-Lopes, V. L. Gillespie syndrome: additional findings and parental consanguinity. Ophthal. Genet. 28: 89-93, 2007.

[2]. Nelson J, Flaherty M, Grattan-Smith P. Gillespie syndrome: a report of two further cases. Am J Med Genet. 1997;71:134-138.

[3]. Wittig EO, Moreira CA, Freire-Maia N, Vianna-Morgante AM. Partial aniridia, cerebellar ataxia, and mental deficiency (Gillespie syndrome) in two brothers. Am J Med Genet. 1988;30:703-708.

[4]. Gillespie FD. Aniridia, cerebellar ataxia, and oligophrenia in siblings. Arch Ophthalmol 1965;73:338–41.

[5]. Wittig EO, Moreira CA, Freire-Maia N, et al. Partial aniridia, cerebellar ataxia and mental deficiency (Gillespie Syndrome) in two brothers. Am J Med Genet 1988;30:703–8.

[6]. Ansari M, Rainger J, Hanson IM, et al. Genetic Analysis of “PAX6-Negative” Individuals with Aniridia or Gillespie Syndrome. Anderson MG, ed. PLoS ONE. 2016;11(4):e0153757. doi:10.1371/journal.pone.0153757.

Figure Legends

Fig. 1: Photo of the right eye showing aniridia with the pupil border of the iris containing a scalloped (festooned) edge and iris strands extending into the anterior lens surface.

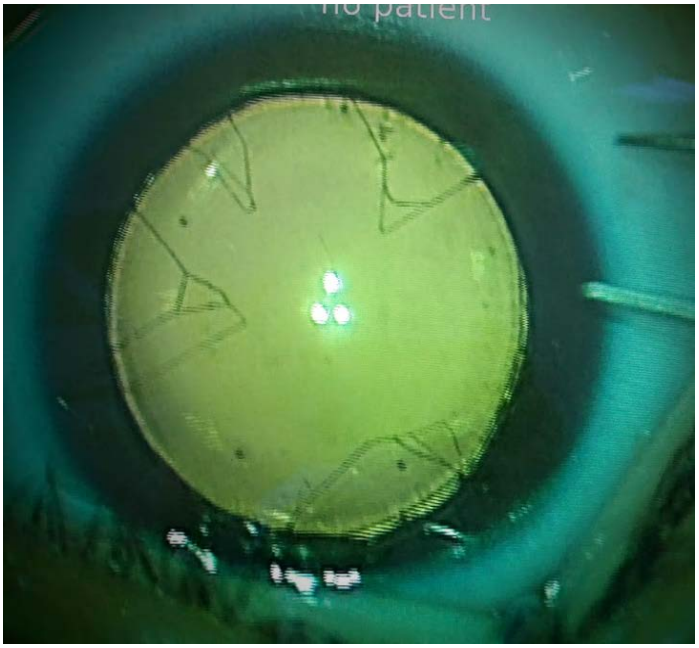

Fig. 2: Abdominal ultrasound showing cholelithiasis without dilatation of the bile ducts.