CASE REPORT

Coexistence of Anal Atresia, Anophthalmia and Intestinal Neuronal Dysplasia Type-A in a Newborn

Hilal Aydın,[1] Atilla Şenaylı,[2]* Fatih Mehmet Kışlal,[1] Dilek Sarıcı,[1] Burhan Köseoğlu,[3] Servet Güreşçi[4]

1 Keçiören Education and Research Hospital, Pediatrics Department, Neonatology Unit, Ankara, Turkey

2 Yildirim Beyazit University, Pediatric Surgery Department, Ankara, Turkey

3 Keçiören Education and Research Hospital, Pediatric Surgery Department, Ankara, Turkey

4 Numune Education and Research Hospital, Pathology Department, Deputy Chief Ankara, Turkey

ABSTRACT

We report a patient with anal atresia, anophthalmia and intestinal neuronal dysplasia type A.

Key words: Anal atresia, Anophthalmia, Intestinal neuronal dysplasia

INTRODUCTION

Incidence of anorectal malformations (ARM) is 1% among all anomalies. ARM has been associated with various anomalies like VACTERL (or VATER), cardiac anomalies, and urogenital anomalies etc. [1]. Congenital ocular anomalies (coloboma of iris, microphthalmia, cat-eye syndrome etc.) have been reported with ARM [2]. Coexistence intestinal neuronal dysplasia (IND) is also rare in Cat-Eye Syndrome [2, 3]. ARM with ocular anomaly has not been reported with IND [4]. We herein report a case of ARM associated with anophthalmia and IND type A.

CASE REPORT

A male newborn, product of consanguineous marriage and weighing 2670g, presented with imperforate anus and anophthalmia (Fig.1). Magnetic resonance imaging proved the patient's anophthalmia. Colostomy was formed before definitive operations for ARM. As a routine procedure, biopsy from colostomy orifice was performed. Pathologic evaluation revealed IND type A (Fig. 2). At one year age, anorectoplasty was performed, and colostomy was closed. After operations, bowel movements and defecation are normal. The boy is on follow-up of ophthalmology as well for ocular problems.

Figure 1: Anophthalmia of the patient. Inset shows imperforate anus.

DISCUSSION

In this report, we presented a case of ARM associated with anophthalmia and intestinal neuronal dysplasia (IND) Type A. Intestinal neuropathies like Hirschsprung's disease have

* Corresponding Author

been reported with ARM. IND is rarely associated with ARM. IND Type A is rare disease, and it is characterized by diminished or absent sympathetic innervations of the myenteric and submucosal plexuses [5]. IND has been reported with Hirschsprung's disease and infantile pyloric stenosis [4]. Anophthalmia is the absence of the eye. Eighty percent of the anophthalmia cases had other malformations [2, 6]. Neural tube defects, facial clefts and limb reductions are some these malformations [6]. Pathogenesis for these associations is not clear [2].

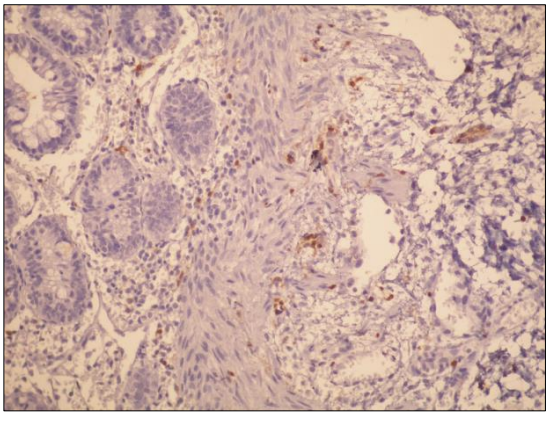

Figure 2: Diminished ganglions and neurons are highlighted by S-100 immunostaining in the sub-mucosa of the colon wall (immunoproxidase x400).

Coloboma of the iris has been reported with ARM [2]. This association of ARM with anophthalmia and IND has not been reported earlier to the best of our knowledge.

REFERENCES

1. Solomon BD, Raam MS, Pineda-Alvarez DE. Analysis of genitourinary anomalies in patients with VACTERL (Vertebral anomalies, anal atresia, cardiac malformations, tracheo-esophageal fistula, renal anomalies, limb abnormalities) association. Congenit Anom. 2011; 51:87–91.
2. Weber FM, Dooley RR, Sparkes RS. Anal atresia, eye anomalies, and an additional small abnormal acrocentric chromosome (47, XX, mar+): Report of a case. J Pediatr. 1970; 76: 594-7.
3. Rosias PR, Sijstermans JM, Theunissen PM, Pulles-Heintzberger CF, De Die-Smulders CE, Engelen JJ, et al. Phenotypic variability of the cat eye syndrome. case report and review of the literature. Genet Couns. 2001; 12:273-82.
4. Jeng YM, Mao TL, Hsu WM, Huang SF, Hsu HC. Congenital interstitial cell of cajal hyperplasia with neuronal intestinal dysplasia. Am J Surg Pathol. 2000; 24:1568-72.
5. Langer JC. Hirschsprung's Disease: Intestinal Neuronal Dysplasia. In: Coran A, (ed). Pediatric Surgery. 7th ed. USA, Elsevier; 2012:1277.
6. Morrison D, FitzPatrick D, Hanson I, Williamson K, van Heyningen V, Fleck B, et al. National study of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland: investigation of genetic aetiology. J Med Genet. 2002; 39:16-22.

Address for Correspondence*:

Atilla Şenaylı,

Ziraat Mah .İrfan Baştuğ Cad. Kurtdereli Sok. No: 10, Dışkapı, Ankara

E mail: atillasenayli@gmail.com

Submitted: 31-05-2015

Accepted: 25-09-2015

Conflict of interest: None

Source of Support: Nil

How to cite: Aydın H, Şenaylı A, Kışlal FM, Sarıcı D, Köseoğlu B, Güreşçi S. Coexistence of anal atresia, anophthalmia and intestinal neuronal dysplasia type-A in a newborn. J Neonat Surg. 2015; 4:44.