CASE REPORT

Congenital hypothyroidism in Axenfeld-Rieger syndrome

Çağatay Çağlar¹, Muhammed Batur², Erbil Seven², Serek Tekin², Tekin Yaşar²

¹Ophthalmology Department, Faculty of Medicine, Hitit University, Corum, Turkey
²Department of Ophthalmology, Faculty of Medicine, Yuzuncu Yil University, Van, Turkey

ABSTRACT

Axenfeld–Rieger syndrome (ARS) manifests varying degrees of anterior segment dysgenesis and carries a risk of resistant glaucoma. It can affect ocular and extraocular structures derived from the neural crest. A small part of the thyroid gland originates from the neural crest, and defects in thyroid origination can result in thyroid agenesis. Thyroid agenesis is a cause of congenital hypothyroidism. We have presented clinical features the first case of ARS associated with congenital hypothyroidism.

Key Words: Axenfeld-Rieger syndrome, anterior segment dysgenesis, congenital hypothyroidism, neural crest, thyroid agenesis

Introduction

Axenfeld–Rieger syndrome (ARS) is mainly characterised by anterior segment abnormalities of the eye, and comprises a clinically and genetically heterogeneous group of conditions with a rare spectrum of developmental disorders involving both ocular and extraocular structures derived from the neural crest. We reported a clinical characteristics the case of ARS with congenital.

Case report

A 12-year-old girl presented at a pediatric clinic to obtain a disability report and was referred to ophthalmology clinic for a routine ophthalmologic examination. Her medical history revealed congenital hypothyroidism and growth retardation. She had no personal or family history of ocular abnormalities. An ophthalmological examination showed that her best corrected visual acuity was 1.0, bilaterally. Her intraocular pressure (IOP) was measured as 13 mmHg by Goldmann applanation tonometer, bilaterally, and the fundus examination was normal, bilaterally. A slit-lamp biomicroscopy and gonioscopy revealed posterior embryotoxon, with strands of peripheral iris tissue extending to the cornea anterior to the Schwalbe’s line across 360º, bilaterally (Figures 1a, 1b and 1c). There was a focal iris atrophy in the inferior section of her right eye (Figure 1c). The patient had obvious maxillary hypoplasia, a broad nasal bridge, hypertelorism, microdontia and hypodontia (Figures 2a and 2b). She also had a significantly short stature and an intermediate level of mental retardation.

The patient has been followed up due to congenital hypothyroidism diagnosis. On her first visit to the pediatric endocrinology clinic, her TSH was >75.0 uIU/mL (0.400-4.00 uIU/mL), her free T4 was 0.441 ng/dL (0.80-1.90 ng/dL), and her total T4 was <1.00 ng/dL (4.50-12.5 ng/dL). The thyroid was non-palpable on physical examination. There was no thyroid gland identified in the thyroid ultrasonography. There was no uptake activity at the location of the thyroid gland in the thyroid scintigraphy (Figure 3). According to this data, the diagnosis was congenital hypothyroidism caused by the congenital agenesis. The patient's karyotype analysis was performed and the result was normal. The patient takes L-thyroxine (100 µg/day) and her congenital hypothyroidism is being followed up by the pediatric endocrinology clinic.

The patient has been followed up for 8 years. In the second year of the patient’s follow-up,
glaucoma was diagnosed in the left eye. Latanoprost 0.005% eye drop was prescribed one drop per day for left eye. The patient's glaucoma is still under control with latanoprost eye drop.

**Discussion**

Axenfeld-Rieger syndrome is mainly characterized by anterior segment abnormalities of the eye, and comprises a clinically and genetically heterogeneous group of conditions with a rare spectrum of developmental disorders involving ocular and extraocular structures derived from the neural crest. It is associated with secondary glaucoma in about 50% of cases, because of peripheral anterior synechiae and secondary angle closure of the anterior chamber angle (1). In our case, the IOP and cup-disc ratio was normal.

Ocular manifestations of ARS involve iris stromal hypoplasia, ectropion uveae, corectopia, full-thickness iris defects, severe iris atrophy and peripheral anterior synechiae (2). A typical abnormality of the cornea is a prominent anteriorly displaced Schwalbe’s line (posterior embryotoxon), which appears as a white ring on the posterior cornea, near the limbus. This tends to be more common temporally and rarely involves the full 360º. In our case, a 360º Schwalbe’s ring existed in slit-lamp biomicroscopy in each eye. Non-ocular manifestations of ARS may include developmental defects of the teeth and facial bones, pituitary anomalies, cardiac disease, oculocutaneous albinism, and redundant periumbilical skin (2-3). Failed neural crest cell migration and differentiation during embryonic
Congenital hypothyroidism based on congenital agenesis has not previously been reported in association with ARS. However, in these cases, mental and growth retardation secondary to hypothyroidism can improve. Therefore, patients with ARS should be investigated for hypothyroidism, as an early diagnosis and treatment of congenital hypothyroidism is crucial to ensuring mental, physical and neurological development.

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References


